# Juvenile Neuronal Ceroid Lipofuscinosis, Childhood Dementia and Education

# Intervention, education and learning strategies in a lifetime perspective



Edited by Stephen von Tetzchner, Bengt Elmerskog, Anne-Grethe Tøssebro and Svein Rokne

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Edited by Stephen von Tetzchner, Bengt Elmerskog, Anne-Grethe Tøssebro and Svein Rokne A step towards an extended curriculum – Conclusions and recommendations from the Erasmus+ sponsored project «JNCL and Education».

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# Foreword

This book presents broad information about education and non-medical interventions for children, adolescents and adults with juvenile neuronal ceroid lipofuscinosis (JNCL). This is a serious disease and most research on JNCL and other NCL diseases concerns medical aspects. The low prevalence of JNCL may have contributed to the limited non-medical research. Educational guidelines for practice and coherent information about the typical course and variation in the development of JNCL have been lacking. Educational practice tends to be based on experience obtained from teaching individual students or from input provided by small support and advocacy organizations. Owing to language and cultural barriers and limited resources, the individual knowledge bases have not been pooled successfully to others who work with persons with JNCL, and written documentation hardly exists. Dedicated teachers and other professionals have lacked both research-based and systematic experience-based knowledge about educational and other non-medical interventions that can support development, learning and skill maintenance in individuals with INCL. There is thus a need for more research to determine innovative and varied approaches within educational and other services to meet the challenges of the disease. Also needed is a strategy to pool current knowledge and disseminate so that all stakeholders can draw on it.

The present book thus builds on existing literature and adds to it by reporting findings from an international project. This project draws on findings from seven countries. It includes findings from a comprehensive survey and interviews with a relatively large number of families and professionals working with this group (e.g., teachers, special educators and residential staff). The project also includes the development of educational tools and strategies and small trials. The authors include researchers and practitioners with many years of experience with working for persons with JNCL, as well as family members. The book gives guidelines for practice and suggestions based on information about the typical course and the considerable variation that exists in learning and development within this group.

The book is intended for teachers, special educators and other professionals working with children, adolescents and adults with JNCL, as well as for families with a member who has JNCL. The book addresses educational and other interventions for individuals who show visual and cognitive decline with onset in childhood, and it may be useful also for families and professionals working with other diagnostic groups with similar problems.

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Batten Disease Support and Research Association (BDSRA), USA Chiltern Music Therapy, UK NCL Danmark, Denmark Department of Psychology, University of Oslo, Norway Finnish Federation of the Visually Impaired, Finland LVR-Johanniterschule, Germany NCL Gruppe Deutschland, Germany New College Worcester, UK

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University Medical Center Eppendorf, Germany University of Rochester Medical Center, USA University of Roehampton, UK Vision Centre Refsnæs, Denmark

We want to thank the participating families and professionals who shared their knowledge and gave of their time. Our sincere thanks go to all organizations and individuals who have contributed to the project with insights, background material, relevant experiences and life stories which form the backbone of the present work.

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# Introduction

1

Stephen von Tetzchner, Bengt Elmerskog, Anne-Grethe Tøssebro and Svein Rokne

The present book addresses learning and coping in individuals with juvenile neuronal ceroid lipofuscinosis (JNCL) from a life span developmental and educational perspective. A life span perspective examines how human abilities and skills grow and decline through development (Baltes & Baltes, 1990). The present book examines teaching and other forms of intervention for individuals with JNCL throughout life, including both early intervention and support in the period after they have left school. The education and intervention for individuals with JNCL include both early intervention and later adaptation, reflecting the growth and decline in skills and abilities in this group (see Chapter 12). The chapters present a variety of teaching tools and strategies designed to support learning and coping, maintenance of skills and abilities, and quality of life from childhood to adulthood. The vision of education and learning as lifelong processes emphasizes the need to choose goals that go beyond the classroom.

### The NCL spectrum

Neuronal ceroid lipofuscinosis (NCL) is a group of inherited, progressive neurodegenerative diseases that affect cells in the brain, the retina, the heart and the skeletal muscles (see Chapter 3). The onset of symptomatology varies from infancy to adulthood, and they share the combined characteristics of retinopathy, dementia and epilepsy (Cooper, 2010; Kohlschütter & Schulz, 2009; Mole, Williams, & Goebel, 2011; Mole & Haltia, 2015; Rakheja & Bennett, 2018). Categorizations of the NCLs have varied through the years (Haltia & Goebel, 2013; Rider & Rider, 1999) and recent classifications include 9–14 genetically distinct disorders (Haltia & Goebel, 2013; Kolschütter & Schulz, 2009; Rakheja & Bennett, 2018; Williams & Mole, 2012), and 360 different mutations (Kousi, Lehesjoki, & Mole, 2012).

This spectrum of diseases has several names. Neuronal ceroid lipofuscinosis is commonly known as Batten disease but there are also variants named after the genes they are associated with. Infantile Batten disease is also called CLN1 disease, late infantile Batten disease is called CLN2 disease, and variants of late infantile Batten disease may be CLN5, CLN6, CLN7 or CLN8 disease. The juvenile form (JNCL), also called the Spielmeyer-Vogt disease, is caused mainly by the CLN3 gene but also by mutations in other CLN genes (see Chapter 3). In the present book, the term juvenile neuronal ceroid lipofuscinosis (and its acronym JNCL) is used, as it is well established internationally. JNCL symptoms were first reported by Otto Stengel (1826), who documented the course of the disease in two sisters and two brothers of apparently healthy parents: the first symptom was loss of vision at an early age, followed by deterioration in cognition and speech, seizures and premature death.

### The course of JNCL

JNCL is characterized by a severe loss of vision which becomes noticeable around age 4–8 years, with a developmental course that includes blindness, epilepsy, speech and language problems, cognitive regression and motor coordination problems. Individuals with JNCL may also experience a number of non-defining symptoms, such as problems with sleep, eating, breathing, pain, and mood (Rokne, 2009). JNCL is a complex disease and the many symptoms and declines entail severe challenges for the individuals with this disease and their families.

JNCL influences all aspects of development but it is vision problems together with dementia that characterize the disease. Visual impairment is usually the first sign and to a large extent contributes to the identification of the disease. However, most individuals with congenital or early blindness have developmental functioning that is comparable to their peers with normal sight. Unlike those groups, children with JNCL experience the onset of dementia not long after the onset of visual impairment. The childhood dementia, meaning the onset is in childhood rather than later in life, has a pervasive influence on development and learning, and compounds the difficulties imposed by the visual impairment. Childhood dementia is not well researched but knowledge from dementia research in general may be useful when planning educational and other interventions for this group. The consequences of blindness and dementia are therefore *the leading theme* that recurs throughout the book.

Figure 1.1 shows the average age at first occurrence of symptoms and difficulties typical of JNCL, based on information from parents whose children had received a JNCL diagnosis and participated in the present project (Appendix A).

There was considerable variation in age at onset, with reports of some symptoms and difficulties occurring mainly within a limited age span, and others exhibiting more variation. Symptoms falling within a more limited age span included the appearance of something being wrong with the child between the age of five and seven years, visual impairment became noticeable between five and eight years, difficulties following mainstream education between six and ten years of age, and memory problems when the child was between seven and twelve years old. The symptoms and problems showing more variation in when they first emerged included problems with speech, gait and balance, which were first observed when the participants were between 10 and 18 years of age, and motor problems with hands and arms, which tended to occur between the age of 14 and 21 years (see Chapter 7).

The onset of some problems and declines occurred in a rather fixed order while the sequence of other symptoms varied more from participant to participant. For instance, visual impairment and memory problems were early symptoms of the



Figure 1.1 Average age at onset of some problems and declines Note: The spotted lines represent standard deviations.

disease; the visual problems usually appeared earlier than the memory problems, while speech and motor problems were observed later. In some participants, problems with spoken language occurred before motor difficulties, while others showed the opposite sequence. However, there was also variation within the motor domain, most participants showed problems with gait and balance before problems with hands and arms.

All symptoms had not been observed in all participants. Figure 1.2 shows the percentage of participants who had shown the symptoms and difficulties related to the disease. Problems with speech, memory and gait and with following mainstream education had been observed in almost all participants. It may be noted that some symptoms appeared relatively late and a lower prevalence may be due to the age distribution in the sample. For example, about half of the participants had problems with fine motor skills (use of hands and arms) but these problems tended to appear rather late, on average around 15 years (see Chapter 7) and many participants had not reached this age yet. On the other hand, behavioral problems were first observed at an average age of eight years but comprehensive behavior problems were reported in only about half of the participants (see Chapter 27).

In addition to the common problems related to JNCL, parents had also observed a number of less frequent problems. Some parents with older children reported that their child had developed hypersensitivity for certain sounds or



Figure 1.2 Prevalence of some observations, problems and declines (percent of total sample) Note: Prevalences will be influenced by the age distribution in the sample.

an inability to cope with certain sound frequencies and noisy environments, or reacted strongly to non-anticipated sounds. Other parents and educational staff expressed that the participants' behavioral and emotional reactions were their biggest daily challenges (see Chapter 27).

# **Evidence-based practice**

There is a general demand that practice should be evidence-based, and that assessment and intervention should have documented effects. Evidence-based practice means that there is a research basis for the assumption that the particular methods used in educational and other interventions for a child or adult are effective and useful (Berninger, 2015; Kratochwill & Shernoff, 2004; Stoiber et al., 2016). Studies where similar individuals are randomly allocated to a group that gets a new intervention or to a non-intervention or intervention-as-usual group, are considered the highest level of evidence. This kind of study is important for comparing different intervention methods or educational strategies. However, it is not always possible to implement such studies with children who have rare diseases. Individual case studies and systematic collection of professionals' experiences with the intervention methods used in practice may therefore be important for developing guidelines for best practice with these children and adolescents. Case studies can go into greater depth and provide insight into the processes underlying the atypical development and the intervention, also in a scientific sense (see Parker & Hagan-Burke, 2007; Yin, 2009). This requires that there is documentation and analysis of current practice, both of which are lacking for many groups of children with rare conditions.

The lack of documentation and systematic evaluation of experiences with different educational strategies and other interventions was an important motivation for the present project on JNCL and education. To our knowledge, systematic collection of experiences from the education of students with JNCL is lacking. Gathering information from professionals who are responsible for the education of a particular group, and from parents, represents a first step toward evidence-based practice. The results presented in this book may not meet the scientific requirements for strong evidence but they represent a first step toward systematization of the experiences of a relatively large group and may inspire further research in this area.

Raising the question whether there exist supportive research results for a particular practice is an opportunity to reflect on the methods used and hence to renew or supplement these methods. Some interventions continue to be useful, others fail the "evidence test" and may be replaced with approaches that

have a better theoretical and empirical basis. New knowledge about children's development and disorders may open up for new interventions but good practice is not only a matter of evidence but of professionals having an investigative and critical stance and reflecting on their own practice (Kratochwill, 2012). One aim of the present book is to encourage this kind of reflection on the approaches used in education and intervention for children, adolescents and adults with JNCL.

# Education as a leading activity from childhood to emergent adulthood

Learning is a core element in adaptation. Learning may be defined as an experiencedbased process which leads to relatively permanent changes in the knowledge, skills and behavior of an organism, and which is not caused by maturation, disease, fatigue or injury (Kolb, 1984). The transfer of knowledge across generations is basic to the evolution of human societies, and the school is society's main organization for providing shared knowledge and competence to all individuals, as well as ensuring the diverse competence society needs (see Chapters 9 and 12). However, humans also learn from their actions and exploration outside school. Much of this learning is social, as more competent members of the society provide guidance toward what is important and relevant in their society (Tomasello, 2009; Vygotsky, 1978). The individual learns throughout the lifespan but the acquisition of new knowledge and skills is especially important in childhood and adolescence. In new situations, adults can to a greater extent use knowledge and skills they already have.

Abilities and learning vary between members of a society and some have problems that imply an atypical course of development (von Tetzchner, 2019). Both the content and method of teaching and the students' ways of learning will vary with the physical and mental abilities and skills of the students. In spite of such differences, educational activities are equally important to students with typical and atypical development, including to students who show regression in abilities and for whom maintenance of knowledge and skills, rather than new knowledge and skills, becomes the major aim.

# **JNCL** and education

Over the last decade, there has been some attention to the processes involved in educating children and adolescents with JNCL, especially after the first international conference on JNCL and education (von Tetzchner, 2006). Some useful approaches appear to be available but there is a lack of evidence in relation to the educational strategies that are currently in use, and there has been no comprehensive evaluation of the strategies that are applied. There are very few publications about JNCL with an educational perspective (von Tetzchner, Fosse & Elmerskog, 2013). Research publications about cognitive functions in children and young people with JNCL rarely mention any implications the findings may have for education. Medical, psychological and educational professionals seem to have focused on the disease itself and its consequences for the individual's development, but not on how it affects participation in education, including both challenges and possibilities. There is thus still limited knowledge about the typical learning paths and variation within this group, indicating a need for larger studies.

The complex picture of different symptoms and problems apparent in the present study (see Figure 1.1.) indicates a need for competence in different areas. Some symptoms, declines and problems were affecting all participants, whereas others were affecting only some of them. The declines are caused mainly by neurological disorders but some problems are related to environmental factors. The two main roles of education and learning are to compensate for declines caused by the neurological progressions, and to reduce environmental influences that may cause secondary problems. The challenges are many and complex, but most challenges can be met by educational measures for maintaining skills and sustaining participation and quality of life for the individual. The main challenge for educators is to provide the right interventions at the right time based on knowledge and experience.

A major focus in this book is the role of dementia in the development of individuals with JNCL and its consequences for education. Awareness of childhood dementia in education highlights the problems related to observing and measuring the effects of interventions. A person with dementia will – independent of age – always show declines and loss of functions even with the best educational interventions. Education has standardized tools and observations for measuring learning effects in students who show progress, but few or no standard instruments for observing the effects of educational interventions for low-frequency groups like students with JNCL, who are showing declines.

Educational services for individuals with JNCL are in most countries organized under visual impairment because visual impairment is a basis for the diagnosis (see Chapters 3 and 9). This situation is also reflected in the organization of the present project. Most of the project members are affiliated with institutions in the field of visual impairment. This implies that the loss of vision is usually met with the necessary competence, even if many mainstream schools have limited competence and are struggling in this area. Declines in other areas of the child's functioning (e.g., speech and language, cognition, and motor performance) are often not met with the expertise needed. Services for children and young people with JNCL need to be supplemented by interventions that are not usually part of interventions related to visual impairment. Education and other interventions for students with JNCL should also be based on an understanding of how different declines and problems interact in forming the developmental course, and of possible measures and strategies that can meet these combined challenges. For instance, emotional and behavioral reactions may be related to the unavoidable visual and cognitive decline, but also to a lack of cautionary and compensatory measures that might have contributed to making the changes less stressful and upsetting.

There is little documentation of the role of education and learning for children and adolescents with JNCL and a need for a larger body of histories about students with JNCL with typical as well as exceptional educational paths. The brief case histories presented in this book illustrate that education may have a significant impact on the life situation for individuals with JNCL. Asking what factors were significant for the long-term educational success of these individuals is a valuable start, but it should be emphasized that those factors mentioned by informants may not be representative for students with JNCL in general. However, they do demonstrate possibilities for individuals in this group and the importance of providing person-centered services (see Chapters 11 and 12). One common factor in successful case histories seems to be that the persons with JNCL established strong interests early in life, interests that could be used in education to promote the development of knowledge and skills. The students' education was characterized by continuity, with each school knowing and building on the work of the former school. There was close collaboration between families, schools and multidisciplinary teams and the resources that were provided were necessary for continuity in the long-term and goal-oriented education. The important role of the parents in the present case histories cannot be overestimated; they were the experts on their own children and the final guarantors for life flow and continuity. Case histories show the importance of starting adult life with a rucksack and an agenda full of activities, interests and skills. Personal style and image must be respected by the people who take over the responsibilities for their adult life. This means that their sheltered workplace and new home should take each individual's strengths, weaknesses and life history into account.

As the present book demonstrates, there is much to learn about the education of children and adolescents with JNCL, and its possible consequences for adult life. Part of the knowledge comes from the observed variation in perceptual, motor, cognitive and language abilities in this and some other diseases with cognitive decline in childhood (Schoenberg & Scott, 2011). However, the experiences of families with members who have JNCL and the professionals who support the children's learning and coping, are important sources of information. Information from parents and educational staff is the empirical foundation of the present project (Appendix A). Research on education usually focuses on teaching and learning during the period of formal education, as defined in each country, but there is a need to include some consideration of the long-term consequences of the education and of the intervention strategies and adaptations that may contribute to positive adulthood. Some studies include emotional and behavioral reactions to educational and other aspects of daily life (Bills, 2011; Bäckman, Santavouri, Åberg & Aronen, 2005). Adaptations provided to the educational settings of people with JNCL must facilitate learning possibilities while minimizing negative challenges. Only then will they contribute significantly to improved learning conditions, better maintenance of skills, less frustration, and hence less severe behavioral and emotional reactions in children as well as adults (see Chapter 27).

### Registry

Due to the low prevalence of JNCL, many teachers and other professionals know only one or two students with this disease. In order to avoid self-fulfilling stereotypes, there is a need to gain information about the variation in the course of the disease and how teachers and professionals can evaluate developmental signs and prepare for the potential progression and regression in the individual student's developmental course (se also Chapters 2 and 10). Today, there are registries for many diseases and disabling conditions, such as for Duchenne muscular atrophy (www.duchenneregistry.org). The data collected here might be a first step toward developing such a registry for JNCL and other CLN diseases. It would be a natural task for national or regional competence centers for JNCL to register and systematize educational strategies and teacher experiences within their geographical area. International collaborative research efforts may compare the development of students with JNCL in different educational (e.g., mainstream and special schools) and cultural settings (see Chapter 9). Professionals and families can collaborate with universities to ensure appropriate theoretical and methodological competence.

# **Overview**

The chapters in this book describe the features of JNCL with an emphasis on vision impairment and early onset dementia, and discuss a range of theoretical, empirical and practical issues related to educational and other supports for children, adolescents and adults with JNCL. Chapter 2 first relates the developmental course of JNCL to developmental theory in general, as well as to Baltes' theory of goal

selection, optimization and compensation, and second, it introduces the concept «zone of developmental maintenance» as a tool for understanding the variation in development among individuals with JNCL and supporting intervention planning. Chapter 3 provides an introduction to medical issues related to JNCL. The chapter is rather short because there is a huge literature on the biological and physiochemical aspects of the disease and the main aim of this book is to provide information about educational possibilities. Chapters 4–7 describe the four main domains related to JNCL: development and decline in vision, cognition, language and motor abilities. Chapter 8 discusses ethical issues related to working with people with disabilities and developmental decline. Chapters 9-12 are about the basic processes related to assessment, planning, organization and implementation of educational and other non-medical interventions for individuals with JNCL, with special attention to proactive, precautionary and hastened teaching and learning. Chapters 13–21 present more specific teaching areas and educational approaches, especially related to reading, writing and mathematics, but also include the use of technology, games, drama and music. An important consideration addressed in these chapters is the need for support personnel to recognize how for each child with JNCL there is a process of moving from independent to interdependent functioning while participating in educational activities and coping in everyday life. Chapter 22 discusses the consequences of JNCL for peer interaction, social life and participation in society. Chapter 23 goes beyond the school years and discusses processes related to the transition from a school life to an adult life. Chapter 24 addresses parent needs and support, and chapter 25 presents experiences of being a brother or sister of a person with JNCL. Chapter 26 describes the important functions of the family associations. Finally, Chapter 27 discusses the behavioral and emotional reactions that may be observed in individuals with JNCL; the relationship between these reactions to the developmental course of the disease and the complexities of the difficult life situations that ensue; and strategies that may lessen the stress and frustration of the persons, and thereby their behavioral and emotional reactions.

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## A Developmental Perspective on Juvenile Neuronal Ceroid Lipofuscinosis

Stephen von Tetzchner

uvenile neuronal ceroid lipofuscinosis (JNCL) is a developmental disorder, with a gradual loss of vision and other functions, beginning in childhood. The pathological biological bases of JNCL (see Chapter 3) constrain developmental growth and eventually lead to developmental decline in most areas. The present chapter presents a theoretical approach to framing the developmental processes involved in the regression, arguing that a developmental way of thinking is useful even when there is developmental decline. This chapter examines how a person with increasing cognitive impairment grows and develops, by addressing multiple aspects and placing them in different frameworks of understanding. The theoretical model and knowledge provided in this book, about the typical course and developmental variation in JNCL, provide a basis for timely and appropriate education and other forms of interventions. This approach may contribute to compensating for functional loss and decline in individuals with JNCL through education, adaptation and support within their «zone of developmental maintenance». This approach may be adequate for children with other childhood dementia disorders.

### The concept of development

Traditionally, development has been defined as an age-related process involving positive changes in the structure and functioning of human beings and animals as a result of interaction between biological structures, psychological states and ecological factors. At the core of this process lies *transformation*: something new emerges, less may become more, simplicity may turn into complexity, and limited skills may evolve into advanced mastery (Overton, 2015). Development towards adulthood implies a greater degree of autonomy and independence from the parents and a successively increasing social affiliation in the society (Keller,

2016). However, from a life-span perspective, developmental transformation also includes reductions of skills and abilities (Baltes & Baltes, 1990).

Development is thus the conglomeration of processes that together lead to an individual's physical, cognitive and personal characteristics, social relationships and roles in society. Change is a defining characteristic of development but in most areas there is both change and continuity. The individual's mastery and understanding of the world change, but change always emerges from a foundation of prior abilities and experiences, and the individual remains the same (Nelson, 2007). The parallel paths of change and continuity are also a characteristic of developmental decline, even if the process of regression differs from *typical development* in many aspects.

There is considerable individual variation in most aspects of development. Typical development is the most common course, with unimpaired functions and ordinary individual differences between children. *Atypical development* is a broad term used to describe all forms of irregular development, such as the normal but unusual language development of deaf children when they learn sign language instead of spoken language, or the different ways of thinking and reasoning that differentiate children with autism spectrum disorder from their peers. Development follows the same basic principles, regardless of whether it proceeds typically or atypically, but while the organism adapts to its environment, the environment must also have properties that allow the organism – with its strengths and weaknesses – to develop. Most children with typical development will show positive development in quite different environments. Children with atypical development have a narrower range of possibilities and are more dependent on an environment that fits their particular strengths and weaknesses (von Tetzchner, 2019).

## Development and disability

Some children acquire skills and abilities early on, while others have late or unusual development in one or more areas. Some have impairments or deficits that may inhibit some or all aspects of development, rendering them unable to perform many actions or requiring them to perform actions in unconventional ways.

In the gap model (Lie, 1996), disability is defined as a gap between an individual's abilities and the abilities that the environment requires to allow participation. Both the individual's abilities and the demands of society change with the individual's age and training, and hence the disability gap may both increase and decrease over time. The gap in functioning between individuals with severe disabilities and those with typical functioning may widen with age (Stadskleiv, 2017). However,

the developmental consequences are not determined by an impairment or deficit alone, but instead, both primary and secondary consequences are related to the support provided by the family, professionals, and society at large. There are aspects of disability that reflect structure and process. The *structural* perspective is a characterization of the child's actual skills and abilities – or his or her lack of skills and abilities. The processual perspective characterizes processes related to both a primary impairment or deficit, and the possible secondary consequences of these. From a developmental point of view, disabilities emerge progressively or regressively as a result of a comprehensive and complex process, where learning is an adaptation to the ecology, culture and life situation of the individual. There is no simple relationship between an impairment and its developmental consequences. Intervention may lose efficiency if designed on the basis of the specific impairments of the child, out of context; rather, design should proceed according to both the child's functioning and his or her physical and social environment. For example, when a child is unable to perform self-propelled movement, it has consequences for the child's exploration, world knowledge, concept development, self-regulation and attachment (Anderson et al., 2013; Campos et al., 2000). Similarly, the memory impairment accompanying dementia not only affects thinking and reasoning, but also many other skills including communication, opportunities for learning and socialization, and maintenance of other skills and abilities.

## Development is transactional

Children are dependent on adults to support their development of abilities and autonomy, and parents usually form the core of children's social environment. However, it is not only the case that the environment influences the child's development, but also that the environment is influenced by the child. For example, extroverted and social children elicit very different reactions from the people in the environment than introverted and shy children (Kagan & Snidman, 2004). Children of small stature are met differently than children who are large for their age (von Tetzchner, 1998). Children who are blind may be listening actively but without facing their parents, and may therefore be perceived as innattentive and uninterested by the parents (Fraiberg, 1977).

Transaction designates the reciprocal interaction between the child and the environment (Sameroff, 2010). The environment influences the child, and the child, in turn, influences the environment. The changed environment without facing the parents influences the child, who in turn influences the environment, and so on. This means that the child's development is influenced both by how the child perceives the environment *and* by how the child is perceived by the environment. People in the environment will be influenced by the child and

adapt to the child's characteristics as they perceive them. Transaction thus has a subjective basis. Transaction is a basic feature of development, whether it involves motor skills, personality, social relationships, emotions, communication, problem solving or other areas.

Parenting styles reflect parents' experience, beliefs about children and childhood, and sensitivity and ability to adapt to their child's needs and abilities. As the child develops, the parents gain detailed knowledge about the general and unique characteristics of their child, the child's possibilities and limitations. However, parents' experience and general beliefs do not usually include children with atypical development, and they may lack knowledge about how to interpret and react to a child who develops differently than most children. Their expectations and interpretations of their child with a disability may therefore be guided by their general knowledge about children, together with the knowledge they have about their own child and the child's disability. Parenting may still be positive for the child, but parenting of children with atypical development often reflects both overestimation and underestimation, because the parents do not have sufficient knowledge about the typical course and variation that the impairment of their child may imply. As the child grows, parents of children with disabilities also get a better understanding of the child's possibilities and limitations. However, it can still be difficult for parents to understand both what their child with a disability can do, as well as what the child cannot do. When parents lack sufficient insight into the child's development, abilities and challenges, their expectations may not match the possibilities and limitations of the child, and they may misinterpret the child's behavior and reactions. They may need to be guided by professionals towards a *redefinition* of the child's behavior and expressions, of his or her biological possibilities and impediments and of how they can create an environment that adapts to both the abilities and disabilities of the child throughout development (Sameroff & Fiese, 2000). Mobility supports the child's exploration, world knowledge, concept development, self-regulation and attachment independent of the manner in which the child moves around (von Tetzchner, 2019). Also teachers and other professionals may need guidance to understand and become able to provide appropriate developmental support to a child with a disability. In particular when the child has a rare disorder, many professionals will have no prior experiences with that disorder, and in addition may spend only a few hours together with the child every week. They may need considerable guidance and support to become able to create a physical, educational and social environment that promotes an optimal development.

# Development may imply both progression and regression

Abilities and disabilities do not come ready-made; instead they are the result of complex interactions between biological and environmental processes over time. Development may include periods with reduction in skills and abilities, and intervention must be based on an understanding of the process. The developmental changes in regression are age-related but the direction is different from developmental progression and reflect complex processes. In particular, the intervention measures needed to support adaptation to weaker physical, perceptual and cognitive abilities in children and young people may be poorly known and understood. It may be difficult to define future goals and make decisions about intervention when regression is observed or expected, and to evaluate the effects of the interventions when the functional level of the individual has become lower.

## Selection, optimization and compensation

Growing up with a disability represents a way of life that in important ways differs from that of most children in the community. Children with JNCL, as compared with their peer group, typically have to find other ways to play and select alternative leisure activities. Some children have few choices for both themselves and the people who are making adaptations to their environment. The differences between the developmental trajectories of children with JNCL and other children, with or without disabilities, are increasing with age.

In the lifespan theory of Paul and Margret Baltes (1990) human development is described as processes of selection, optimization and compensation (SOC). These theoretical concepts are particularly useful for explaining development in individuals who are lacking or losing many core abilities. *Selection* concerns the individual's choice of relevant and achievable goals, while *optimization* is the process of forming and maintaining the means to achieve the goals that have been selected implicitly or explicitly. *Compensation* is the use of new strategies to maintain functions and goals that are no longer sustainable in ordinary ways, for example due to physical weakness and illness, which may prevent or delay some of the functional decline (Baltes, 1987, 1997). The model has been applied mainly to developmental processes in elderly adults, but it includes development throughout the lifespan, including early development and development in individuals with negatively progressing conditions (Baltes & Smith, 2004).

The concepts of selection, optimization and compensation represent a useful theoretical framework in the present context because they highlight important aspects of the developmental process in individuals with declining functioning in core areas. For example, a child or adolescent with JNCL may together with parents and professionals select actions and activities that can scaffold «learning in participation». Optimization may include guidance of adults and peers in the environment, and compensation may include special training as well as adaptation of the classroom and educational strategies. From a developmental perspective, the aim of early intervention is not primarily to «repair» functions but to initiate and give direction and support to the developmental processes, preventing decline and supporting coping maintenance. For example, when a child is unable to perform self-propelled movement, a wheel chair provides access not only to movement itself, but also movement that results in participation opportunities.

## The zones of proximal development and developmental maintenance

Children gradually learn to master new things. Many of the things they are unable to do independently, become possible in collaboration with adults and more competent peers. The things children are unable to master on their own but are able to accomplish in collaboration with others are within what Vygotsky (1962) calls their zone of proximal development. The beginning and end of the zone of proximal development are defined by what a child is able to achieve, respectively independently and with help. The point at which children no longer master a task on their own marks the beginning of the zone. When a maximum amount of help from more competent peers and adults does not enable the child to attempt to solve a task, the task lies outside the child's zone of proximal development.

It is within the zone of proximal development that learning can take place, and adults must be able to introduce new «tasks» and adapt their mediation to the child's zone. If a child encounters a task with a degree of difficulty that is beyond this zone, the child will not understand the task and thus will not be able to contribute to its solution. If the task is too easy, solving it will not contribute to the child's development or changes in their zone of proximal development. For tasks located within the zone, the strategies contributed by adults are termed *scaffolding* (Wood, Bruner & Ross, 1976). Scaffolding corresponds to the principle of *guided participation*, that is, that children develop while participating actively in culturally valued activities with the guidance, support and challenge of children and adults who vary in skill and status (Hundeide, 2003; Rogoff, 1990).

Scaffolding is mediation: adults attempt to clarify what is required to solve a task, limit those aspects of the task that the child is unable to master – for example by holding an object stationary while the child tries to take something from it – and shield the child from distractions that can draw attention from the task at

hand. As the child develops, the help and support are gradually reduced, and once the child is able to master a task independently, scaffolding is no longer needed. This means that adults guide the child towards tasks at the child's own level *as well as* offer help that the child actually benefits from. Too little help can prevent children from being able to solve the task. Too much help can reduce children's trust in their own ability, because the help provided by adults also reflects their evaluation of the child's competence (von Tetzchner, 2009). Both the task and help must lie within a child's zone of proximal development if adults are to contribute to the child's development. The transactional influences are working through the adults' sensitivity to the child and their ability to create an environment that the child can cope with and learn from. Children with disabilities have fewer social opportunities and more constraints in everyday life than most children, but their development is still facilitated by participation in social activities.

The concept of zone of proximal development applies to developmental progression, and the aim of scaffolding is to promote the child's future independent mastery. This aim cannot be applied to developmental regression. For an individual who is showing developmental decline, guidance and support therefore have a different function than for children showing progression. The aim of adults' help is to contribute to maintaining the individual's ability to manage tasks that he or she can no longer master independently. Because scaffolding may support prolonged self-initiated performance and autonomy, it is within the individual's *zone of developmental maintenance* (see Figure 11.1, page 193). When the action is performed only by the adult without any contribution from the individual with developmental decline, the task is outside the zone of maintenance. Like in the zone of proximal development, maintenance through interdependence (see Chapter 16) depends on the knowledge and sensitivity of the adult in recognizing and providing the individual with the help that he or she needs, not too much and not too little.

Scaffolding reflects the transactional nature of development, being characterized by the adult being sensitive and giving help and support that are adapted to the skills and abilities of the individual. For children and young people with developmental decline, the scaffolding strategies applied by parents and professionals will reflect how they perceive the abilities and limitations of their child and the adaptive nature of ordinary or compensatory strategies used to maintain or support a particular function. An individual with JNCL may need help solving immediate challenges, but the main aim of scaffolding of individuals with JNCL is to delay the decline and maintain their functions and coping as long as possible, by increasing the scaffolding when needed. According to social constructivist developmental theories, adults enable children to acquire writing, arithmetic, and other cultural cognitive tools. For individuals with JNCL, the cognitive tools developed early in life may play an important strategic role in maintaining functions and delaying later decline (see Chapter 14).

Scaffolding is often perceived as a «natural» rather than educational process, emerging from the interactions between children and their caregivers. For individuals with JNCL, intervention may also represent a scaffolding activity, for example when it promotes communicative success in social interactions that are not easily guided in ordinary situations. Moreover, for children with JNCL, the developmental process will have to be more planned and the scaffolding more explicit and direct than for children without such a comprehensive disability. Scaffolding in the zone of maintenance is integrated with the processes of selection, optimization and compensation.

## The role of planning in atypical development

A common feature of atypical development is the need for planning. Although nearly all children attend schools with planned education, children with impairments are more dependent on detailed plans than children with typical development (see Chapter 11). The choices made by the family are closely interwoven with decisions taken by professionals, and often made through dialogue (see Chapter 24). The environment must be arranged individually and plans may be necessary for creating opportunities and reduce the restrictions on participation in activities that typically characterize the environment of individuals with JNCL (see Chapter 16).

Good planning requires sufficient knowledge of the developmental processes and the consequences impairments may have in the short and long term. Integrating the theory of Baltes and scaffolding, one important task of professionals is to guide individuals with JNCL and their families in selecting and optimizing goals. All children and adolescents thrive with goals that challenge, as long as they are comprehensible and achievable. However, for children and young people who show decline, goals are often *not* developed on the basis of a thorough assessment or systematic observations of the child's current abilities, up-to-date knowledge about the condition and professional reflection. The result may be a lack of appropriate compensating measures and goals. The goals may be either too repetitive and boring or too new and complex, sometimes leading to passivity or challenging behavior in the child. Plans and goals should include precautionary measures and be based on an anticipation of how JNCL is likely to affect the development and learning of the child in the future.

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## Medical Aspects of Juvenile Neuronal Ceroid Lipofuscinosis

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uvenile neuronal ceroid lipofuscinosis (JNCL) is a brain disease with symptoms usually emerging slowly in late preschool age or in the early school years. The disease influences the brain, leads to epilepsy and deprives children of key abilities, such as vision, intellectual capacity, motor ability and language, described in other chapters of this book. Eventually, the disease leads to a decline in self-help skills and a shortened life span. The sequential loss of skills follows a characteristic order and contrasts with the typical development in all areas during puberty and adolescence. People who are caring for or involved in education of children and young people with JNCL need to be aware of the opposite processes of development and decline that are simultaneously at work in these young people (see Chapter 2). Understanding the medical basis of the disease and how it affects a child or young person's abilities will make it easier to react appropriately to the numerous challenges caused by the disease, both in everyday life and in education, which is the most important activity from late childhood to emergent adulthood. The impaired quality of life relating to JNCL varies considerably with the phase of the disease and the presence of other medical issues.

## Definition of JNCL

Neuronal ceroid lipofuscinosis (NCL) is a group of different genetic disorders with many common features, both in respect to symptoms and underlying biological features. JNCL refers to an NCL disease that is «juvenile» and starts in early age. «Neuronal» means that neuronal cells (nerve cells) are mainly affected by the disease process and «ceroid lipofuscin» (Latin for «waxy, fatty and brownish») refers to a peculiar material that is found in large quantities in all cells and is characteristic of all NCL diseases.

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Classic JNCL has been known for a long time, with the first description dating back to Stengel (1826). Other names for JNCL are Batten disease or Spielmeyer-Vogt disease, but using historical designations is not always adequate. For example, in parts of the Anglicized world, the term Batten disease is often used to designate the whole spectrum of NCL disorders, including forms that manifest in infants or in adults. Due to recent progress in genetic research, the medical classification of NCL disorders is currently based on the genes involved and the age of disease onset (Table 3.1). In this book, the term juvenile neuronal ceroid lipofuscinosis (JNCL) refers to all forms of NCL diseases with a juvenile onset.

## Genetic causes of JNCL

All types of JNCL are caused by mutations (alterations) in various genes, creating different variants or alleles of the DNA material responsible for the hereditary transmission of personal characteristics. In the vast majority of children and young adults with JNCL, the mutated gene is called CLN3, and they may be said to be having juvenile CLN3 disease. There are other forms of NCL disease with a juvenile onset caused by mutations in CLN1 or CLN2, and very rarely in other genes (see Table 3.1).

There are other medical conditions with similarities to JNCL, such as the mucopolysaccharidoses (MPS disorders) or Niemann-Pick disease type C (NPC). These conditions differ from JNCL in the absence of visual loss and a more pronounced involvement of organs outside the nervous system (Schoenberg & Scott, 2011).

Inheritance of all JNCL forms is autosomal recessive, which means that the individual has received mutated genes from both their father and mother. As human beings carry a double set of most genes, a person with only one such NCL gene variant (a carrier or heterozygous person) does not usually develop symptoms of the disease because the second, non-mutated, gene compensates for the defective NCL gene. A child who has inherited a defective CLN variant of the NCL gene from both parents, either homozygous (when the mutations are the same) or compound heterozygous state (when the mutations are different), cannot compensate for the pathological function of the NCL gene, and therefore will develop the disease. One of the consequences of this form of inheritance is that a family can have several children who are affected by the disease. In the family described by Stengel in 1826, all four children developed the disease. A child will have a 50 percent probability of inheriting one abnormal gene, which would make the child a carrier who is unaffected by the disease. There is a 25 percent probability of the child being born with two non-mutated genes and hence

Disease	Age at which first symptoms typically appear						
Name according to mutated gene	At birth (congenital)	Before 1 year (infantile)	2–4 years (late infantile)	5–12 years (juvenile)	20–30 years (adult)		
CLN1 Disease		Х	Х	Х	Х		
CLN2 Disease			Х	Х	х		
CLN3 Disease				Х	х		
CLN4 Disease					Х		
CLN5 Disease			Х	Х	х		
CLN6 Disease			Х		Х		
CLN7 Disease			Х				
CLN8 Disease			Х	Х			
CLN10 Disease	х			Х	х		
CLN11 Disease					х		
CLN12 Disease				Х			
CLN13 Disease					Х		
CLN14 Disease		Х					

Table 3.1 Classification of currently known NCL diseases according to genes and age of manifestation

of not being affected by the disease or being a carrier. When the child develops JNCL, both parents are carriers, and the risk that a sibling will be affected by the same disease is 25 percent. Professional genetic counselling, which includes discussing the various possibilities of prenatal testing, is part of the management for a family where a child has been diagnosed with JNCL.

## Disease mechanisms in JNCL

Genes contain information that is necessary for the structure and function of the body. A disease-causing mutation of a gene disables the production of an important molecule, frequently a protein. The lack of this protein then leads to a disturbance of the delicate biochemical network of cells. In the case of JNCL due to a CLN3 defect, the mutation most frequently consists of a large *deletion*, that is, the loss of a piece of the DNA strand that forms the gene. The size of a deletion is measured in kilobase (kb, 1000 base pairs of DNA), and the typical deletion in CLN3 disease has the size of about one kb. Some individuals have smaller mutations, usually

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so-called missense or nonsense mutations where a single part of the DNA is not missing but incorrectly sequenced. The symptoms and disease progression of this group may differ somewhat from the majority of individuals with JNCL.

The relevant protein produced under the influence of a non-mutated CLN3 gene is called the CLN3 protein. The size of the CLN3 protein, its chemical structure and location within cells have been characterized and the protein appears to be an important protein because it emerged very early in the evolution of living organisms. It can be detected in a large number of organisms as well as in many different cells of the human body. It is disappointing, therefore, that despite extensive research, the true function of the CLN3 protein has not been fully clarified. It is a structural component of the biological membranes that surround



Figure 3.1 Microscopic picture of blood cells from a young person with JNCL

In the center is a single abnormal white blood cell (a lymphocyte). The many vacuoles (white holes) in the cytoplasm (the brighter part of the cell) are evidence of the storage disease. Originally the holes contained stored ceroid lipofuscin, which was lost during preparation for microscopy. The many round grey shadows in the neighborhood of the lymphocyte are normal red blood cells (erythrocytes).

all cells and individual working parts of the cell (and is therefore a transmembrane protein). One such membrane is that of *lysosomes*, small specialized compartments (organelles) within cells. Lysosomes are the organelles responsible for degrading and recycling used material. If lysosomes do not work properly, undegraded biochemical waste builds up inside cells, and the specific function of a cell is impaired. In the case of JNCL, as in other NCL diseases, lysosomes function poorly and become filled with material that should have been degraded, and this material has given the name to these diseases: ceroid lipofuscin. Because of these mechanisms, the NCL diseases belong to the larger group of lysosomal storage disorders.

The abnormal storage material is found in almost all cells of the body, but most cell types apparently do not seem to be disturbed by this process. Figure 3.1 shows a white blood cell that looks abnormal with Swiss cheese-like holes in it, which contain storage material. Among the many cells of the body, however, there are two cell types that are extremely sensitive to the deficiency of the CLN3 protein: light-sensitive cells of the retina and the neuronal cells within the nervous system, mainly in the brain. Although these cells are able to work normally during the first years of life, they work less well later on and eventually die, a process called neurodegeneration. All medical symptoms of JNCL are a consequence of this process, many details of which have not yet been clarified. The general medical hallmark of JNCL, as of most other forms of NCL, is the combination of visual loss through damage to the retina and a progressive brain disease characterized by epilepsy and dementia.

## **Diagnosis of JNCL**

Once JNCL is suspected, making a diagnosis is usually not difficult. However, diagnosis is often delayed for several years after the onset of symptoms; a recently published survey suggested this takes four years on average (Dulz et al., 2015), a delay that has not been much reduced in recent years and is unacceptable. One reason for this delay is the lack of awareness of JNCL among ophthalmologists, who are usually the first to see a child with the beginnings of visual failure. JNCL is the most common cause of severe visual impairment with onset between the age five and fifteen years. Although there are other retinal disorders to consider, a child of school age who begins to have visual problems due to an abnormality of the retina should be suspected of having JNCL until it is proven otherwise.

A first diagnostic test is the microscopic examination of blood cells for the presence of characteristic vacuolated lymphocytes (see Figure 3.1). Technically, this test is relatively simple as it starts with a routine blood smear, but it requires

Defective gene	Type of mutation	Remarks		
CLN3	Classic deletion (1 kb)	A large majority of individuals with JNCL have this gene defect. The basis of most medical (Åberg et al., 2011) and educational research (von Tetzchner et al., 2013).		
CLN3	Missense mutations (may be in combination with the 1 kb deletion)	A rare condition. May have a protracted clinical course (slower progression of the disease than in individuals with classical deletion) or later onset, with only some symptoms (Licchetta et al., 2015)		
CLN1	Missense	The disease may start relatively late, visual loss may not be so prominent in the beginning as in individuals with the CLN3 gene (Khan et al., 2013). A potentially treatable enzyme deficiency.		
CLN2 Missense		Larger variability of the order in which symptoms appear. Epilepsy may occur early, visual loss may not be the first symptom (Kohan et al., 2012). A treatable enzyme deficiency.		

Table 3.2 Gene defects in the major forms of JNCL

specially trained personnel who are mainly found only in specialist medical centers. When a high number of lymphocytes with typical vacuoles are found, the diagnosis of JNCL is almost certain. Molecular genetic analysis of the CLN3 gene can be used to detect the most frequent typical deletion in this gene. If the deletion is not present, other mutations in the same gene must be looked for or other genes analyzed, mutations of which can occasionally cause JNCL (see Table 3.2). In most areas, molecular genetic analysis is recommended as the standard procedure for diagnosis.

For the ophthalmologist, the best way to handle the problem is by sending the child to a pediatric medical center with experience in genetic metabolic disorders. Although the necessary tests can be carried out almost anywhere, pediatric experience is desirable for discussing the test results. If results are negative, other rare metabolic or neurological diseases will have to be considered.

A positive test result for JNCL is a matter of great distress for the family. Talking about a disease like JNCL to a family for the first time – preferably to both parents – needs a doctor and other professionals who are adequately informed (Reed et al., 2015). Many follow-up discussions on the nature and prognosis of the disease will be needed with a range of specialists and professionals. Parent organizations provide support and advice through the disease process and it is important to have JNCL teams involved from the outset (see Chapter 9).

## Medical treatment of JNCL

JNCL is an incurable disease. For some other forms of NCL that are caused by deficiencies of enzymes, different experimental therapies are in a preclinical stage, while enzyme replacement therapy has already been successful in children with the CLN2 disease (Schulz et al., 2018). JNCL caused by CLN3 mutations is not amenable to such an approach. The reason for this is that the deficient CLN3 protein is not soluble and cannot be replaced in cells with relative ease. Presently such techniques do not work for a protein like CLN3 that is an integral component of membranes.

However, the lack of curative therapy at the present time does not mean that individuals with JNCL cannot be treated for many of the issues arising during the progression of the disease. While most of the treatment options are shared with other neurodegenerative diseases, their application may require a thorough understanding of the disease and how it progresses.

## Common problems during disease progression

In view of the characteristic progression of JNCL caused by CLN3 mutations, it has been suggested that the clinical course can be described as a sequence of phases or stages (Kohlschütter et al., 1988; Marshall et al., 2005). There is an overall pattern of loss of functions and abilities, typically with an initial period of mainly visual failure, followed by years with gradually increasing cognitive decline and epilepsy, a growing complexity of problems as motor and language functions decline, and a final phase leading to a comprehensive need for help. However, there is much variability in when cognitive, language and motor problems appear, even in children with the same gene mutation (Lebrun et al., 2011). Moreover, although the phases are typically related to age, they do not always appear at the same time, and the age indications below should be considered approximate. The phases overlap to some degree and the problems caused by the disease may vary considerably. With age, individuals with JNCL may experience a variety of medical problems, including epilepsy, problems with sleep, nutrition and circulation, as well as pain and hallucinations.

## Phase 1: Isolated visual failure (4 to 8 years)

During the first years of life, children with JNCL show apparently typical development. The onset of visual problems is usually noted around the age of five to six years, but it often takes several years for a definitive diagnosis to be made (see Chapter 4). One reason may be that ophthalmologists can have difficulties

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recognizing that this poor vision is not due to an abnormality of the refractory system of the eye, but of the retina. Examination of the eyes will eventually show severe and characteristic changes (Figure 3.2) but these are less pronounced in this phase of the disease. Electroretinography – a more demanding technical investigation – will early show that the retinas are damaged and do not produce electrical responses when exposed to light (extinguished electroretinogram). Frequently, a misdiagnosis of retinitis pigmentosa is made. This is a disease that leads to blindness but not to the same consequences as a brain disease such as JNCL. Optical coherence tomography (OCT) is a newer non-invasive procedure that will show a characteristically abnormal structure of the retina (Dulz et al., 2015).

Early in development, as vision deteriorates, children with JNCL show a characteristic "overlooking" behavior that can be easily recognized by non-specialists. While the center of the retina is already destroyed, some vision is still possible using the periphery of the retina. So when a child with JNCL is asked to look at you, he or she will often look at a point above or on the side of your head.

The loss of vision tends to occur rather uniformly and rapidly in individuals with JNCL, resulting on average in legal blindness by nine to ten years of age. However, there are noticeable exceptions (see Chapter 4). During this first phase of JNCL, there are usually no other significant medical problems.



Figure 3.2 Ophthalmologist's view of the light-sensitive retina at the back of the eye in two young people with JNCL

The photograph on the left side shows a "bull's eye" phenomenon in the area of the macula (the spot of greatest visual acuity) in the center, an abnormally pale optic disc (the entrance of the optic nerve) and thinning of blood vessels, indicating decay of the retina. The photograph on the right shows irregular accumulation of blackish pigment, which can lead to the misdiagnosis of retinitis pigmentosa.

# Phase 2: Emerging intellectual difficulties (6 to 10 years)

During this phase, it slowly becomes clear that the disease is not restricted to the eyes. Initially, a teacher may observe that a child is still active as normal and has no problems with assignments requiring the use of language, in sports or social communication, but develops unexpected difficulties in mathematics (see Chapter 12). Thinking and talking in particular seem to undergo some changes in children with JNCL in this phase (see Chapters 5 and 6). Eventually, the child will begin to find following lessons so difficult that special educational support and help are required (see Chapter 12). For a while, these difficulties may be attributed to issues of coping with visual loss and may not be regarded as being caused by the dementia process. In some children, behavioral changes (see below) may start in parallel to the cognitive decline.

# Phase 3: Increasing medical problems (9 to 14 years)

#### Epilepsy

A seizure disorder (epilepsy) is a regular component of JNCL (Kohlschütter et al., 2014). The degenerative processes in the brain often lead to abnormal electrical activity or abnormal excitability of the cells during this phase of the disease. The abnormal electrical activity causes muscular convulsions and altered mental states. The first seizure usually occurs at around ten years of age; in the present survey (Appendix A) the first seizure was observed on average at 10.4 years (Sd = 3.0) but with a range from two to nineteen years. Family and teachers should be informed and prepared for this to happen. A number of more or less pronounced types of seizures can occur in JNCL.

A typical and the most dramatic type of seizure is the tonic clonic (used to be called «grand mal») seizure. Without warning, the individual falls, loses consciousness and shows some kind of abnormal movement. The limbs may be stiff (tonic) in the initial phase of the seizure, or move jerkily (clonic). This type of seizure is a combination of the loss of consciousness and unusual motor activity, and implies that the whole brain is in a state of altered electrical activity – a generalized seizure. The stiffness of all muscles of the body during the initial phase of the seizure can hinder normal respiration and lead to a bluish discoloration of lips and skin (cyanosis). After a few minutes, the abnormal movements and the cyanosis subside. The individual falls into a deep sleep, from which he or she awakes with a feeling of exhaustion. Whilst seizures cause great concern, a generalized tonic-clonic seizure is not usually a life-threatening event. In children and young people with JNCL, such seizures usually last only a few minutes and tend to stop without medical intervention. When a seizure takes place, it helps to turn the person on the side, move items to prevent accidents and observe calmly until the person becomes fully awake and responsive. When a seizure follows this pattern and lasts only a few minutes, emergency transportation to hospital is not usually necessary, but an appointment with the general practitioner or a pediatric neurologist should be made.

Other types of seizures, such as «absence» seizures or «partial» seizures, can be hard to recognize because they may just involve moments of non-responsive behavior or suggest daydreaming. The electroencephalogram (EEG) is a helpful method to determine the presence of subtle seizures and to characterize them.

The propensity of a child or young person for repeated seizures is quite variable. It can be reduced by antiepileptic drugs, but when seizures occur only rarely and can be managed, starting continuous antiepileptic treatment after the first appearance of seizures is not always necessary. The disadvantages of such a treatment needs to be balanced against the desired benefits for each individual. There are excellent antiepileptic drugs to be used in JNCL. As the brain in JNCL is subject to a process of degeneration, the way it reacts to commonly used drugs may not be the same as in children with other conditions (Kohlschütter, Schulz & Denecke, 2014). Some anticonvulsants may be suitable for individuals with JNCL (valproate and lamotrigine), while others may have negative effects on the disease course and should be avoided (carbamazepine, phenytoin, vigabatrin) (Schulz et al., 2013). Some drugs are more prone to produce side effects such as speech disturbance (topiramate) or agitation (levetiracetam and topiramate). Certain drugs used for children with epilepsy may have unusual side effects in individuals with JNCL (Larsen & Østergaard, 2014).

Generally speaking, seizures early in the course of JNCL usually respond well to treatment with antiepileptic drugs, but they will not disappear completely. Later on, seizures may take on a treatment-resistant character. Doctors should bear this in mind and give parents and professionals sufficient information and advice on the possible options so informed choices can be made. Appropriate treatment of epilepsy may be developed with a carefully controlled approach, starting initially with a single drug in low dosage, later using higher dosage. If ineffective, the drug may be changed or combined with another one. A treatment is satisfactory when seizures are suppressed and no side effects present. If this cannot be achieved, it is often better to tolerate some seizures than to experience toxicity from using too many different drugs. Ideally, the management of a seizure disorder is in the hands of a pediatric neurologist or other clinician who is in contact with an NCL Center. Observations by teachers may be helpful to recognize frequent subtle seizures, which may significantly interfere with the child or young person's attention and thinking. Side effects of antiepileptic drugs, such as excessive sedation, may also be detected at school. With the help of such observations, the dosage of a drug can be modified to provide greater benefits.

### Behavioral changes

With time, a child with JNCL realizes that things are different. Apart from the loss of vision, other things that had been previously mastered become more difficult, and the difference in achievements to peers is more noticeable (see Chapter 27). Mood changes, emotional instability and obsessive-compulsive-like behavior (Adams et al., 2006) become noticeable to other people. Frustration can result in depression or aggressive behavior. Help from an experienced child psychologist or psychiatrist may be needed. Poor medical management of epilepsy may add to these problems and should therefore be monitored carefully.

#### Sleep

A striking observation is the apparent loss of a normal day-and-night rhythm in children with JNCL (Lehwald et al., 2016; Telakivi, Partinen, & Salmi, 1985). Many children and young people with JNCL develop the habit of staying awake until late at night. In the present survey (Appendix A), the average age when sleep problems first appeared was 11 years but variation was large, with a standard deviation of 6.1 years and a range from one to 23 years.

Sleep problems can be disturbing or annoying to the whole family so some form of sleep inducing-medication may be needed. Melatonin is a popular drug in this situation, but does not seem to be very helpful (Hätonen et al., 1999). Alternatives include chloral hydrate, but alterations to a daily routine or lifestyle, as well as a possibly suboptimal antiepileptic treatment should also be considered. Sleep problems may relate to anxiety or other psychological factors (see Chapter 27). Good sleep hygiene should be encouraged. Sometimes a 24-hour EEG is helpful to detect epileptic activity during sleep, as this can add to sleep disturbances and may be managed by improving the anticonvulsive treatment.

#### Decrease of motor abilities

In late childhood or early adolescence, minor motor problems may become noticeable (see Chapter 7). In this phase, children with JNCL begin to appear clumsy and to lose their balance more easily than previously. Some everyday activities (dressing, undressing etc.) requiring fine motor abilities may take more time. In a child who has learned braille, these difficulties may interfere with reading.

## Phase 4: Puberty, adolescence and young adulthood: Multiple functional losses (15+ years)

## Puberty and sex-related issues

Somatic maturation during puberty proceeds without obvious difference. There are some sex-related issues that have not yet been fully clarified. In a large cohort study, the disease showed a slightly more rapid progression in females (Cialone et al., 2012). In another study, several girls were described as having acne and hirsutism, a few had hormonal abnormalities (elevated androgens in blood) and abnormal (polycystic) ovaries. It is possible that some of these abnormalities were related to effects of antiepileptic drugs (Åberg et al., 2002). Menstruation may present problems in personal hygiene because of blindness and decreased mental and fine motor abilities. Girls may experience more frequent and heavier seizures or an excessive increase of tension and nervousness around menstruation. In such situations a gynecologist should be consulted. As well as preventing pregnancy, menstrual suppression – usually by injection of hormones – can be helpful.

### Language

In this phase of the disease, verbal communication becomes increasingly difficult due to a combination of problems: cognitive difficulties are increasing and at the same time the motor apparatus required for speaking becomes more severely disturbed. Frustrated by a willingness to communicate but not being able to talk as easily as before, young people with JNCL may develop a peculiar and characteristic way of speaking. Their expressive language becomes more simple in content, it sounds soft and slurred, and often becomes understandable only to persons who are familiar with the person with JNCL (see Chapter 6). Some young people with JNCL appear to be in a hurry to get their idea over to their partner. Their speech contains many repetitive elements, stuttering or repeating of words or short sentences, and may include apparently meaningless phrases. The age at which language becomes hardly understandable is very variable, ranging from eight years to more than 20 years (Lebrun et al., 2011). Comprehension of language persists longer than expressive speech, which may contribute to frustration and aggressive behavior, as a reaction to not being understood. There are several approaches to help both young persons with JNCL and their caregivers to communicate better (see Chapter 13).



Figure 3.3 Typical posture of an adolescent with JNCL

## *Major motor problems, loss of ambulation*

Starting at the age of 10 to 12 years, some children with JNCL develop a peculiar «extrapyramidal» motor disorder that resembles Parkinson disease. In others, this arises later on (Järvelä et al., 1997). Symptoms of this disorder are hypokinesia (decreased bodily movement), rigidity (stiffness) of muscles, stooped posture (see Figure 3.3), shuffling gait and impaired balance. The symptoms can sometimes be temporarily ameliorated by anti-Parkinson medication (dopa or similar drugs) (Åberg et al., 2001). Some young people with JNCL become wheelchair-bound at around 16 years of age, but the age when this happens is very variable (from 11 to 26 years) (Lebrun et al., 2011).

## Decrease in intellectual abilities

Visual loss and speech impairment may make cognitive assessment difficult beyond the age of 15, but dementia may become more severe and include decline in memory, attention, emotional control, and general reasoning abilities (see Chapter 5). There is also likely to be a decline in independence skills like mobility, feeding and communication. The need for support among individuals with

JNCL can be likened to the care needed by older people with Alzheimer disease or other forms of dementia. The aim of support in this area is to provide reassurance to the young persons and their relatives, and to avoid that they become isolated and inactive.

## Behavioral, psychological and psychiatric issues

Children and young people with JNCL may show many behavioral problems and psychiatric symptoms (Adams et al., 2006; see Chapter 27). In a group of individuals with JNCL aged between 9 to 21, the most common symptoms reported by parents and other caregivers were problems with social interaction, thinking, attention, aggressive behavior and some less well defined somatic complaints

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(Bäckman et al., 2005), as well as anxiety and depression. While similar problems are frequent in individuals with intellectual disability (Myrbakk & von Tetzchner, 2008), they are often recognized late in young people with JNCL and treatment is often insufficient. Hallucinations are frequent in the late phase of JNCL (Lanska & Lanska, 1993). A hallucination is a vivid experience of something that appears to be located in external objective space, typically an episode of several minutes duration, and perceived by the subject as real. It is important for caregivers to be aware of the individual's sense of reality of the experience (see Chapter 25). Hallucinations may be harmless, even pleasant. The individual may be pausing without apparent motive or movement, or may smile without any comment. Quite different and potentially dramatic are hallucinations with an unpleasant content. A hallucinating person may have the subjective experience that the house is on fire, he or she might say (or think) that immediate action is required or try to run away. When this happens in the classroom and does not stop spontaneously, it can be distressing for all involved. Reassuring words like «Look, nothing feels hot in the room, there is no smoke,» may not be sufficient to calm the person down. The situation may develop into a psychiatric emergency and require the use of strong sedative medication and further psychiatric management.

In this phase of the disease, young people with JNCL are generally less able to tolerate a change in their environment. They feel safer, more comfortable and show more emotional stability in familiar environments and in the presence of people they know. Such considerations may be of value when decisions relating to education and accommodation have to be made.

## Other somatic problems

#### Nutrition

There is no need for a special diet. Constipation should be avoided, as this can become a chronic problem in later stages of the disease.

#### Circulation

Children and young people with JNCL frequently have cold hands and feet without abnormal findings when examined. Later in the disease progression, the heart may be affected (Østergaard, Rasmussen, & Mølgaard, 2011), primarily resulting in a slowing of the heartbeat. In exceptional cases, late in the course of the disease, surgical insertion of a cardiac pacemaker can significantly improve well-being. In adolescents, a specific treatment for a heart condition is not usually necessary, but the heart should be checked as part of the medical follow-up.



Figure 3.4 Results from a study of 25 individuals with JNCL with identical mutations (classical deletion) in the CLN3 gene

Scoring: (3) normal function, (2) slight problems, but readily recognized, (1) severe problems, and (0) total loss of function. The shaded area represents scores between the 10th and 90th percentile. The diagram illustrates that the vision impairment occurs quite uniformly, while the loss of language and motor functions is more variable (Lebrun et al., 2011, p. 1255). The dotted red lines represent the course in an individual with a particularly fast loss of language and motor functions. The second individual showed some improvement in motor and language functions after the implantation of a cardiac pacemaker at age 30 years.

#### Pain

When a young person with JNCL has lost the ability to communicate successfully, any situation where he or she expresses unhappiness or pain without apparent cause, raises difficult questions. When the problem does not subside spontaneously, a doctor should look for possible abnormalities in organs before analgesics are prescribed.

## **Concluding comments**

Following the individual trajectories of JNCL over many years offers a striking observation: while the loss of vision proceeds quite uniformly and is very similar in all individuals, the decline of language and motor abilities take a more variable course with large individual variation (Figure 3.4). The reasons for this are not known, nor why there are differences between typical courses in males and females. Understanding the reasons for this might help to improve medical treatment. It cannot be ruled out that variable quality of the medical treatments used may contribute to these differences. It is therefore very important to carefully monitor treatments and the disease progression in all children and young people with JNCL throughout their life.

There are further medical issues that can arise during the disease progression that are not addressed within the scope of this book. Families should know that specific medical experience with JNCL has developed so that whatever their child may experience, expert advice is available (see Appendix B).

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## **Development and Loss of Vision**

Bengt Elmerskog and Stephen von Tetzchner

Perception is the ability to distinguish and identify sensory information, to direct and sustain attention to various aspects of the environment, and to lend meaning to them. Children use their senses to explore the physical and social world, establish a basis for action, and to monitor and regulate their own actions. The sensory systems facilitate interaction with other people. Vision is the sense that provides the most information about the environment (von Tetzchner, 2019).

#### Visual impairment

Visual impairment is defined as a reduced ability to perceive and interpret visual sensory stimulation beyond what may be compensated with glasses. The impairment may consist in reduced visual acuity or field of vision, or a combination of these two functions. Visual impairment ranges from "normal or mild" to "(total) blindness" (Table 4.1). Normal vision is usually described as 6/6, meaning that the person with best correction (glasses) at a distance of six meters can see the same as most people can at that distance. Moderate visual impairment ranges from 6/18 to 6/60, indicating that the person only at a distance of six meters can see what people with normal sight can see at 18 to 60 meters. The British legal definition of blindness is 3/60, which means that the person needs to be at a distance of three meters to be able to see what people with normal sight can see at 60 meters. The US definition is 6/60. "Near total blindness" indicates some light perception while "total blindness" implies that the person has no light perception (World Health Organization, 2010).

Mild visual impairment is common and can in many cases be corrected with glasses (Wiener, Welsh, & Blasch, 2010). Approximately 1–2 percent of school children have reduced vision with functional consequences. Many children with severe visual impairment have additional disorders, particularly in mobility,

Categories of visual impairment		Worse than	Equal or better than	
0	Mild or no visual impairment		6/18	
1	Moderate visual impairment	6/19	6/60	
2	Severe visual impairment	6/60	3/60	
3	Blindness	3/60	1/60	
4	Blindness (near total)	1/60	Light perception	
5	Blindness (total)	No light perception		

Table 4.1 Categories of visual impairment

Note: Degree of visual impairment is described as degree of vision related to typical vision. Average normal vision is usually described as 6/6, meaning that the person can see the same at a distance of six meters as most people do. Moderate visual impairment ranges from 6/19 to 6/60, indicating that the person can see at six meters what people with normal vision can see at 19 to 60 meters.

hearing impairment, intellectual disability (Geddie et al., 2013). Although rare, JNCL continues to be the most common cause of blindness in many countries in children aged 5–15 years (Mole et al., 2011). There is much research on the development of children with congenital or early acquired blindness. Less is known about the developmental consequences of blindness acquired in middle childhood (age 6–12 years).

## Consequences of visual impairment

The visual sense is so basic to human functioning that severe visual impairment will cause difficulties in many areas of development. Many ordinary activities of daily living become difficult to master, such as pouring liquid into a cup, finding things and orientating in the physical environment (Brambring, 2007; Dale & Edwards, 2015; Fraiberg, 1977). For people with low vison, access to visual information may be enhanced with technical aids, improved lighting, and magnification, whereas individuals with blindness often must learn to do things in a different way to cope in the environment (see Chapter 16). Even a minimum of light perception may make it easier for the person to acquire many daily living skills.

Early visual impairment will typically affect motor development and physical status (see Chapter 7), and children who are visually impaired may be less physically fit than their sighted peers (Aslan, Calik, & Kitis, 2012; Skaggs & Hopper, 1996).

Independence in activities of daily living may be challenging for most individuals with severe visual impairment, particularly those activities involving movement from one location to another. An early- or late-acquired blindness will always entail an increased dependence on others, as well as special needs for learning and physical adaptations (Wiener et al., 2010). For instance, for persons with total blindness, orientation in space may depend on stable placement of objects and of topographic patterns in the surroundings that can be detected by touch or hearing (see Chapter 16).

Most ordinary social activities for children require vision. Severe visual impairment will restrict possibilities for participating in soccer, bicycling, window shopping, and so forth. Although many children walk or bicycle to school, children who are blind may need to go to school by car or be accompanied by an adult. Children with normal sight may explore their surroundings visually on their way from a starting point to a destination. Children's movements, like play, can exist for their own sake without the need for some goal (e.g., reaching a destination). Thus, like play, they constitute important learning elements in children's development (Bodrova and Leong, 2015; Lillard, 2001; Piaget, 1951). In most cases, children who are blind face barriers to initiating and participating in such activities and there may be consequences for their overall development (Fraiberg, 1977).

Metaphorically, Gibson (1979) describes perception of the environment in a person who is blind as a "bubble" or "tunnel", while a person with normal sight perceives the environment as a "field", "spectrum" or "map". Further, sighted people can perceive spatial information in a single glance, while individuals who are blind need to explore, synthesize, reconstruct and memorize to obtain similar information about the physical environment.

Visual information may to some extent be compensated with auditory and tactile information. Tactile information, for example about an object landmark, depends on the object of investigation being reachable by hands or body. For people who are blind, orientation in space therefore requires directed attention and more cognitive resources, and patterns or structures in the environment that enable them to move independently (see Chapter 16). Independent goal-oriented movement from place to place requires anticipation of learned landmarks and an understanding of environmental features that cannot be perceived with touch or hearing. Knowing where one is in physical space, where objects are in relation to each other and to self, and how these relations change with self-movement, represent life-long challenges for people who are blind (Tellevik, Storliløkken, Martinsen, & Elmerskog, 1998; Wiener et al., 2010).

Moreover, the independence and autonomy of the individual may be negatively influenced when caregivers, in an effort to help or to rush through activities, complete tasks for the individual with blindness, sometimes leading to learned helplessness (Dodds et al., 1991). Such behaviors from caregivers may hinder efforts of individuals who are blind to learn about the world through the remaining senses, particularly hearing and touch (Willings, 2017). For persons who are blind, independent living, without assistance from sighted helpers, is limited to familiar environments; however, independence outside these areas will almost always involve support from sighted helpers. People who are blind therefore need to learn (and dare to and are motivated to learn) how they can use sighted people in the surroundings as tools to achieve personal goals. Some authors define such self-governed strategies as "interdependence" (Tellevik & Elmerskog, 2001; see Chapter 16).

There is evidence that people who are blind may develop stronger memory skills that compensate for the absence of vision in developing and maintaining a social and independent lifestyle (Withagen, Kappers, Vervloed, Knoors, & Verhoeven, 2013). For instance, features in the environment that cannot be perceived by vision, owing to the impairment, must be mapped and memorized using information from the other senses, like touch or hearing. Such behaviors require extensive and time-consuming exploration and memory (see Chapter 16).

### Early and later vision loss

The consequences of visual impairment depend on age of onset of visual loss. Visual impairment that is present from birth (congenital) will have a greater impact on development and learning than visual impairment acquired later. A great deal of an infant's learning about the world is through the visual modality. Learning by observation is considered an important way of learning but is limited for children with congenital severe visual impairment. Social development is also affected, because children who are blind are not able to pick up on non-verbal cues or make eye contact. In addition, children who are blind may appear disinterested (their attentive mode may look different from that of sighted peers), resulting in reduced sustained social interactions with peers (Perez-Pereira & Conti-Ramsden, 1999).

The surroundings will in many cases appear less stimulating for children who are blind compared to children with sight (Adelson & Fraiberg, 1974). Loss of vision will thus have a negative impact on motor development because the child is lacking the visual incentive to move toward things, and this deficit may cause inhibition to move for fear of the unknown (Brambring, 2006; Freeman & Cannady; 1971; Levtzion-Korach et al., 2000). Getting to know the environment is essential for cognitive development, and exploration requires both the ability to move and an incentive to move towards particular objects and people. Without vision there may be little incentive to explore things that require close inspection and contact and cannot be appreciated from afar. Thus, reduced exploration may influence both motor development and conceptual development.

Children with congenital blindness have little or no visual experience, and so they build up their understanding of the world from non-visual experiences.

Children with later onset of visual impairment, like children with JNCL, have already learned about the presence of people and objects in the environment and how to use vision to orientate in the world. Unlike children with congenital blindness, they have acquired typical everyday skills in the same way as most children and hence have a different basis for learning new skills later in their development.

## The role of hearing in visual impairment

Hearing has the same functions in blind as in sighted children, but in addition hearing to some extent compensates for visual loss (Elmerskog, Martinsen, Storliløkken, & Tellevik, 1993). Getting to know the immediate environment and being able to orientate in the physical surroundings are core human functions. In the development of mental spatial representations, sighted children use geometric cues and landmarks. Geometric cues include surface, direction, distance, angle and the like, which depend on the viewer's perspective (egocentric cue). Landmarks are external cues and include characteristics such as wall color and placement of windows, doors and objects, and are independent of viewer location (Ferrara & Landau, 2015). Children who are blind need more time to explore and form mental representations of space based on their experiences with locomotion, nonvisual strategies and landmarks, and the need for time may have consequences for their development of exploration and independence (Schmuckler & Gibson, 1989; McAllister and Gray, 2007). Individuals with congenital blindness use hearing to develop mental spatial representations from sounds, but their representations may be based more on egocentric cues - anchored in own body - while sighted individuals proceed to use more external cues (Vercillo, Tonelli, & Gori, 2017). Although hearing provides important information about the direction from which a sound is coming, it gives less precise information about its distance from the listener, and this influences the spatial map of individuals with congenital blindness (Kolarik et al., 2017). Despite the ability to hear, acquired blindness will cause major changes in the individual's life and limit personal perspectives and control of the surroundings.

Some children with congenital blindness develop outstanding listening skills, such as echolocation and perfect pitch. Such abilities are most prominent in individuals with onset of visual problems in early childhood (Dingfelder, 2005; Wan et al., 2010). Echolocation is the ability to detect location of objects by sensing echoes from stomping the foot, snapping the fingers, or making clicking sounds. Children who are blind can identify objects' location, size, and possibly even material composition from a relatively small distance by using echolocation. Good skills in echolocation make the orientation skills of people who are blind more efficient, and these skills are developed through active and independent exploration (Elmerskog et al., 1993). In addition, verbal explanations and descriptions about things and events in the surroundinga can provide richer, supplementary information. Yet despite these rich compensations, most information that may be available through vision will not be available to persons who are blind. Specifically, sighted people are typically using visual references in communication, which may not be useful for people without sight. For instance, it may be useful to apply egocentric references (relative to the position of the person) rather than topographic references in communication with persons without sight (see Chapter 16).

Moreover, auditory information gives a different conception of the environment than vision. Vision gives much information simultaneously, provided there is enough light, including information about the current spatial relationship of things and people in the environment, and how they move. Many objects do not make sounds and may be out of the perceptual field, inaudible, most of the time. Perceived stability will depend on the sound being continuous or repeated. Localization and identification of a sound are two separate processes in hearing (Gibson, 1966). For instance, a scratchy sound created by a person moving on a graveled road can be identified and localized at the same time for a person with vision. A person who is blind can localize the sound through hearing but may not be able to determine the source of the sound unless it is recognized as a sound that has been heard before.

Many people who are blind show exceptional skill in using sound to navigate in space and explore the physical and social environment. However, in some ways, the information provided by vision or by sound may create somewhat different impressions of the environment.

## Visual impairment in individuals with JNCL

The first symptom of JNCL is typically visual loss due to retinal degeneration between four to ten years of age (see Chapter 3). Children with JNCL show decreasing visual acuity and reduced luminance and chromatic contrast sensitivity. They often have large central retinal visual field defects (scotoma) and may compensate for these by using para-foveal or para-macular fixation techniques. This means that the child uses part of the peripheral visual field rather than the central part, for example by fixating above, below or next to the natural foveal fixation point (Augestad, Fosse, & Didrichsen, 2008). Most of the participants in the present study (see Appendix A) showed normal development prior to onset of visual problems. A few parents reported some cognitive or behavioral problems prior to the visual impairment, but none of these symptoms led to a JNCL diagnosis.

In the present survey, parents had first become aware of their child's visual problems at an average age of 6.2 years (SD 1.7, range 2-12) (see Figure 4.1). A little later, at around 7.8 years (SD 2.0, range 4-14), the visual impairment began to have a major impact on everyday functioning. Around the age of 10.7 (SD 3.4, range 5-20), the child had become blind. The average time period from when the first symptoms emerged to when the children had become blind was rather short, around 4½ years. However, there was still considerable variation in the following variables: (a) age at onset of visual problems, (b) age when these problems made a significant impact, and (c) age when the child became blind; and there were exceptional cases with much earlier or later age of onset. Despite the rather short average time span, the results indicate a larger variation in visual decline among children with JNCL than has usually been assumed. This finding is important when starting to plan for the education and environmental adaptation for a child in this group. Many children with JNCL utilize their vision actively for orientation and in leisure activities throughout the early school years, and they may use residual vision effectively for learning purposes until the age of 9–10 years, in some cases well into the teens or even after the age of 20 (see Chapter 12).

Children with JNCL have several years of visual experience when the visual decline starts. The development in this period constitutes a foundation for how they think and act in the world. Children with JNCL know what the world looks like, they have developed an understanding of spatial concepts and space based on visual experiences, they know what colors are, and they have developed



Figure 4.1 Visual decline in JNCL

interests and preferences in a similar manner as their peers. The content of their conversation after the onset of blindness illustrates the continuous role of their visual experiences. Colors of clothes and dolls were important for many children after the onset of blindness, they were using visual references and concepts when describing the environment, and they used gestures and other vision-dependent non-verbal expressions to underline their own meanings like sighted children do. In fact, in the present survey some parents and staff reported that the visual references based on memories were so pervasive that the children and young people with JNCL believed that they had seen objects or events after they had been diagnosed with total blindness. Moreover, there were no reports of the use of echolocation or other outstanding auditory abilities often observed in children with congenital blindness.

The results further emphasize the difference in functioning between children with congenital blindness and children with JNCL. Almost no children with JNCL showed problems with passivity, self-stimulation or self-understanding in early life as do children with congenital blindness. However, many parents found that their child's life became more passive after he or she became blind, and this was more prominent after the age of 16, several years after the onset of blindness. This may suggest that problems with passivity were not primarily caused by the visual impairment. Emerging dementia and motor problems, as well as reduced access to activities with friends, may have contributed to the development of a more passive life style (see Chapter 21).

Individuals with JNCL and other forms of early acquired visual impairment must learn how to cope with the world in new ways. This coping requires mental and cognitive effort, and it may be easier for children to begin learning how to cope while their cognitive capacity and ability to learn are as good as possible (see Chapters 12 and 16). The difficulty lies in the finding that cognitive decline (dementia) in children with JNCL evolves not long after the emergence of severe visual impairment (see Chapter 5). Cognitive abilities and difficulties are important considerations for children's education. However, the findings of the present survey suggest that to some extent, the visual impairment seemed to over-shadow possible cognitive problems in the early years, and cognitive problems received limited attention from the schools and educational authorities, or even from JNCL counselors. In fact, cognitive and visual decline seemed rather to be approached as two discrete, unrelated phenomena leading to fragmented intervention measures.

Of special importance is how people in the surroundings respond to the child's emergent visual problems and special needs. A compassionate response may result in over-eagerness to help or assist the child in many situations, to do things for the child that the child is able to do herself, or to wait with useful educational initiatives until the situation has "calmed down". It is important to
emphasize that besides the gradually increasing visual problems, most children with JNCL do not experience big changes in connection with getting the diagnosis. Their life goes on as usual and most children in the present study were not aware of their own diagnosis at this point in time. However, the child's diagnosis will have a major impact on parents and others who are informed about it.

We experienced that we received a new son after the diagnosis was delivered to us. Our old son with all expectations about the future were gone in a glance and replaced with a new son with alternative expectations for the future.

Reactions from people the environment, transactionally caused partly by the child's visual problems and changing behavior (see Chapter 2), may impose an indirect impact on the child's everyday life (see Chapters 22 and 27). Teachers and other professionals may also show exaggerated compassionate reactions. It is a risk that such reactions will interfere or impede the development or maintenance of independence in young persons with JNCL. For instance, a child or adolescent with JNCL who originally had a high degree of independence, may become accustomed to things being done on his or her behalf. However, it may support maintenance of independence and autonomy when young people with JNCL learn to use sighted people as tools to achieve personal goals in unfamiliar environments. Such self-governed strategies may be defined as interdependence (see Chapter 16).

### Conclusions

Visual loss is one of the defining symptoms of JNCL and the visual problems that typically begin to emerge in early childhood are in most cases the main basis for the diagnostic process. There is considerable research on the development of children with congenital blindness, and this research represents one important source for understanding the development of children with JNCL. Nevertheless, there are significant differences between the development of children with congenital blindness or onset of blindness in infancy, and the development of children with gradually increasing visual impairment later in childhood. In addition, the visual problems interact with the emergent cognitive problems in the development of children with JNCL. A necessary basis for goal-oriented and adapted education is an understanding of how the visual problems work together with the cognitive problems in development, learning and participation. In many cases, adapted education will necessitate inclusion of an extended curriculum for children and adolescents with JNCL

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# Dementia in Adulthood and Childhood

Per Kristian Haugen, Stephen von Tetzchner, Judith D. Oxley and Bengt Elmerskog

Defention of the progressive nature, in which there is disturbance of multiple higher cortical functions, including memory, thinking, orientation, comprehension, calculation, learning capacity, language, and judgement (World Health Organization, 2018). Thus, dementia is a general term that encompasses a loss of memory and other acquired mental abilities that are severe enough to interfere with many aspects of daily life. Together with visual impairment, dementia is a core aspect of juvenile neuronal ceroid lipofuscinosis (JNCL). This chapter discusses specific aspects of dementia, including how problems with attention, memory, and other cognitive features are present in children and young people with JNCL.

The developmental trajectories of individuals with intellectual disability and dementia differ from typical development (Shapiro & Klein, 1994). The trajectory of intellectual disability reflects steady growth but has a lower curve than typical development, and the gap widens with age. The developmental trajectory of individuals with JNCL and other forms of childhood dementia initially follows the typical course for a shorter or longer interval, and then starts to decelerate and fall behind typical development, and eventually regresses below the individual's former level (Figure 5.1). There are significant individual differences in the symptomatology and development only. The figure also illustrates why traditional methods for evaluating educational practice may not be useful for students with childhood-onset dementia.

Elderly people with adult-onset dementia may experience perceptual problems, postural instability, tremor, slowness, rigidity and other motor problems, as well as emotional or personality changes in the form of depression, anxiety or deviant social behavior (Finkel, Burns & Choen, 2000; Podell & Torres, 2011). Dementia thus influences the main functional domains and includes cognitive,



Figure 5.1 Developmental trajectories of individuals with typical development, intellectual disability and childhood dementia (after Shapiro & Klein, 1994)

The lines designate typical development, the development of children with intellectual disability and the development of children with dementia and JNCL. Development for this last group of children proceeds normally at first, then comes to a stop, and then declines.

behavioral and motor symptoms. The same features are apparent in dementia with onset in childhood (Schoenberg & Scott, 2011; Shapiro & Klein, 1994). Visual development and impairment in JNCL are discussed in Chapter 4, motor development and impairment in Chapter 7, language in Chapters 6 and 13, and emotional and behavioral problems in Chapter 27. This chapter focuses mainly on the cognitive aspects of dementia.

### Dementia

Dementia is caused by physical changes in the brain, usually degeneration in the cerebral cortex, the part of the brain mainly associated with thinking, memory, action-planning, and personality. Common dementia diseases are Alzheimer disease, vascular dementia, dementia with Lewy bodies, Parkinson disease and Huntington disease (Alzheimer Europe, 2013; Engedal & Haugen, 1993; Harvey, Skelton-Robinson & Rossor, 2003). Childhood dementia is rare and less known, but several diseases imply a decline in cognitive functioning in childhood after a period of typical functioning, some with onset after a short period of normal functioning, others with onset later in childhood or adolescence (Schoenberg & Scott, 2011; Shapiro & Klein, 1994). Regardless of age, dementia represents a major challenge for the individual with the disease, and for his or her family.

Dementia is usually classified as either primary or secondary. In *primary dementia*, cognitive decline is the main symptom. Most of the primary dementia disorders, such as Alzheimer disease or frontotemporal dementia, are of a progressive nature. The symptoms emerge over time, from mild cognitive difficulties to complete dependence on assistance in all aspects of daily life. Dementia without additional symptoms is most common among older people.

In *secondary dementia*, cognitive decline is one of several symptoms and usually appears in a later phase of the disease. Secondary dementia may be related to brain tumor, Parkinson disease, acquired immune deficiency syndrome (AIDS), normal pressure hydrocephalus or subdural hematoma. Some rare neurodegenerative, autoimmune or inflammatory disorders will primarily affect people under the age of 45 (Kelley, Boeve, & Josephs, 2009). In diseases with onset in childhood, dementia is often regarded as secondary and part of a larger set of symptoms, such as in Nieman-Pick disease (Mengel et al., 2017) and mucopolysaccharidose disorder (Shapiro, Escolar, Delayney, & Mitchell, 2017). Although JNCL usually starts with symptoms of visual impairment, visual impairment is not in itself a cause of dementia. The prominent role of the cognitive decline and other features of dementia over the course of the disease suggest that dementia in JNCL might be considered primary.

A diagnosis of dementia requires that a person exhibits changes in functioning caused by a documented brain disease. There must be a decline in memory together with a reduction in one or more other cognitive functions, such as language, attention, abstraction, judgement or executive functions (Stopford, Thompson, Neary, Richardson, & Snowden, 2012). The reduction in cognitive capacity must be of such a degree that the individual is less able to manage daily life. Consciousness is not clouded and the condition must have lasted at least six months. The impairment of cognitive function is commonly accompanied, and occasionally preceded, by deterioration in emotional control, social behavior, or motivation (American Psychiatric Association, 2013; World Health Organization, 2018). The decline in memory is most evident in the learning of new information, although in more severe cases of the disease, recall of previously learned information may also be affected. In other cognitive abilities, the decline implies a deterioration in judgement and thinking, such as planning, organizing and in the general processing of information, but the person is aware of the environment (Aalten, Van Valen, Clare, Kenny, & Verhey, 2005).

It is common to describe three levels of severity. In mild dementia, the decline in cognitive abilities causes impaired performance in daily living. In moderate dementia in adulthood, the individual can perform the functions of daily living, such as shopping and handling money, but only with assistance. In severe dementia, cognitive functioning is characterized by an absence, or virtual absence, of intelligible creation of ideas (Naik & Nygaard, 2008).

In the early phase of dementia, symptoms vary considerably, depending on type of dementia. In addition, even in the same type of dementia, there may be major individual differences in symptomatology. For example, although, most persons with Alzheimer disease primarily have difficulties with memory, people with other forms of dementia may show language difficulties, apraxia, visual perception difficulties or failure in executive functions in the early phase (Koedam et al., 2010). Similar differences are apparent in childhood dementia (see below), thus emphasizing the need for careful observation of the young person with dementia to assess the overall profile of relative strengths and weaknesses (see also Chapter 10).

The following sections address three cognitive domains affected by dementia: attention, memory, and executive functioning with a view to providing basic descriptions of each, and an account of how they collectively and individually contribute to competencies and decline of competencies in education and general functioning of the young person with JNCL.

#### Attention

Attention refers to being aware of, focusing and processing information from the environment. In everyday life, the individual is continuously exposed to people, things and events in the environment. To understand and make sense of what is happening, the individual needs to select which stimuli are relevant and should be processed. Research suggests there are four different kinds of attention, each of which depends on a different neural network, or set of networks (Peterson & Posner, 2012; Posner, Rothbart, Sheese, & Voelker, 2014). Focused attention is the ability to maintain awareness over time directed at one task or event and respond specifically to one stimulus. It is believed to be mediated by the *alerting* network (Peterson & Posner, 2012). Selective attention refers to the ability to avoid stimuli that can distract from the focus of attention, such as cars driving by, closing of doors, or people having a conversation nearby (Lezak, Howieson, Bigler, & Trane, 2015). It is believed to be mediated by an *executive network*. Alternating attention requires ability to disengage and reengage one's response to stimuli in the environment (Parasuraman, 1998), and is suggested to be supported by the *orienting network*. *Divided or distributed attention* is the ability to maintain awareness of several aspects of the environment or objects at the same time. It can imply doing several things simultaneously (e.g., as when many things happen at once), or keeping track of multiple ideas simultaneously (e.g., keeping track of several elements in a verbal message) (Perry & Hodges, 1999). Distributed attention appears to rely on all three types of networks (i.e., alerting,

executive, and orienting). In summary, attention is a complex cognitive function that underlies social, educational, and daily living skills.

### Problems of attention

Attention deficits are classified as a separate group of disorders (American Psychiatric Association, 2013) but problems with attention are apparent in many developmental disorders (Kerns et al., 2015). These problems are not only a matter of regulating attention better or worse than other children, but also of what children and adolescents pay attention to (Burack et al., 2016). For example, studies have found that children with autism spectrum disorders differ from their peers in terms of which aspects of a situation serve as the focus of their attention (Fan, 2013; Keehn et al., 2013). This finding suggests differences in their alerting network. Difficulties with dividing or distributing attention are often one of the first signs of dementia (Stopford et al., 2012) perhaps due to the complexity of these forms of attention. Attention and distractibility may be affected by problems related to vision (Tadi , Spring, & Dale, 2009).

### Memory

Memory represents the preservation or retrieval of knowledge and of events that took place a shorter or longer time ago, often many years back in time. Memory is an essential element in all learning and necessary for the management of everyday tasks and social interaction. Memory allows the mind to use earlier experiences and makes it possible for the individual to anticipate the future and create continuity between the past, present and future. Memory problems appear in many developmental disorders (Peterson, Jones, Stephens, Gözenman, & Berryhill, 2016), and decline in memory functions is a defining feature of all forms of dementia.

### The memory systems

The human memory consists of several elements with different functions. *Working memory* stores information for a very short time (seconds or minutes) and processes or organizes the information, as when searching for something or remembering a telephone number while dialing. Working memory has limited capacity. The functions of working memory are related to attention, the here-and-now and what the individual is aware of in the moment. At any given time, working memory contains what is needed in the moment. The material

held in working memory may be transferred to long-term memory or become forgotten after a few seconds. Working memory registers information from both the environment and long-term memory, and thus helps tie together new and already stored information and forms an integral part of all learning and thinking (Cowan, 2014).

Long-term memory has two parts: procedural memory and declarative memory. Procedural memory contains action schemas, how to perform something that is learned, including automatized actions such as bicycling, swimming or reciting the lyrics of a popular song. *Declarative memory* is divided into semantic memory and episodic memory. Semantic memory comprises general knowledge, facts, what the person knows, such as the names of capitals in Europe or the fact that an English mile is 1609 meters. *Episodic memory* is about the experience of the self and the quality of one's own memories. It includes such things as details from a conversation one was involved in the day before, what happened at the appointment with the doctor a week ago, or the summer vacation enjoyed many years ago. Episodic memories also have qualities such as color, shape, image, motion and sound. The content of episodic memory implies a feeling of authenticity, of "having been there", and of personal participation and involvement. The life history or *personal narrative* of the individual is made up of episodic memories (Daselar & Cabeza, 2008). Some researchers believe that semantic and episodic memory constitute two different memory systems, whereas others maintain that all the content of the declarative memory is semantic but that events experienced by the self have an added emotional quality (Parkin, 1997).

Long-term memory stores information over time, in some cases throughout life. It is considered to have unlimited capacity and constitutes the individual's personal and cultural knowledge base. However, to make use of what is stored, the individual must be able to relate the content to reality. Retrieval from memory can be accomplished through recall or recognition, such as when recognizing a voice or an object, or recalling relevant information when it is needed (Schneider, 2015). Recall is a conscious process of recreation or reconstruction, the individual tells or shows what was observed or experienced, and the focus of attention and emotional state will to some extent determine what is being recalled. By contrast, recognition can proceed unconsciously, as when something the person perceives elicits memories. However, forgetting and unlearning are also important functions of the memory system. They are necessary, enabling the child or adolescent to adapt to constantly changing environments, instead of continuing to act as if the environment remains the same (Bauer, 2014; Cuevas, Rajan, Morasch, & Bell, 2015).

### Memory and dementia

In everyday life, the selection and processing of information, and the meaningmaking process, may make heavy demands on working memory. Impairments in these skills, which reflect deficits in working memory, typically represent early signs of dementia. In addition, problems with episodic memory may be apparent early in dementia, and memory for recent events is often more affected than memory for personal events from the past. Semantic memory, often relatively intact in the initial phase, may nevertheless appear reduced in mild and moderate dementia. Procedural memory is usually the least affected in the early phases of dementia (Hodges, 2000).

The use of recall strategies is usually consciously executed and demands allocation of internal attentional resources, whereas the use of recognition strategies can proceed automatically, without any conscious effort, and dependent on the exploitation of external clues to retrieve memories. However, a person can actively recruit recognition strategies to help in memory retrieval (e.g., purposefully scanning shelves in supermarket for items one forgot to put on the shopping list, or on the list one forgot to bring!). Recognition strategies are preserved longer in dementia than recall strategies, and change in recognition is usually first noticeable in the moderate and severe phases. Recall is usually affected early also in mild degrees of dementia, and may decrease rapidly as the disease develops (Helkala, Laulumaa, Soininen, & Riekkinene, 1988; Van Liew et al., 2016). The name of a former classmate at primary school may be very difficult to recall, even if he or she is readily recognized in the old class photograph. It may be possible to observe for example that someone has heard a voice or seen a face, even when it is clear the person has not consciously registered that recognition has taken place.

The fact that recognition tends to be more intact than recall is of great importance when adapting the environment and encouraging people with dementia to be mentally and physically active. An environment with a limited number of familiar people, familiar surroundings, and avoidance of unnecessary relocations and changes in the environment or in the regular routines, will contribute positively to stability and security, and thus to increased activity and well-being in individuals with dementia (Kitwood, 1997; Woods, Thorgrimsen, Spector, Royan, & Orrell, 2006).

It is a common belief among relatives and professionals that people with dementia are not capable of new learning because of the extent of their memory deficits and the need for memory in learning. However, both clinical observations and research show that people with dementia may have preserved learning abilities (Bird, 1998; Bragin et al., 2009). For instance, studies have found that people with dementia benefit from learning programs as evidenced by improved processing

speed and executive functions related to planning and organization (Kawashima et al., 2015). Moreover, there is considerable individual variation among people with dementia, with a continuum of decline from relatively intact learning capacity to increasingly reduced capacity in the moderate and severe phases of dementia. Even people with quite severe dementia who move to a nursing home will often learn to find their way around and to differentiate between the care staff they meet daily and modulate conversation and interaction in ways that reflect some limited capacity to adjust to new conditions (Müller & Mok, 2018).

# **Executive functions**

Executive functions constitute a foundation for the individual's learning, planning and performance of everyday tasks at home, in school and at work (Blair, 2016; Carlson, Zelazo, & Faja, 2013). These functions consist of several elements, including attention and working memory. They start to develop in preschool age and continue developing throughout childhood and adolescence, and some functions even into adulthood (Best, Miller, & Jones, 2009). Since executive functions are not automatized, they require more of the individual's cognitive resources than actions that can be carried out on "automatic pilot" (Diamond, 2013). It is common to distinguish between "hot" and "cold" functions. Cold executive functions include cognitive processes, while hot executive functions involve emotional and motivational factors. It is difficult for an individual to plan and make appropriate decisions when motivation and emotional involvement are too high, but also when they are too low.

### Executive functions and dementia

Decline in executive functions affects planning and organization of everyday tasks, and this is apparent in the early phase of dementia, maybe due to the reduced memory and attentional capacity, which affects the ability to reason, plan and perform actions and solve practical tasks in daily life (Kirova, Bays, & Lagalwar, 2015). In mild dementia among adults, the first signs of executive difficulties may be in handling money, washing clothes or making hot food. In moderate and severe dementia, there are increasing problems with carrying out personal activities in daily life such as getting dressed and undressed, washing oneself and eating (Giebel, Sutcliffe, & Challis, 2015).

The decline in executive functions makes it difficult for the individual to get an overview of the situation he or she is in, to distinguish between relevant and irrelevant information, and to understand complex or unexpected situations. Decline in executive functions may also imply reduced ability to suppress and control impulses, change perspective, and make sound judgements. It affects the individual's ability to learn new skills and establish an independent life.

#### **Dementia in JNCL**

Visual impairment is usually the first noticeable sign of JNCL, although there is also evidence that a decline in cognition and language may sometimes be apparent before the visual problems have become notable. The relationship between the two is complex, and cognitive decline is usually slow during childhood, thus making it difficult to pinpoint which comes first, and how the two are related (Laabs, 1988; Santavouri et al., 1988). So, although loss of vision is typically the first to be noticed, it gradually becomes clear that the disease is not restricted to vision. A lag time of approximately two years often separates the onset of visual impairment and the onset of cognitive decline, but in some cases decline proceeds in parallel (see below). There appears to be a relationship between visual and cognitive decline, in that both cognitive abilities and vision influence the ability to recognize places and orientate in the surroundings (Cameron, 1941).

Prior research shows that children with JNCL usually show typical cognitive development and participate in age-appropriate activities in kindergarten and the first school years, except for physical activities that require adequate vision (e.g., football or running). Lamminranta and associates (2001) found that from age six to ten years, the average verbal IQ on the Wechsler Intelligence Scales for Children (WISC) decreased from 88 to 72, but there were considerable individual differences. After the early school years, reports indicate that children with JNCL tend to have difficulties following lessons at school. However, the difficulties may be attributed to coping with the visual loss and therefore not perceived as a cognitive decline by people in the environment. Seizures are present in most individuals with JNCL, and EEG findings reflect slow deterioration in brain function as the illness progresses (Kirveskari, 2001). On the average, learning difficulties become apparent at the age of 8–9 years, epilepsy at 10–11 years, and speech and language problems at 12-13 years (Uvebrandt, 2006). Visual loss and speech impairment may make assessment difficult, and because cognitive assessment of children who are blind is typically based only on verbal scales, it may be difficult to distinguish cognitive abilities from language abilities.

In the present survey, some parents reported specific memory problems already when the child was 4–5 years old, but the average age when memory problems was first observed was around ten years. At 14 years, nearly 90 percent of the adolescents with JNCL had memory problems (Figure 5.2). Around ten years, they

also showed problems learning and remembering new things (Table 5.1). However, the parents reported an average age of just under eight years for problems with following mainstream education, suggesting that general cognitive problems were noticeable earlier than the specific memory problems. Functioning that is indicative of working memory problems is often apparent at an early phase of dementia (Kessels, Overbeek, & Bouman, 2015), and it is likely that it was problems with working memory that were affecting learning and school performance. However, the contribution of visual impairment to problems in following mainstream education should also be considered. Young students with JNCL must learn new behaviors to cope with their new situation (see Chapter 16), and they are challenged to do so at the same time as learning new skills is becoming more difficult. Moreover, mainstream educators usually have limited experience with teaching students who are blind. Also given the relevance of visual information to attention, particularly in young children, any decline in attention ability will add to the list of factors that challenge learning. Taken together, this situation helps explain how multiple variables compound to amplify the decline in function observed. Some of the low sub scores observed in intelligence test performance (IQ) will be due to lack of new learning (especially in the early phases), while other scores might reflect a loss of earlier acquired knowledge, or perhaps loss of ability to allocate appropriate attention in the absence of vision. This explanation is in line with other researchers who have reported that children with JNCL develop deficits in working memory which limit their ability to learn new things (van Delden, 2009). Working memory is assumed to be important for the acquisition of language and of reading



Figure 5.2 Percentage of individuals at each age from 4 to 18 years who are reported to (a) have problems with following a mainstream curriculum and (b) have problems with memory

	Mean age	SD	Range
Problems with memory	10.5	3.9	4-23
Problems learning and remembering new things	10.7	4.1	4-27
Cognitive decline compared to peers	9.1	3.0	4-25
Problems recalling planned events of no personal significance	13.0	5.6	5–30
Problems remembering planned events of great personal significance	16.6	4.2	6–24
Problems establishing interest in new things	14.4	4.9	6–28
Problems remembering what just happened	16.0	5.5	5–28
Problems remembering daily routines	15.3	4.8	7–30
Problems remembering names of important persons	14.9	5.5	6–27
Problems remembering and maintaining strong interests	17.2	5.2	6–27
Problems maintaining previous knowledge and memories	11.7	4.4	4–25

Table 5.1 Mean age, standard deviation and age range when problems became noticeable according to parents, among individuals who have shown such problems

and writing (Baddeley, 2003; Montgomery, Magimairaj, & O'Malley, 2008), and reduced working memory capacity has been suggested to make reading acquisition difficult for students with JNCL (Lou & Kristensen, 1973; but see Chapter 14). Studies have reported that on average, children with JNCL scored lower on a subtest measuring digit span than on other subscales of the WISC-R (Adams et al., 2007; Lamminranta et al., 2001). This interpretation is further supported by the fact that in the present study, cognitive decline compared with the development of peers was noted around the age of nine years.

In the present survey, however, there was considerable variation in the parents' reported age for emergence of problems with cognition and memory. The standard deviation for first problems with memory was nearly four years, which – assuming the age distribution followed the normal or Gaussian curve – means that around two-thirds of the group showed the first memory problems between age 6½ and 14½ years, and the lowest age was four and the highest 23 years. Table 5.1 shows that memory for events that had a strong significance for the individual (i.e., possibly high episodic content) on average was maintained longer (16.6 years) than memory for less important events (13.0 years). Similarly, problems maintaining previous knowledge and memories was on average observed at 11.7 years, while problems remembering and maintaining strong interests were observed at an average age of 17.2 years. This emphasizes the importance of finding educational material that is interesting and promotes participation in activities that the young person with JNCL finds engaging. Young people with JNCL may

to some extent enjoy repetition, but activities that are not intrinsically engaging may lead to lower activation of memory and less participation. Engagement thus has a dual function: increasing interest and mental activity while simultaneously preventing inattention and passivity.

Table 5.2 shows that the intercorrelations between different measures of memory were found to be in the moderate-to-high range (r = .43 - .96), suggesting that the individual dementia paths followed similar tracks, even if the decline started at somewhat different ages. The parents were asked to indicate the social impact of memory problems on a 5-point scale from no negative effect (1) to very high negative effect (5). An average score across ages of 3.1 (SD = 1.1) and a moderate correlation (r = .37, p<.01) suggest that the impact increased only slightly with age and the increasing degree of severity.

Last, accounting for the cognitive difficulties of dementia is crucial for the children's educational situation and needs. Dementia together with reduced vision can significantly weaken the possibility to understand and control the surroundings. Difficulties navigating the environment due to memory problems become more severe when the individual cannot make use of visual cues (Watkinson et al., 2014). However, there was no evidence prior to the present project indicating that

	1	2	3	4	5	6	7	8	9	10	11
1		.58	.72	.76	.54	.48	.58	.77	.43	.69	.57
2	.58		.63	.68	.58	.61	.62	.61	.56	.63	.48
3	.72	.63		.87	.75	.72	.63	.75	.58	.70	.71
4	.76	.68	.87		.74	.66	.70	.78	.56	.69	.64
5	.54	.58	.75	.74		.90	.82	.84	.81	.89	.84
6	.48	.61	.72	.66	.90		.86	.88	.69	.83	.74
7	.58	.62	.63	.70	.82	.86		.81	.70	.81	.84
8	.77	.61	.75	.78	.84	.88	.81		.69	.85	.86
9	.43	.56	.58	.56	.82	.69	.70	.69		.87	.84
10	.69	.63	.70	.69	.89	.83	.81	.85	.87		.96
11	.57	.48	.71	.64	.84	.74	.84	.86	.84	.96	

Table 5.2 Pearson correlations between age when different signs of memory problems first appeared (all correlations are significant, p<.01)

 First memory problems, 2) Cognitive decline compared with peers, 3) Problems maintaining earlier knowledge and memories, 4) Problems learning and remembering new things, 5) Problems remembering names of important persons, 6) Problems remembering strong interests, 7) Problems establishing new interests, 8) Problems planned recalling events of no personal significance, 9) Problems recalling planned events of great personal significance, 10) Problems remembering what just happened, 11) Problems remembering daily routines memory problems had been explicitly considered together with visual impairment by educational researchers, educational authorities, schools or JNCL counselors. Visual impairment and cognitive decline seem to have been approached as two separate phenomena. When adaptation does not consider the interaction effect of the visual impairment and cognitive decline, the result may be non-optimal physical and social environmental adaptations.

### Dementia and practice related to JNCL

The main consequence of dementia is reduced cognitive capacity, and research on dementia has been concerned mainly with deficits and decay. Less attention has been given to the two issues of how people with dementia experience and evaluate their own situation, and how people in the environment can support and contribute to maintenance of intact resources and abilities. Although people with dementia, by definition have a brain disease, they may still respond actively and adapt to the disease (Cheston & Bender, 1999). Research on dementia is thus not about persons who passively develop symptoms but rather about persons who try to cope with a gradually changing life situation and control their life as the disease progresses (Beattie et al., 2004; Johannessen & Möller, 2013).

The aim of this chapter is to provide information to support families and professionals. A clear knowledge of the typical onset and course of dementia in individuals with JNCL, as well as the variation that exists, can help in several important ways. First, it can provide parents and professionals with information that can identify the help and support that a child or young person with JNCL may need; second, it emphasizes not only that help is needed, but also that help can be utilized; last, it can contribute to create awareness of the individual's zone of *developmental maintenance* (see Chapter 2), within which support can accomplish the most. The concept of dementia, viewed as creating this zone of developmental maintenance, is a foundation throughout this book in seeking to understand and support individuals with JNCL and in discussing educational tools and strategies, as well as social relations and emotional reactions. To preserve identity, self-esteem and function from day to day, persons with JNCL and other types of dementia will try to maintain a balance between the desire to maintain status quo and to adapt to the changes that occur. The challenge for parents and professionals is to see the possibilities and opportunities, even when many cognitive functions are in the process of being reduced or have been lost. People with dementia become increasingly dependent on adaptation of the environment to meet their needs. It is important that parents and professionals who help and support individuals with dementia are attentive to the pace of the decline and to what kind of help the individual wants and needs, how he or she wants the help and when they want it (Clare, 2005). The ultimate aim will be for professionals and parents to promote maximum development and maintain use of abilities and resources throughout the course of the disease despite its constraints.

The changes in the brain of individuals with JNCL that lead to dementia constrain the possibilities for new learning and management of daily activities. As the disease progresses and the changes become more evident, the person's need for help and care increases. However, a singular focus on the brain disease may lead to overlooking possibilities that can promote growth and development, as well as possibilities for maintaining abilities and skills. There are powerful positive effects accruing to participating in activities that are of personal interest and to active engagement in motivating activities. Persons with dementia have residual abilities that should be respected: they can convey how they feel, they can share their own insights, they have resources to master, and they can benefit from other people listening, supporting and giving advice. In summary, they have important residual capabilities.

A JNCL diagnosis implies many challenges for the individual and the family. The focus will inevitably change from acquiring new competence to maintaining already established skills as the disease progresses (von Tetzchner, Fosse, & Elmerskog, 2013). It is a difficult but essential task to create an environment that is both stimulating and secure for individuals who have a reduced capacity not only for new learning, but also for coping with the new challenges posed by the ordinary daily activities (e.g., secondary to changes in functional vision). Providing appropriate support requires insight and interest in the individual's situation. Persons with dementia rely not only on receiving knowledge and help from others, but also on the helpers' commitment and engagement. Those who are responsible for providing support (e.g., organization and adaption of measures) are likely to find these responsibilities demanding, but at the same time both challenging and meaningful.

Childhood dementia constitutes a theoretical and practical framework for educational interventions where it is possible to consider windows of opportunity to meet the ongoing and anticipated declines and challenges. It is all about the foundation for providing interventions to promote the best possible quality of life for persons with JNCL. Children, adolescents and young adults with JNCL and other types of dementia disorders are in need of an adapted curriculum compared to other persons of same age. The zone of developmental maintenance (see Chapter 2) and the principles of selection, optimization and compensation (Baltes, 1987) are among the useful tools for practitioners who are working with children and young people with JNCL. The educational strategies described in several chapters in this book (hastened learning, precautionary learning, skill-based learning, participationbased learning and life flow) are based on knowledge about the typical course and variation in JNCL, the zone of developmental maintenance, and Baltes' model.

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# Language Development and Attrition in Juvenile Neuronal Ceroid Lipofuscinosis

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Language development may be defined as the process by which children come to comprehend and use linguistic forms and structures through interactions with more competent children and adults in a variety of social and cultural circumstances (von Tetzchner, 2019). Language distinguishes humans from other species, and is the basis for the child's entry into society. Human language consists of linguistic symbols that represent social conventions to draw attention to specific people, things, events and ideas, and a grammar, which allows the symbols to be arranged in patterns according to certain conventions – sentence structures – that create meaning beyond the individual symbols themselves (Tomasello, 2006). Language makes advanced communication possible about a range of topics for a variety of purposes, as well as reflection on the past and the future, creating continuity between past, present and future. Written language allows for the organization and storage of knowledge beyond the limits of each individual's memory (see Chapter 14).

Language has the same functions in children and young people with juvenile neuronal ceroid lipofuscinosis (JNCL), but their developmental course is unique in that it typically includes both language growth and attrition, and hence implies a need for measures that can support and maintain communication (see Chapter 13).

### Language comprehension and production

Language use implies both comprehension and production, and the two processes make different demands on the user. When inferring meaning from language produced by others, the child has to recognize the words' perceptual patterns (visual, auditory or tactile), the communicative intentions, and the social patterns of the situation. Language production requires a purpose or meaning and an ability to articulate, that is, mastery of the mechanics of speech or another mode of production. Expressive language may represent an initiative to communicate, relaying opinions, emotions, interests and stories, or a response to communicative initiatives from others, such as answering questions in a dialogue. Language comprehension and production may follow the same or somewhat different developmental courses (Bloom, 1974; Bates, Bretherton, & Snyder, 1988). Language attrition may affect both comprehension and expression, initiation and responding, but in different ways, at different times, and at different speeds. In typical development, children learn to master the speech sounds and phonemic contrasts of the language, as well as the syllable shapes and prosodic patterns. This developmental period will include errors observed from the onset of first words and into school age. Attrition of speech ability might appear in the form of persisting developmental errors (such as final consonant deletion of words or difficulty with particular sounds), or complete loss of contrast between sounds (the child produces different words that sound the same to the listener; e.g., go and *goat* both sound like *go*). Speech attrition can also affect the fluency, resulting in stuttered speech.

#### Language development in children with JNCL

There is a significant body of research on language development in children with congenital blindness (e.g., Fraiberg, 1977; Perèz-Pereira & Conti-Ramsden, 1999; Pijnacker, Vervloed, & Steenbergen, 2012; Tadic, Pring, & Dale, 2010). Despite the lack of visual information, children who are blind without additional impairments show a relatively normal language development, although many words may have a somewhat different conceptual content and usage than in sighted children. For example, "look" may not refer to vision but to experience in a more general sense (Landau & Gleitman, 1985). Nonetheless, language is the main source of information about social factors and the physical world, and early language skills are one of the strengths of many children who are blind. Moreover, their social interaction is mainly based on language use, and many children who are blind are rather talkative.

There is less research on language development among children who become blind in middle childhood. The onset of visual impairment for most children with JNCL occurs between 5 and 8 years of age (see Chapter 4). Their early language development therefore proceeds in a typical way, with no signs of the upcoming language problems. They use visual cues in the same way as other sighted children, and this is reflected in memory, conceptual structure and language use. This means that almost all children with JNCL are in a normal process of language development at the time when visual impairment and emerging dementia start to affect their language development and language attrition begins.

# Language attrition in JNCL

All individuals with JNCL will experience language attrition but both the age when language problems become noticeable and the nature of the problems vary considerably. In particular, it is important to distinguish between comprehension and expression, and in general, expressive language seems to be more affected and at an earlier age than language comprehension. The expressive problems make it difficult for the family, peers and others to understand what the person with JNCL is trying to communicate, and they may have to rely on contextual factors when interpreting what the person is saying. Familiar people will understand more than communication partners who do not know the person so well.

# Expressive language

Table 6.1 shows the age when decline became observable for core aspects of language attrition in the present study (see Appendix A). The average age when problems with speech became noticeable was 13 years and ranged from 2 to 25 years. Similar results have been reported in other studies (Bendixen, 1996; Kohlschütter, Laabs, & Albani, 1988). Among the 91 participants in the present study who were reported to have shown speech problems, six showed the first signs of language problems at the age of five or earlier, while five were 21 years or older when these symptoms first appeared. None of the speech problems were found in 100 percent of the participants. Fifteen children and young people had not developed unclear speech at the time of the surveys and interviews. For some this may be due to young age but the results also reflect the considerable differences in age of onset for the diverse speech problems. One parent described her daughter's development in this way:

Speech slowly left her. She went from being able to speak at will, to speaking when spoken to, to short phrases, to one word response as yes or no, to complete non-verbal (...) but she can still make noises if she is agitated or uncomfortable.

The decline of speech in young people with JNCL is complex. The pattern of speech problems differed considerably among the participants in the present study, and most had more than one speech problem. Articulation problems were common, leading to reduced speech intelligibility, that is, the partners' recognition of word forms. Eighty-three participants were reported to have developed speech that was imprecise but usually intelligible to unfamiliar people at an average of 14 years. Sixty-eight participants had developed speech that was unclear and not usually

Table 6.1 Number of individuals who show core expressive language and speech problems, and average age when observed (N=111)

Observed speech (S) and expressive language (EL) problems	n	Mean age of onset if occurred	SD	Range
First sign of speech problem	91	13.0	4.7	2–25
Stuttering (S)	47	10.8	4.3	4–20
Using wrong words (EL)	36	12.6	4.8	6–26
Word finding problems (EL)	83	12.7	4.8	4–25
Mumbling (S/EL)	54	13.9	4.2	5–24
Only short sentences (EL)	56	17.2	3.9	10-25
Limited vocabulary, same words repeated (S/EL)	48	18.2	4.1	10-27
Voice weak and difficult to hear (S)	24	20.6	4.3	13–26
Problems calling attention with speech (S)	38	18.2	4.1	10–30
Only single words (EL)	42	20.6	3.9	12-30
Imprecise but usually intelligible to unfamiliar people (S)	83	14.0	4.3	3–25
Unclear and not usually intelligible to unfamiliar people out of context (EL/S)	68	16.8	3.7	7–26
Speech no longer comprehensible (S/EL)	32	20.3	3.6	14–30
No production of speech (S/EL)	23	22.5	3.6	15-28

comprehensible to unfamiliar people outside a known context at the average age of onset of 16.8 years, while the average age for having developed unintelligible speech was 20.3 years. Twenty-three participants had stopped producing speech at an average age of 22.5 years. Some participants were unable to use speech to gain attention from others by an average age of 18.2 years, and in some participants, the voice had become very weak and utterances almost inaudible by 20.6 years. The communication partners' problems with comprehending unclear speech increase in noisy conditions. However, for all the symptoms, the age range was considerable. For example, age of onset of imprecise but usually intelligible speech ranged from 3 to 25 years, and age when speech had become incomprehensible ranged from 14 to 30 years. Several parents described this process:

*The pronunciation of many words become almost the same. Very often other people misunderstand.* 

The words simply don't come out, even if she keeps trying.

Similar observations were made by staff:

*His speech is very slurred. People who do not know him do not understand him. He repeats single important words until the counterpart guesses the correct word.* 

Together with imprecise but intelligible speech, problems with word finding were most frequent. Eighty-three participants showed this symptom at an average age of 12.7 years, which is a common feature in developmental language disorders (Best, 2005; Messer & Dockrell, 2006) and dementia (Bang, Spina, & Miller, 2015; Klima & Kuca, 2016; Rohrer et al., 2008). Use of wrong words was observed around the same age but this was much less frequent (36 participants). There are similar reports of idiosyncratic use of words without showing awareness of it, and odd constructions like «confirmation ingredients» and «world champion dancing dress» (Chaffey, 1987; Laabs, 1988; von Tetzchner, 2006). Idiosyncratic use of words make the person's expressive language difficult to understand.

Dysfluency (cluttering or stuttering) is considered a typical feature of language development in children and young people with JNCL (Gayton, 1982; von Tetzchner, 2006) and was observed early (average age 10.8 years) but only for 47 participants. Mumbling was also a relatively early feature (average age 13.9 years) for 57 participants, about the same proportion as for dysfluency. The intelligible expressive vocabulary may decrease and repetitions of the same words had become prominent by an average age of 18.2 years (table 6.1), as described by one parent:

#### Repetitious (has to start again many times). Often he is stuck with the first word.

Information from bereaved families, which covers the whole life span of 33 participants with JNCL, indicates that stuttering never occurred in 16 of these participants. The results indicate that speech dysfluency is quite common in young people with JNCL but also that about half of the people with the disease never develop dysfluency, although they have other speech problems. Similarly, only 19 (57%) of the bereaved parents told about complete loss of intelligible speech in their child at some point in time. This indicates that a considerable proportion of adolescents and adults with JNCL retain some intelligible speech throughout life, and that they at least to some degree can make themselves understood by their family and others who know them well.

Problems with sentence construction were observed in many participants, typically with onset in late adolescence. About half of the participants used only short sentences, observed by an average age of 17.2 years and about one third had begun to use mainly single-word utterances, observed by the age of 20.6 years.

There are very few detailed descriptions of the expressive language development and attrition in children and young people with JNCL. To our knowledge, there is only one linguistic study. Chaffey (1987) analyses the expressive language of her daughter Christine. The first signs of language attrition were problems with finding words, especially nouns. Around age 16 years, she started to stutter and often repeated what the other said, maybe reflecting problems with comprehension or word finding. Sentence structure was mainly maintained to 18 years, when Christine started to make errors in negation. She also began to make errors in sentences with complex syntax, simplified the sentences and used fewer syntactic and more analytic constructions. Her utterances involving negation comprised both syntactic and semantic relations, but they were arranged in unusual manners. When making a sentence, she seemed to start with a positive sentence (which initially is simpler) and then add a simple negation, such as All animals with fur she can stand not instead of «she cannot stand any animals with fur» (allergic) or Always I want not you leave instead of «I never want you to leave» (translated from Norwegian). Many of Christine's sentence constructions may also be found in the language of younger children, and are also typical of language attrition in general (Chaffey, 1987). Even when each word is intelligible, unusual syntactic constructions may make the meaning of the person's utterances difficult to understand.

The decline in expressive language constitutes a major problem when persons with JNCL try to convey meaningful contributions to the dialogue. Many children with JNCL are rather talkative, and may experience comfort from conversations about past experiences (van Delden, 2009). However, even when they speak a lot, partner comprehension may be limited:

*Speaks very much, knows and uses a lot of words, but is no longer understandable.* 

Children and young people with JNCL are not always aware of their own speech and expressive language problems and may struggle to understand why conversation partners do not give instant answers to their conversational initiatives, as described by parents and staff:

For a long time she thought that her speech was clear, that the others didn't hear well. It took a long time before she became aware that her speech wasn't that easy to understand.

She was not conscious of her speech problems.

The huge variety found in the present and other studies of expressive language in children and adults with JNCL emphasizes the need to adapt support and intervention to individual abilities and challenges in both early and later phases of the disease.

### Language comprehension and expression

Reduced language comprehension is a core element of dementia (see Chapter 5), and language comprehension will decline also in children and adults with JNCL. Childhood dementia may particularly affect comprehension of abstract concepts and complex procedures. However, when topics are concrete and related to familiar everyday issues and the person's own experiences and personal interests, information from parents and professionals suggest that comprehension may be preserved even when intelligible speech is lost:

*She understood language well at the age of 22 even though she could hardly express herself.* 

We always felt that she continued to understand a great deal even though she was unable to speak back to us.

Although language comprehension is affected by the emerging dementia, it is much better retained than speech production. In fact, comprehension of vocabulary may continue to increase even after cognitive functions have started to decline. Hearing acuity is usually not affected by the disease, but comments in the present study indicate that some young people with JNCL experience problems in processing auditory information or might become hypersensitive to sounds and find noisy environments disturbing and unpleasant. Still, hearing and language comprehension are their most important resources for understanding their environment and participating in it.

In the present study (Appendix A), parents were asked to compare their child's language comprehension in relation to expression both at the time of the survey and retrospectively from age seven years. The survey included factors that relate to expressive language, speech, or skills that could reflect a combination of these competencies. Figure 6.1 shows that comprehension and expression initially were considered equal and that comprehension gradually became relatively better than expression with age. At the age of 7 years, comprehension and expression were considered equal in 82.5 percent of the participants. By the age of 22, only 6.7 percent of the participants were considered having equal comprehension and expression and express



Figure 6.1 Comprehension of speech in relation to expression (percent of responses)

seemed to retain an ability to speak but had problems with comprehension and probably also expressive content.

In the questionnaires and interviews, parents and professionals described comprehension as better than the expressive ability.

*Language comprehension was not his major problem. Language production was getting difficult when he was getting older.* 

*She listened to audio books and her comprehension was good until around age 14–15. Then she started to regress, preferring younger books and more repetition.* 

The results thus support observations from clinical experience that the decline in speech production and expressive content is more pervasive than comprehension of spoken language (Elmerskog & Fosse, 2012; Gombault, 2010).

Both language comprehension and production are influenced by familiarity with the situation. Persons with severe visual impairment depend on non-visual contextual cues, such as what other people say and sounds from ongoing activities in the environment but will miss cues that require vision. The cognitive decline may lead to fragmented experiences, which in turn make language comprehension fragmented and lacking in coherence (von Tetzchner, 2006). For young people with JNCL, being in a familiar environment will enhance comprehension and facilitate communication. As the disease progresses, their comprehension becomes increasingly dependent on being in a situation that is familiar and does not contain too many new elements or the kinds of noise that the individual finds distressing because of susceptibility to sensory overload. An adapted school environment that is stable and predictable is therefore important also for communication. In adulthood, the same applies to activity centers and work places.

The gap between comprehension and expressive spoken language communication also suggests that young people with JNCL may benefit from augmentative and alternative communication (AAC) intervention (see Chapter 13).

### **Observations and assessments**

Both formal and informal assessment of individuals with severe visual impairment and dementia may be challenging and require adapted material and situations (see Chapter 10).

Language is important for all aspects of communication and daily life. When language is impaired, the promotion of communication and language is always an important target in the curriculum. Assessment of comprehension and use of language are therefore important elements of educational practice. There are various tests and checklists for evaluating language competence, both comprehension and production (e.g., Goldman & Fristoe, 2015). Many of the tests are based on picture recognition or naming and therefore are not suited for children and young people with visual impairment (Eirin & Koenig, 1997). When children's vision precludes use of such tests, alternate assessments can be obtained through use of inventories of spoken words drawn from a sample of speech in which the child is actively engaged in activities that include known words (toys, actions, people, etc.), and from parent diaries of spoken word attempts of known words (Bauman-Waengler, 2016; Velleman, 2016). For example, use of scripted activities helps assure a known context for children with highly unintelligible speech (Lund & Duchan, 1993).

Receptive language is difficult to evaluate and assess (Bishop, 2006). Tadic and associates (2010) used different tests of expressive and receptive language (e.g., CELF-3, CCC-2) with a group of children with congenital blindness, and found that a few subtests could be used for children with no vision, but also that these test results would have reduced validity.

Receptive language skills may to some extent be inferred from responses to questions or how instructions are followed, but these are typically based on spoken answers, visual discrimination or motor performance. In such observations, it may be difficult to distinguish between receptive and expressive language competence, as well as motor performance (Graven, 2018).

Expressive language is also difficult to assess formally in individuals with JNCL, but speech and problems related to expressive language are easier to observe and describe than comprehension. The different aspects of language decline in JNCL follow a recognizable course, although there are individual differences concerning time and speed of symptom occurrence. An alternative to formal assessments is to observe a young person with JNCL in a range of settings and record their language use and their responses during observations. This can be done for example annually or semiannually with standard procedures in situations that are familiar to the person.

Observations of receptive language require interpretation and analysis. It is necessary to record all attempts to elicit understanding or response, as well as words and other communicative expressions of the individual. It may be useful to observe interaction with several people, both familiar and unfamiliar partners, as the quality of the interactions may reflect the extent of the partner's personal knowledge and insight into the communicative means of the person. Registration and discussion of all cues and expressions are important also to avoid attributing intentions and attitudes that the person with JNCL does not have (Grove, Bunning, Porter, & Olsson, 1999).

#### *Educational Development Observation (EDO)*

EDO is an observation checklist for assessing educational needs and possibilities developed for use with children and young people with JNCL. Table 6.2 shows the language and communication part (see Chapter 10 for more information about the EDO). Use of the EDO for evaluation of people with JNCL should include input from a professional with comprehensive knowledge about both JNCL and EDO. In communication and other domains it is an aim to start intervention before symptoms develop, because this may allow the child to develop skills that he or she will need but not will be able to develop in later phases of the disease. The principles of hastened and precautionary learning are important when learning for the future (see Chapter 12). A professional with expertise in JNCL will have knowledge about future declines in communication and language and can guide parents and professionals in preparing the child for these declines in best possible way. The checklist should not be used solely for mapping impairment or symptoms. Rather, the intention is to guide the support team to identify the best possible interventions to compensate and optimize the individual's social participation and quality of life.

Table 6.2 The communication and language part of Educational Development Observation Tool(EDO)

#### C1. Speech intelligibility (mark one alternative)

- 1) Speech is understood by all, special attention from others is not required
- Someone who knows the person well may not need to ask for repetition/clarification but someone who does not know the person well, may sometimes have to ask for clarification of a few words
- 3) Speech is unclear and not usually understandable to unfamiliar listeners out of context
- Someone who knows the person well will understand simple, single words only (e.g., yes/no, etc.).
- 5) The person's speech cannot be understood by anyone

#### C2. Speech (mark all that apply)

- 1) Speech is normal (note, if speech is normal, no other options should be marked)
- 2) The person stutters
- 3) The person mumbles/has poor articulation
- 4) The person has problems finding the right words
- 5) The person uses wrong words when trying to express something
- 6) The person is only able to express short and simple sentences (3-6 words)
- 7) The person is only able to express single words
- 8) The person's voice is weak and difficult to hear
- 9) The person has very limited vocabulary, uses the same words again and again
- 10) The person has problems with calling on other people's attention by speech
- 11) Other, please describe:

# **C3. Expressive communication modes: What expressive communication modes are used?** (mark one alternative)

- 1) Mostly oral communication
- 2) A combination of oral communication and signs/gestures/body language
- 3) Mostly communication via signs/gestures/body language
- 4) Mostly communication via technical/electronic aids or/and objects of reference
- 5) The person has no or almost no expressive communication

# **C4. Expressive communication: When using expressive communication alternatives, is communication understandable?** (mark one alternative)

- 1) Irrelevant, no need for communication alternatives
- 2) Communication alternatives are imprecise but usually understandable even for unfamiliar recipients
- Communication alternatives are unclear and not usually understandable to unfamiliar recipients out of context
- 4) Communication alternatives are unintelligible even to those who know the person well
- 5) No communication alternatives are used despite the need

C5. Language: Language comprehension versus language production/expressive communication (mark one alternative)

- Language comprehension is the same as language production (normal). The person shows an equal correspondence between what the person is able to express (for instance by talking, signing, body language, using technical aids etc.) and the person's comprehension of what other people are expressing
- 2) Language comprehension is slightly better than language production. The person is from time to time showing that he/she understands more than she/he is able to express. The difference is evident but does not have a major impact on everyday living
- 3) Language comprehension is better than language production. The person is on a regular basis showing that he/she understands more than he/she is able to express. The difference has an impact on everyday living.
- 4) Language comprehension is evident, but there is no expressive communication. The person has no or almost no expressive communication but it is evident that the person from time to time understands what others are expressing
- 5) There is no expressive communication and it is difficult to assess if there is any language comprehension. The person has no or almost no expressive communication and there are no or almost no signs that the person understands what others are expressing.

**C6.** Expressive communication extension. Estimate the number of words, gestures, signs, objects of reference etc. that are actively used by the person (mark one alternative)

- 1) Normal for age
- 2) Above 100 signs, words, objects of reference etc. but not normal for age
- 3) Between 21 to 100 gestures, signs, words, objects of reference etc.
- 4) Between 1 to 20 gestures, signs, words, objects of reference etc.
- 5) No or almost no expressive communication

After having filled in the information above, discuss and describe the consequences for education and non-medical service provision, and recommendations for interventions.

### Consequences of language loss

For persons with severe visual impairment, social interaction is mainly based on language use. Although individuals with JNCL develop severe language problems, language continue to be an important tool for social participation and well-being. Comments in the present study indicate that the need for and wish to be in interaction and dialogue with other people are not reduced by the disease. Young people with JNCL may become frustrated by not having the ability to participate socially in the same way as they used to. Their communicative initiations are often frequent despite pervasive speech problems. The loss of speech and reduction in comprehension cause significant frustration and stress in both the young person with JNCL and in the family (von Tetzchner, Fosse, & Elmerskog, 2013). Persons who have problems with communication and language – irrespective of diagnosis – often experience misunderstandings and frustration (von Tetzchner & Martinsen, 2000). People with JNCL experience the additional strain of losing skills that they earlier mastered with ease.

During the course of the disease, people with JNCL become increasingly dependent on support. Many will need assistance from others to communicate successfully and participate in social activities. Conversations become more tiresome and frustrating, both for the person with JNCL and the communication partners. These difficulties will affect the person's social life and total life situation and make spontaneous communication with peers and others difficult. The problems with language and social interaction may create frustration and elicit despair, anger and other emotional reactions (see Chapter 27). They want to communicate as much as before but do not have the ability to participate in the same way as they used to.

When the parents in the present study were asked to evaluate the extent of the negative social consequences of speech problems on a scale from 1 (no negative effect) to 5 (high negative effect) (see Appendix A), 43 percent of the parents answered that speech problems had a high or very high negative effect on social life. No parents answered that the problems with speech did not have any negative effect on social life.

The central role of language and communication in all aspects of life indicates that research should give priority to developing interventions that may compensate for the deterioration of language by strengthening or at least supporting the existing communication skills for as long as possible.

Any discussion of intervention must address the need to choose and implement measures in advance of symptom development to make the best possible foundation for meeting future declines. Of necessity, when symptoms are evident – one must also decide what to do next. The goal is to prolong the period with fluent communication for the person with JNCL. The EDO can potentially be a helpful tool when making decisions for today and for the future.

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## Motor Development and Loss in Individuals with Juvenile Neuronal Ceroid Lipofuscinosis

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Physical actions are voluntary movements with intentions and goals, which allow humans to overcome gravitational forces, to plan, coordinate, perform and evaluate actions, and to create new physical and social opportunities for action. The ability to locomote is a core element of human adaptation and motor skills develop through maturation and practice (Hadders-Algra, 2018; Piaget, 1983). Individual differences may originate in biological differences as well as environmental and cultural factors (Adolph & Robinson, 2015; Houwen, Visscher, Lemmink, & Hartman, 2009). Children and young people with juvenile neuronal ceroid lipofuscinosis (JNCL) show gradual changes in motor abilities and activity. These changes appear to be related to motor decline caused by the disease but also to visual impairment and emergent dementia.

#### Motor functions and impairment

*Gross motor skills* involve the large muscle groups, such as body posture and walking, balancing, climbing, swimming and crawling. Independent locomotion is important for action and participation. Gross motor skills enable exploration of objects, people and places, and thus influence the whole psychology of the child (Anderson, 2018; Anderson et al., 2014; Campos et al., 2000). The basic gross motor skills develop during early childhood but the quality of movements and motor coordination continue to improve into young adulthood. When gross motor skills are established, they are usually maintained even after longer periods of non-use. For example, walking and biking do not require much practice once they have been learned and mastered (Galahue & Ozmun, 2006; Stallings, 1973).

*Fine motor skills* involve the use of smaller muscle groups and make it possible to manually handle and explore things. They include movements of the wrists, hands and fingers, for example for pointing, gripping, reading braille or Moon,

playing the piano, dressing and painting. Many fine motor skills are present in early childhood but fine motor skills continue to develop into young adulthood. Fine motor skills require more exercise than gross motor skills to be maintained, they will to a larger extent be lost if they are not exercised. For example, playing the piano on a high level requires continuous exercises (Stallings, 1973).

Physical activity usually has associated health benefits. Gross motor activities are influenced by the individual's physical condition and health, the strength and endurance of muscles and the uptake of oxygen. They support health because they require substantial energy, while fine motor activities do not confer the same health benefits. One's physical condition and motor abilities may mirror one's lifestyle, reflecting whether muscles are exercised on a regular basis (Stallings, 1973; Tremblay, Colley, Saunders, Healy, & Olsen, 2010; Van Duyn et al., 2007).

*Physical impairment* may be caused by a bodily abnormality or a neurological condition that causes partial or total loss of the function of one or more body parts, such as movements of the limbs. Physical impairment can for example cause muscle weakness, poor endurance, lack of muscle control, reduced co-ordination, or total paralysis. Physical impairments are evident in neurological conditions such as cerebral palsy, muscular dystrophy, Parkinson disease and JNCL. An extreme form of physical impairment is the locked-in syndrome, where the individual's control of muscles is lost while cognitive functions are not affected by the disease (Haig, Katz, & Saghal, 1987). Developmental coordination disorder (dyspraxia) is a neurological condition that affects the person's ability to plan and coordinate physical movements, balance, posture and motor actions, but does not involve paralysis or motor impairment in the conventional sense (Gibbs, Appleton, & Appleton, 2007).

A loss or decline of motor function can consequently be influenced by bodily and neurological motor impairment, while reduced practice will impede learning, development or maintenance of motor skills. A sedentary lifestyle will contribute to reduced motor skills. To promote the best possible motor function, education should focus on skills in the zone of proximal development (i.e., those that a child is on the way to master) or, if the child is showing regression, on skills within the zone of developmental maintenance (i.e., skills that can be maintained with adaptation and help) (see Chapter 15). This focus will also support the best possible health in children, adolescents and adults with JNCL.

Being physically active does not come easily when children and young people have severe motor impairments. People with physical impairments are often not sufficiently supported – or even hindered – in participating in physical activities, due to lack of environmental adaptations and individual support (Gibbs, Brown, & Muir, 2008). Severe motor disabilities make performance of aerobic exercises difficult, that is, exercises that make one perspire and breathe harder, and the heart to beat faster (Læssøe, 2011). Even moderate impairments tend to imply a sedentary lifestyle and limited social and societal participation, which again affect the person's physical condition and health. Severe motor impairments may thus have a negative effect on development in general, health and participation.

Physical barriers in the environment may represent significant hindrances to being physically active for people using wheelchairs, but also for people with coordination or balance problems. Steps, curbs or a rough ground make independent movement not just difficult, but aversive if falls occur, and should be removed or adapted when possible and required. Physical play and sport activities, such as climbing or playing soccer, can contribute positively to motor development. Such activities are usually not accessible for children with severe motor disorders (Gibbs et al., 2008).

Minimal use of some gross motor movements may in addition have a negative effect on the joints and the tendons attached to the muscles. A joint that is not exercised or maintained over time will become stiffer and the range of movement in the joint will become reduced or even arrested. Contractures (permanent shortening of muscles, tendons, or ligaments) may entail pain when performing daily activities such as dressing or change of position in bed (Jeremiassen, 2016).

### Decline of motor function and activities in individuals with JNCL

Most children with JNCL have typical motor development in the early years. At the time of diagnosis, children with JNCL can walk, jump, and run normally, and they do not have any problems with fine motor skills. However, with age, abnormalities in gait and slight reductions in voluntary coordination of muscle movements and motor functions controlling speech become noticeable (Østergaard, 2016) (see Chapter 6). Some children with JNCL develop an extrapyramidal motor disorder that resembles Parkinson disease around the age of 10 to 12 years (Järvelä, 1997), with hypokinesia (reduced bodily movement), rigidity of muscles and joints, shuffling gait, reduced balance and a stooped posture (see Figure 3.3 in Chapter 3). In a Finnish study of 53 persons with JNCL, the average age at the first sign of such Parkinsonian type of walking was 13.7 years (Santavouri, Heiskala, Westermark, Saino, & Moren, 1988). Some of the symptoms can sometimes be temporarily ameliorated with anti-Parkinson medication (Åberg, Rinne, Rajantie, & Santavuori, 2001).

JNCL affects both gross and fine motor activities. In the present study (Appendix A), parents were asked when they first observed problems with gait and use of the hands and arms. Figure 7.1 shows that for most participants,

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Figure 7.1 Percentage of children who first showed problems with gait or hands and arms at the different age intervals

gross motor problems appeared earlier than fine motor problems. Gross motor problems often appeared in early adolescence, gait problems were first observed at an average age of 14.1 years, while problems related to the use of the hands and arms typically appeared in the late teens (average 15.5 years).

The later stages of the disease may include increased rigidity, slowness of movement such as slow steps with flexion in hips and knees, a shuffling gait with shortened steps, reduced arm swings and a forward-flexed posture. Involuntary movements are also reported. Many persons with JNCL need a wheelchair in late adolescence or early adulthood but there is considerable variation in the group (Østergaard, 2016, 2018). Not all persons with JNCL lose the ability to walk. A Finnish study found an average age of 17.3 years for losing the ability to walk (Järvelä, Autti, Lamminranta, Åberg, Raininko, & Santavuori, 1997). A Danish study found that females became dependent on a wheelchair at the average age of 17 years (range 13.8–20.6), while males became dependent on a wheelchair at 20.2 years (range 14.3–22.1) (Nielsen & Østergaard, 2013). Motor dysfunctions include chewing and swallowing difficulties and may be observed from the late teens. To ensure adequate intake of nourishment, feeding with a gastric tube may be required (Østergaard, 2016).

The results indicate that problems with upholding a healthy lifestyle due to motor impairment are likely to emerge around 11 to 13 years in young people with JNCL, unless met by appropriate measures. In the present study, a few individuals with JNCL were able to walk long distances around the age of 25 years. They showed no evidence of extrapyramidal motor disorder, abnormalities in rigidity, or chewing and swallowing difficulties, and also had no major difficulties in the performance of fine motor skills. The variation in the onset of decline in fine and gross motor skills thus seems to be considerable in individuals with JNCL.

#### Visual impairment

Visual impairment will usually influence participation in motor activities, and severe visual impairment may entail a sedentary life style. Sight is important for exploring the world from a distance and visual observations motivate exploration of places and objects and participation in physical activities like moving around, participating in sports, and for stimulating us to learn and refine motor skills, for instance by observing others (Fraiberg, 1977). For instance, children who are blind may not be aware of a red ball on the floor and thus not start playing with it or experience the thrill that comes from noticing a tree in the garden that invites climbing. Lacking incentives to explore the surroundings are likely to have negative influences on motor activities and development in children who are blind.

Children who are blind move less to explore, and their motor skills develop later due to a lack of visual incentives for movement (Freedman & Cannady, 1971). Moreover, in children with congenital or very early-acquired blindness there are significant structural changes in the brain, which may be beneficial for attention and acting, but they also imply some maladaptation to the sensory information that is accessible (Singh, Phillips, Merabet, & Sinha, 2018). This does not happen to the same extent when visual impairment starts later. There are thus significant differences in neurological and adaptive resources between the developmental courses of individuals with congenital or early onset of blindness, and individuals who lose their sight in middle childhood or later.

Individuals with JNCL, who start to lose sight during preschool years and become blind in middle childhood, have mainly used visual cues for orienting and continue to do so after the visual problems appear. Yet, they may underperform and function more like sighted people who are blindfolded (Schinazi, Thrash, & Chebat, 2016; see Chapter 16). The link between loss of vision and loss of motor skills needs to be considered. Children and young people with JNCL can no longer rely on the perceptual cues that they used earlier in life. Even if met with appropriate measures, they may also start to lose some motor skills due to lack of incentive as well as increased insecurity.

Students who are blind need more cognitive resources and learning to orientate themselves than do sighted peers (see Chapter 16). A Norwegian study found that cognitive-based orientation was difficult for blind children before the motor actions were automatized, and concludes that problems in orientation may have a negative

influence on motor performance in this group (Tellevik, Storliløkken, Martinsen, & Elmerskog, 2007). Performing daily activities demands more energy for people who are blind and the need to be fit might even be greater (Buell, 1984). Poor posture due to a sedentary lifestyle is relatively common among blind children and persons with severe visual impairment with no other disabilities they tend to show a lower level of fitness than sighted peers. They have a higher rate of obesity, higher degree of muscle weakness and lower cardiovascular endurance than peers of same age (Aslan, Calik, & Kitis, 2012; Lieberman & McHugh, 2001; Weil, Wachterman, McCarthy, Davis, O'Day, Iezzoni, & Wee, 2002). In addition, children with severe visual impairments are often not expected to pursue a full range of life goals (Lieberman & McHugh, 2001). The lack of expectations among people in the environment may also lead to reduced motor activity and furthermore to obesity and other health risk factors (Weil et al., 2002).

Children who are blind are more dependent on adult helpers than their peers who are sighted. This reliance in turn may create another barrier for children's engagement in motor activities; adults' preferences and priorities do not always align with those of children. The adult world is formed by preplanned schedules, movements are done with a high degree of time-efficiency, and adults will in many cases avoid situations where they become dirty and wet, which frequently characterize the explorative activities that usually are stimulating for children and young people. Children who are blind and their adult guides will often move in accordance with the adult culture, taking the shortest and most efficient way between two locations. However, this need for time efficiency also reduces the total amount of physical activity for everyone (Elmerskog & Fosse, 2013).

The difference in movement and physical activity between children who are blind and children who are sighted may be considerable, affecting the long-term physical status, gross motor development, and the personal development of children who are blind (Houwen et al., 2009; Elmerskog et al., 1993; Fraiberg, 1977). There is a significant risk that early blindness may lead to a sedentary lifestyle. Such a lifestyle should be prevented to support physical and mental health, through appropriate adaptation of physical activities, guidance and support (Chapala et al., 2017).

#### Effect of motor and visual impairments on activities

Children and adolescents with severe visual impairment are often encouraged to focus on activities requiring fine motor skills (Cheadle, 1994) which make small demands on orientation ability (see chapter 16). Many individuals with congenital blindness become good at playing the piano, reading braille or dressing, which mainly require the use of hands and arms.



Figure 7.2 Percentage of gross motor and sitting activities as main activities at different age intervals

In the present study (Appendix A), the parents were asked what kind of activities their children were mostly occupied with at different age spans. Figure 7.2 shows that the relative participation in gross motor activities decreased gradually from age seven to twenty-two years compared to sitting activities. There is no doubt that the reduced physical activity and increased passivity in early life are related to the visual impairment but also other factors, such as needs for increased



Figure 7.3 Percentage of participants with onset of visual impairment of major impact and onset of gross motor problems at different age levels

support and adaptations in the environment. Figure 7.3 shows the age when the visual impairment had a major impact on the participants' functioning and the age of onset of gross motor problems. The visual decline had begun to have a major impact on everyday life at an average age of 7.2 years among the participants, while total blindness occurred around the age of 10.7 years (see Chapter 4). Onset of gross motor decline appeared significantly later, around 14.2 years on average. There are reasons to believe that the early decreases in participation in gross motor activities among individuals with JNCL have other causes than the later declines in motor skills.

The participants in the present study were on average affected by a severe visual impairment between age 6 and 9 years (see Chapter 4). The initial physical inactivity thus seems to be related to visual decline rather than loss of motor functions. Visual impairment affects agility, speed, balance, orientation, independence and so forth. The participants stopped taking part in gross motor activities like playing soccer, climbing trees and playing involving gross motor activities that children between the age of 7 and 13 years participate in are heavily dependent on vision, and these are difficult to perform for children with JNCL. The reduction in participation in gross motor activities shown in Figure 7.2 reflects a change in lifestyle forced by the visual impairment. The following quotation from a parent illustrates this:

Our daughter of 11 years of age stopped doing physical activities when she became blind three years ago. She was more or less exempted from doing P.E. with others at school, there were no alternatives for her to participate in sport activities in our town. She prefers doing smooth activities, like singing, listening to music or audio books or playing with her dolls (she also loves activities like drawing). She became physically inactive, it is difficult for us to encourage her to do physical activities, partly because she cannot share such activities with her sighted friends.

Children with JNCL require adaptations of accessible gross motor activities and the environment, and support from adults. Schools have an important role in promoting and supporting a physically active lifestyle in these students. In the present study, many of the participants first attended mainstream schools that had limited experience with students who have visual impairment and little insight into how this disability may affect the students' daily living (see Chapter 9). Many schools make good efforts to promote the students' academic performance but the comments in the surveys and interviews did not indicate that the schools were initiating measures to prevent or meet the students' growing sedentary lifestyle, for example with proactive, precautionary or enhanced teaching (see Chapters 10 and 12). For instance, almost none of the parents and staff reported that extra resources were used at school for adapted physical education in early school age.

There are thus reasons to believe that the early decline in participation in gross motor activities often reflects a life-style problem related to the visual problems. This emphasizes the importance of distinguishing between an impairment and the developmental consequences of the impairment, consequences which are influenced by a range of other factors (von Tetzchner, 2019). The motor impairment (and possibly other disorders) that emerges as JNCL progresses increases the decline in participation in activities involving gross motor skills. The results in Figure 7.3 indicate that promoting a best possible motor development and maintenance plan for children and young people with JNCL requires a multidisciplinary approach, in particular between specialists with competence in visual impairment and motor impairment.

# Physical activity, cognition and childhood dementia

There is a general positive association between an active physical lifestyle and cognitive functioning. Physical activity contributes positively to maintain functions in elderly people with mild and moderate dementia (see Chapter 5). There is therefore interest in the potential impact that interventions leading to improved fitness and exercise might have on the maintenance of cognitive function and learning in persons with JNCL.

#### Physical activity and cognition

A number of studies have observed a positive influence of physical exercise on cognitive functioning and academic performance in both children and adults without dementia (Donnelly et al., 2016; Sofi et al., 2011). For example, a sedentary lifestyle was linked to poorer reading skills in the first three school years in 6 to 8 years old boys according to a study from Finland (Haapala et al., 2017). Results from other studies indicate that higher levels of activity or fitness enhance thinking, concentration and academic performance, and students with good fitness obtained higher scores on standardized tests of cognition and brought home better report cards (Coe, Pivarnik, Womack, Reeves, & Malina, 2006; Coe, Peterson, Blair, Schutten, & Peddie, 2013). Coe and associates conclude that there is enough evidence to support a greater provision of physical activity into the school curriculum. Other studies have found that young adults who run or

participate in other aerobic activities maintain memory and other cognitive skills better in middle age. For instance, one study found that young adults (on average 25 years old) with a physically active life style, who were running or participating in aerobic activities, showed better cognitive functioning about 25 years later (age 43 to 55 years) (Zhu et al., 2014).

The assumption is that physical activity influences the brain (Zhu et al., 2014). One explanation is that moderately intense exercise can increase the size of the hippocampus, an area of the brain involved in learning and memory, which in turn leads to better thinking and problem-solving, more focused attention and improved learning. It is the endorphins the brain releases during exercises that help children to improve mood, energy levels and even sleep. It is argued that physical activity and exercise are significant for children's health, wellbeing and development, and that physical activities should always be considered when promoting children's development, in particular in children with illnesses and disabilities.

An active physical life thus seems to promote learning and cognition in children and adults, although many factors influence cognition and learning (Wang, Xu, & Pei, 2012), and the statistical effects of physical activity are small or moderate (Donnelly et al., 2016; Keeley & Fox, 2009; Singh, Uijtdewilligen, Twisk, van Mechelen, & Chinapaw, 2012; Tomporowski, Davis, Miller, & Naglieri, 2008).

#### Physical activity and dementia

Physical activity seems to both reduce the risk for dementia and contribute to maintaining cognitive functioning in elderly people with mild and moderate dementia (Aarsland et al., 2010; Sofi et al., 2011). A Finnish twin study found that physical activity in midlife (age 24–60) seemed to decrease the risk of dementia in old age, 29 years later. The participants who were engaged in leisure-time physical activity at least twice per week had a lower risk of dementia than individuals who were less active (Iso-Markku et al., 2016). Other studies present similar findings, that adults with an active physical lifestyle have lower risk for Alzheimer disease and other forms of dementia than less physically active persons (Laurin, Verreault, Lindsay, MacPherson, & Rockwood, 2001; Rovio et al., 2005).

Most studies have compared people with different life styles across the lifespan but there is also some evidence that intervention with physical activity in elderly people may be beneficial. In one study, elderly people who had memory problems and were at risk for Alzheimer disease were allocated to an intervention group who did 20 minutes of extra physical exercise every day or a control group who did not do any extra physical activity. After six months, there was a small improvement in performance on cognitive tasks in the intervention group compared to the control group, and the difference was maintained 12 months after

the experimental intervention was stopped (Lautenschlager et al., 2008). There is thus evidence that increasing physical activity also later in life reduces the risk for dementia (Lautenschlager, Cox, & Cyarto, 2012).

### Dementia, physical activity and JNCL

The studies cited above indicate that physical activities to some extent can prevent cognitive decline and dementia among elderly and middle-aged people. Childhood dementia is a core characteristic of JNCL (see Chapter 5) but a literature search for research about childhood dementia and physical activities yielded no results, probably due to the low prevalence of such diseases. Professionals and lay people have rarely heard about JNCL and childhood dementia, and this seems to be the case even for professionals in special education and social services. Moreover, the relevance of research evidence from elderly adults has not yet been acknowledged by the educational and social services that are responsible for children, adolescents and young adults with JNCL. However, it is likely that physical activity and an active lifestyle will have similar positive effects on cognition, learning and wellbeing in persons with childhood dementia, including persons with JNCL. The following quotation supports such an assumption:

My son with JNCL is 16 years old. He lived a rather inactive life until he was 14. We noticed this was not good for him. His physical condition was low, it was difficult to engage him in activities, he showed low spirits close to depression, and his behavior changed from day to day. It was not easy to deal with him. We decided to involve him in physical activities on a daily basis. After some struggle in the beginning he learned to love it. Today we do physical exercises one hour per day. He asks for the activities, his mood has improved and is more stable than before, and he is actually doing better at school.

Even if this is the experience of just one young person and his family, it seems important to follow it up in research and practice.

### Conclusions

Both visual impairment and motor impairment will make participation in a healthy lifestyle more difficult. Individuals with JNCL typically become visually impaired in the early school years, while gross motor problems become observable in early

teens and fine motor problems in the later teens. However, there are considerable differences in how and when impairments appear in individuals with JNCL. The disease causes loss of motor skills in individuals with JNCL, but motor performance is also influenced by lifestyle and factors in the environment. Education cannot heal or stop the progress of JNCL but can certainly affect the individual's lifestyle and participation. On the basis of research with children and adults with typical development, as well as adults with mild and moderate dementia, and comments in the present study, it seems justified to suggest that a physically active lifestyle should be a mandatory goal and integrated into Individual Education Plans and Habilitation Plans for all individuals with JNCL, from childhood to adulthood. Promoting a healthy lifestyle entails a focus on compensation and should proceed in accordance with the principles of proactive, precautionary and enhanced learning and stimulation (see Chapters 11 and 12).

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## Ethical Issues Related to Working with Individuals with Juvenile Neuronal Ceroid Lipofuscinosis

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E thical issues related to people with disabilities have received growing attention in many professions, especially issues related to life quality and rights to intervention and compensatory measures to create equal opportunities in spite of differences in abilities (Blackmer, 2000; Leahy & Szymanski, 1995). Most countries have laws that give children with disabilities equal rights to receive appropriate education. However, children and young people with disabilities are still at-risk for being left out of social and societal participation. It is an ethical prerogative for professionals to prevent or remedy such situations, and the ethical issues here are therefore about the beliefs and actions of professionals.

Most professions experience ethical dilemmas, such as lack of competence or inappropriate allocation of resources, and situations where there are contradictory ethical reasons for taking conflicting and incompatible courses of action (Howe & Boele, 2018; Knauss, 2001; Shapira-Lishchinsky, 2011). Applied professional ethics refers to a broad system of principles of conduct that guide and regulate the actions of people in a professional role, which are usually taught as part of the professional education (Jacobs & Hartshorne, 2003). Professionals facing ethical dilemmas have three main guideposts to help choose between right and wrong: laws, professional codes of ethics, and personal values and beliefs (Darden, 2014). The professions' national and international ethical guidelines are designed to help them in such situations (Clark, 2012; Clemente, Espinosa, & Urra, 2011; Conley, 2013; Gauthier & Petifor, 2014; Glosoff & Cottone, 2005; Leach & Leong, 2010; McNamara, 2011). However, ethics go beyond the regulations of the ethical guidelines; in fact, the spirit of the guidelines is not only to avoid doing wrong but rather to have a positive impact on the life of the student, client or patient (du Preez & Goedeke, 2013; Knapp & VandeCreek, 2004). Moreover, moral acts transcend the legal rights of the individual. Acting ethically does not merely imply following a legal rule but rather a human duty in a Kantian sense (Wood, 2007), where *respect* for the individual's dignity is added to the acknowledgement of the law. Most important is the ability to see a person with a disability as a person instead of as an expression of an impairment. For the professional, transcending the values rather than the laws of society is the essence of advocacy.

#### Ethical reflection and professional practice

All educational and clinical work with children, adolescents and adults involve a certain degree of intrusion into their privacy and family life. The most fundamental ethical issue is whether assessments and interventions are sufficient to benefit the child and the family (Glosoff & Cottone, 2010). The challenges of the family and the person with juvenile neuronal ceroid lipofuscinosis (JNCL) make them vulnerable (see Chapter 24), a particular ethical responsibility therefore rests on professionals working with this group to facilitate cooperation and dialogue. Parents must be given sufficient insight into appropriate goals and methods and the children must be given enough information to understand their educational needs and social situation (Glosoff & Cottone, 2010). On a macro level (Bronfenbrenner, 1979), the cultural attitudes and attribution of worth to people with disabilities will be reflected in how the agencies are organized and collaborate (Knapp & VandeCreek, 2007). The dilemmas emerging from the education and habilitation work should be subject to ethical reflections and within a larger professional group when such a group is needed for a broader discussion of appropriate goals.

It is an ethical necessity to protect children and young persons with disabilities from incompetent and non-appropriate practices and to make sure that the measures implemented are relevant (Falvo & Parker, 2000; Glosoff & Cottone, 2010). There is a risk that the moral standards that are applied to people with disabilities are lower than for other citizens, in intervention, social interaction and other aspects of life (von Tetzchner & Jensen, 1999). The basis of such risks is not only in the person's disability but rather in the person's relationship with the professionals. However, symmetry in such relationships is difficult to achieve, particularly when the expressive means of the person with disabilities are reduced. The asymmetry implies a relationship with an uneven distribution of power, which has implications for professional practice.

Terms are the tools of ethical professional reflection and the distinction between Aristotle's terms praxis and poiesis may be useful when planning and evaluating education and habilitation work (Skjervheim, 1964). *Praxis* (or primary morality) refers to the everyday acts, acts which involve interaction with other human beings, both in private life and elsewhere. They are not valued according to the results they produce, but whether they are following the cultural standard, habit, and tradition. They concern how one conducts one's life in relation to people who are close and toward strangers. Praxis is also a matter of honesty and of acting truthfully in real life. In praxis, the value of a person with severe disabilities, who may not be able to contribute to the common good in the community, cannot be considered less than that of people without such disabilities.

*Poiesis* (or secondary morality) refers to acts which have a meaning beyond themselves, which seek to achieve certain goals in the future, such as education and habilitation work. They are valued according to their intention as well as their final result. This is an important element, because the final results of education and habilitation measures are not possible to predict precisely, neither growth nor decline. The right poiesis acts are those that lead to the desired state and are performed in a professional sense.

For professional practice, the relationship between praxis and poiesis is a core issue. According to Skjervheim (1964), it is praxis which makes poiesis possible, and the morality of everyday acts is thus the basis for the ethical status of professional practice. The fact that poiesis depends on praxis implies that it is the *praxis* of the person that determines the moral status of the situation and, through reflection, the ethics and the intervention to be performed. There is no clear divide between the ethics of everyday life and professional ethics: How professionals act in their everyday life should also be reflected in their professional life. This also reflects that professionals do not approach their practice in a moral vacuum. They have a background of personal beliefs that becomes a part of their professional work and duties.

Skjervheim (1964) expresses concern that this relationship between praxis and poiesis is being overlooked, that poiesis has become primary in education. He points out that one cannot do in the technical realm what one cannot do in the ethical. Poiesis and technical acts must be based on praxis and ethical reflection. This is illustrated in his view on developmental psychology and upbringing: To act technically means to act on the basis of a calculation of what will happen if one does this or that. Upbringing – or parenting – belongs to the general human praxis, where the attitude behind the acts is more important than choosing the right means in the technical sense. But where the attitude is decisive, it is not primarily a question of psychology, but of ethics. According to Skjervheim (1964), goals of intervention – which belong to poiesis – can only be achieved if they are based on praxis. However, in discussions of ethics related to disability from a psychological and educational perspective, the *praxis* perspective may be difficult to detect. Focus is typically on the quality of the work of the professionals from a technical perspective, for example, whether teachers have selected the right goals and teaching methods or strategies. These discussions of ethics differ little from professional discussions in general. Although professional issues are basically ethical, the focus tends to be on the technical aspects, and ethical dilemmas may be hidden behind questions related to form and execution.

In a similar vein, Tranøy (1994) distinguishes between *morals* and *ethics*, where morals concern what is good and bad, and right and wrong, in everyday acts of life. Ethics comprises a more explicit, systematic and elaborated treatment of moral issues. Implicit in this distinction is the fact that ethics is related to consciousness or awareness based on reflection on moral phenomena. These phenomena are part of the living of everyday life, as well as of professional practice, and highlight the reciprocal relationship between practice and everyday life. Professional ethics are reflections on the moral implications of professional practice. In this sense ethics is a bridge between the two aspects of being a professional person: the professional side with its technology or *poiesis* on the one hand, and the everyday-life experiences or *praxis* on the other. At the same time, it mirrors the reciprocity between the life of the professional and the life of the person with disabilities, their mutual lifeworlds.

In the professional literature, ethical considerations are primarily related to the roles of the professionals but morality is in the interchange between the professional life and the supported life of the person with disabilities. The moral of professional actions is a question of whether the individual with a disability is treated as a person with dignity. For professionals, the person with a disability is neither a family member nor a person they may feel they belong to, a fact that makes a significant difference in the relationship, compared to family and other people who are close to the person with a disability. In the professional and scientific literature, people with disabilities are typically mentioned as "patients" – in a sense "objects" – although the patient role is only one of many roles in human life. The transformation from object (or patient) to subject implies seeing the individual with a disability as a person, not as a set of symptoms of a particular disease or impairment, and accepting that the person's limitations and differences still constitute a basis for a life with meaning.

When a person with JNCL is losing the ability to communicate (see Chapter 6), it influences life in more ways than just self-expression. It will change the interaction and how the person is perceived and treated, also when alternative means of communication are provided. A truly moral professional practice will depend on the acceptance of expressions of the person with a disability as authentic, even when communicative means are limited. This means that it is perceived as an actual expression of the individual's thoughts (Habermas, 1983). To acknowledge another person's communication as authentic also implies an acknowledgement of what the other person says as having value and worth taking into consideration, not only in order to achieve a particular goal, but to reach a shared understanding. Accepting a person also means acknowledging the person's inability to express meaning in a conventional manner and attribute meaning and decision to all his or her acts, also the unconventional ones. However, although it

is an act of intended acceptance, it creates a risk for attributing goals, wishes and ideas that the person does not actually have, and for erroneous interpretations of the person's behavior and intentions. Krivohlavy (1996) describes a seriously ill woman who whispered *water*, and a care worker brought a glass of water to her on the assumption that she expressed a wish to drink. However, she did not drink the water. She was asked whether she had said *water* and confirmed this. Only when the care worker went to her side and followed her gaze did he understand the meaning of the utterance: she was looking at a flower. When the water in the glass was poured into the vase of the flower, she smiled her acknowledgement. In order to understand the woman, it was necessary – as in all true communication – to take her perspective.

When people have severe cognitive and other disabilities, it may be necessary for professionals to guide and assist their choice-making, and in some situations, take decisions on behalf of the person in collaboration with the family. However, even with the best intentions professionals may take over and dominate the choices of the person. Respect for the autonomy of others and their freedom to do as they choose is a core element of ethical action (Knapp & VandeCreek, 2004; Raines & Dibble, 2011). Building the person's autonomy or self-governance therefore is a basic element of ethical professional work. A person may need help and assistance but should not be unnecessarily controlled by others, even if personal limitations may require interdependent rather than independent choice (see Chapter 16).

The understanding, respect and value that are the foundation for ethical practice also imply acknowledging that education is the normal situation for children and adolescents with JNCL. This may include the use of mainstream education as long as – but not longer than – this can provide meaningful activities and inclusion. The person should be in familiar environments to reduce the effect of dementia and declining eyesight, planning should be for a satisfying today and tomorrow's necessity, with possibilities for pursuing personal interests, goals and a good life. Yet there is very little research about education for children and adolescents with JNCL. This may reflect that their education is considered as being of little value, even if the school is a central element in their life, as it is for children and adolescents with typical development. If valued as poiesis, their future contribution to society may be little, and hence the value of the education because it may not lead to a profession or working life in the ordinary sense. However, when seen in the perspective of praxis, the education of children and young people with JNCL has its own value and equal to that of others. From this perspective, schools should provide positive challenges rather than an "easy" life with little possibility for meaning-making. In the present project, there were comments indicating that teachers and other staff found it difficult to make appropriate plans, find the right content of the education, or giving education at all (see Chapter 11). These comments demonstrate the need for ethical reflection about human values and the good life (Barker, 2011).

# Understanding the lifeworlds of people with disabilities

Professionals' understanding and attitudes toward people with disabilities will be a basis for their ethical reflection and practice. Their education and competence will influence their decisions but also less well-founded assumptions about brain training or "neuromyths" (Ansari, 2015). The professionals' understanding of an individual or group of individuals should take the whole life situation into account and not be based on a few features, such as the symptoms of a disability. All aspects of the person and the person's environment are integral to the identity and meaning-making of the person, the strength and challenges of the person, culture, history, and the societal organization and structure (Bronfenbrenner & Morris, 2006).

There are two typical ways in which a professional may develop a false understanding of the situation of persons with disabilities. Firstly, the professional may keep distance and see the situation from a narrow technical-professional perspective. The result may be an approach that focuses on instrumental achievements and less on understanding the person's sometimes idiosyncratic meaning-making. The second way is when professionals focus on their own feelings. For some, the perception of people with a disability appears to be partly based on their fear of seeing themselves in a similar situation (see Tomkiewicz, 1996). This may lead to a false feeling of being empathetic and able to evaluate what a good life is for that other person. This in turn increases the probability that the professional's own views and emotions will be attributed to persons with disabilities as if they did not have their own life. It is necessary for professionals to see both the person with a disability and themselves as equal partners in the relationship. Similarly, when talking about people with disabilities, politicians and others may show false empathy by saying that «all people are a little disabled or have special needs», thereby expressing a lack of acceptance of a person who is truly disabled and disregarding or minimalizing the everyday practical problems and challenges related to having a severe disability.

These possible conceptions of persons with disabilities have in common that they are not based on a true dialogue. The professional may not have listened to the person with a disability or looked for details in his or her lifeworld. Only through a broader perspective and appreciation of their everyday challenges, it is possible for professionals to obtain an authentic impression of the situation of the disabled person. Such an insight is necessary for understanding what a meaningful life in atypical circumstances might be, and hence for planning and implementing educational and other interventions for people with JNCL or others who are losing abilities. Of importance here is the professional's realization that the learning and developmental progress of the person cannot be used to measure the quality of the work of the professional – even the best educational adaptations and interventions cannot stop the progress of symptoms in persons with JNCL.

These ethical reflections point to a wider perspective. For example, children and young persons with JNCL have different reactions to a difficult life, and anger, aggression, depression and apathy – when the person understands and manages less, and hormones rage as in puberty – should be met with *understanding*, if not with accept. Emotional and behavioral reactions need to be interpreted within the life situation of the person – a context characterized by increasing life confusion. The reactions should not be perceived as "bad" behavior and it is important to remember that the "challenge" in challenging behavior rests in those who are challenged, not in the person whose behavior elicits the challenge (Emerson & Einfeld, 2011; von Tetzchner, 2004). A similar understanding based on ethical reflection may apply to collaboration with parents. Parents of children with JNCL experience a difficult life situation, resulting in varying moods that from time to time may make positive and effective collaboration difficult. In such situations, the professional should not be aggrieved but show sympathy, understanding and acceptance – and openness if and when parents change.

#### Disability, ethics and human rights

Ethical reflection is a foundation of planning and implementing education and other interventions and is closely related to human rights as expressed in the UN Universal Declaration of Human Rights because human rights have underlying moral imperatives (see Gauthier & Pettifor, 2014; Nickel, 2007). In this context, reflection on ethical practice and human rights is about the values guiding the education and habilitation of people with JNCL and others with extensive needs. It is about what is best for the person with JNCL. The interventions that are selected should be satisfying today and useful for tomorrow. Childhood has a value of its own and represents a way toward adulthood.

In this perspective, a person is not just a composite of different parts but an integrated whole with continuity and change. Similar formulations may be found many places in this book. However, the demands on intervention imply choices that may give rise to dilemmas that do not have any obvious solutions. For example, participation is a core concept in ICF (World Health Organization, 2001) and habilitation work, but may represent a dilemma when the person does not want to engage in activities which others consider necessary for a better future life. Much habilitation work is done on a very personal and private level, entering into other people's home. The following statement, based on the views of many parents in the current project (Appendix A), shows the importance of considering pro-active learning:

*It was too late to start with the learning program; the cognitive decline had gone too far.* 

Some professionals considered it unethical to address a problem, which was not yet present. They also expressed doubts about the student's motivation to learn something he or she did not need in their current situation.

*I am not sure it is fair to make plans when you know the outcome of the disease.* 

*It feels wrong to make plans for the future when nobody knows what is going to happen.* 

Precautionary or pro-active teaching was also sometimes perceived as *odd*, in particular when the students were not aware of what to expect from their own diagnosis. However, parents and professionals also gave good examples of pro-active learning for students with JNCL (see Chapters 11–14). It was a matter of enthusiasm and dedication by people in the planning group, and how the teaching activities were accomplished and organized.

Participation in education that is based on the student's inability to take active part in an activity may pose a severe threat to students with JNCL and represents an ethical dilemma (Mortensen, 2006; von Tetzchner & Jensen, 1999). An Individual Education Plan (IEP) should always express the importance of education. Maintaining educational and social participation will be one of the most important requirements in IEPs for children and adolescents with JNCL. The participatory perspective should be explicitly formulated in IEPs for all students with JNCL, as well as in long-term Habilitation Plans (see Chapter 11).

Confidentiality is essential, but so is information. This is a sensitive issue with diseases like JNCL. Educational and other interventions may involve many persons and different settings. Both professionals and classmates may need to know something about the problems they see that the student with JNCL is struggling with. In addition, awareness of the possible negative consequences of not introducing proactive or precautionary teaching makes it necessary to discuss how and when to talk about the future, and what should be told, for example in relation to the need for early learning of braille (see Chapter 14). In such a situation it is necessary to ask «what is really needed to be known by whom» (Kjønstad, 2010; Lindén & Rådeström, 2008). To solve this kind of dilemma there must be time for discussions between the family and professionals. There are no "right" answers to such questions and dilemmas, but it is important to find a practical consensus.

In the same vein is the question of keeping the disease secret from the individual with JNCL and people in the environment. This has been an ongoing discussion among professionals with little parental participation (Elmerskog & Fosse, 2012). The only acceptable solution to this dilemma is simple: it is the parents who have to make such a decision on behalf of themselves and their child, without their decision being questioned or reviewed by professionals. This is illustrated in the present project, where two pairs of parents had made different decisions and both expressed that their decision was a successful one (see Chapter 25). Each family has built their own context and decisions are made within this context. However, this does not mean that professionals should avoid such delicate themes but should contribute with objective information and their experiences when asked, expressing a clear attitude that such issues are for each family to decide and cannot be divided into right or wrong solutions.

At an organizational level, it is important to obtain a consensus between the family and the services regarding education and intervention for the person, especially what the family should and can expect of the services. When decisions to be made involve professional issues and values, it is also an ethical issue how the decisions will affect other services and persons, and who is entitled to make the decisions. Sometimes it may seem to be solely a professional decision but in reality, it is more of a moral decision; it is about values and attitudes, such as when there is a choice between a specialized institution relatively far away from family and peers or a more mainstream solution nearby the home (Hesselberg & von Tetzchner, 2016).

#### Ideal and pragmatic services

Ideal educational and other intervention measures are based on professional sensitivity to the family and the student (Hesselberg, 2017). For example, the amount of professional follow-up needed and the form this should have, will be one of the first joint decisions of the family and the professionals. The aim is to establish an understanding where parents and professionals share expectations to the collaboration and can raise the issue if these expectations are not met. In

addition, knowledge and competence of teachers and other staff should be sufficient and predictable. There must be local knowledge as well as knowledge about the disease and what to do. Poiesis implies being trustworthy and committed to the ethical stance *and* to strategies that are proven to be useful – in this way, the evidence base is a theoretical and empirical foundation of both practical work and ethical reflection. Professionals should have authority, which means that their knowledge should be substantial and presented in a believable way. Importantly, they must know the limits of their own knowledge and where to get help with things they do not know well enough or are unable to manage themselves. This implies knowing the limits of their responsibility, what decisions they can and cannot take. The ethical stance requires routines for making joint decisions about education and special education, and about providing challenges and care (Raines & Dibble, 2011).

The need for ethical reflection may be most evident in working with plans for the future. The Habilitation Plan and the Individual Educational Plan (see Chapter 11) need ethical reflection to ensure that plans are based on praxis and poiesis. Habilitation Plans involve personal and private matters, results from assessments, and decisions affecting the whole family. Professionals must acknowledge the variation between individuals with JNCL, for example related to age, problems and circumstances of life, support and counseling. Ethical reflection is needed for making plans for education and intervention, but plan-making in itself also contributes to raising awareness of the need for such reflection. Ideal and pragmatic services seek to establish an *alliance* between the family and the professions, with collaboration, engagement and insight into the ethical dilemmas that may challenge the overall plan and sub plans (see Chapter 11).

#### **Concluding remarks**

Working with children and young people with JNCL and their families may imply many challenges and ethical dilemmas. An ethical practice implies searching for the balance between the focus on the life today and the needs of tomorrow, and between positive learning challenges and social participation. Ethical dilemmas are always difficult, this is why they are dilemmas, but the answer is not neglect but ethical reflection. Morals, even if applied to the individual, are basically social, reflecting the norms and values of the society. Moral development is not conceivable outside a community of some kind. It is, however, not enough to follow the habits and traditions of the society, it is these that the individual has to take a stand on and transcend. It is possible to reflect on ethics in solitude, but ethics can only be cultivated in a social context and through discourse. To ensure that interventions have an ethical foundation, discourse should be facilitated between various professionals, family, and other significant persons in the life of the person with JNCL. Communication with the person, even if severely and profoundly impaired, should also be part of this process.

Reflecting on ethical dilemmas is an important element of working with education and habilitation for children and young people with JNCL. However, ethical reflection is not only about the basic existential questions but also about a fundamental respect for the individuals with JNCL and their families, as well as for those who are doing the everyday interventions, from assistants to specialized professionals. One's ethical stance and attention to reflection and reflectivity are equally important for the responsibilities of determining practical adaptations of everyday activities and of participating in discussions about life choices. Both responsibilities require knowledge, understanding and sensitivity to be addressed in a systematic and good way.

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### Organization of Education and Counseling

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The organization of the educational system is a foundation for the education of all students. However, it is especially important for students who have a disability because they need educational structures and functions that are tailored to their needs and possibilities. While most children will develop and learn under varied conditions, children with disabilities depend on a narrower set of structures and functions in order to develop and learn optimally (von Tetzchner, 2019). The organization also varies in geography and size, culture and history, and the national traditions and ideas about education.

The present chapter presents elements from the educational systems in five countries (Germany, Scotland, Finland, Denmark and Norway) that are relevant for the education of children and young people with juvenile neuronal ceroid lipofuscinosis (JNCL), including some case stories. In all the countries, support for the family and collaboration with parents are important parts of the work (see also Chapter 24). In the present project (Appendix A), the percentage who attended mainstream school was gradually reduced with age. More than 80 percent of the participants with JNCL had attended a mainstream school in grade 1 and 2. About half had attended a mainstream school in grade 5, and in the higher grades (8+), more than 80 percent had attended a special unit or school.

# Germany: Educating children and adolescents with JNCL in Hamburg

Germany has a size of 357,170 square kilometers and a population of 81 million (2014). It is a federation of 16 states and Berlin is the capital with 3.5 million inhabitants. Hamburg is a state and the second largest city with a population of 1.8 million. It is located in the north of the country.

The Education Centre for the Blind and Partially Sighted (BZBS) in Hamburg has provided education for children and adolescents who have JNCL for several decades. A JNCL working group was established many years ago to organize and monitor the education of this group of students. The work presented here is related to students starting school, transitions (changing class or school and leaving school) and collaboration with parents.

#### Starting education

The course of the disease varies significantly, and the school experiences and careers are also quite diverse for students with JNCL. Most of the students with JNCL who have attended BZBS began their education in a mainstream school. The eye condition usually became noticeable during their first two years at school (see Chapter 4). In this challenging situation, the dilemma arises as to where the child will receive the best possible education. The options were either that the child stayed in the mainstream school with special educational support or moved to the Education Centre for the Blind and Partially Sighted.

The decision about whether it is best for the child to move to a special school should be carefully reviewed and it is essential that it is made on the basis of a close and trusting relationship between counselors, special education teachers, the ordinary teachers and the parents. Experience in Germany indicates that an immediate change of school, especially and preferably in the early years (primary school), is most effective. Depending on the course of the disease and the child's condition, a further decision is made about placement in BZBS: whether the child attends an ordinary class or a special needs class (for students with multiple disabilities). Both may be a good choice, depending on the needs of the individual child.

The parents initially find themselves in a seemingly inexplicable, threatening and extremely stressful and challenging situation. Embroiled in the difficulty of coming to terms with the diagnosis of visual impairment, they encounter new changes which at first seem to be related to the emerging blindness. Parents live through the fear and unhappiness of their child and find themselves in a state of exceptional stress.

The immediate changes experienced by the child at this time make him or her increasingly anxious. The child tries to cope, compensate or even hide what is happening. At the same time, the people around the child are also experiencing insecurity, uncertainty and perplexity, as well as feelings of guilt. The changes in the child's daily life, such as starting school or changing school, and an odyssey of visits to hospitals and other institutions, imply numerous stressful and frightening situations. It usually does not take a long time from the initial observations of the child's deteriorating vision until changes in behavior and loss of motor functions become noticeable. Learning challenges typical of children with JNCL are identified by the special education teachers and the final diagnosis is soon made. The diagnosis of JNCL is a huge shock to parents and places them under enormous pressure. It is difficult for them to come to terms with the situation. They need to consider how to support their child and at the same time look after their other children, as well as deciding how and when to explain the situation to the child and the siblings. The issues of how to inform others and speak about the illness need consideration and are challenging to all parties involved. When a child is transferred to a school for the blind, it is important that the school is aware of the personal situation of the child's situation are important, including the home situation and the atmosphere in the home and other parts of the immediate environment.

To this end, the school offers many opportunities for dialogue with the parents, where the support options and forms of help available are outlined without exerting any pressure. The child's lessons are modified to address the changes which may not yet have substantially affected the teaching and learning process.

The speed and extent of the changes in behavior, cognitive decline, reduced motor functions, speech problems and onset of seizures vary, and these may appear together or in succession (see Chapters 3–7). However, irrespective of the course of the disease, the emergent declines have serious effects on the child with JNCL and impact every area of the child's life – family, social life and school.

The child or young person will often be keenly aware of these changes and will ask «What is happening to me?» and then «Why?» They may become extremely distressed, show fear and despair, and at the same time go through periods of denial and suppression, not wanting to acknowledge what is happening.

As the cognitive functioning of the child or young person is gradually affected they become less capable of a realistic appraisal of their own situation and the options to manage these changes will be fewer. Parallel to the changing reactive behavior, the disease also may imply changes in behavior and character that bring additional problems to the group situations. Young persons with JNCL tend to develop an insistence and reliance on specific routines and patterns of behavior, which can be very personal and idiosyncratic. It is not always possible to accommodate these behaviors in the class, and this may lead to stress and conflict in the classroom.

It is necessary to look at each child or young person individually and constantly review the organization and teaching content to determine where they can be best supported. A move to a special needs class should always be thoroughly discussed with all parties involved (parents, current and new teachers) and the transfer should be carefully prepared and implemented.

#### Moving to a special needs class

Many students with JNCL are becoming sensitive to change and moving to another class should be considered only when absolutely necessary. It is important to reduce the stress of having to adapt to new conditions, places and people. A gradual phasing-in of such changes seems to be the best way to reduce the possible strains. The student will be introduced to the new class in the course of several visits and the duration of the visits will gradually be increased. When entering the new student group, the student with JNCL will gently be introduced to the new peers and to new but interesting media and materials, such as the computer program *Sarepta* (see Chapter 19).

The student may observe a discrepancy between his or her own ability level and that of the new classmates, and this could prove problematic as the level of the class might be clearly lower. However, this can also have a positive and motivating effect, as the student with JNCL may feel less pressure to perform or feel superior. At this time, the student may have considerable skills and competence that can be used in the classroom. When it is possible, the student can continue having lessons in a favorite school subject (such as sport or music) in the former class and thus maintain his or her existing social relations.

Generally, the student with JNCL will become less perceptive of the situation and less aware of the changes and declines. Teachers must be attentive to signs or behaviors in the students, in order to respond appropriately to their changing needs and ensure that the right help and support are provided in an atmosphere that is peaceful, secure and comforting.

#### Cooperation between school and home

Naturally, the teachers, special educators and other members of the school staff cooperate closely with parents. Sufficient time needs to be allocated to collaboration with parents as it is evident that the education and support provided to the student with JNCL in school are greatly determined by the quality of the cooperation and interaction with parents.

How the communication between school and parents is conducted and how important facts are relayed will depend on several factors. It is the school's experience that the relationship that is formed between school and parents is greatly influenced by the personalities involved. Some parents are very approachable and desire regular communication, while others may prefer more sporadic contact.
The location of personal meetings should, at first, be decided by the parents according to their wishes. Inviting parents to the school, at least during the first period, may trigger uncertainty and anxiety. One well-tried approach is the «round-table talk», where a psychologist invites the parents to a meeting to discuss specific issues with family members and class teachers, and then moderates the meeting. The team members are thus relieved of the problem of how to manage the discussion and what they sometimes refer to as an «emotional chaos», allowing them to focus on the issues discussed in the meeting.

Meetings can involve several parents and teachers, which facilitates the exchange of information and experience. This may lead to a continuous exchange of updated information and a network directory, an exchange of day-to-day experiences, expectations and feasible outcomes. Importantly, such meetings offer time and place for expressing anger and grief, as well as for sharing the fun and enjoyment that can be encountered in daily classroom situations.

It is not a question of who has the highest level of professionalism with respect to the care of and relationship with the student. The parents are always

	The following core statements form the ethical basis for our pedagogical and therapeutic mission. They express our commitment to an authentic, professional approach. The code commits us to a serious and honest management of the individual characteristics and condition of every child with JNCL, both in our day-to-day actions and in every discussion and consultation. Implicit to this code is open and honest communication between all staff in this institute and the parents, based on trust, respect and acceptance.						
	Every child has the right to a fruitful life and the chance to develop his/her personality as fully as possible.						
	The rights and needs of a child with JNCL are of no lesser value and are equal to the rights and needs of other pupils.						
	We ensure that the dianity of the child is observed and tolerate no belittlement or disrespect						

Our mission is to make every day as fruitful and meaningful as possible.

We encourage and foster the child to help it develop its full potential within his or her capabilities.

We accompany and support the child through the process of change and deterioration.

We are committed to ensuring that the child is part of the community and group experience for as long and as far as possible.

We protect the child from the risk of becoming lonely and isolated.

The child has the right to know the truth. Working closely with the parents we gradually reveal the truth of the situation.

While we will never lie to a child, we will also never deny him or her any hope.

Table 9.1 The school's statements for guiding an ethical practice

the center of all decisions. Together with the school, they are responsible for achieving agreement concerning responsibility and authority. An essential part of such agreements, or alliances (see Chapter 10), is consultation on informing all the persons and groups involved with the child about JNCL. Through long experience with working with children and adolescents with JNCL, BZBS has accumulated basic know-how which has been formulated in writing as an internal ethical consensus or code of conduct, which is binding for all educational and other personnel. These ethical principles are the basis for a code which governs professional behavior and treatment of the students with INCL and their fellow students, cooperation with families, cooperation within the institution and with other schools, specialized agencies and cooperating institutions and partners (see Table 9.1). The application of this internal school code to other agreements, which define mutual commitments and responsibilities, has proven useful. The statements and how they are applied on a day-to-day basis in work are constantly reviewed and scrutinized. The work of BZBS is carried out within the context of a continuous exchange and mutual support with other specialist disciplines (see also Chapter 8).

#### Transitions and leaving school

Schooling generally finishes at the end of the statutory school-leaving age, although sometimes an application might be made to end it earlier or beyond the maximum statutory age limit, depending on the development of the child, his or her life situation, and the family's circumstances, all of which are important factors in this process.

A place in an institution after leaving school will usually be made available at short notice to persons with JNCL. This offers new perspectives and opportunities for the individual, as the transition from school to further training is rare for persons with this condition. In such cases it is necessary to prioritize the preparation and encouragement of the person and facilitate a smooth and unbureaucratic transfer (this could entail leaving school mid-term or before the end of statutory school education).

The following principle applies: schooling should be organized as flexibly as possible. Any decision about subsequent suitable vocational training should be made on an individual basis taking the person's needs and abilities into account. Depending on the individual, the most likely options are a special day school or training in a workshop for people with disabilities. Students participate in various vocational assignments during their last three years at school, and thereby individual interests and abilities are identified, nurtured and developed with the aim of finding appropriate workplaces. Other factors are also considered, including the living and care situations, the situation of care givers and support staff and opportunities for

retreat and relaxation. During the vocational assignments members of the staff of the Center (BZBS) talk to staff from the potential workplace about JNCL, with the aim of facilitating a job offer. It is important to ensure continuity and as far as possible avoid future changes of workplace, group and support persons.

If a suitable future workplace or occupation is found, the trainer is offered the opportunity to meet and observe the student in the classroom in order to gain a better understanding of the future trainee in his or her familiar and adapted surroundings. Similarly, the Centre offers to send the class teacher to the new training place to facilitate the transition and provide support and advice at the early stage. An additional period of vocational experience at the future job or workplace while still being a student at the school has also proven useful in helping the student to prepare for the future life.

The entire process of seeking sheltered employment takes place in collaboration with the parents. The issues that need to be addressed and the specific approach are decided very much on an individual basis, asking questions like: How much support do the parents want? Will vocational activities be combined with a residence? Does the workplace need to be close to the person's residence?

Our experience over the years indicates that the transition of a student into employment is a challenging time for parents of children with JNCL. Following a fairly long period of relative peace and stability during the school years, parents again need to make important decisions for the future. They are again confronted with the degenerative nature of their child's disease and this will shape their actions and decision process.

# Educational concepts and aspects of the schooling and support of students with JNCL

The review of educational work with children and adolescents with JNCL by the working group of the Center was presented in 2010, describing four core elements and principles: a) Tasks and objectives, b) teaching content, c) equipment and materials, and d) approach and methods. The first three of these elements are presented below.

# Tasks and objectives

The staff of BZBS accompanies and supports children on their educational journey and ensures their dignity. Core principles of BZBS are *attentiveness, support* and *sincerity*. These constitute the heart of BZBS's work. BZBS has undertaken an obligation to cater for each student's educational needs and not make premature assumptions about the students' educational possibilities and limitations. BZBS try to attain the right balance between educational challenge, encouragement and support. The staff at BZBS should have insights and perspectives related to normality, typical development and atypical development based on an understanding of disabilities and disorders. BZBS should acknowledge and understand that declines and regression are to be expected in children with JNCL. This acceptance should not exclude the possibility of providing engagement and opportunities to the students, enable their participation in society, or making every day enjoyable and meaningful. This understanding will guide the staff of BZBS when they are planning and implementing educational programs. The programs should be specific and support the following steps:

- BZBS will support the student's development by encouraging them to acquire the highest possible knowledge and skills.
- BZBS will help the students to retain and maintain acquired competence and skills as long as it is possible.
- BZBS will seek to ensure the maintenance of the students' best possible physical and mental state and health.
- BZBS will support the students' reminiscence and the maintenance of memories of special importance to them.

# **Teaching content**

Education for students with JNCL is based on a profound contradiction between development and regression. This dichotomy is however less pronounced today, due to the diversity of learning options and attention to individual needs resulting in individualized education programs.

The students should remain in inclusive settings as long as possible, that is, within their peer group, and receive the same offers and activities as others. However, the overall aim is to promote the best possible learning, development and maintenance of skills and capabilities.

An Individualized Education Plan (IEP) will be needed when the condition of the student with JNCL deteriorates. The mainstream curriculum will at some point not meet the student's special needs. Catering for the individual needs specified in an IEP requires a broad understanding of the student's life and his or her interests and preferences (see Chapter 11). However, the educational content is not determined solely by the student's interests and preferences for subjects and themes. Education must also address goals and learning areas selected by objective means, such as maintaining independence, active participation and social relations.

Group learning and activities become easier to achieve if the special interests and skills of the student with JNCL can be integrated into the activities of the class. As the disease progresses, the student may retreat into strong personal interests, which may function as escapes from the everyday reality. These escapes or behaviors may dominate the student's life. Sometimes the student's personal interests are so special and different from those of others that integrating them in the class activities becomes impossible. There may be situations when students should be encouraged to discover and develop interests to be used as escapes for meeting difficult realities in life. However, this may be difficult to achieve. Even the best plans may not result in achievements, as illustrated in the example below.

The beautifully designed furnished doll's house did not prove to be as successful as expected. The girl with JNCL, who we assumed would be eager to play with the house and the dolls, simply found the house to be stupid.

A varied musical curriculum is important (see Chapters 17 and 18). Other areas are role play, theatre, games, working with materials, sounds, dance and movement (see Chapters 12 and 21). These areas can be used to support feelings, expressions and communication.

Program content also includes sport, swimming, activities of daily living, physiotherapy, occupational therapy, mobility and orientation. In early phases of the disease, these activities will be used to support further learning and development, and in the later phases to maintain skills and capabilities. Many of these areas will also support participation in social settings and activities and will help students to cope with everyday challenges.

Sitting in a cozy corner together, cuddling, talking, changing positions, movement, enjoying going outdoors and fresh air are simple (yet valuable) experiences that can be easily achieved at the school grounds, on shopping trips, walks, excursions and the like.

The students may need psychological support to cope with their own situation and attain emotional stability. It is vital that the class has an atmosphere of openness, trust, safety and comfort in which a student can speak about feelings without anxiety and fear. Being open about feelings is positive for everyone, students as well as teachers, and on equal terms. This is illustrated in this example:

Students with JNCL can use a memory bank where they can store ideas, thoughts and reminiscences. The boxes will be filled with different thoughts and memories, and they can be emptied and refilled again, dependent on the student's needs for reflection and storing memories.

Explaining bodily changes for students with JNCL requires skills and strategies. It is important that the students do not feel isolated and alone when experiencing changes and declines. There are children's books and CDs that effectively address fear and anxiety, limitations, loss and separation, death and dying. This is illustrated in this example.

The book Seelenvogel (Snunit & Golomb, 1991, 1999) contains excellent images that illustrate different human qualities, concerns and aspects of life. Through role play and cuddling we used the themes of the books to make a reality play. During the session, a girl with JNCL was able to put aside her inhibitions and express freely about her anger with having this disease.

# Material and equipment

It is not always possible to know what material and equipment there might be a need for as the interests of the students are constantly changing. It is a good idea to provide teachers with a collection of much-loved and tried materials and accessories. For example, unconventional materials can be used for creative work:

One boy had a special love of shoes of every kind, so the school janitor built a shoe cupboard for the whole class. The boy who loved shoes was able to share his interest with the class and the shoes became a source of discussions.

Memorable and favorite items may be stored for the future, even if the student at present is not showing particular interest in the object. It is useful to compile a collection of some kind – including unconventional objects – for every child. The «treasure chest» is a much loved and extensively used place to store memorable objects, loved items, and mementos. Some children have a treasure chest at school and at home. It may contain old toys and CDs (it is advisable to make copies!). The possessions and objects can be used for refreshing or reviving memories in the future. Students seem to like objects that can produce sounds, and sound recordings of their own voice is often a success. Sarepta is a software program that offers diverse opportunities for work and play. It can be customized to the individual and the changing needs and abilities of the person (see Chapter 19).

# Scotland: Aspects of the Scottish Education System

Scotland is part of The United Kingdom. Scotland has a size of 78,282 square kilometers and a population of 5,4 million (2017). The capital is Edinburgh with a population of 450,000.

In Scotland, provision has been made by the Scottish Government through the Education Act (2000) which stipulates that individuals with additional support needs, such as students with JNCL, should be able, as a right, to attend their local mainstream school. Further the Education Act (2004) was introduced in order to provide explicit and legally binding support for children and young people identified as needing additional support within the school setting. Both Acts grant children and their parents important rights which affect education. The 2004 Act was amended by the Education (Additional Support for Learning Scotland) Act 2009 to include support from other professionals, to ensure that young people are provided with all the necessary support to help them work towards achieving their full potential within a mainstream school.

Under the Education Act 2009 (Additional Support for Learning Scotland), parents and/or anyone working with a child who concludes that the child has additional support needs, can request that the local education authority investigates to establish if this is indeed the case. If so, the education authority is legally required to make "adequate and efficient provision" to meet the identified additional support needs of the child. This process is underpinned by another framework initiated by the Scottish Government – "Getting it right for every child" (GIRFEC), which puts the child at the center of any identified initiative or plan involving other professionals. Together with "The Curriculum for Excellence", GIRFEC aims to support and improve outcomes for all children who have been identified as having additional support needs. These policies, however, at present, offer no template for provision within a mainstream setting for young people with JNCL. Therefore, the Royal Blind School is just one model of possible and potential provision as educational authorities could assert that these individuals' needs could be met in their own learning support provision within a mainstream setting.

After a process of assessment, if a child or young person is identified as having complex or multiple additional support needs, a Coordinated Support Plan (CSP) is compiled to ensure that the support for learning can be coordinated across a range of agencies. The CSP is a legal document and an educational psychologist of the education authority is responsible for monitoring, reviewing the plan, and ensuring that all set targets are being met annually. Depending on the financial resources of individual local authorities within Scotland, the path to securing a placement in a school of choice by parents and others is not always guaranteed. In some situations, if parents are determined to secure a place of their own choice, rather than that of the local educational authority, then a tribunal takes place, which can be an arduous and demanding process, with no guarantee of the desired outcomes for the parents.

The Royal Blind School in Edinburgh is a specialist center of provision of education for young people with visual impairment. Local education authorities provide referrals and ensure funding for day or residential students with a range of additional needs. Since there is a wide range of expertise on site, young people have access to experienced staff trained in supporting young people's individual needs, including qualified teachers of the visually impaired, trained support staff, occupational therapists, physiotherapists, speech and language therapists, and habilitation and orientation specialists. There is also access to the latest assistive technology, small teaching groups, and specially tailored programs of learning through the implementation of Individual Education Plans (IEPs). The Royal Blind School also provides outreach support to other teachers and support workers throughout Scotland through its "Learning Hub", via for example short online video programs and training days in school. Staff in school continues to be involved in provision for students with JNCL and work through dedicated computer application technology and in-service training sessions within school, outside conferences, both as participants and facilitators.

Presently in Scotland, young people deemed to have an additional learning need, such as persons with JNCL, do have a choice of provision. However, the options may not always directly meet the needs of the young person. For some individuals an inclusion unit may provide the necessary support whilst for others a specialist school placement may be more appropriate.

#### Case story: Residential school placement in Scotland

Ideally, children would attend their local school and engage and participate in their local community. With children who have additional support needs, and specifically children with JNCL, this is not always possible. Parents want their child's needs met in an environment which supports their learning and wellbeing. It is not an easy decision to opt for a residential placement. And even when the decision has been made, funding is not always easy to access. The Royal Blind School offers weekly and part week residential placements. All residential care workers are qualified, and the service is regulated by the Care Inspectorate. Residential care workers, education staff, therapists and habilitation specialists are offered regular in-service training on

JNCL. They are all encouraged to promote excellent collaborative practice and to share innovation in their approach to the individual pupil.

This case study is to highlight for families and professionals the role of residential staff in supporting children and young persons with JNCL and their families.

Adele joined the School after it had been noted, at her local school, that there had been a significant deterioration in her vision. She was diagnosed with JNCL shortly after this. In discussion with the parents, the local education authority and the school, a weekly placement was decided as the best option for Adele. Factors that were emphasized included home circumstances, geographical location of the family, access to expert staff with knowledge and experience of JNCL. The parents visited the school and met with senior managers, including the head of care and other staff who would be working directly with their daughter. Adele was allocated a key worker who was the main contact point for the parents regarding all residential matters and a teacher to liaise regarding educational matters. The key worker and teacher met regularly to pass on relevant information and ensure that communication with the home was effective.

The key worker would attend team meetings and ensure that Adele's daily observation file was up-to-date, pass on information to relevant staff and write monthly reports. The file would contain a detailed Care Plan and IEP, daily observations, monthly reports, notes from liaison meetings, medical information, information from other professionals, updates on *INCL from in-service sessions, BDFA and conferences attended by one of* the deputy head teachers. The documents would detail, for example, the need for a smaller, quieter living skills area for Adele to allow her to maintain independent living skills as long as possible, Adele's interests and aptitudes, and strategies for dealing with mood swings. The staff used a communication diary and, in accordance with the wishes of the parents, telephoned regularly, daily if requested. Adele's mother was supported by a close relative and later on by a family friend, both of whom kept in regular contact with the school. *Her mother was able, if she wished, to meet with the staff any Monday or* Friday by coming to school with Adele's transport. Though welcome to come at other times, transport was not easy and time consuming. Parents are also welcome to come to class to see strategies in practice.

Like all pupils at the school, Adele had reviews at least once a session and more often at times of transition. These were attended by school staff, local authority professionals and the family. Adele attended some of her reviews, being prepared and supported by her key worker and teacher.

*As she entered her teenage years, Adele began to have quite dramatic* mood swings. These had, at times, a significant impact on her attitude and behavior. She could be very stubborn regarding personal care and angry at her peers, not wanting to engage with them and staff. This was a pattern that was happening at home at weekends and her mother found this very challenging. The staff supported Adele by allowing her time and a quiet space. *Her parent felt supported with strategies that had been tried successfully at* school. The residential staff were responsible for the residents in their care from around 4 pm until 9 am the following day. This is much longer than with any other group of staff. Of course, strategies that worked at school did not always work at home, and vice versa, but the communication with the parents was vital to ensure the best possible support for Adele. The regular meetings with the educational staff also ensured that new ideas could be discussed and taken forward with time allocated to evaluate suitability and effectiveness of any adopted strategy. Adele's teacher and key worker collaborated with the family to build a memory book. This was a precious document to Adele and her family, as well as being of great use to new staff and for Adele's transition post-school. Over time Adele was also subject to hallucinations and would become extremely agitated in the evening. As *her communication skills deteriorated, life became very challenging for her* and, naturally, for her mother and close family.

The residential staff and especially Adele's key worker observed to identify what worked well for Adele. She liked familiar people around her when she was distressed. She loved music and that continued to be an area she connected with throughout her time at school. Adele enjoyed having her nails manicured and the company of younger peers in the residential area where she was living. Animals were of great interest to her. Any changes to her routine had to be gradual and by maintaining her interest Adele could be supported by staff to try new things or meet a new classmate or member of staff. This stimulation was seen to have a beneficial effect on her determination to keep trying. Throughout her teenage years, the staff supported Adele with the technology she used in school and encouraged her to practice her braille skills and maintain her social life. Key to Adele was music; even when her speech became unclear and she stuttered, she was able to sing clearly. She also enjoyed certain stories that she was happy to hear time and time again.

The bond that was built between Adele and the residential staff, the parents, the teacher and the support staff lasted as the staff continued to visit Adele after she had left school.

# Finland: Valteri Centre for Learning and Consulting

Finland has a size of 338,430 square kilometers and a population of 5.5 million (2015). Helsinki is the capital with 621.000 inhabitants.

Valteri Centre for Learning and Consulting is a national center operating under the auspices of the Finnish National Board of Education (FNBE). FNBE is subordinate to the Ministry of Education and Culture and its organization and functions are set in the legislation. Valteri consists of six units in different parts of Finland: Mikael, Mäntykangas, Ruskis, Onerva, Skilla and Tervaväylä. According to the Law of Education, Valteri has the nationwide responsibility to administer and develop consultation and support related to inclusive education in municipalities and arrange education and rehabilitation for some children with special needs at each Valteri unit.

Valteri professionals, such as consulting teachers, class teachers and rehabilitation professionals, have considerable competence and experience related to autism spectrum disorders, neuropsychiatric disorders, disorders of language and communication, hearing, vision, mobility and motor functions, neurological diseases or other chronic conditions, and multiple needs. Valteri staff has extensive multi-disciplinary cooperation with schools, municipalities and universities, as well as with various federations and associations for persons with disabilities.

The overall aim of Valteri is to provide equal opportunities for learning and development to all students. According to the role as a specialized center, Valteri supplements municipal and regional educational and habilitation services by offering a wide range of support services. The services may target the needs of individual students or the needs of an entire working community, municipality or region. The form, content and practical implementation of support services vary according to the individual needs. The most common services are counseling and consulting services for day-care centers and schools. These include periods with support for assessment, education and rehabilitation services for children and young people with special educational needs, the production of supplementary material for students with special needs, as well as production of publications and teaching material for professional staff. Valteri also organizes training courses or packages and seminars for professional staff working with students who have special needs.

The services provided for students with JNCL and their teachers and other professionals are offered nationwide from the Valteri centers. According to the Law on Financing Education and Culture, the services from Valteri are free for guardians and partly paid by the education department of the municipality. The rest of the costs are covered by the State.

Valteri-Onerva has appointed a special education teacher who works nationwide as a counselor who is supporting the education of students with JNCL. This person has a close cooperation with central hospitals, the NCL professional group at the Finnish Federation for Visually Impaired and the Finnish NCL Family Association.

To start the pre-school path for a child with special needs and challenges, the family might contact the counselor, or the counselor may contact them. After discussion with the parents and having received their permission, the counselor contacts the rehabilitation worker in the regional hospital, the child's school teacher and the other professionals needed to get a comprehensive picture of the situation and to be able to plan how to proceed. The aim is to build up a supportive multi-disciplinary team for the needs of the child with JNCL.

Support for the pupil, teacher and multidisciplinary team at school level also takes place during the consulting visits. The consulting teacher observes the situation at kindergarten or school class and supports the teachers and others to proceed with the specific needs in education. At the end of the day, the parents, the multi-disciplinary staff and the rehabilitation workers usually have a joint meeting and share the observations of the consulting day. After each visit the consulting teacher writes a summary of the meeting, which is delivered to parents and the multi-disciplinary team around the child.

Valteri-Onerva also organizes in-service training for teachers and teaching assistants working with students with JNCL. The themes addressed and the extent and implementation of the training, as well as the costs, vary according to the type of training.

Special courses for students with JNCL are organized to support their education. These courses are mainly organized at Valteri-Onerva as group sessions for one school week. The students are gathered mainly according to their school grades once or twice a year. Each course includes individual assessment of special needs, such as functional vision, braille skills, computer skills, and mobility and orientation skills. The students are also given individual guidance and practice in adapted study techniques. The course package comprises teaching, learning materials, rehabilitation, leisure time activities, board and lodging. After the course, a multi-disciplinary report with possible recommendations for equipment and educational strategies, is sent to the parents, the school, and often to other parties involved.

According to the individual situation, it is possible to apply for school placement at Valteri-Onerva special school. The responsibility for organizing education rests primarily with the municipality, and negotiation between the local

educational authorities and the family is therefore needed before an application can be sent to the state special school. Valteri-Onerva also requires the child to attend the assessment course before accepting a possible school placement.

Whether services for a student with JNCL and his or her parents and teachers are organized locally or on the national level, it is crucial to focus on the multi-disciplinary work. The Finnish Federation for the Visually Impaired has an NCL working group. This team offers thorough professional support for families in cooperation with schools and rehabilitation units. The Finnish NCL Family Association is also an important part of this cooperation. As a peer support, they provide guidance and meetings for families as well as updated information on their internet pages.

### Denmark: The Danish NCL Team

Denmark has a size of 43,090 square kilometers and a population of 5.8 million (2017). Copenhagen is the capital with 1.3 million inhabitants.

The multidisciplinary NCL Team in Denmark has six members: one social worker, two special education consultants, one doctor and two parents representing NCL Danmark (the Danish NCL Family Association). The multidisciplinary aspect is very important, from the time of the diagnosis. The close collaboration between medical, pedagogical and social counseling services, in conjunction with parents, continues throughout the life span. Parents of individuals with JNCL are valued for their expertise. The multi-disciplinary approach is important because the provision of services is based on observations and ideas informed by insights from many perspectives. The methods applied by the professionals are continuously discussed and elaborated with the team's parent representatives (Spielmeyer-Vogt Team, 2015).

# Organization of team activities

In addition to its ordinary tasks, the NCL Team is concerned with organizational and professional issues. The relationship between professional knowledge and parental expertise is important, as well as the ways to maintain effective and sustainable networks on many levels: the children's network, the parents' network and the professionals' network. Counseling and supervision are provided locally in the municipalities where the children and their families live. The NCL Team emphasizes the importance of establishing a professional network around each child with JNCL, involving local services and parent participation. In Denmark, the municipalities are responsible for the care of people with special needs, including those with rare diseases. The welfare system in Denmark allows parents to choose to keep their child with JNCL (or another form of NCL) either at home or in a public residence. Whatever the choice, the person with JNCL is guaranteed to receive an equal amount of public support. Young adults with JNCL receive a social pension beginning when they reach the age of 18 years. Most young adults with JNCL have their own apartment connected to the parents' home. The municipality provides pedagogical and nursing staff in their homes, as well as in schools and activity centers.

The NCL Team represents a supplement to the services offered by the municipalities and is organized and funded by the government. The team has nation-wide responsibility and is located at Synscenter Refsnæs, a regional center for visual impairment (see also Chapters 24 and 26).

#### The purpose of supervision

The NCL Team offers supervision and guidance to staff in the parental home. The Team also serves staff in schools, residential homes and activity centers. The competence of the local staff related to JNCL differs considerably. Some staff members have never received supervision and guidance while others are more experienced and have received supervision and attended courses.

The starting point for supervision is of particular importance. The nature of the disease can be overwhelming for staff with no experience. Early supervision will focus on possibilities, good teaching and quality of life, rather than on the disease. Early supervision is concerned with building a good culture and awareness of possibilities and appropriate initiatives, including the separation of professional and private feelings. The supervision will support the staff's communication with parents. Staff in parental homes or institutions will sooner or later meet the parent's dilemmas or feelings related to having a child with JNCL. Such situations will require professionalism by the staff and the NCL Team can support the staff in such situations. This is illustrated in the following story:

A staff group at a day care center communicated that they were worried about the contact between a mother and her son. During visits they observed the mother sitting passively beside the son doing nothing. There were no physical or communicative contact between the mother and her son. The staff questioned if the mother had any interests in her son.

The supervisor of the Team asked the staff members if they had discussed the situation openly with the mother and they answered no. The supervisor also asked if there could be other circumstances that should be considered *in relation to their observations. The staff members had never made such considerations.* 

In agreement with the staff, the supervisor from the Team talked with the mother about how she experienced the visits. The mother replied that she often became sad during these visits, because she did not have the same close and good contact as she used to have with her son. The supervisor asked if there were any particular causes for this change. The mother replied that she had been afraid of touching her son, because it might trigger epileptic seizures and anxiety.

The Team organized a meeting with the mother and the staff. It became evident that the mother had a need for guidance from the staff. The meeting resulted in a better and closer relationship between the son and the mother, and she became encouraged to obtain physical contact and communicate with her son as she had done in the past.

After the meeting, the staff became more concerned about how to interpret and meet parents' feelings. They would approach parents instead of making their own speculations. The following question became important: how would you like to be met in a similar situation?

The Team also participates in planning in the later phases of the disease. The purpose of the services is to relieve parents of the burden of practical care, so they can relate to their grief in peace, to the extent that is possible. This service is highly appreciated by the parents. To be a member of the Team entails more than just being a consultant to parents and staff. The relations between the Team and families become close over the years, and participation in difficult situations becomes a mandatory issue.

# Meeting people's emotions

The Team's supervision is based on consideration and reflection. Supervision will focus on the process and understanding of actions, relationships, professionalism and individual responses when a child or young person is affected by losses. The supervision will also address the staff's and the parents' emotions that may affect the work with the person with JNCL. The supervisor's most important task is to act as a catalyst when emotions are affecting the daily work with the individual with JNCL. The Team meets the staff every third month on average, to share their experiences from the daily work. Evaluations show that these meetings are important to maintain the staff's spirit and engagement in the daily work.

# Case story: Learning for Life

This story is written by a mother of a daughter with JNCL named Sofie. The usual length of education in Denmark is 13 years (primary and secondary school), but Sofie was not able to conclude her education in a traditional way.

Our daughter Sofie got problems when attending her 12th year of school because of seizures, bad days and lack of educational content. We, the parents, had to find an alternative educational solution for her. This short story is about this alternative school solution, tailored to meet the needs of our daughter Sofie.

*After some discussion the municipality finally agreed to pay for homebased education for Sofie. This included full time staff (two persons) and miscellaneous costs, such as transportation. The home-based education saved money for the municipality and worries for us. Sofie was given full time support with two assistants from 8 am to 4 pm, Monday to Friday.* 



Sofie and her educational assistant.

*We, the parents, in collaboration with the staff defined the main goals for Sofie's education:* 

- Education and learning should be based on Sofie having a meaningful life with a strong focus on educational themes of importance for her.
- The educational activities should be based on selected themes.
- The school days should be structured, that is, follow a strict program to emphasize predictability.

It was decided to have planning and evaluation meetings every second week to monitor education and create the best possible educational life for Sofie. Examples of selected themes were seasons, animals, favorite food, family and friends, love, the world, outdoor life, religions etc. These themes were converted to educational activities, for example:

- The world theme was converted into specific activities, such as salsa dancing, African music, Thai food and stories about explorers.
- An animal theme was about lions. This theme was converted into activities like going to the Zoo and "seeing" a lion, roaring like a lion, watching and listening to the film The Lion King, smelling a lion, touching a toy lion, and making a lion steak.
- The outdoor life theme was converted into the following educational activities: Visiting the Queen's castle, beach tour, barbeque in the garden, visiting the rose garden, and taking a walk in the forest.
- The religion theme was converted into activities like learning and singing hymns, praying the Lord's Prayer, going to church, commune with crispbread and squash, baking, visiting Camilla's cemetery and eating fish and bread.

One theme with many different activities could last for a week or more. The themes were addressed in a rather fixed schedule to promote best possible predictability as shown in Figure 9.1.

The home-based education worked very well for Sofie. She was engaged from early morning to late afternoon, and she learned a lot. The devoted staff became very familiar with Sofie and her special needs. Sofie's last years of education became a success for both Sofie and us, the parents. 160 | JNCL, childhood dementia and education

Date	12th	13th	14th	15th	16th
	September	September	September	September	September
	Monday	Tuesday	Wednesday	Thursday	Friday
Theme	Theme	Theme	Theme	Theme	Theme
	Africa	Africa	Africa	Africa	Africa
08.00-09.00	Breakfast,	Breakfast,	Breakfast,	Breakfast,	Breakfast,
	plan for the				
	day and song				
09.00-10.00	African music				
10.00-11.00	and dances				
11.00-12.00	Prepare	Prepare	Prepare	Prepare	Prepare
	African lunch				
12.00-12:45	African lunch				
12.45–13.00	Cleaning the kitchen				
13.00-14.00	Tour to Zoo				
14.00–15.00	to watch				
	and listen				
	to African				
	animals	animals	animals	animals	animals
15.00-16.00 Summar	Summary	nary Summary	Summary	Summary	Summary
Compulsory activities	of today's activities and massage				

Figure 9.1 Sofie's school schedule for one week.

### Norway: The structure and function of services

Norway has a size of 385,180 square kilometers and a population of 5.3 million (2017). Oslo is the capital with 674,000 inhabitants.

The public services in Norway are based on a law that says that all citizens have the same right to services, independent of who they are and where they are living (Hesselberg, 2011). However, equality in services is difficult to achieve in a country like Norway because the population is scattered and the number of inhabitants in the municipalities varies from 500 to 674,000 inhabitants.

There are three administrative levels in Norway: The municipalities are responsible for kindergartens, primary and secondary schools and special education, including special education for adults. Health services on the local level include health centers, physiotherapists, general practitioners, community care institutions and school psychology agencies. The counties are responsible for transport, dentists, secondary education with school psychology services, and education in different institutions. The national level, the State, is responsible for specialized health services, centers for rare diseases, special training centers, centers for special education, labor and welfare administration, technical aids and special interest organizations. Persons with JNCL are receiving services from all these levels.

Most children with JNCL in Norway attend mainstream schools. There are very few special schools in Norway and no special school for students with visual impairment. Some mainstream schools have special units for students with special needs, which may include for example children with intellectual disabilities, severe motor impairment or autism spectrum disorders. Adults with special needs may be provided with special education after assessment, recommendations and case considerations by the local or regional educational authorities. This right to education is not limited in time or age.

The developmental course of JNCL creates challenges for the health and education department of the municipalities when providing education and other interventions. The local professionals will meet a person with a rare progressive disease they have never heard about. Life-long changing needs will require commitment and collaboration between the parents, the schools and other public institutions. This collaboration is particularly challenging because it cannot be based on a standard system but must be based on a person-centered approach (see Chapter 11).

The services have to adapt to the needs of persons with JNCL, which are increasing with age, and there are considerable differences between persons in this group. Different institutions are responsible for providing the services and competence building needed to support persons with JNCL. The local and specialized services must be coordinated, and there is usually a formal agreement on responsibilities and cooperation. There is a need for support and guidance for those who are close to the person with JNCL, but also for other people in the environment.

# The workshop model

All services must be based on sufficient and relevant knowledge. Knowledge about JNCL is limited in general and is not usually found among professionals working in the municipality or in county-level institutions in Norway. Building up local knowledge and competence is a core issue in provision of educational and other services for individuals with JNCL.

A person-centered approach requires two types of insights: knowledge about the person with JNCL and knowledge about JNCL. Knowledge about JNCL is mainly found on a national level, in specialized centers or resource centers. Some of these institutions are providing outreach services, such as Statped.

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Knowledge about the person is well represented locally, in the family, the school, and sometimes within local or regional educational counseling services. Statped Midt in Trondheim has specialized competence related to education of students with JNCL and is usually responsible for building up the necessary local competence when a new child is diagnosed with JNCL. Statped midt cooperates with the family, the local school and pedagogical-psychological services, and other local or regional services. It is necessary to establish a close and secure relationship and efficient ways of communication, tailored to the needs of the child or young person with JNCL. Figure 9.2 describes a workshop model for collaboration that has proved useful in many cases.

There is always a risk that the challenges related to JNCL can seem overwhelming for staff receiving competence-building services. The workshop model allows the inclusion of bottom-up strategies where competence building and guidance to some extent is based on everyday situations, situations that are successful as well as with negative experiences. The workshop model and the bottom-up strategy invite the participants to set the agenda of the workshop and address topics that are important to them.



Figure 9.2 The workshop model for person-centered collaboration

Experiences with applying this model indicate that the specialized professionals need to spend some time in the school and the home prior to the workshop. This provides an additional perspective as the JNCL specialist may observe and understand things that people with less experience with the disease are not likely to see. Further, the specialist observing the child or young person with JNCL in familiar surroundings and in different situations may get a more accurate understanding of the issues that are going to be discussed in the workshop.

The workshop usually takes one working day. If a longer workshop is planned, it may be difficult for the family to find time and for organizers to get all the relevant professionals together. Leaders and administrators may not be able to attend the whole workshop but are often able to come for the summing up and allocation of responsibilities. A written report with conclusions and recommendations is always provided by the JNCL counselor after the workshop.

In addition to workshops, Statped Midt organizes 3- to 4-day centerbased courses for teachers, other staff and parents. An advantage of attending center-based courses is the possibility of meeting specialists who would not come to local workshops, and of meeting other professionals who are working with other individuals with JNCL. However, the attendance of center-based courses is often limited, possibly due to the expenses they represent for the municipalities.

The one-day local workshops seem more manageable for schools and other institutions. They are often attended by ten or more participants: teachers, residential staff, assistants and parents. A workshop thus reaches a substantial group of the people around the person with JNCL. Workshops are usually managed by one or two professionals who have specialized on JNCL. An important argument for local competence building is the fact that one can concentrate on the issues related to just one individual, which is the foundation of the person-centered approach. The courses have a more general content and may not meet the knowledge needs of all the parents and professionals who attend the course.

The Norwegian experiences with applying the workshop model are good. However, the model requires competent counselors who can meet the local needs, which may vary considerably with the person's age, development and declines, as well as interests and personality. It takes time to build such competence and it requires access to the diverse and various fields of specialization that are involved in the education and habilitation of this group. There is at present no formal education specializing in the educational needs of persons with JNCL. The Norwegian national educational competence on JNCL is built through in-service training.

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# Assessment and Evaluation

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E valuation is an ordinary part of all education, in the form of grades or qualitative feedback. The assumption is that feedback helps students understand what they need to learn and guides them toward learning strategies that may support their learning (Baird, Andrich, Hopfenbeck, & Stobart, 2017; William, 2018). Assessment goes beyond an evaluation of the student's actual school work and is to a larger extent a help to those who are responsible for the education. An assessment gathers information about how the student understands, thinks, feels and acts in relevant areas, and on the characteristics of the physical and social environment, including relations within the family and with peers and adults outside the family. The information should indicate whether an individual has difficulties in areas requiring treatment, special education or other measures, or if changes need to be made in the individual's environment. The function of assessment is thus to establish a foundation for making decisions about intervention, including the decision that intervention is not necessary.

Assessment is a process that is usually initiated on the basis of a concern and a referral, and these will guide the assessment. The referral identifies the concern; the function of the assessment is to find out *what* the problems might be and *why* the child is showing these problems. When the diagnosis of the child or young person is known, knowledge about the typical course and variation in symptoms within the diagnosis will guide what domains to assess. In addition, assessment will build on the strengths and weaknesses observed in earlier assessments. The developmental and learning histories of the individual give information about the problems encountered by the individual and how the interventions functioned, including, for example, to what extent they led to increased knowledge and independence, or enabled a student with declines to continue doing things with help. They also inform about growth and decline in various domains, which may be helpful in adapting new intervention measures to the individual's present functioning and assumed functioning in a near and more distant future.

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Although children and young people with JNCL share many of the same difficulties, there are also significant differences between their developmental patterns. For example, not all individuals show a full range of known JNCL problems, and the symptoms are not manifested at particular ages or in set order. This diverse profile is evident in case presentations discussed in many chapters of this book. Thus, to regard individuals with JNCL as a homogeneous group would mask important differences in their possibilities, limitations, their coping mechanisms, and their respective need for support and environmental adaptations. Assessment is therefore essential for capturing these important differences so they can be used to customize shorter and longer term treatments and life plans even if the overall long term course of the disease is the same.

A review of assessment in general is beyond the present chapter (see for example Brassard & Boehm, 2007; Crick, 2007; Dombrowski, 2015; Sattler, 2008a,b; Williams & Hill, 2012). Instead, this chapter describes only the basic assessment strategies and processes relevant to the assessment of individuals with JNCL. It also presents a checklist for observing developmental progress and decline in these individuals the *Educational Development Observation Tool (EDO)*, which has been designed as part of the JNCL and Education Project.

#### Assessment strategies and processes and JNCL

Assessment may include direct and indirect measures. Direct measures come from tests and observations while indirect measures are based on information from structured conversations or standardized interviews with parents, teachers or others about the student's functioning in specific areas. The conversations provide important information about the student and the family but also contribute to making professionals, teachers and parents acquainted with one another and to establish an *alliance*, that is, a shared understanding of the student's strengths and weaknesses and the types of measures that may be useful. The conversations also provide information about the student's physical and social surroundings, about the parents' and the teachers' opinions about earlier and present intervention measures, and about resources and deficits in the student's environment. Each profession has its instruments and strategies for assessment. Feedback to parents and teachers from assessments and observations is also typically mediated in a conversation (Hesselberg & von Tetzchner, 2016).

In standardized interviews, there is a tendency for the professional to ask all the questions, and indeed to set the agenda through their prechosen question set, and for the parents' role to be relegated to answering those predefined questions. Structured and semi-structured conversations are more open, thus allowing and encouraging both parents and professionals to introduce topics freely, according to how the topics arise in the conversation. A guiding principle for conducting both conversations and interviews is the importance of creating an open and secure situation for all participants. This principle is particularly important when the parents know more than the professionals, which is often the case with rare disorders like JNCL (see Chapters 24 and 26).

An observation procedure provides a goal-oriented perception of what one or several individuals are doing. Observations conducted in the home or at school can give the professional an impression of the student's functioning in ordinary situations, including levels of attention, performance, effort, social functioning and participation. It can be useful to observe the student in both structured and unstructured situations, and together with both peers and adults. There are some checklists with predesigned categories for recording children's play and activities (Barton, 2010; Kelly-Vance, Ryalls, & Gill-Glover, 2002), but these may not be valid when the child has a disability (e.g., blindness or motor impairment) that interferes with performance in ordinary activities.

If testing is part of the assessment, then the tests must be appropriate for the level of the individual, and should be evidence-based (Hunsley & Mash, 2007; Bruce, Luckner, & Ferrell, 2018). Moreover, the validity of the instruments used in the assessment of a child or adult with a diagnosed or suspected disorder must be documented for that particular diagnostic group. In fact, the use of assessment instruments that are not appropriate for the person who is assessed is considered a major breach of professional ethics in school psychology (Leach & Oakland, 2007; Mendes, Nascimento, Abreu-Lima, & Almeida, 2016).

Although tests are designed to be suitable for most individuals, it may be necessary to adapt the test situation and the material being used to the perceptual and physical abilities of the individual (Zebehazy, Zimmerman, & Zigmond, 2012). Assessments of individuals with severe visual impairments are often based only on the verbal part of the test. Tasks consisting of images with a missing item, categorization based on shape and colors, puzzles and checkerboard patterns are unsuitable for children with poor vision. Many tasks used to measure language comprehension require ability to follow instructions that involve locating items on a table or in the immediate surroundings. These tasks also represent challenges for individuals who are blind. Some individuals with JNCL have motor problems, which will influence their ability to follow instructions that require motor performance. Moreover, problem-solving will vary with the familiarity of the material (Wason, 1977). A test with material that is very unfamiliar to the individual may be less useful to measure the individual's skills than tasks with more familiar material (see also Chapter 12). The problems related to test assessment of children with visual impairment are expressed strongly by Erin and Koenig (1997): «The use of standardized tests with students who have visual disabilities is fraught with complex problems. Even with stringent and thorough adaptation, such tests alone rarely provide sufficient information for making important decisions about students with visual disabilities» (p. 311). However, there are also tests with non-visual non-verbal tasks for assessment of children with visual impairments (Smith & Amato, 2012; Zebehazy et al., 2012).

Any special arrangements and adjustments that are made during testing must be taken into account when interpreting the results. When testing takes place under specially tailored conditions, the norms may merely provide a guideline, since they are usually based on a representative group of children with typical development. Children with atypical development are rarely included in norm-referenced tests. Consequently, the norms may not apply and should be used with caution; nonetheless, they may still be useful for optimizing the children's learning situation.

Norm-referenced tests are used for ranking individuals and comparing them to their peer group. When it has been established that performance is much lower than expected for the age, the norms serve little purpose. The test scores in themselves are less important than the implications they may have for educational strategies (Lichtenberger, Mathen, Kaufman, & Kaufman, 2009). Criterionreferenced tests are used to assess what an individual can and cannot manage, with the aim of establishing what kind of training he or she needs to acquire the missing skills. Criterion-referenced test results thus indicate how closely the child is to skill mastery in some domain, instead of how the child compares to the peers. For students with JNCL, criterion-referenced tests may be useful in assessing degree of decline and amount of help needed.

Traditional use of tests is often referred to as "static", because the tests measure knowledge and skills at a particular point in time. By contrast, from a social constructivist perspective on cognition, assessment should record not only which tasks the child does and does not master (i.e., static appraisal), but also the learning process itself. Thus, from this perspective, assessment must be dynamic; dynamic assessment implies identifying the child's zone of proximal development – what the child is able to do with help, and how much help is needed to solve a given task (Jeltova, Birney, Fredine, Jarvin, Sternberg, & Grigorenko, 2007; Lidz, 1997). Dynamic assessment does not merely measure the individual's skills at a given time but what the individual can do with help and learn under standard conditions; thus, norms must be used with caution. Static assessment a way to assess independent function, and dynamic assessment a way to assess interdependent function and the kind of and amount of help that are needed, thus assessing the zone of developmental maintenance (see Chapters 2 and 16).

Cognitive assessment should never rely on tests alone. According to Erin and Koenig (1997), «The results from an assessment, whether standardized or

not, must never be the sole source of information on which to base any important educational decision» (p. 313). Test results should be collated with observations of how the individual copes with familiar and unfamiliar everyday situations. The teacher's observation may be useful but there are some limitations. For example, due to the low incidence of severe visual impairment in school-age children, teachers and other professionals in mainstream schools often feel inadequate in evaluating students with visual impairments (Loftin, 1997). Observations of the student in the classroom and other familiar surroundings may be useful in seeking to assess possible cognitive decline or dementia. However, observations are time-consuming and a short observation may not provide a full impression of the variation in the individual's functioning. A checklist may function quite well and take less time.

Checklists comprise questions about skills, behavior and everyday functioning, and have become a common element in assessment. They are usually completed by parents, teachers and other adults, but checklists can also be self-completed by older children and adolescents. A checklist can also provide a good basis for conversations about the child's functioning. In particular, in relation to behavior, there are often significant differences between the answers provided respectively by the parents, teachers, and the child or adolescent (De Los Reyes & Kazdin, 2005; Rescorla et al., 2014). However, it is important that parents should not be responsible for evaluating their child's development alone – they see the child through their special "parent glasses" and even the glasses of the mother and the father can be quite different (Seifer, 2002). Checklists can be part of the parent interview and contribute to a dialogue about the abilities of the child and the features of the environment (Rosenbaum & Valsiner, 2011).

In addition, checklists are used by professionals to structure their own observations. An advantage with checklists is that they give information about the everyday functioning of the individual, but like tests, it is important that they concern actions, skills and activities within domains that are relevant and categories that are appropriate for the person. Checklists may be a useful approach in assessing individuals with JNCL provided they are suitable for individuals who are showing visual and cognitive decline. The Educational Development Observation Tool (EDO) is especially designed for this group (see below).

Giving tests and completing checklists represent only the first stage in the assessment process. The main task is to analyze the results, describe functional profiles with strengths and weaknesses, and compare test results, observations and information from interviews and conversations. For example, the declines in language and cognition do not always start at the same point in time. Some children with JNCL develop speech problems (i.e., hypokinetic dysarthria) early while their cognitive functions, including linguistic skills underlying expressive and receptive language remain seemingly unchanged for some time. For other children with JNCL, it is the other way around, they have problems with language while their speech is clearly intelligible. This implies that non-verbal cognition, expressive and receptive language, and motor speech functioning must be evaluated separately. When cognition is compromised, there will be difficulties with receptive and expressive language (i.e., the symbolic code, vocabulary, ability to understand ideas in complete sentences, etc.), but speech itself need not be affected. Alternatively, problems with motor speech production can mask an adequate language system. Problems with articulating speech should therefore not be interpreted as an indication of cognitive problems or decreased language comprehension. An individual with hypokinetic dysarthria might also simplify how ideas are put into words, in the interests of being intelligible to others. This simplification process can make it appear that the speaker lacks age-appropriate vocabulary or grammar, when in reality, the simplification is strategic, thus reflecting adequate cognition and judgment.

It is important to compare the individual's strengths and weaknesses as they emerge in a structured test situation with the individual's functioning in structured and unstructured activities at home, at school and in less familiar places. Dynamic assessment strategies may shed light on the processes of decline, thereby supplementing other observations used to assess the amount of help the individual needs to cope in various situations.

Educational initiatives for individuals with JNCL must build on an alternative principle compared to more traditional initiatives within special needs education, because decline, not progress, is expected over time. Standard evaluations focus on



#### AGE IN YEARS

Figure 10.1 Hypothetical development with and without proactive intervention

successive improvements in personal performance and growth. This focus is also important for individuals with JNCL, but evaluation must be based on the extended perspective. Having JNCL will always imply declines and losses affecting the individual's level of performance and functioning. Results from the present JNCL project (Appendix A) indicate that appropriate educational initiatives may delay the onset of the descending curve on functions and slow down the rate of decline, thus leading to longer maintenance of functions and skills. The developmental trajectories in Figure 10.1 illustrate this extended perspective. The figure shows a hypothetical difference between a person with JNCL's growth and decline with and without proactive or preventive measures. The lower line illustrates growth and decline with no proactive measures while the upper line shows growth and maintenance with proactive measures.

# The Educational Development Observation Tool (EDO)

The Educational Development Observation Tool (EDO) is designed to give an overview of eleven central themes or domains that should always be considered in planning for persons with JNCL. The EDO is a tool that can be used for long- and short-term assessments and interventions, and as a background to facilitate the development of the Individual Education Plan (IEP) and the Habilitation Plan (see Chapter 11). The central themes identified in the EDO are vision, communication, literacy, social life, gross motor function, fine motor function, physical activity, behavior, memory and attention, independence and autonomy, interests and equipment. These themes can be met by different interventions. Some themes included in the EDO, such as expressive communication are addressed from a long-term perspective, because the use of speech and written communication will decline sooner or later; other domains must be considered in the short term, and the EDO helps identify what can be done now to meet known future challenges.

The EDO tool is tailored for individuals with JNCL and includes assessment domains and suggestions for interventions based on the assessment findings. The tool focuses on individual functions and environmental factors. Parts of the tool can be used for other groups with disabilities, for instance for children with other diseases involving dementia. The main aim of the EDO is not mapping declines or symptoms but identifying the best possible interventions to optimize quality of life despite all challenges. The EDO's three broad objectives are (a) to support educators, social workers, residential staff, parents and others in achieving the best possible quality of life for individuals with JNCL, (b) to identify interventions to meet current or future challenges, and (c) to create a focus on possibilities instead of barriers. The EDO should be used to support development, learning, maintenance and teaching in and beyond the classroom, and within and beyond school age.

The EDO has been developed as a part of the *JNCL and Education Project* (see Appendix A). The body of experiences obtained from the use of the EDO was limited when this book was published, and it is assumed that the EDO will undergo further development and refinement in the coming years.

#### Use of the EDO

The EDO is an interactive document. It is extensive in size, but rather timeefficient to administer when one knows how to use it. It is recommended that it be used annually as a preparation for the next operational year, such as the next school year. It is particularly important to use the EDO prior to major transitions, such as transitions between schools, or the transition into adult living.

The tool should be completed by the parents together with a team composed of educationalists, social workers, JNCL specialists and possibly others who know the person well. The time constraints under which parents and professionals operate are important considerations. Thus, the JNCL specialist should have sufficient knowledge about the EDO and be proficient in its administration, including being able to complete the tool efficiently, ensuring that the assessment does not take more time than necessary.

The assessment part should be based on the person's current optimal functioning, that is, when the person with JNCL has a "good day" in an optimal context. However, it is also important to know how the person functions during periods when he or she is affected by seizures or a negative mood, or is in unfamiliar or nonpreferred settings.

Some questions or topics of the EDO may not be considered relevant at all times, depending on the person's current situation. The decision to leave out topics or questions should be made in close collaboration with the JNCL specialist. Another important role of the JNCL specialist is to ensure that the team consider early educational interventions that are not currently obvious, but that might be beneficial in the future. The JNCL specialist should provide guidance about expected future symptoms, and how to prepare the person with the JNCL disease to meet these symptoms in best possible way.

JNCL includes a series of declines in cognition, mobility, vision, communication and independence. Educational interventions cannot stop the declines, but education can optimize interventions and compensate for some losses and problems caused by the declines. Children, adolescents and young adults with JNCL should – for as long as this is possible – follow the ordinary curricula and

participate in everyday, socially appropriate activities even if these are not clearly illustrated in the EDO. The EDO focuses on educational areas that are specific to individuals with JNCL and that will require interventions that are not applied in the education of the typically developing population. The EDO is based on educational strategies and principles described earlier in this chapter and other chapters of the book, such as hastened learning, proactive learning, skill-oriented learning, participation-based learning and life flow.

# A brief introduction of the content

The EDO has 12 parts or domains of particular importance for individuals with JNCL. All parts are important, but all questions may not be important at all times. The questions or topics in each part should give a picture of the person's present situation (the observations) followed by an analysis of the consequences for education and non-medical service provision (the interventions). The questions may sometimes be difficult to answer but should still be elaborated as well as is possible. An overall objective when using the EDO is to ensure that persons with JNCL will receive appropriate services and interventions at the right time.

*Part One: Demographic information.* The team collects personal details such as name, age, where the person lives, school and who participated in the completion of the EDO.

Part Two: Vision. The vision of children with JNCL is normally assessed by ophthalmologists. This part gives a brief description of the visual function that may be of importance for non-medical interventions. The change from being a person with functional vision to become a person who is blind is a difficult transition. Such concerns as losses (e.g., friends, favorite activities) and the growing need to complete tasks in new ways must be considered by educational planners. It may also be desirable to prepare the child for the forthcoming visual impairment by using pro-active learning in certain areas. The onset of visual impairment is typically evident when the children receive the JNCL diagnosis. Visual impairment may imply the selection of some new goals and new adaptations. With the onset of blindness, more goals may need to be revised, and optimization and compensation are likely to be comprehensive and wide-ranging, and hence requiring insight into both blindness and the other features of JNCL. Preparation for blindness implies making the student's surroundings blind-friendly, by removing hazards, marking important places, and providing the environment with tactile landmarks, shorelines and so forth (see Chapters 4 and 16).

*Part Three: Communication.* The JNCL disease will – eventually – include severe problems with speech. The person's language comprehension will often remain better than the speech intelligibility. Education should consider how to

support current or forthcoming speech problems by using enhanced and proactive learning. Both speech and language therapy and the use of alternative and augmentative communication forms (AAC) should be discussed with a JNCL specialist in an early phase of the disease, even if the student's speech at this time is functioning well in all situations (see Chapters 6 and 13). However, there is a need to collect more evidence and build competence from systematic training of AAC for students with JNCL. This is an important area where future research is needed.

*Part Four: Literacy.* Reading and writing are pivotal skills. Most children with JNCL will at first learn reading and writing through the visual modality. Many of them are eventually offered learning of tactile reading and writing. Delaying the introduction of tactile reading and writing may make learning difficult due to the concurrent, progressive cognitive decline. It is advisable to consider tactile reading and writing when the child's learning capacity is at its best, even if the child's sight still is sufficient for visual reading and writing. It is recommended that decisions about reading and writing instruction are made in close collaboration with JNCL specialists (see Chapter 14).

*Part Five: Social life.* Social life is of great importance for everyone, but it can be affected negatively by the many symptoms of JNCL (e.g., declines in vision, communication and motor skills; and possible changes in mood and behavior). Problems will, to some extent, reflect shortcomings in the environment and may be reduced when environmental adaptations are made to meet the person's social and learning needs. The ability to participate in social settings can be facilitated by inclusive interventions, by using the interactive team model, by providing support and other environmental measures (see Chapter 22).

*Part Six: Gross motor function.* All persons with JNCL will be affected by gross motor declines. The motor declines may restrict participation in certain life situations if not met by appropriate interventions. Interventions (e.g., physical adaptations in the environment, provision of supporting materials, technical aids etc.) will reduce the effects of the declines. Anticipating forthcoming motor declines is the best way to ensure that each person with JNCL can achieve a good outcome (see Chapters 7 and 15).

*Part Seven: Fine motor function.* Because persons with JNCL become blind, they come to rely upon use of their fingers to explore the world. Fine motor coordination is a necessary skill for performing and participating in every-day activities such as eating, cooking, reading, and so forth. In the early phases of disease, while children are still capable of learning and mastering new skills, fine motor skills should be trained and developed. As the disease progresses, fine motor functions should be maintained to highest possible extent, and limitations should be addressed through the provision of customized task adaptations.

*Part Eight: Physical activity.* Motor problems and visual impairment will affect options for being physically active. Physical fitness is therefore an important goal (see Chapters 7 and 15). Being physically active is of special importance for individuals with the JNCL disease because through such activity, overall functioning can be preserved and declines mitigated. It is desirable that persons with JNCL are physically active every day. Physical activity includes participating in sports and dancing, and it can be incorporated into daily routines such as walking to school. Physical activity should be supplemented with physiotherapy when needed. It is important to be in good shape to meet future motor declines. The IEP and Habilitation Plan should include a training program and regular activities that involve physical performance that is adapted to the motor functioning of the person.

*Part Nine: Behavior, anxiety and mood.* Behavior problems, worries, anxiety, problems with mood and so forth are known challenges for some, but by no means all individuals with JNCL (see Chapter 27). These problems often occur after the onset of severe visual impairment and in combination with dementia. Challenging behaviors, worries and anxiety can be difficult to handle and may in turn affect learning and interaction with peers. Such conditions may subside if appropriate measures are taken. The IEP and Habilitation plans should include plans for environmental adaptations as well as appropriate reactions from family and staff.

Part Ten: Attention and memory. Attention and memory are two vital components for learning and performance. All individuals with JNCL will sooner or later have problems with their attention span and the ability to remember new things. Long-term memory is considered to be an important resource for persons with JNCL, whereas working memory is more affected by the disease. Thus, there is a need to address such problems with educational strategies that are appropriate at each phase of the disease (see Chapter 12); moreover, it is important that teachers and educational staff are alerted to potential for memory problems and receive the training needed for implementing useful strategies as the need arises. Examples of interventions include: providing enough repetition, structuring, and clear information; asking closed-set questions instead of open-ended questions; and implementing memory lists.

*Part Eleven: Independence and autonomy.* In their early years, persons with JNCL are just as independent as their peers. The declines occurring later will decrease their ability to act independently. Developing and maintaining independence should always be considered within education planning whenever possible. Autonomy must be reconceptualized from the perspective of what it means to maintain some sort of autonomy in the face of the many and inevitable functional declines. The interactive team model (see Chapter 16) provides a suitable framework within which a team should successively compensate for the inevitable

decrease of independence caused by different declines. This model, illustrated in the EDO, focuses on what the person can participate in when supported by a helper. Early planning and preparedness are important for maintaining independence by teaching new behaviors and making adaptations in the environment (see Chapter 16). The planned interventions should facilitate an understanding of the environment that is based on earlier knowledge and available sensory information.

*Part Twelve: Interests and equipment.* This part can be used to identify activities important for the person's quality of life and ensuring best possible life flow. The part can also be used to specify equipment that has a positive impact on the person's life situation.

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# Planning Education and Intervention for Individuals with Juvenile Neuronal Ceroid Lipofuscinosis (JNCL)

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A n essential element for developing and sustaining modern societies is the existence of a system of education for all. Schools are important societal institutions for giving children and young people the knowledge they need, and in accordance with the needs of society (Ballantine, Hammack, & Stuber, 2017; Hesselberg & von Tetzchner, 2016). All modern societies therefore have both an organizational structure for education and general plans for all levels of education (i.e., primary, secondary, colleges and universities). During early school years, most individuals attend the local primary school where the curriculum has little scope for personal choice, and any choices tend to be made by the parents. In higher school grades and university, there are more opportunities for individual choice among the courses offered, and because the children are getting older, they are given more latitude in making their own choices.

While a large majority of children and adolescents follows the ordinary or mainstream curriculum, some students and young people have disorders and disabilities that necessitate a more individualized educational course. However, like other students they also build knowledge and skills, although usually at a slower pace or in atypical ways.

The educational situation is very different for students with JNCL. While their knowledge and skills in the first years increase more or less similarly to their peers, the perceptual, motor and cognitive declines will gradually reduce their ability to search for, process and convey information. Their understanding of the physical and social world, their own needs and their current and future possibilities will gradually decrease (see Chapters 4–7). They will need more help in communication and everyday functioning. They become dependent on extensive individualized planning and less able to actively participate in the planning of their own life content and maintain their personal autonomy (see Chapter 16 on interdependence). The family and service providers will have to make the most important decisions and assume planning responsibility. 180 | JNCL, childhood dementia and education

Educational planning is concerned largely with preparation of appropriate content and organization. Making plans for the educational course of a person with JNCL requires comprehensive knowledge about the typical course of the disease, the individual variation (see Chapter 2) and the person's developmental history and current functioning. Selecting goals and making adaptations require thorough knowledge about the person's strengths and weaknesses, needs, interests and wishes. Planning should be based on assessment of all relevant domains (see Chapter 10). Development within each of these domains may follow its own course and may vary among individuals with INCL. Each domain should be assessed separately: the person's functioning in one domain cannot be used to make assumptions about the person's functioning in other domains. For example, speech problems should not be used as an indicator of language comprehension or cognitive functioning. However, consideration of the person's needs must account for all domains individually and collectively. The educational plan will include activities that optimize the individual goals and interventions that ensure the necessary compensations to achieve the selected goals (see Chapters 2 and 12). Predicting the exact outcome of the interventions is never possible, and although plans should be based on a long-term aim, there may be frequent revisions of the plans and the goals that have been selected.

#### Person-centered planning

Students with typical development adapt and make their choices in accordance with the standard educational system. This implies that they have to adapt to *system-centered* plans, and the educational challenge is how to facilitate this process. Students with severe and complex disabilities are unable to adapt to the standard system, they need person-centered educational measures and interventions, a system that adapts to their needs (Meadan, Shelden, Appel, & DeGrazia, 2010; O'Brien & O'Brien, 1999). This in turn requires a person-centered approach. Person-centered planning has a different goal than standard planning. The basic question is what the system or society can do for the individual with a disability or disorder to obtain best possible learning outcome and secure the individual's rights and possibilities. A person-centered plan should be *proactive* and *realistic* in form. Choice of personal goals and interventions should lead to an optimal outcome within a realistic framework. The plan should optimize current and future possibilities of development and growth and be of help to avoid unnecessary problems and barriers at present and in the future. This might lead to some strain on the existing system, due to the needs for an individual-oriented organization, special competence or other resources, which might imply extra-ordinary costs.

Person-centered planning requires close collaboration between different agents who together have the knowledge required for making an optimal plan. Person-centered planning requires detailed knowledge about the person, which means that the family – and the person himself to the extent possible – will have a leading role in the planning process. Together, the family and the professionals representing the different agencies constitute a planning or responsibility group. It is a group rather than a team because parents are part of it. Here, it is called *a Responsibility Group* because the main task of the group is to allocate responsibility for the different areas to agencies or persons, thereby ensuring that it is clear who should do each of the tasks that have been decided by the group. If successful, the person-centered planning process will promote team building and cooperation. When the plan is completed, all the members should know their tasks and responsibilities.

In the present context, there are two types of person-centered plans that are relevant for people with JNCL: habilitation plans and education plans. Habilitation plans are usually comprehensive and include many life domains with a long-term perspective, often several years. Educational plans concern education and school life in a broad sense and usually have a one-year time perspective.

## **Habilitation Plans**

The long-term plans for people with disabilities have been given different names, such as the Habilitation Plan, the Rehabilitation Plan, the Individual Plan or Individual Care Plan. In this context, the term Habilitation Plan will be used. The Habilitation Plan is mainly intended for people who have complex and comprehensive needs requiring support and adaptation in many life domains. The use of the Habilitation Plan is recommended by the authorities in many countries, and is a legal right in some countries, like in Norway where the habilitation plan currently is named Individual Plan (Norwegian Directorate of Health, 2018):

Anyone in need of long-term and coordinated health and care services is entitled to have an Individual Plan. The Plan should be prepared on the request from the person or his/her guardians. The local government service authority is responsible for developing the Plan.

The Individual Education Plan may be an integrated element of the Habilitation Plan. However, the Habilitation Plan is a holistic plan that goes beyond education, covering all relevant aspects of the individual's life related to social, educational, behavioral, recreational, residential, vocational, and medical needs. It is a written plan of needs, interventions and actions, which outlines the goals and objectives selected by the individual, the family or the Responsibility Group, as well as the adaptations and compensations needed to achieve these goals and objectives (Baltes, 1997).

The Habilitation Plan may build on a positive and realizable vision of the person's functioning and achievements some years into the future, constructed and agreed upon by the members of the Responsibility Group. The Habilitation Plan lists the actions needed to fulfil the future vision described in the plan. These may include more short-term and detailed plans for the coming year (Hesselberg & von Tetzchner, 2008; Jeglinsky, Brogren Carlberg, & Autti-Rämö, 2014; King & Meyer, 2005).

The Responsibility Group should meet on a regular basis to evaluate existing goals and interventions, revise, reformulate or abandon long-term goals and decide on necessary changes to adaptations and interventions. An important function of the Habilitation Plan is to coordinate intervention efforts and ensure collaboration between different parties. The Habilitation Plan is person-centered and should function to empower the person and the family and ensure that the interests of the person are in the center of the discussion when goals are defined and selected.

The main purpose of the legislation related to the Individual Plan in Norway is to ensure that persons with social, psychosocial or medical needs are offered coordinated habilitation and rehabilitation services that can promote health, functioning, coping, participation and equality. Another purpose is to contribute to strengthen the interaction between service providers, service receivers and relatives. According to the authorities, the individual's own wishes should be emphasized. However, the personal goals and interventions described in the Individual Plan are not legally binding for the authorities.

The effect of a Habilitation Plan depends on the implementation of the measures that have been agreed upon. Habilitation Plans are sometimes criticized for being a mere formality with no real impact on the person's services. In spite of good intentions, the plan may be put in a drawer and forgotten, or the goals may be lost in the stress of managing everyday routine tasks. In the present study (Appendix A), some parents found that the habilitation plans functioned more as directives from the authorities than as a tool to promote this individual's goals:

The authorities have prepared a standard outline of the plan with significant limitations regarding content. It makes the plan more or less useless for us, the plan does not cater for our daughter's comprehensive needs. The plan is not the voice of our daughter, it is more a directive or overview of what we can or cannot expect from the society today and in future. Actually, this quotation shows a violation of the intentions behind habilitation plans. The Habilitation Plan is supposed to be an overall plan to promote the best possible development, life situation and services for the person. It should include measures for making physical adaptations in the new environment, building necessary staff competence, and ensuring continuation in activities that are significant for the person and the social network. A Habilitation Plan should always have a calendar function for describing planned and achieved actions on a time line, when an initiative was going to take place, who was responsible for initiating and coordinating the initiatives or actions, and when the goals were achieved or revised.

# **Planning transitions**

A transition may be defined as an important change in education, school setting or other life domains. The Habilitation Plan is an essential tool for planning transitions, for example from preschool to primary school, from primary to secondary school, or from attending school to attending a vocational activity center in emergent adulthood.

In some countries, students with JNCL are attending mainstream schools through all grades, although they may spend an increasing part of their time in smaller groups or special units. For students with JNCL attending special schools, there may be fewer transitions, as many special schools cover the full or most of the educational course. Students in mainstream schools therefore tend to be more affected by transitions than students in special schools. In some countries, students with JNCL first attend mainstream schools, then change to special schools for students with visual impairment, and finally move to schools for students with comprehensive needs (see Chapter 9). These transitions usually involve changes of teachers, staff, locations, classmates and sometimes changes of boarding facilities.

Some transitions may imply new rights or withdrawal of rights, for instance rights related to special needs education. The transition from school to not being part of an educational system in emergent adulthood (see Chapter 2) is a major life event for all persons with JNCL, independent of country and educational system. Education is a major structural element in all young people's life and usually has an even more central role for young persons with JNCL. In a successful transition, the role of the school must be replaced with sheltered workshops or similar, and planning and implementing this change is an important task for the Responsibility Group (see Chapter 23).

For students with typical development, a transition may be a positive experience, with new friends, new challenges, new teachers and maybe new opinions of the student's proficiencies. The situation may also be positive for students with disabilities, but for many students with learning problems, and for those with JNCL in particular, a transition can be challenging. The competence of the individual and the staff's knowledge of goals and future needs may have to be rebuilt. Planning of transitions is therefore essential. Transitions are complex and if not thoroughly prepared, they may end in chaos and despair. The *educational flow* (se Chapter 16) may stop if competence and necessary compensatory equipment are not in place when students move to a new school. Students may experience a long time with restricted learning opportunities and loss of meaningful activities. Transitions should be thoroughly prepared and described in the Habilitation Plan because this plan has a long-term perspective. However, a separate short-term plan of action for the transition itself is often needed. The main objective of such a plan is to ensure continuity in learning and functioning, and to prepare the next school or service providers for meeting the individual with special needs in best possible manner (Elmerskog & Fosse, 2012). Moreover, transitions may involve domains other than education, such as health, social participation and activities outside school. A Habilitation Plan can also include needs that are not likely to be relevant in the short term but will be important in the future. For this reason, a Habilitation Plan should cover a period beyond the next transition.

#### Individual Education Plans

The Individual Education Plan (IEP) is a tool for planning and implementing education. An IEP is needed when the mainstream curriculum is not sufficient to meet the individual's educational needs (Mitchell, Morton, & Hornby, 2010). It typically has a time horizon of one year, meaning that it should be evaluated and revised at least once every year. However, the IEP should be in line with the goals and objectives described in the long-term Habilitation Plan.

The IEP is person-centered as it defines the educational needs and priorities for a single student (Keyes & Owens-Johnson, 2003). Students with learning disorders and disabilities are candidates for an IEP and many countries require an IEP when a student needs special education services. The IEP should address the student's learning challenges and needs, strengths and weaknesses, and include a specification of educational goals. It is a working document that may identify teaching goals and methods that are modified from the age-appropriate curriculum, or that represents an alternative educational content. It is a tool for the student, the parents, the school and all who are supporting the student to achieve the defined goals. An IEP should have clear and detailed descriptions of short-term goals, objectives, methods and resources needed. The right to have an IEP is legally binding in most western countries. It is strongly recommended that parents are involved in preparing the IEP (Andreasson & Carlsson, 2013; MacLeod, Causton, Radel, & Radel, 2017).

Selection of goals is a core element in Individual Education Plans. The plans describe areas that should be given priority and reflect selection, optimization and compensation in relation to the student's goals and development (see Chapters 2 and 12). In the IEP, selecting goals also means selecting educational activities. For example, when tactile learning becomes important as vision deteriorates, the IEP should identify activities where the student can use the hands for exploration and creation. This is a characteristic of IEPs for students with JNCL. The plans include activities and learning based on anticipated declines that do not seem relevant at the time of planning but are important for the future.

The goals specified in the IEP should be proactive and optimistic, but also realistic, achievable and concrete (Elmerskog & Fosse, 2012; Pretti-Frontczak & Bricker, 2000). The educational goals should be specifically described because general formulations can make it difficult for teachers to decide which activities will most likely promote the selected goals. At the same time, the IEP should have a flexible content that can be evaluated and adjusted through the year. Evaluations may imply re-selection, adjustments and compensation.

# Individual Education Plans for students with JNCL

Like the Habilitation Plan, the IEP should be based on assessments, knowledge about the disorder or disability and knowledge of the student. It should contain specified cognitive and physical goals and activities, which will give the student appropriate challenges. Experiences for example from New Zealand and Norway suggest that the IEPs for students with JNCL sometimes have a large proportion of goals related to entertainment, enjoyment and rest, and fewer related to the promotion of learning and development (Elmerskog & Fosse, 2012; Williams, 2008).

The activities chosen to realize goals should be based upon considerations that students with JNCL with increasing age will need more time and support to perform an activity or achieve a goal. Important issues for this group are what is necessary for new learning or for maintaining knowledge and skills, how the knowledge is going to be used at present and in the future, and how the students can use their knowledge and skills in everyday life outside school. For example, as the disease progresses, academic skills will gradually become a smaller part of their curriculum, while activities related to maintaining and managing activities in everyday life, such as going to the canteen, using the swimming-pool or cooking simple food, will take a larger part of the student's school time (van Delden, 2009). Students should be helped to remain independent as long as possible, even if the

time needed to accomplish the tasks increases. The declines will however sooner or later necessitate that autonomy to a greater extent is maintained through *interdependence*, where activities are performed together with another person (see Chapter 16). Learning by participation is therefore typically emphasized in the IEP of students with JNCL. There is a continuous process of learning to cope in new ways and with gradually more help.

The IEP will always include directions for the structure and organization of the teaching and the arrangements and adaptations of the classroom and other settings in school. The description of the organizational structure should extend beyond the student's classroom and include all parts of the school that are relevant for the student's education and social life. The educational measures should aim to ensure social affiliation, motor and cognitive learning and stimulation. Students with JNCL in mainstream schools will over time fall behind their peers in most subjects and may spend more time in a smaller classroom with a few other students with disabilities. This classroom should have a central location in the school and near the rest of the mainstream class to facilitate interaction with classmates and other peers.

An important function of the IEP is to contribute to maintaining appropriate expectations to the student. Expectations should neither be too high nor too low. The IEP should be written by teachers and other professionals who know the student with JNCL well in collaboration with the parents. In the present study (see Appendix A) many parents found the expectations to the student's educational possibilities too low. This might be caused by fears of failure in reaching educational goals or by an exaggerated focus on medical issues where the fatal and inevitable nature of the disease tends to overshadow educational possibilities. The IEP should state explicitly that it is neither harmful for the student, nor a reflection of failure, if a reasonable learning goal is not achieved as anticipated. However, to omit a learning goal or action, that could be attained if training were provided, can prevent students from developing their potential and reduce the zone of developmental maintenance (see Chapters 2 and 5). The present study indicates that many students with JNCL achieved learning goals far beyond what was anticipated in the beginning (see Chapters 12 and 14).

An IEP that focuses on what the student is unable to do rather than the educational possibilities may pose a severe threat to a student with JNCL. The IEP should express a clear belief that educational participation is important for development, activity, learning, cognitive maintenance and stimulation. Maintaining educational and social participation should always be essential elements in IEPs for children and adolescents with JNCL. The participation perspective should be explicitly formulated in IEPs for students with JNCL.

# Reflections on educational planning

In the present study (see Appendix A), parents and staff were asked about the participants' IEPs. The IEP was more common than a Habilitation Plan, probably due to the fact that an IEP is legally required in many of the project countries. When the parents (N=107) were asked about their child's age when receiving an IEP for the first time, 72.9 percent specified the age (mean age 8.3, SD 2.4, range 4–16). Fourteen percent answered that they did not know, while 13.1 percent answered that their child had never had an IEP (N=69). The staff informants were asked only if the student had an IEP and according to them, 82.6 percent had an IEP, 8.7 percent did not have an IEP, while 8.7 percent of the staff said they did not know.

Among the parents whose child did have an IEP, 26.1 percent received the IEP before the school year started, 47.7 percent received the IEP after the school year had started, while 5.7 percent answered that the IEP was finished so late that it had no consequence for the education (Table 11.1). Parents were asked if the IEP was ready for use prior to the start of the new school year, but most IEPs were not ready at that time, partly due to the staff finding the planning difficult because of the nature of the disease or for bureaucratic reasons.

We as the parents have always requested the IEP to be ready before the beginning of the school year, the new IEP should be developed at the end of last school year... We would always ask the first week of school (August) and would generally get one mid to late October. The school considered it too much paperwork to do a full IEP at the right time.

*The IEP was always late in secondary school, the IEP had no practical consequences for our child.* 

Table 11.1. Time of completion of the Individual Education Plan (IEP), according to parents whose children had received an IEP, and staff

	Parents (N=88) %	Staff (N=61) %
In due time before the new school year started	26.1	18.0
After the person had started the new school year	47.7	62.3
Too late, no practical consequences for education	5.7	1.6
Don't know	20.5	11.5
No IEP	-	6.6

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*The first IEP was way too late – it took about 1½ years from the learning and educational problems were identified until we had an IEP.* 

Parents and teachers emphasized a need for support from JNCL specialists when developing an IEP:

*IEPs are so important for us parents. It must be a written document, and followed up in collaboration between teachers, parents and JNCL supervisors.* 

*The work with IEP and the collaboration became good when specialists from the School for the Visually Impaired in Jyväskylä were involved.* 

JNCL specialists and parents are important contributors of IEPs, it is wrong to expect teachers with minimal knowledge about JNCL to do this important work without guidance from supervisors with sufficient knowledge.

In addition, 20.5 percent of the parents did not know when the child's IEP was completed, indicating that they did not participate in the work with the education plan. The staff informants were asked the same questions as parents (Table 11.1). The fact that 11.5 percent of them did not know if the student had an IEP suggests that the staff who were interviewed had not been engaged in the work with the IEP.

Several parents were critical to how the IEP was handled by the educational authorities:

*The IEP was denied by the educational authorities in the municipality, my child has not been given an IEP so far.* 

*The local authority (Primary Care Trust as it was then) took limited interest most of the time, no IEP.* 

We wanted an IEP for our child, but I felt our local school district didn't want us around and they tried very hard not to work with us, we had to fight for everything we wanted.

Comments from some staff members indicated that they found it difficult to plan for the student with JNCL. Some even considered all planning unnecessary or unethical for students with JNCL because of the course of the disease, which is apparent in the following quotations: *This process with the Individual Educational Plan has been changing and it is difficult to comment on the impact of the IEP on something that is evolving.* 

*I* sometimes feel the work done with IEP is wasted time. Suddenly the condition is so different.

*I am not sure it is fair to make plans when you know the outcome of the disease.* 

*It feels wrong to make plans for the future when nobody knows what is going to happen.* 

IEP is not possible because of the disease.

IEP for this school year is delayed because of the child's illness.

Teachers and other staff members also underlined that the IEP had room for modifications and revisions when needed. However, there was no evidence that education and IEPs were based on the habilitation plan. Educational measures seemed mainly to be based on the current situation. There was little or no attention to the progression of dementia and the need for precautionary or hastened learning and educational life flow (see Chapter 12). These concepts imply a need for «planning for tomorrow's needs» (proactive learning and teaching), and a need to teach to an automated level of mastery so that ultimately, the skill thus taught can be executed with minimum demands on cognitive monitoring.

Parents and educational staff were asked about the impact of the IEP on the students' learning, development and maintenance of skills. The results showed that 61.2 percent of the parents and 53.9 percent of the staff evaluated the impact to be high or very high, while 15.7 percent of the parents and 23.1 percent of the staff found the impact moderate, and 11.2 percent of the parents and 4.6 percent of the staff found little or no impact. The parents and the staff thus had quite similar evaluations of the impact of the IEP.

Parents and staff were asked about the parents' role in the development of the IEP. Forty-four percent of the parents and 33 percent of the staff answered that the parents had been important contributors to the IEP. Most countries emphasize collaboration with parents in the development of the IEP (MacLeod et al., 2017). However, the parents' comments about IEP and the relatively low degree of parent contribution indicate that the current practice needs improvement. In fact, several parents suggested that the IEP was not used primarily for achieving the best possible education for their child but rather to achieve other goals:

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	Parents (N=89) %	Staff (N=65) %
No impact	2.3	0.0
Low impact	8.9	4.6
Moderate impact	15.7	23.1
High impact	29.2	27.7
Very high impact	32.6	26.2
Don`t know	11.2	10.8
No IEP	-	7.7

Table 11.2 Impact of the IEP according to parents whose children had received and IEP, and staff

*I am sitting with a feeling that the IEP was done to satisfy the authorities, not the needs of my child.* 

The educational authorities demand that IEP is written according to the needs of the authorities. We feel we use a lot of time on paperwork to satisfy the authorities instead of our child.

Some parents found the IEP too vague, it was not really possible to find out what would be happening in school, which the following quotations indicate:

*I* do regret that we were not specific enough about the amount of time and sessions for the music therapy, speech therapy and hydrotherapy.

The IEPs were vague in the targets but vital to ensure that resources are in place and funded.

The activities defined in the IEP were not always implemented:

When BDSRA came out and helped with our latest IEP a suggestion she had was to implement objects that would be significant to our child for transitions (a steering wheel to go to the bus, another object when it was time to go to specials, and so forth). To date, we have not heard about them implementing this and his IEP was completed in September.

However, most parents and staff expressed the view that educational planning is important for students with JNCL and has a positive impact on learning, development and maintenance of skills. Parents (N=112) were asked to appraise the information they had received from different contributors during the first year after the diagnosis was confirmed on a scale from 1 (no contribution) to 5 (very high degree of contribution). Parent organizations received the highest mean score (3.6), followed by other parents in same situation (3.4) and competence centers (3.3). The local educational authorities got the lowest mean score (1.4). These results document the need to include JNCL specialists early in educational and other planning.

The staff was asked to appraise the role of different contributors to their own competence building related to visual impairment and JNCL. Collaboration with parents got the highest mean scores (3.8 for visual impairment and 3.7 for JNCL), followed by competence centers (3.2 for both visual impairment and JNCL). The local educational authorities got the lowest mean scores (2.1 and 2.0).

These results probably reflect that JNCL is a very rare disease and that parents, teachers and other staff did not find sufficient competence in the local services. As part of the present project, a literature search was performed, and the result was almost exclusively articles from the medical domain. This indicates that JNCL and childhood dementia are practically unknown in both the educational and the social services. The abscence of adequate knowledge may lead to lack of initiative, disillusionment and discouragement, because professionals become likely to focus mainly on the negative medical aspects of the disease and not on the educational possibilities and interventions. It is noteworthy that information from the parent organizations (see Chapter 24) was considered important by many parents, emphasizing the importance of having contact with other parents, for both the educational planning for their own child and for supporting other parents.

Parents were further asked about the impact learning during school years had had on adult living, on a scale from 1 (no impact) to 5 (very high impact). The two school subjects with the highest average were physiotherapy (4.1) and Music/music therapy (3.9), followed by physical education/training (3.5) and social gathering with other students (3.5). The lowest scores were for mathematics (1.9) and nature studies (2.1) but even if these domains had less apparent impact, they represent general knowledge in society and participating in such classes is part of inclusion.

# Principles for making plans for persons with JNCL

Educational planning for persons with JNCL differs from planning for most groups who need special education. Plans usually focus on growth in knowledge and skills, on identifying the zone of proximal development and learning potential, and appropriate adaptations and educational strategies (see Chapters 2 and 12).

This is also important for individuals with JNCL but in addition, planning must take the possible emerging declines and losses into account. Planning should include adaptations and educational strategies in areas with independent and interdependent functions within the zone of developmental maintenance (ZDM). Figure 11.1 shows learning and development in two hypothetical persons with JNCL, who have different areas of strength and weakness, and hence need different interventions. The figure illustrates that functions may be maintained to different degrees when independent function is reduced, but maintenance depends on appropriate help and support.

The standard planning procedures that are used for students in general should be utilized as far as possible for students with JNCL, meaning that they share goals, activities and themes with their peers in an inclusive setting. However, because students with JNCL require new educational measures as the disease progresses, there will be a need for planning for lower functioning in the future. Interventions that build on growth are within the zone of proximal development, while interventions preparing for or following decline are within the zone of developmental maintenance, applying Baltes' (1997) principles of selection (choice of relevant and achievable goals), optimization (forming and maintaining means to achieve the goals) and compensation (new strategies to maintain functions and goals that are no longer sustainable in ordinary ways) (see Chapters 2 and 12). This will include developing an extended curriculum and interventions in areas of special importance for each student. This will usually involve areas and strategies that ordinary teachers and other staff in mainstream educational settings are not familiar with and should therefore be emphasized in the IEP and the Habilitation Plan.

The «The Participation and Performance Dilemma» (Gray & Hollingsworth, 1999) is illustrated in Table 11.3, adapted to the education of persons with JNCL. The table shows four possible scenarios: In Scenario 1, the individual is performing and participating in an activity. This scenario represents the general situation in education. In Scenario 2, the individual is not performing but still participating in the activity. This scenario is rather uncommon since education typically is based on both participation and the student's performance in the activity. However, Scenario 2 may represent a goal for individuals with JNCL. The declines may make performance within the activity difficult, while assistance, adaptations and interdependence may make participation and engagement possible, for example in mathematics or art (see Chapters 12 and 16). Scenario 3 does not represent a desirable situation and may be a result of poor planning. For instance, the person has learned to use braille but is not provided with materials that enable him to use his tactile reading skills, because the staff at the person's new residence is not familiar with braille and not aware of the person's abilities. In Scenario 4,

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Figure 11.1 Zone of Developmental Maintenance (ZDM)

The figure shows independent performance and the zone of developmental maintenance (what a person is able to do with help) for two persons at two points in time, and for eight knowledge and skill domains. The persons differ in knowledge and skills and the differences between the domains are shown as variation in "width" and "length". The developmental courses of the two persons differ also somewhat from Time 1 to Time 2. In domain 1, both independent performance and ZDM have decreased from Time 1 to Time 2 in the two persons, although somewhat differently. In domain 2, independent performance has decreased and the ZDM increased correspondingly in both persons. They have lost independence but have maintained the function with help (interdependence). In domain 4, the ZDM of Person B has decreased (can do less with help), but there is no change in the person's independent performance. For Person A neither independent performance nor ZDM have changed in this domain. In domain 7, independence was reduced in both persons from Time 1 to Time 2, but the ZDM remained the same in person A and increased in person B. Consequently, the two persons have different strong and weak domains and show different developmental declines between Time 1 and Time 2.

	Ability to do activity	Inability to do activity
Participation	<b>Scenario 1</b> Assessment shows Performance and Participation	<b>Scenario 2</b> Assessment shows No Performance but Participation
No participation	<b>Scenario 3</b> Assessment shows Performance but No Participation	<b>Scenario 4</b> Assessment shows No Performance and No Participation

Table 11.3: The Participation and Performance Dilemma (based on Gray & Hollingsworth, 1999)

the activity has been terminated because it was now outside the person's zone of developmental maintenance and thus not relevant for the person anymore. However, Scenario 4 may also be a result of poor planning and too little support.

The scenarios in Table 11.3 raise several questions. If a situation is described as Scenario 2 or 3, one may ask what initiatives are needed to achieve Scenario 1 (if desired and feasible). If the situation is described as Scenario 4, one may ask what initiatives are needed to achieve Scenario 1 or 2 (if desired and feasible). There is always a risk that low performance will lead to the assumption that the activity is not suitable for the person, and subsequently to non-participation. Participation must therefore be the core element in planning for maintaining or improving skills and quality of life in persons with JNCL, also when independent performance is limited. Participation is about personal engagement and should not depend on the ability to perform. An important objective in the planning work is to promote initiatives that will allow persons with JNCL to participate in desirable activities and provide adequate support and adaptations to make participation possible.

For persons with JNCL, participation may depend on proactive planning. In the later stages of the disease, persons with JNCL may need extensive support from helpers. The need for individual support in all daily activities should be described explicitly in the plan. Preservation of personal engagement, participation, skills and quality of life all depend on appropriate support. The guiding questions of planning will thus be: What can the person with JNCL do and participate in when supported by a helper, and what help and support are necessary for maintaining performance and participation?

# Core issues in educational planning

The educational system is based on expectations of acquisition of knowledge, skills and attitudes in predefined domains. The student is expected to achieve defined goals, for instance within literacy and mathematics in accordance with a curriculum. For students with JNCL, the processes of planning and implementing educational interventions will undergo comprehensive changes during the individual's life time (see Chapter 12).

*Participation* in educational and social situations is important for students' learning and quality of life (Centre for International Development and Training, 2001). No curriculum can be compared to what children and young people learn through participation in everyday situations and special events, that is, by being engaged in situations where things happen. Participation-based learning is an implicit general goal in society where participation is taken for granted (United Nations, 2018a,b). It is usually assumed that participation eventually will be maintained by the individual. Participation-based learning facilitates implicit learning, that is, the acquisition of knowledge and skills that occur incidentally, without planning and without personal awareness of the fact that learning is taking place (Frensch & Runger, 2003). Planning for participation in everyday activities is therefore a way to promote implicit learning and development in students with JNCL in all phases of the disease. Students with JNCL may to some extent be able to ensure participation in the early school years but will need help to participate in activities with implicit learning when explicit learning becomes more difficult to achieve. Possibilities for participation-based learning should be included in IEPs of all students with JNCL and become more emphasized with age. Planning to promote participation is therefore about creating a life agenda for the student, with specified meaningful activities from early morning to bedtime, based on knowledge about the student's interests and possibilities. Maintaining and securing educational and social participation through participation-based learning is a central element of interventions for students with JNCL, irrespective of how they function.

*Precautionary or proactive teaching and learning* mean the teaching of skills that the student does not need at present but will need in the future (see Chapter 12). Teaching augmentative and alternative communication when the student's speech is still intelligible is an example of early learning to later problems, in this case when speech is no longer intelligible to others (Chapter 13). Early learning of how to operate a computer and software programs adapted for individuals without vision will prepare the student for a future loss of vision. However, it is unusual in special education to prepare for future losses of skills. Some of the parents and teachers in the present project described resistance to proactive teaching activities. Some teachers thought it was unethical to address a problem which was not yet present (see quotations from staff and parents, Chapter 14). There were also doubts about the students' motivation to learn something when they were unaware of their need for it. Proactive learning was perceived as somewhat odd, in particular when the student was not aware of the diagnosis. However, the project also found

good examples of proactive teaching and learning in students with JNCL (see Chapters 13, 14 and 16). It depended on the enthusiasm and dedication of the people in the planning group, and on how the teaching activities were organized. The ethical dilemma related to proactive teaching is illustrated by the following statement, based on many quotations from parents in the current project: *It was too late to start with the learning program, the cognitive decline had gone too far* (see also Chapter 9). In *hastened learning,* intensified teaching is planned to accelerate the student's learning in selected and complex areas which require best possible cognition, when this is considered advantageous in the longer term (see Chapter 12).

Maintaining *life flow* means to have a certain degree of continuity (see Chapter 16). This principle is of special importance for planning of transitions, which often can threaten the life flow of the individual. A dementia perspective acknowledges that the person's history and experiences, as well as skills, interests and social life should be addressed in the IEP and Habilitation Plan. The major transitions may be experienced as dramatic, and if not considered in the plans, may create chaos in the person's life and negatively influence the person's mental health (see Chapter 27). The JNCL project has disclosed examples of severe breaches of the individuals' life flow in connection with transitions.

The principle of life flow is always important in planning for people with JNCL but will become more decisive with age. Transitions should be planned and prepared in close collaboration between the family, the person when possible and all relevant service providers. However, this is not always the case, as evident in the following quotations from parents:

When he transitioned from elementary school to middle school, the person in charge of special education recommended he be placed in a classroom that focused on daily living skills as opposed to regular subjects like math and science. This was actually done without our knowledge. He was very bored.

We as parents always had to organize the transitions on our own. We never received any support.

*The transition to adult living was difficult, we had to accept what was there, the needs of our son was not important.* 

The transition to adult living is further discussed in Chapter 23.

# A planning tool for JNCL

Children and young people with JNCL should have similar plans, educational measures and life contents as their peers when this is possible and to their benefit. Participation in ordinary activities and sharing of knowledge and goals with others will promote social inclusion and participation in society. However, children and young people with JNCL need more than the standard plans. The unique and different perspectives on planning for students with JNCL are anticipation of future challenges and consideration of possibilities despite these challenges. Anticipating future challenges and implementing measures to meet these challenges can be a very emotional process, especially for parents but also for staff. Such issues must be handled with great respect and empathy by all involved. Professionals should all the same be able to make plans for future challenges and possibilities and act accordingly with or without parent participation. Assessment is necessary for planning to be based on the best possible knowledge. The Educational Development Observation tool (EDO) is designed to give an overview of themes that should always be considered in planning for persons with JNCL and thus constitutes a knowledge foundation for the IEP and the Habilitation Plan. The main themes in the EDO are vision, communication, literacy, social life, gross motor function, fine motor function, physical activity, behavior, memory and attention, independence and autonomy, interests and equipment (see Chapter 10). From a long-term perspective, the EDO is making it possible to address domains, which will decline sooner or later. From a short-perspective horizon, the EDO helps see what can be done at present to meet the future challenges. It is particularly important to use the EDO prior to major transitions, such as transitions between schools, or the transition into adult living.

# Conclusions

Making plans for a child, adolescent or adult with JNCL requires knowledge about JNCL and the individual. The overall needs are comprehensive and wide-ranging, and it is necessary to involve JNCL specialists and parents in the planning of education and other interventions. Observations and discussions must be initiated at the right time according to a timetable based on an anticipated progression of dementia. The ongoing or anticipated symptoms caused by the disease should represent a working agenda for the Responsibility Group. What is not done today may be difficult to achieve tomorrow. The development of dementia sets the framework for educational planning and interventions. Education cannot stop the disease from progressing, but education can meet the symptoms with selection, optimization and compensation measures.

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# Early and Later Educational Intervention

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The education of individuals with juvenile neuronal ceroid lipofuscinosis (JNCL) will depend on the competence in the educational system, as well as the society's general traditions in mainstream and special needs education. A search for research on planning and implementation of education for students with JNCL yielded very little, and there are hardly any studies about the impact and efficacy of educational practices for students in this group (von Tetzchner, Fosse, & Elmerskog, 2013; Williams, 2008). Educational systems differ a lot between countries, also among the countries participating in the present project (see Chapter 9). There are a few published guidelines for teachers about education for students with JNCL (Bills, 2011; Elmerskog & Fosse, 2012). In general, students with JNCL follow the same curriculum as peers for as long as possible. Some countries have special schools and institutions with considerable experience in working with students who have JNCL, in other countries students in this group attend mainstream schools with no prior experience in teaching a student with JNCL.

In recent years, two general principles have been emphasized in special education: selection of individual educational goals and individual adaptation. The teaching should meet individual needs and utilize individual strengths, and this applies to students with JNCL as to all other students.

Individuals with JNCL develop childhood dementia, which is dementia with onset in childhood (see Chapter 5). There are several small diagnostic groups with cognitive and other declines in childhood (Schoenberg & Scott, 2011) but the number of children is small and childhood dementia is a rather unknown concept in education, also in special schools and schools with children who have degenerative diseases. Compared to education and special education in general, students with childhood dementia require a significant change of practice, and the selection of educational goals and individual adaptations is partly based on different assumptions and premises.

The education of students with JNCL must be adapted to the present and future changes in their learning abilities, to what the students are able to do and learn in the present situation and in the near and distant future. The period before the onset of decline in a domain may be considered a «time window» or «window of opportunity» for the student to learn particular skills for later use. For instance, the educational strategies will change from utilizing residual vision to relying on auditory and tactile information, and educational strategies may be directed at precautionary learning (see below) to meet future needs (Elmerskog & Fosse, 2012).

All students need continuous changes in education to meet their knowledge, abilities and skills, mostly as a result of their intellectual growth. Students with JNCL need continuous educational adaptation to meet both growth and decline in their abilities. This means that teachers need knowledge about the functional consequences of the disease and the educational strategies that may support learning and coping (Elmerskog & Fosse, 2012; Uvebrandt, 2006). Because of the limited knowledge, there is a need for systematic experience and developing new strategies. The model proposed by Baltes and Baltes (1990) may be a useful tool for selecting goals, optimizing methods for reaching educational goals, and developing new ways to compensate for functions that are slowly deteriorating (see Chapter 2).

This chapter describes changes in practices through the educational course of students with JNCL, from a focus on academic subjects to more practical domains. Academic subjects may include mathematics, writing and reading, physics, native and foreign languages, geography, and history. Some require more reasoning skills while others require an ability to learn facts. Reasoning is usually difficult for students with cognitive problems. Remembering facts can be easier for students with JNCL, in particular when facts are related to an area they are interested in (Elmerskog & Fosse, 2012). Practical domains may include physical education, woodwork, home economics, domestic science, and orientation and mobility. These are often associated with how to do tasks and procedural learning (see Chapter 16).

Several chapters in this volume include examples of educational adaptation. At the end of the present chapter, the adaptation of two school subjects is described. Mathematics represents an academic subject while drama is a practical school subject, facilitating the achievement of personal, social and cultural identity.

# Adaptive education and special needs education

Education for children and adolescents with JNCL usually includes both adaptive and special needs education. In *adaptive education* the ordinary curriculum is made accessible for students with special needs (Fasting, 2013). For instance, learning how to read the clock is a goal within the mainstream curriculum, and can be adapted to students with visual impairment by using a tactile or auditory clock, and by introducing quarters of an hour instead of minutes in the early intervention when needed. Similarly, braille or text-to-speech programs may make textbooks accessible for this group of students.

Simon was working with addition and subtraction, using an electronic workbook on his computer. The text in the workbook was read aloud with synthetic speech, and Simon wrote his answers into the book with a braille keyboard. These adaptations made it possible for Simon to work with the same material and having the same goals as his classmates.

*Special needs education* may be defined as the adoption of educational goals and associated strategies not usually applied with children of the same age group. In this definition, special needs education will always imply a change in curriculum. For instance, learning the mobility route from the classroom to the schoolyard would be a goal within special needs education for students who are blind because it is not part of the ordinary curriculum. A student with JNCL will not share this education goal or the training with sighted peers in a mainstream setting.

The distinction between adaptive education and special needs education is not always clear. For example, literacy is a main goal in both mainstream education and special needs education but different modalities and strategies are applied. Teaching of tactile reading and writing strategies is not part of the ordinary curriculum and sighted peers do not usually participate in the training needed to learn braille or Moon.

# The child's rights and needs

In many countries, adaptive education and special needs education are legal rights in the educational legislation. Irrespective of the need for either of these rights, it is a commonly held view that education of students with typical development and students with disabilities should consider the students' strengths and limitations, as well as possibilities and barriers in the educational environment (Elmerskog, Storliløkken, & Tellevik, 2008). It is important to know the child, the social environment and the culture (Rogoff, Dahl, & Callanan, 2018).

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# Three educational phases

The education of students with JNCL can be divided into three main phases. The results of the present study (Appendix A) indicate that the average age when students with JNCL begin to show problems with learning and remembering new things is around 10–11 years, but also that the age of onset of cognitive and other declines vary considerably within the group (see Figure 1.1). The phases therefore do not follow age but the students' developmental declines and general functioning, and the length of each phase will vary.

# The phase with no or mild dementia

At the time of diagnosis, cognitive problems of students with JNCL are usually not very pronounced and they may be able to follow the mainstream curriculum for some time (Elmerskog & Fosse, 2012; Uvebrandt, 2006). The present survey found that some of the skills and interests that the students develop early in life will last for a considerable length of time. This early phase is therefore important for building interests and skills that may be used in education and everyday life in the later phases of the disease.

### *Flexibility and variation*

Participating in class during the early school years facilitates the exposure to ordinary school subjects and themes. Mainstream education implies flexibility and variation, which will benefit both the child with JNCL and the classmates. The teaching focuses on new knowledge and skills and switches between different subjects several times each day. This period of relatively intact cognitive functioning represents an important learning window for students with JNCL and is of high importance for capacity building and for establishing individual interests.

Adam is very interested in dinosaurs. He has a lot of knowledge about the different species. The interest was kindled early in life and has been an inspiration both for learning and remembering facts and developing writing skills. Every day, Adam is writing facts and stories about dinosaurs on his computer with a braille keyboard.

Most children with JNCL follow the ordinary curriculum as long as possible, acquire new knowledge and skills, and develop concentration and problem solving ability within many subjects and themes. Learning is to a large degree explicit, skill-oriented and focused on capacity building.

### Learning through vision and visual memories

From an educational perspective, it is a huge advantage that students with JNCL have had normal eyesight in the first years of life. Reference to their visual memories allows them to think visually and they will associate early visual experiences with experiences based on tactile or auditory perception later in life. For example, prior visual experience facilitates students' early learning of mathematics.

School beginners with JNCL experience a gradual deterioration of visual functioning, but some students retain residual vision for several years (see Chapter 4). During the first years, children with JNCL can usually use visual learning strategies to some extent. They learn many daily living skills through visual observation, such as washing the hands or turning on the radio or television. Teachers must monitor the student's visual functioning carefully, in order to know when it is necessary to introduce strategies based on other modalities.

#### *Learning in a social setting*

Being a part of a class or a group and participating in the same educational and social activities as peers is highly motivating for most children. Students with JNCL will eventually need technical aids and special learning materials that make them appear a little different, such as magnifying devices with closed circuit television (CCTV) and equipment for reading and writing braille (see Chapter 14). Writing braille on a Perkins brailler or on a braille display with synthetic speech feedback creates noise and may be a distraction for others in the classroom; therefore ways should be found to minimize distractions and facilitate full participation. The use of head-phones will reduce the noise but make it more difficult for the student to hear instructions from the teacher and other important sounds in the classroom. Such adaptations may interfere with inclusion goals and should only be used when necessary. It is important to organize the classroom and monitor the activities in a way that ensures the best possible working environment both for the student with JNCL and the classmates.

Susan had some residual vision and used CCTV to magnify texts and illustrations in the textbooks. She was dependent on this equipment to do her school work. However, she had never felt quite comfortable with these technical aids because she in this way became different from the classmates. To prepare for future needs it was time to introduce Susan to braille. Teachers were concerned about how she would react when becoming even more different by learning and practicing tactile reading. To «normalize» braille the whole class was introduced to braille reading and writing during a visit from the national competence center. The classmates read braille letters using vision. The introduction of braille became a success for Susan because of the enthusiasm the other students expressed. *Comments like «This is the most exciting thing I have learned for a long time!» from a classmate made a big difference for Susan's attitude to braille.* 

Reducing noise, making the environment physically inviting and allowing other students to learn to use the special equipment will contribute to inclusion and social affiliation for students with JNCL.

#### Precautionary learning

Precautionary or proactive learning implies that children are introduced to new skills they will need in the future but not in their present situation. The gradual cognitive decline in JNCL will make acquisition of complex skills increasingly difficult. Complex cognitive and tactile skills, like reading and writing braille or Moon, should therefore be introduced as early as possible, preferably very soon after the diagnosis is confirmed (see Chapter 14). The time window for learning tactile reading is in early childhood when the child's learning capacity is best. However, the present survey found that many students with JNCL were not introduced to braille before their visual impairment made visual reading impossible (see Chapter 14). By this time, the time window for learning braille might be closed because of the student's cognitive decline.

Some skills are pivotal in the sense that they establish a foundation for further learning and functioning, for example literacy skills and basic mathematics, and such skills should be automatized when possible. This automatization has been described within the concept of *hastened learning* (Tellevik, 2008, see below). Automatization means skills are mastered so well that they require little conscious effort. Automatization requires massed training and many repetitions during the learning period. Automatized skills are also resistant to decline because they require little cognitive capacity. Promotion of pivotal skills should therefore be given priority in the early education of children with JNCL.

#### Hastened learning

The principle of *hastened learning* concerns intensified instruction to accelerate learning in selected areas. The underlying assumption is that the student's current capacity enables learning of skills that might be difficult or impossible to learn in a later phase, such as writing and reading or playing the piano. Hastened learning may also be used to facilitate the development of early interests that might be of importance in later phases, such as music and sports (see Chapters 14, 16, 17 and 23). The aim of hastened learning is thus to achieve a level of performance that will require little cognitive capacity or strain when it has been acquired, and thus represent good cognitive economy (Tellevik, 2008). Hastened learning will usually require explicit teaching of skills.

### Selection, optimization and compensation

For most students with JNCL the selection of goals in the Individual Education Plan (IEP) in this period should reflect the same range of subjects and themes as for their classmates. There are however, some additional domains that should be prioritized because of the visual deterioration, such as tactile reading and writing (see Chapter 14), mobility and orientation, and autonomy (see Chapter 16). The demand for special teaching can feel overwhelming to teachers responsible for implementing the training. In addition, teachers might perceive that the demands on the students' learning capacity exceed an acceptable level. When prioritizing is necessary, there should be a focus on knowledge and skills that will be useful for the student both at present and in the future (see Chapter 11).

In this process, Baltes' (1997) principles of selection (choice of relevant and achievable goals), optimization (forming and maintaining means to achieve the goals) and compensation (new strategies to maintain functions and goals that are no longer sustainable in ordinary ways) may be applied. Optimization for achieving the individual goals may imply new activities and strategies both for the teacher and for the student with JNCL. For example, staff may need to mediate visual information by giving the necessary information verbally and the student may have to learn to use technical equipment in order to read and write a tactile alphabet. Activities related to teaching route planning and mobility will ensure successful locomotion on the school premises and greater autonomy (see Chapter 16).

Like other students, students with JNCL learn best when the educational situation is exciting and positively challenging, and when the content is meaningful and matches their skills and interests (Norris & Closs, 1999). Ensuring a proactive and functional IEP for a child with JNCL requires the joint efforts of the school team as well as support from specialists with competence in visual impairment and JNCL (see Chapter 11).

# The moderate dementia phase

As students with JNCL become older, the need for individual adaptation gradually increases. Their ability to maintain attention over time, listen to the teacher and remember spoken information decreases. The students benefit from teaching involving personal experiences and meaningful activities with sensory information from multiple sources, such as in the example below.

Lisa was a student in secondary school and attended an ordinary class. The teacher was telling the class about the different nutrients in dairy products and Lisa was struggling to keep attention and remember what the teacher

was saying. The teacher therefore chose another approach for Lisa. Together with Lisa, she prepared an interview with ten questions about dairy products and made an appointment with the local dairy for a visit and interviews with members of the staff. Lisa was given a tour around the dairy, where she could taste and smell some of the products and listen to the production sounds. Lisa was able to complete her task with an adult assistant. Learning became much easier for Lisa, she could work with the same educational goals as her classmates, but in her own way and speed.

Lisa's story may be an example of activity-based cognition (see below) and demonstrates the importance of meaning-in-activity for learning. It is an example of interdependence (see Chapter 16), because Lisa and the teacher worked together with tasks that were within Lisa's capacity for learning, in her zone of proximal development (see Chapter 2).

### Simplifying goals

In this phase, it will often become necessary to simplify learning goals. This may be facilitated through optimization and compensation, rather than abandoning goals. For example, instead of teaching technical information about food nutrients, a student with JNCL may be taught which food sorts are healthy and which are not. Using personal experiences may support attention and memory, such as when learning how to make a weekly dinner menu or ensure a healthy diet for the family pet. When cognitive abilities decline, it is important to use topics of special interests and/or personal experience as foundations for learning. Evidence from other diagnostic groups suggests that dementia makes learning more difficult, but that learning is still possible (Clare & Jones, 2008; Clare et al., 2000; De Vreese et al., 2001; Kessels, Remmerswaal, & Wilson, 2011). However, it is likely to require frequent repetitions to refresh and maintain memories, more adaptation and more support.

The deterioration will not follow the same pace in different domains. In the present study (Appendix A), the mean age for onset of symptoms differed considerably between domains, for example between declines in vision, speech and gross motor functions (see Chapters 4, 6 and 7). Learning new skills may be possible in some areas but not in others.

### Project work as educational strategy

In project work, students work together to produce a joint result and this result depends on students having different responsibilities. Project work makes it possible to utilize individual strengths and preferences, both when choosing themes for the project and when choosing tasks for the student with JNCL. This strategy also ensures participation of the student with JNCL in a community of learners and being part of a social setting (Rogoff, Matusov, & White, 1996; Rogoff, Paradise, Arauz, Correa-Chávez, & Angelillo, 2003). Project activities may for example be applied to practical-aesthetic subjects such as woodwork and music. They may involve planning meetings where tasks and responsibilities are distributed among the participants, information gathering and making decisions about how to present the results. Several school subjects may be integrated into one project.

Ethel was interested in handicraft and had recently started to attend a handicraft workshop at the school together with her special education teacher. This was a useful preparation for adult life, because it was an interest that may be pursued well into adulthood. At the workshop, Ethel participated in making various items for decorating and practical use in the home. Some of the items were included in a sales exhibition, while other items would become Christmas gifts. Ethel's first task was to decorate Christmas cards to follow the gifts and be part of the sales exhibition. The teacher put small colored pieces of paper in front of Ethel, who crumbled them into paper balls, dipped the paper balls in glue and decorated the cards. Ethel still remembered colors and had conversations about colors with other people in the workshop. She wanted to use the best color combinations when she decorated the cards. The teacher helped Ethel find the colors she wanted. Another task was putting labels with product name and price on the products for the sales exhibition.

#### Changes in goal selection

Studies of the elderly population show that systematic educational effort may delay cognitive decline in dementia, given that the educational activities and tasks are manageable for this population (Baltes, 1997; Bird, 1998; Kawashima et al., 2015). Supporting maintenance is also an important aim of education for individuals with JNCL. There will gradually be less time spent on building new knowledge and skills, and more time spent on maintaining already established knowledge and skills. There are time windows for developmental progress and maintenance of skills. The role of the zone of proximal development, where education and support facilitate new learning and development, will gradually diminish. The zone of developmental maintenance however, where education and support contribute to maintaining knowledge and skills, will become more prominent (see Chapter 2 and 11). Appropriate daily cognitive stimulation is a part of the maintenance process. Goals can involve practical tasks, such as mixing ingredients when baking a cake, or more intellectual activities, such as discussing which cake ingredients are to be put

on the shopping list. Studies of elderly people with moderate dementia have found that participation in practical and cognitive activities lead to better performance of everyday tasks and increasing independence (Loewenstein, Acevedo, Czaja, & Duara, 2004). Similar strategies may be used with children and adolescents with JNCL, for example by stimulating and activating memories with familiar objects and sound recordings (Gylfason & Jóhannsdóttir, 2006; see Chapter 19).

#### Optimization and compensation

In this phase, children and adolescents with JNCL become gradually more dependent on help and support from other people and interdependence becomes more important in everyday tasks (see Chapter 16). However, participation seems to decrease when independence levels drop. Although help and support may be available most of the time, the survey found that social and societal participation becomes sparse (see Chapter 22). If activities are omitted from the weekly schedule, instead of being adapted to the person's abilities, this is likely to lead to a general reduction in activity level, less stimulation and a more passive life. For students with JNCL an important issue is to maintain activities and participation levels also when independence subsides. Optimization by forming and maintaining the means and activities to achieve individual goals should therefore be continued. It is further important to maintain the expectations of people in the environment even when extensive support is required. Education without expectations will be a degrading experience for most students, including students with JNCL (Elmerskog & Fosse, 2012).

Philip was 11 years old and his speech was starting to become slurred. One of the goals in Philip's Individual Education Plan was to maintain and strengthen his articulation skills as long as possible. A speech and language therapist visited the school once a week to give him training and give supervision to the teachers. The training program was followed-up by the teachers.

Because participation is an educational goal for students with JNCL, educational activities should be evaluated with regard to whether they support participation in activities that are manageable and meaningful for the student (Elmerskog & Fosse, 2012; Elmerskog & Storliløkken, 2006). Taking part in meaningful activities may have a considerable impact on the student's learning, cognitive maintenance and quality of life.

In this phase, the need for a clear daily and weekly structure will become evident (Elmerskog & Fosse, 2012). Structuring may be defined as a sequential organization of activities and tasks (see Chapter 16). A recognizable structure will

give the student an overview of the activities and may contribute to a feeling of security and control over the situation. If the student is able to read the daily schedule independently or interdependently, this may enhance feelings of self-efficacy and autonomy. Clear structures are also useful when adapting work situations or activities during leisure time (Elmerskog & Fosse, 2012).

Maintenance can be further enhanced through repetitions with some variety, and the students' personal knowledge and skills may be maintained through frequent refreshments of memory (Woods, Spector, Jones, Orrell, & Davies, 2009). The IEP should specify appropriate learning and maintenance goals, and how these can be achieved, for example how often repetitions and refreshments of memories of distant and recent events should be initiated by the teacher. Many individual education goals may be maintained through interdependence and participation (see Chapter 16). Over time, however, there will be skills that cannot be sustained, like writing in the example below.

Rachel was a young adult. She had been fascinated by fairytales and fantasy stories all her life. She had a rich imagination and a strong urge to communicate her thoughts. For some years she had been able to write her stories herself using a braille keyboard, but recently fine motor decline had made her writing pace slower and the number of spelling mistakes larger. Now her writing skills did no longer match her need and wish to express herself in writing. This activity of so great importance was maintained with compensatory measures. Rachel now conveyed her stories to the assistant who typed them on Rachel's computer. In this way, Rachel produced the stories and could listen to them via synthetic speech output, and she could share her stories with others.

# The phase of severe dementia

Sustaining an active and stimulating life in order to achieve best possible life quality requires shifts in focus during the severe dementia phase of the disease. Skills will deteriorate in spite of efforts to maintain them, not abruptly but gradually over time. The zone of developmental maintenance replaces the zone of proximal development, and over time even the zone of developmental maintenance will shrink (see Chapter 2 and 11). Interdependence becomes clearer and participation in personally meaningful activities will gradually become the main source of communication and cognitive stimulation (see Chapter 16). When the time window for learning and maintenance begins to close, participation in meaningful activities may still be maintained through individual support from family and staff. Hearing is less affected by the disease than the other sensory systems and the auditory system may function throughout life (Elmerskog & Fosse, 2012). The present study indicates that comprehension is better preserved than production of speech (see Chapter 6). The auditory sense and perception of speech are therefore important resources. In the phase of severe dementia, it may still be possible for persons with JNCL to perceive and understand environmental sounds and verbal information, such as descriptions of visual features, at least in relation to routines, everyday activities and other familiar events.

#### Optimization and compensation

Participation in activities with others will usually contribute to increasing learning and skill maintenance. Individuals with JNCL should therefore be in a rich speech environment to enhance and maintain their comprehension of the spoken language. They should be encouraged to listen and to speak when possible, as illustrated in the example below.

Richard was 18 years old and spent two days every week at a work place for persons with learning disabilities, together with a school assistant. Richard and the assistant worked interdependently: the assistant placed the material in front of Richard, and Richard's job was to disassemble a small piece of building equipment into two parts, while the assistant did the rest of the task and put the finished work away.

Around the work table was a group of 4–5 people, some of them very sociable and talkative. There was a continuous conversation and many themes were discussed. Richard had problems retrieving and articulating words, but he participated actively in the conversation by listening intently. Sometimes he would suddenly repeat a word or a name that someone had just said. Although it was usually very difficult to understand Richard's speech, he was understood because his utterances were relevant in the context and the words he said were repetitions of words that everyone had just heard. In this way, Richard participated in the conversation, both by listening and talking.

The presence of word-finding problems is a characteristic of severe dementia (see Chapter 5). In the story above, Richard was able to recognize words in the conversation and repeat some words now and then, but he was not able to bring new information into the dialogue. Asking open-ended questions to individuals with severe dementia is not likely to lead to relevant answers and may create stress and a feeling of shortcoming in individuals with JNCL. Instead, answering by choosing between a few possible answers depends more on word recognition,

which is easier than production (see Chapters 6 and 13). Students experiencing more difficulty can also be offered binary choices, a strategy that provides models of two possible answers, thus circumventing the need to retrieve words. Studies show that receptive vocabulary actually may continue to expand when other cognitive skills have started to decline (Adams, Kwon, Marshall, de Blieck, Pearce, & Mink, 2007).

For schools and other services, dealing with skill decline constitutes an unusual situation. There is a risk of a loss of participation opportunities provided to adolescents and adults with JNCL. For example, when their skills decline or they lose the ability to initiate actions for themselves, they might not be offered stimulating activities and situations. In addition, decline of skills may be misinterpreted as loss of interest and lead to non-participation.

Barbara was 20 years old and attended a work center for persons with learning disabilities. One of her favorite tasks was to make knitwear using a knitting machine. Barbara had a lot of strength in hands and arms and she was very proud of this. For a long time, she managed to control her movements perfectly when using the knitting machine. But eventually, problems occurred. Barbara was no longer able to regulate the strength needed to administer the knitting machine. Her arm movements became too abrupt and this destroyed the knitwear. The work center therefore decided that Barbara could no longer make knitwear on the knitting machine.

In the example above, the young woman lost a favorite work activity because of declining skills. Barbara and her assistant could have cooperated in doing the hand movements needed for knitting, and the favored activity and the associated social participation might have been maintained. Instead, compensational measures or interdependence were never considered. The next story illustrates compensation.

Brian was a young adult who had been interested in music all his life. When he was younger, he had been quite skilled at playing the piano. He gradually lost the motor control that was necessary for his playing. To compensate for this and help him to continue producing and performing his own music, he was given a computer program where he could compose music by choosing from a pool of free sounds and loops. With this program, Brian put together different instruments, sounds and rhythms together with helpers, and in this way he composed his own music. The computer program enabled Brian to maintain his interest and ability in performing and producing music, even if he could no longer play the piano. His assistant gave the help and support that was necessary for keeping Brian engaged in music-making. Brian made all the creative choices needed to make the music. His music is shared and available for others on YouTube.

Ian and his assistant went by bus to an activity center five days every week. Each Tuesday Ian spent three hours in the kitchen together with a small group of other young people. The group collaborated in preparing food, and each group member had a different task. One of Ian's tasks was to administer the food processor. Some days Ian was more attentive and active than other days. On active days, he was able to find and push the on/ off button himself, other days he needed to do this task together with his assistant. When they had turned on the food processor the assistant set a timer that would ring when it was time to turn the food processor off. On active days Ian shouted out when the clock rang to alert the others that the time was up.

Kitchen activities include a variety of tasks and sensory inputs. All group meals are social events, and when the cooking was finished the group would eat together around the table. For Ian, eating was restricted due to medical issues, but he could still have a little taste of the food. After the joint meal, Ian and his assistant brought food to other parts of the center. Most people will show enthusiasm when a nice young man offers them freshly cooked food, so there was a lot of positive feedback and enjoyable communication at this endpoint of the activity.

It is not clear if and when a person in this phase is no longer able to learn new skills or remember new information. Clinical experience suggests that learning is becoming restricted to areas of special interest and significant events in the individual's life. However, participation in diverse life situations may still be possible and is a prerequisite for learning and maintaining knowledge and skills, for creating interest and engagement, and for being part of the social and cultural community.

# Mathematics education and visual impairment

Mathematical skills are used in many everyday situations. A practical understanding of mathematics enhances the individual's independence, and a basic understanding of mathematics is necessary for full inclusion in school and society. However, when experience is limited, mastery of mathematical skills
may depend on how these skills are learned, and they may not be mastered in all situations. One study found that 9–15-year-old street vendors in Brazil were able to solve mathematical problems when they sold fruit on the street, but not in relation to other activities, even if the problems by and large required the same level of mathematical skills (Nunes, Schlieman, & Carraher, 1993). Similarly, British children who have learned to work out pre-formulated math exercises may encounter difficulties when confronted with practical math problems in text form (Desforges, 1998). This indicates that mathematical reasoning may be grounded in concrete experiences, and at least in the early stages is activity-specific rather than context-independent knowledge (von Tetzchner, 2019). This may have implications for teaching and maintenance of mathematical skills.

Mathematics is an important school subject for all students, including students with severe visual impairment. Many students with blindness follow the grade curriculum when individual adaptations are sufficient and adequate. This however depends on teachers who have relevant knowledge, competence and access to necessary support (Rosenblum & Smith, 2012). Teaching mathematics requires mathematical and pedagogical competence (Ball, Hill, & Bass, 2005; Silverman & Thompson, 2008). Teaching mathematics to children with severe visual impairment depends on three elements that require additional competence. Firstly, teaching must be based on an analysis of mathematics as a discipline to ensure that it really is mathematics that is taught in class and not other school subjects. Secondly, severe visual impairment requires teaching methodology that compensates for the visual impairment, and the teacher must be familiar with this methodology. Finally, the teaching strategies must be adapted to the level of the individual student, to ensure an optimal curriculum (Ostad, 1982).

Children learning mathematics need to acquire some basic skills, including identifying differences and similarities, categorizing, ordering objects and quantities and understanding the principle of conservation (i.e., that a given amount or weight remains the same irrespective of the elements' size or shape). Basic skills also include the understanding and use of concepts that describe physical orientation of objects, such as «top,» «bottom,» and «beside» and concepts to compare quantities, such as «more», «less», and «the same.»

It is widely recognized that visual impairment highly influences learning of mathematics (Dick & Kubiak, 1997). Learning of basic mathematical concepts incidentally is largely based on the use of vision. Learning mathematical concepts without vision requires planned and formal teaching adapted to the individual's abilities and problems. «For children who are blind, direct teaching of mathematical concepts is essential; that is, the development of concepts must not be left to incidental learning» (Koenig & Holbrook, 2000, p. 374). However, many teachers find it difficult to teach mathematics to their students with visual impairment, because much of the material used with the sighted peers is not useful. During the first school years, teaching mathematical concepts to children with typical development is mainly based on visual illustrations. Most of this material is difficult or impossible to adapt to tactile usage. To ensure that students with visual impairment have equal access to mathematics, teachers need at least a basic understanding of how mathematical concepts and operations may be presented and explained to students without sight. Coordination of teaching for both a child who is blind and the sighted classmates is important to ensure the child's inclusion in the class. This requires close cooperation between the main teachers and a special education teacher with specialization in visual impairment.

In spite of difficulties, many children with visual impairment do well in mathematics. One study of 248 students with visual impairment born between 1967 and 2001 found that 141 of the students (56.9 %) were taught mathematics on grade level. However, the study found considerable variation between students with different diagnoses: «Students with diagnoses related to the central nervous system had a comparatively higher risk of not attaining their normal grade level in mathematics» (Klingenberg et al., 2012, p. 93). Similarly, Erin and Koenig (1997) found that different causes of visual impairment lead to different spectra of symptoms, and that diagnoses with a neurological origin tended to affect both vision and learning in general.

## Teaching mathematics to students with JNCL

Children with JNCL are born with normal vision and during the early years, they develop mathematical concepts in the same way as sighted children. Because of this, their understanding of mathematics concepts will be based on visual experiences when they start school. As their vision declines, they need gradually more adaptation to compensate for the loss of vision, although the conceptual foundation from the years with full vision gives them a good basis for further learning.

In the present survey (Appendix A), seventy-seven percent of the parents found that the mathematics education had a low impact on their child's daily life after school age. Mathematics had still been an important part of their curriculum. A small study of children and adolescents with JNCL found that most of them mastered all four arithmetical operations (i.e., addition, subtraction, multiplication and division) for a period of time, but only addition and the ability to count were still mastered in adulthood (Åberg, 2001). The present survey found that a few adults with JNCL were able to apply mental calculation in some situations, for example when playing cards or counting money. This indicates that mathematical skills may be maintained within some activities, in line with the assumption that mathematical skills may be grounded and maintained in specific activities (von Tetzchner, 2019). One 25-year-old man with JNCL was reported to master all four arithmetical operations by using mental arithmetics. Another young man with JNCL was able to calculate with the help of a calculator. He had shown special interest in mathematics from early school years. This demonstrates that special interests may be utilized for teaching mathematics (Elmerskog & Fosse, 2012; see Chapter 16).

Students with JNCL differ when it comes to personal interests, strengths and abilities. Some of them have a persistent interest in mathematics. They show strong intrinsic motivation for exploring mathematical tasks and acquiring mathematical skills, which is a good precondition for learning. Knowledge about their challenges related to vision loss and about future decline should not reduce the teaching of mathematics to students with JNCL. On the other hand, as with children with typical development, there are children and adolescents with JNCL who are not interested in mathematics and find it boring or difficult. The content of the curriculum and the pace of teaching should be adapted to each student, so that their learning potential can be fully utilized.

## Key skills

Children with JNCL have experiences and ideas about space and shape, and they have elementary knowledge about arrays and numbers. They have learned many words and concepts related to quantity and calculations. Children typically develop intermodal and amodal perception early in life (Bahrick, 2004) and get rich intermodal experience in construction play and other activities. They have counted objects and gradually understood that the last counted number is the number of objects in the set as a whole. Many students with JNCL have already started developing these key skills before the onset of visual decline. This gives them a good starting point compared to children who are born blind.

When children with JNCL start school, their understanding of the key mathematical concepts should be assessed before starting teaching mathematics. The tactile sense will become increasingly important when vision declines and students with JNCL should be given opportunities for learning to use the tactile sense to explore groups of objects with different characteristics with their hands. Access to physical manipulation of objects in the environment will enhance students' understanding of basic concepts (Csocsán, Klingenberg, Koskinen, & Sjöstedt, 2002). The tactile sense may be used by students with JNCL when counting and matching numbers to arrays of objects as well as finding matching quantities of objects that differ in size, shape and weight, although it may be noted that also blindfolded sighted children have difficulties with non-visual recognition (von Tetzchner & Martinsen, 1980). The serial aspect is important for counting forwards and backwards, knowing the next number, matching one-to-one in counting, and counting in ones, fives and tens (Koenig & Holbrook, 2000).

During the first grades, sighted students make extensive use of pictures to enhance comprehension. Students who are blind must instead have access to tactile materials. For children with JNCL these key skills can be learned and supported by using tactile illustrations, bead strings or objects, or using vision for children with enough residual vision to locate and recognize the materials. Matching numbers to arrays of objects often requires extended learning opportunities in addition to adapted materials. Knowing number facts by heart will facilitate further learning. Number facts may for example be: 4 = 4+0, 3+1, 2+2, 1+3, and 4+0. The multiplication table is another example. Automatization of number facts will help students to do calculations with more ease and efficiency, and the skills may be more resistant to future cognitive decline.

To adapt teaching to the needs of students with severe visual impairment, schools need support from visual impairment specialists, because ready-made materials are sparse. Mathematics is a complex domain and students with JNCL should learn basic mathematics skills early.

From she was little Sally liked numbers and mathematics became a favorite subject at school. During the first school years Sally learned a lot of number facts within addition, subtraction and multiplication before cognitive decline made further learning difficult. She continued to do calculations using this knowledge. Her mathematical skills declined in line with her cognitive decline. Addition was most resistant to decline. Eventually, she was no longer able to bridge ten but she could still do calculations like 12+5. Doing calculations remained one of her favorite activities.

Students with JNCL will probably never be able to do advanced mathematics. Therefore, the basic mathematics curriculum for the first school years is the most important. Skills acquired early can continue to be part of the individual repertoire for many years, like for Sally above.

## Linear algorithms for students with JNCL

An algorithm is a procedure and a specific way of writing mathematical problems. For sighted students, algorithms are often written using several lines to place the 1s in the 1-column, the 10s in the 10-column, and so on (see Figure 12.1). This is

_	_	_	_
+ 27	+ 17	+ 42	+ 22
12	43	31	57

Figure 12.1 From a visual workbook

functional for students who can see. When tasks are written in a book, students can get an immediate overview over the tasks on the page. Visual reading makes orientation on the book page very flexible.

For students with little or no vision, presentation of tasks like in Figure 12.1 will make orientation on the book page and doing calculations more difficult. For braille readers, tasks are most easily accessed by reading one line at a time. Algorithms should be linear, at least when teaching basic mathematics. Advanced braille users can learn several algorithms for doing arithmetic calculations, like the Nemeth Code (Nemeth, 1972) or the Unified English Braille (Cryer, Home, & Morley, 2013). Students with JNCL, however, might be better off continuing using the linear algorithms first learned.

Figure 12.2 shows one way a student with visual impairment might solve a task writing the calculation on one line in 8 dot braille (see Kacorri & Kouroupetroglou, 2013; see also Chapter 14). For children with JNCL with emergent cognitive decline the use of linear algorithms should be sustained.



Figure 12.2 15+29 = 30+14 = 44 in 8-dot braille

## Mental math

«The ability to calculate mentally with efficiency is an essential skill for all students, but especially for students who are visually impaired» (Koenig & Holbrook, 2000, p. 388). Algorithms for sighted students include notation for every step in the calculation to support memory, like when bridging 10. Doing calculations without the use of vision is more taxing on working memory. Below are a few examples that show how students who are visually impaired may solve some mathematical tasks with the help of mental calculation. These strategies can of course be used by all students, not only students with visual impairment.