The Rian Club.

in the spirit and mood of the people surrounding you, and you saw that something was seriously wrong. The way we looked at you, the way we tried to care for you. The situation naturally challenged your need for security and predictability. Our observations did not come from great knowledge of developmental psychology or deep insight into attachment theories; parents can observe how circumstances and conditions are affecting their child. Our family situation had a significant impact on your early childhood.

DAUGHTER: *Yes, one might say that. As far as I can remember, I have always been sensitive and aware of the atmosphere and feelings around me. I have been afraid of being alone, afraid of being abandoned. Of course, I remember few specific or concrete events from this time, but my body recalls them somehow. It is reasonable to assume that as parents you were not able to be 100 percent there for me at all times psychologically or physically. You were in shock. In shock from knowing that Henning would undergo a change from being a normal and healthy boy to having a long and sometimes very stressful illness. That it would last for 32 years was absolutely unthinkable at that time. In addition, you did not know initially whether I was healthy or if I would face the same destiny as Henning. For over two years you lived with this uncertainty, before you tested me and found out that I was not in danger. Although I did not know this, I obviously felt something in the atmosphere around me.*

I have a daughter now who is three years old. People say she is exactly like me, both in how she looks and the way she behaves. She is very attentive to my emotions, if I am sad or not feeling well. She has given me new insights and understanding about my early life, when you (the parents) underwent the most comprehensive and profound crisis of your life.

Father: As time went by, your ability to adapt to ongoing changes in life was always a challenge. You did not have anything to compare to, your situation was so different from others, but you coped in a natural and very impressive way. You were in a situation where you had to develop skills and ways to care and support Henning. You undertook responsibilities unknown to most children of your age. In addition, you had to cope with our worries and strains. We were not able to hide the truth from you, or what would eventually happen to Henning, and what the future would bring.

Daughter: *As I remember it, you started to give me little hints at an early age about what was going to happen to him. Later I understood that I had been fortunate. Fortunate because you included me in your lives and did not put me on the sideline. I feel I have always been an important member of the Rian club. There have always been enough hugs and love in our family. The openness, trust and faith you showed me have strengthened my relationship with you. I feel we are so close, we can talk about anything today. Almost! When I was ten years old, you and Mom told me the truth about Henning's diagnosis. I was aware that he was blind, had epileptic seizures and eventually would be dependent on a wheelchair. One day when Henning was away, we rented a movie called "Lorenzo's Oil". It was about a boy who had a progressive disease. Like most American movies, it had a happy ending. They found a cure for his terminal diagnosis. After we had returned the movie to the video shop, we sat in the car and chatted. I said I was happy that Henning will only be confined to a wheelchair and not die because of his disease. You told me: «In reality the boy in the film did not survive the disease. He eventually died.» The whole truth came out. I remember we were sitting in the car – it was Autumn and it was raining. You told me that the way Henning spoke would eventually become more difficult to understand, and he would probably not live much longer than 20 years of age. What I was told about Henning's time of death was fortunately wrong, but many pieces of the puzzle fell into place, things I had been wondering about.*

The time that followed was tough. I tried to hold my tears back when I was at home, but I cried every day at school. Fortunately, I had long hair so nobody could see me crying. For a long time you tried to establish contact between me and the health system. You wanted me to talk to a professional about things that

were difficult to talk about at home. You did not manage this before my teacher noticed the problem and contacted the local psychological-educational agency. Sadly, I did not get any meaningful support from them. The first psychologist suddenly retired from his job, and I was not able to establish a close relationship with the next one as the chemistry between us just was not there. Neither did you have any close friends or family that could act as support to me. I could not tell anyone what I knew about Henning. Finally I shared the secret with my best friend. She could bear it for a while, but I suppose I became too intense, too cautious, too precious, too close, and too sad. I felt I was the only one in the world with this grief. That nobody understood me. At home, I was no longer the little sister of Henning. I was Henning's big sister. Intellectually I had overtaken him a long time ago.

Father: As you became older, Linn Sophie, how did you feel about your life with us, life in a different family? What advantages or limitations were there? We had contact with others in the same situation, but how did you find it?

Daughter: *Meeting others in the same situation was very important. My best friend had a big brother who was the same age and had the same diagnosis as Henning. I became very fond of him. Unfortunately, they lived a long way from us, so we could not meet often. We did not talk about sickness and sadness much when we met, but we had a very special relationship because we were both in the same situation. It was good to know that I was not the only one in the world experiencing and facing this.*

The job of raising me was certainly not easy for you and I do not feel that you asked that much of me as a sister or daughter. I suppose you felt you could not ask me to do things Henning was unable to do. I finally realized that this was wrong; there had to be more expectation on me than on Henning. Today, as an adult, I think you should have made more demands on me than you did, because I am still a bungler and muddler today!

There were some limitations having a brother like Henning. It was not possible for you to let me participate in all the usual sports and activities. You had more than enough on your plate. It was just not possible for you to do additional things as other parents did, like selling cakes, or tickets for the local handball club or brass band. However, it was possible for me to participate in ballet dance. I loved it, but at the same time I missed being a part of the community that other girls in my class were part of. On the other hand, I experienced many positive things that other children my age never had the opportunity to be a part of. For instance, we went on tours with the family association you were members of,

and on many exciting tours to Mediterranean countries. Resource centers for JNCL, like Frambu (Competence Centre for Rare Diagnoses) and Tambartun (Special Education Centre) also organized meetings. These gatherings gave me great experiences my friends never had the opportunity to have.

Father: The fact that Henning lived at home for almost twenty years certainly influenced your formative years. We tried to have an open house, and many children came and went on a daily basis. How did you find this?

Daughter: *When my best friends came to visit me, Henning usually joined in, but sometimes we wanted to be alone without him. Henning could be demanding at times. When he did not feel well, for example, he would sit in his room shouting: «I'm a lonely little blind boy.» I was just not able to compensate for his need to have friends. I would meet up with one of his classmates and ask him if he could visit Henning. Sometimes he said yes. However, the reality was that most of Henning's many so-called good friends were just friends in his imagination.*

Being with Henning was often tiring. His incessant talking was exhausting. He talked about dinosaurs, Indians, Elvis and all the plastic figures he had in his collection. He had hundreds of smurfs, he was particularly fond of the inhabitants of Duckburg (Walt Disney). He talked without drawing breath. At times it was impossible to have a conversation with him; it was always on his terms. You – my parents – could not hide being tired by Henning's behavior. When I saw it getting too much for you, I would take Henning out of the living room, and start reading a book to him. I am told that I did this before I had even learnt to read. We cannot deny that the pressure on us at times was overwhelming. I remember I once drew a painting showing you, Daddy, standing and pointing at the door. Henning was on his way out through the doorway while Mom was crying. When you were very tired, you could be tougher and stricter. This made me afraid that you were ready to have Henning leave our home.

Father: As you became a teenager, Henning became more and more affected by the disease and we all had to deal with new challenges. You continued to be a great carer, but you also needed to break away from us, like other young people of your age. Did this raise any issues for you?

Daughter: *I remember I wished that the mysteries behind Spielmeyer-Vogt disease (JNCL) would be understood and the problems solved. Maybe there could be a cure for Henning's disease? Maybe in the future I could do medical research and find a treatment or cure? As the years went by, I realized that this was just a pipe dream.*

Sister Linn Sophie and brother Henning.

When I became a teenager, I was still very loyal to you. But I also had a strong need to rebel like other young people. Our closeness meant that you discovered most of the crazy things I did. Well – almost everything! Henning had become psychotic by this time. In his reality, I was no longer his little sister. He was dead, we were all dead, he felt he was losing all his body parts. We had to help him pick up his fingers and put them back in place. There was no smile. No pleasure. Henning was given strong medication and he was "gone" for a long period. After almost two years, the medication was phased out and a wonderful thing happened. Somehow Henning woke up to life again. The psychosis was gone and he became the good, kind version of Henning we had missed so much for so long. But with one main difference – he used far fewer words now. It was sad to see him gradually lose abilities. I do not remember much from this time and I think I was probably more concerned about myself at this age.

I see myself as a positive person, but in a melancholy emotional state. Eventually, I realized that I could not put my life on hold. Nobody knew how long Henning was going to live. When I was 19 years old, I went to Bali to study for three months. I was very worried that something would happen to Henning

while I was away. You, Mom and I had the same feelings and concerns, which caused a bad conscience because I had left him. I tried to compensate for this by sending many sound clips and postcards to you all.

After returning to Norway, I continued to be 400 kilometers from home as I started my nursing training. During my three years of study, I travelled home every other weekend to see Henning. It was important for me to see him because I really wanted to, not because I felt I had to. Then I moved even further away – 1200 kilometres – to Tromsø. The worry continued when I later moved from Tromsø to Oslo, 550 kilometers from home, for further training as a children's nurse. I had particularly difficult times when I thought that Henning must die soon so we would be able to "breathe" again. I loved Henning so much, but to continually worry about the future, without knowing when and how it might end, was just terrible.

Henning and I had a very close relationship. Mom and Henning used to phone me when I was away. Although Henning had lost his speech, he was more talkative than anyone with similar communication issues I have ever met. He spoke to me in his own language, and it was easy for me to translate this into «I love you and I miss you, sister.» These phone calls made my life much easier. But the very best times were when I was at home and could lie in the crook of Henning's arm, and he would be smiling and laughing at having his little sister in his arms.

Father: We have often wondered how it was for you growing up with a brother like Henning. What do you think the consequences have been, and how does it affect your life today?

Daughter: *Our life with Henning has made me different to other people. I have been aware of this for some time. My opening line when meeting new people was often: «Hello, my name is Linn Sophie and I have a brother who has a rare disease called Spielmeyer-Vogt.» The responses and reactions varied a lot. Some people were curious and asked more. Others just fled. Why was it so important for me to tell them about Henning? It must be allowed to share that he took a lot of space in my daily life. The feeling of being different to others gave me an enormous need to explain my life to them. It is clear that this has scared away some people over the years. I understand this now, but it hurt me every time I felt abandoned by others. I lost many friends because of this. It took time for me to create a distance from these feelings, and to find my own clear identity as Linn Sophie. I now find it easier to share less, but it can still be difficult. Even after his death. I am still so proud of my wonderful brother! Maybe I have been influenced by debates in society about attitudes to people with special needs,*

which make me feel I have to explain to everyone how wonderful Henning was. To hear that people like Henning are seen as burdens on society, vegetables, or a waste of money makes me sad and angry. Who would I have been without Henning? Henning shaped me. He made me what I am today. I am very cautious. I am sensitive and vulnerable. But I am also a caring, kind and cheerful person.

Henning's final phase of life was difficult. In retrospect, I am very grateful you involved me in all the care and nursing decisions. At times I had strong opinions about life-extending measures and resuscitation. You allowed me to express my thoughts. Addressing such ethical dilemmas was difficult for you, me and other caregivers who helped him at the end. It was important for me not to be Henning's nurse during this difficult time even though I had professional competence in the field. I just wanted to be his sister.

Henning has taught me to appreciate the small things in life and to realize what is important. Henning is not here anymore but he will never be forgotten. His 38 years long life had a decisive and definitive effect on me and on many others. He really lived his life; he rejoiced in his way of living and lived in a way that many could learn from. His headstone bears the following inscription: «Thank you for everything you taught us about life and love.»

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26

Family Organizations and Advocacy Groups

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At times our own light goes out and is rekindled by a spark from another person. Each of us has cause to think with deep gratitude of those who have lighted the flame within us.

Albert Schweitzer

Family advocacy groups and other groups safeguarding the interests of individuals with juvenile neuronal ceroid lipofuscinosis (JNCL) play a key role in enabling their members to cope, and thrive in the midst of a severe diagnosis. When parents learn of their child's diagnosis of JNCL, they often are in shock about how this will change their child's life and that of the rest of their family. They ask questions like *How will I care for my child? How did this happen? Why didn't I know how to protect against this? What does this mean?* As the early stages of shock begin to fade into the routine of the "new normal," more questions arise. These big life questions, as well as the more practical questions about how to manage, are most often best answered by other parents who have walked this road with their children. The overarching goal of family organizations and advocacy groups is to help all families facing the disease feel less isolated and to know that others understand their day-to-day worries. They are helped to move from despair to living, from shock to action, and to finding joy again. Parents help each other learn how to travel a new road that was not on the original map for their family. In some countries, the family groups have grown to raise much-needed funds for research and provide educational, social and other support services.

Advocacy groups are organized in many countries to provide special emphasis on the life of the children in school and vocational settings. Here the term "school and vocational setting" is used broadly and includes post-secondary learning, quality of life and learning needs that go beyond the immediate classroom.

This chapter presents advocacy groups related to individuals with JNCL, their philosophies as well as how some advocacy groups provide services to

families and children with JNCL. The chapter includes the parents' associations of Norway, Denmark, Germany, UK, USA and Finland, and a representative of The Royal Blind School in Scotland (Internet addresses can be found at the end of the chapter).

Norsk Spielmeyer-Vogt Forening (The Norwegian NCL Family Association)

Norsk Spielmeyer-Vogt Forening (Norwegian NCL Family Association), or NSVF, was inaugurated in 1980. In 2018, it has more than 320 members, including most of the 33 children and young adults with JNCL or other NCLs in Norway. Day-to-day business of the Association is managed by a board, elected annually by the annual meeting of the members. In addition, several parent members are engaged in project work and other vital tasks for the Association. Work thus carried out is done *pro bono*.

NSVF is mainly concerned with the «living with» aspect in a life-time perspective, and initiates a number of activities aimed at improving life as it is lived by the families. Every year, there are several events, where parents and other family members meet informally to discuss the various challenges of living with the disease and to provide a place for social interaction among the family members. One of the events is a free holiday during the summer at a holiday resort in the southern part of Norway. Here children and young people with JNCL and their families and service providers can enjoy a week together away from the daily grind. In the autumn, there is a long family week-end – Thursday through Sunday – at a center near Oslo belonging to The Norwegian Association of the Blind and Partially Sighted. Professionals are engaged to facilitate activities such as swimming, boating and fishing in the nearby lake, wall-climbing, ATV driving, horse riding, obstacle routes, and a number of other challenging and enjoyable tasks for the children and young people with JNCL. In addition, there are various entertainments at night; bingo and quiz are always big hits!

The Association initiates and participates regularly in a number of projects. For several years, NSVF has worked with the Oslo University Hospital to provide a national clinical service for children and young people with JNCL. The hospital is now a National Resource Center for NCL, where physicians and parents may obtain guidance and counseling related to NCL diseases. In 2016, NSVF and Oslo University Hospital published *Guide to symptomatic treatment of neuronal ceroid lipofuscinosis*, which can be downloaded in Norwegian and English from https://www.nsvf.org/Om-NCL-sykdommene/Medisinsk-informasjon-og-oppfølging/Medisinsk-veileder/.

NSVF's website provides free expert advice to families on topics related to legal rights within the medical, educational, social and psychosocial aspects of living with a child who has JNCL. This service is possible through the Association's collaboration with a law firm which is paid an annual retainer to keep these pages up to date.

NSVF collaborates with Norway's main service institutions in the fields of medicine, education and social work. The Association has regular formal meetings with these organizations, aiming at streamlining services provided to families. This group has developed a set of formal procedures for relaying information about the diagnosis and supporting the family in the first period following this disclosure (see Chapter 24). NSVF has a close cooperation with Statped, a national service for special needs education. The need for early intervention is recognized by this institution, which provides specialized support to schools and other educational institutions throughout Norway.

NCL Danmark (The Danish NCL Family Association)

NCL Danmark (previously Dansk Spielmeyer-Vogt Forening) was founded in 1983. In Denmark there are presently 23 children and young adults diagnosed with NCL. They include one with CLN1, one with CLN2, one with CLN8 and 20 with CLN3 (JNCL). The Association has about 300 members.

The goal of the Danish NCL Association is to provide possibilities for parents to meet and talk with each other about the many issues and challenges they meet in daily life with a child who has NCL. The parents meet three times each year. One weekend is for the general meeting with various courses for the parents. There are nice surroundings and entertainment to promote relaxation and socialisation among parents. One weekend is arranged by the Danish Batten Team (see below) where different problems and issues are discussed. Professor John Østergaard has always participated in this meeting, informing about the latest news in medical research and practice related to NCL. Finally there is one social family weekend with children, support staff, siblings and grandparents. The parents know each other well and there is a very close personal network within the group.

The children and young adults with NCL meet six times each year. This includes one week winter holiday, one week summer camp, one weekend in Legoland (an amusement park in Denmark), the family weekend and two weekends in spring and autumn organized by the Association. These weekends and holidays are very special for the children and young adults with NCL. They know each

other from the very beginning and become best friends. Everyone participates, and there is no differentiation between children, adolescents and adults.

In Denmark, the municipalities and the Government are responsible for providing care services to people with special needs, including people with a rare disease. The social welfare system enables families to choose either to keep their child with JNCL at home during adolescence and adulthood, or live in a sheltered home. In both cases, the young adults will get the help and support they need, and a social pension from the age of 18. At present, most of the young adults are living in their own apartment connected to their parents' home. For their support and care, the municipality provides pedagogical and nursing staff in their home, as well as in schools or activity centers they are attending.

A father's meeting with NCL Danmark

My way to handle mourning is to do things. Just after receiving the diagnosis I wanted to do something that was good for my son. I insisted on all sorts of experiences. We should have a fire and roast sausages. I didn't care whether my son found this okay, I had to feed him with experiences that he could store in his memory. So we did all kinds of things.

The counselor from our Team told about a computer program that could help my son, and in October, 1½ months after the diagnosis, I was on my way to Kalundborg to take part in a course about the program. I knew there would be two other parents present. It was both good and anxiety-provoking.

On the road to Kalundborg I got sick. I had severe stomach pain and had to throw up. I pulled over and thought I'd better turn around, now when I felt so bad. After a serious discussion, I agreed with myself that I was hit by an unknown and irrational fear; that I had to stop thinking and just go on.

In the course room, I sat with 25 other people and looked around. I looked to find the two most sorrowful faces (to find the parents who were present). I didn't find them!

Late afternoon after my first day on the course, I was sitting in the smoking room with my pipe and the door opens. Two women came in and asked whether it is me who is Carsten, the new boy's father. They did not look particularly mournful, but in return they knew exactly how I felt. They were not afraid to make me sad, which meant they could ask me about everything and even help where it was hard for me to explain. It was an indescribable relief.

At this event, a new and life-long learning process started. The very first thing I learned in interactions with parents who were in the same situation,

was that you can have a good life, even if you have a child with JNCL. We eat and drink (a little too much), we laugh and cry, we enjoy and we become wiser. For me, the other parents with children who have NCL are my family. This is true in every way and also includes the parents I have not met yet. In the NCL family there are some you like a lot and some you only meet occasionally. In the family you can talk about everything and you will bear with everything.

The Danish NCL Team

In Denmark there is a unique expert team for NCL, The NCL Team (the Team). The Team mainly exists due to the initiatives of a strong and determined parent association, NCL Danmark. During the 1990s, the Association succeeded in getting the Team started as a temporary project financed by the Government. Today the Team is permanent, financed by the Government.

The Team is cross-disciplinary, which enables it to handle the very complex issues of families with a member who has NCL and to see the problems from different perspectives. There are six members in the Team: one social worker, two special education consultants, one doctor and two parents representing NCL Danmark. The social worker is primarily responsible for social and legal issues in relation to the authorities. This includes the primary contact with social services regarding housing, transport, relief services, pensions and many other relevant tasks. The social worker is also the coordinator of the daily function of the Team. The educational consultants help the families with issues related to school, support staff and after school facilities, and they provide supervision for all professionals working with the children. Each year, the social worker and the two educational consultants organize a three-day course for support staff working with NCL.

The Team's medical doctor is in direct contact with all the children, and provides medical examinations at Skejby Hospital. The doctor is reachable practically around the clock and also makes home visits when needed. If a child with NCL is hospitalized, the doctor always stays in close contact with the local hospital, which in most cases has no experience with NCL. Furthermore, the Team doctor is in regular contact with the local doctors. Most of them have never heard about the disease before. The doctor, as well as the rest of the Team, collects knowledge about all aspects of the NCL diseases.

The parent representatives are elected by NCL Danmark. They are the family experts and know the many complex problems that may arise, because they live in it, live with it and they understand it. They know how it is for the family to be informed about their child's diagnosis, to experience grief, to realize that the child

has problems at school or needs to change school, that there is a need for support in the home or for moving to a residential home, to get a handicap accessible car, and to deal with hundreds and hundreds of meetings with the social services and other public institutions. As Team members, the parent representatives can influence professionals to react and handle the various cases in a more specific and relevant manner, and without losing the human touch. Like the rest of the Team, the parents are bound by professional confidentiality. This is important, because the work of the Team concerns very personal issues. The Team members do their job with great respect for the families they support.

The primary intention behind the Team was to create a support and counseling system for children who had recently been diagnosed and their families. The team has established a set of formal procedures for informing families about the diagnosis (see Chapter 24). It maintains cooperation with the families and with the local educational, health and social authorities that are involved in the daily lives of the children with NCL and their families. The Team is supporting children and adults with NCL and their families throughout life.

NCL-Gruppe Deutschland e.V. (The German NCL Family Association)

In 1977, parents and educators at a school for blind children in Hamburg formed a «community of interests for the advancement and care of children and adolescents with Vogt-Spielmeyer syndrome», the predecessor of NCL-Gruppe Deutschland e.V. In December 1989, NCL-Gruppe Deutschland e.V. was established for children with all forms of NCL diseases and their families throughout Germany. Today, NCL-Gruppe Deutschland e.V. has about 400 members and in 2018 there are eight members with CLN1, 15 with CLN2, 25 with CLN3, one with CLN5 and two with CLN6. Several other families with a child who has a NCL disease get help from the Association without being members.

NCL-Gruppe Deutschland e.V. is a family association and its important functions include providing mutual support, to be there for each other after the diagnosis, to bring the families back on a solid track, and to foster exchange of information and connections between the families who have a child with NCL. The Association provides advice, assistance and support in issues of everyday life as well as special requirements, to the families, their relatives and friends, professionals, teachers and other people who are interested in NCL. This is done through personal conversations and long telephone calls, by email, via the Association's official website and closed group on Facebook, and by distributing information material.

Smaller meetings are organized in four regional chapters, according to the needs and wishes of the members in each region. There is one annual seminar only for mothers and another one only for fathers, with expert speakers on different topics and enough time for the participants to talk and share their thoughts and experiences. For several years, an annual weekend meeting has been organized for siblings of children with NCL, where they can have fun, exchange their feelings and talk about everything in their own peer group.

In contrast to these meetings, the Association's annual conference is for the whole family. The children with NCL and the siblings have their own program, and are taken care of by dedicated caregivers and supporters – with highlights like a visit of a fire brigade with several fire engines. The parents can attend different workshops, talks by scientists, medical and social law professionals, and exchange experiences with other families. They have the opportunity to talk about the needs of their children with experienced doctors from the NCL Counseling Center in Hamburg. The Association works closely with this hospital unit, and families go there regularly with their child with NCL for medical examination and help.

Very important are the two ten-day holiday camps that are organized each year, each with ten children with NCL and lots of dedicated caregivers. The children with NCL can do the same things as other children, teenagers or young adults during this free time, which is different from their ordinary daily life. Over the years, they get to know each other well and they are really looking forward to these camps. The activities may include cinema and concert visits, shopping, swimming, barbecue, bonfire, listening to and making music, or visit to a leisure park, including a roller coaster ride.

Through its participation in the present ERASMUS+ project on education and JNCL (the JNCL and Education Project), NCL-Gruppe Deutschland e.V. seeks to improve education and communication for children with JNCL and give caregivers and teachers better insight into the special needs of the children with JNCL. Germany is a federation with 16 states, each with a different school system, and it is a hope that the ERASMUS+ project will help provide the best education for children with NCL in all the states. Moreover, to achieve the best outcome of education, an early diagnosis of NCL is essential. Through several activities, the Association endeavors to increase knowledge and awareness about the disease and thus the probability of early detection and diagnosis and thereby to provide the best possible support and therapy in daily life and school. Information material is sent to hospitals, pharmacies, therapists, students and doctors. NCL-Gruppe Deutschland e.V. tries to inform journalists, has information stands on fairs, in banks and town halls, and organizes events in several German cities on the Rare Disease day at the end of February. The Association also tries to get a foot into the symposia for doctors, to provide information that might raise their awareness not

only of the challenges imposed by NCL diseases, but the potential for a rich life despite the consequences of the disease.

Apart from providing public education services to diverse audiences, the Association is increasingly involved in NCL research, international networking, and legislation. Although the Association recognizes that coping with NCL diseases is an ongoing reality, it also seeks to draw attention to how people with these conditions, together with their families, can participate in satisfying life experiences.

Batten Disease Family Association (BDFA)

The BDFA is the only UK National Charity that supports families who have a child, adolescent or adult with JNCL. Although the team is small, the BDFA supports families across the UK, providing holistic support to ensure that children and young people with JNCL receive the care and support services that they will need in different phases of the disease.

The role of the support and advocacy team is to interact with families from the point of diagnosis and throughout life. This can be through visits to the family home, via emails or phone calls and accompanying them to large multi-disciplinary team meetings. Often families are not given much of an explanation of the disease from the hospital and are often left with feelings of shock and loneliness. The BDFA provides more detailed information to families and can give anonymized examples of other family situations that may be similar to their own to provide answers to difficult questions and to help families feel that they are not alone.

The BDFA provides holistic, person-centered practice for families and individuals. Not only does the BDFA look at the needs of the child but also the needs of the wider family around them and the needs of the professionals working with them. The BDFA aims to empower and build resilient families to be able to share their views at meetings which sometimes have over fifteen professionals attending, and to help build a solid support network around them. Families often have medical questions that sometimes their local team struggles to answer. The clinical nurse specialist of the BDFA is able to talk to families if their local team cannot provide sufficient answers to their questions.

It is important for families to feel empowered when making decisions about their child's care as well as being able to make others aware of their child's needs. The BDFA offers all families support by providing an information packet called a *family folder*. This folder gives a brief medical description of the different symptoms of the disease and information on education, social care, research and genetics. The feedback from families about the folder has been positive. Many take the folder

to meetings with professionals who know nothing about JNCL and parents take it with them if they unexpectedly have to go to hospital in an emergency, so that the staff there can quickly learn about the symptoms and management of the disease.

The BDFA provides emotional support to the family and the extended family through a free phone helpline. This helpline is also used by professionals who work with young people with JNCL for emotional support and for advice, signposting and guidance. School visits form a significant part of the support and advocacy program. Often, the children with JNCL are in mainstream school when they are diagnosed. The BDFA works with these schools to provide specialist training on JNCL and to provide practical strategies that the schools can use. This may include behavioral strategies, information on JNCL and visual impairment, and how to work with young people with dementia.

Social isolation is often an issue for families with a child or adolescent who has JNCL. Often families feel alone as they do not know of any family in their area with a child with the same disease. Social media play a huge part in reducing that isolation for the families, and the BDFA often puts families in contact with each other or adds them to various secure discussion groups on websites for parents of children with JNCL.

The BDFA holds an annual family conference which is free for families to attend. Professionals from across the UK also attend the conference to speak with families about education, research, social care, and health care. Families value this time to be able to get together with professionals and with families they may have only met through social media. The BDFA recognizes that for most families this is not easy. Parents with children who were recently diagnosed will see young people who are much further in the disease progression than their own child. Families with older children will see children who are in the initial phases of the disease, and bereaved families may be attending the conference for the first time without their family member with JNCL. However, despite being at different stages, they value the time that they spend together, sharing their experiences and gaining knowledge from others who have been in the same situation. For bereaved families, there is space for reflection in the remembrance room.

At the conference, the BDFA provides a complete program of care and activities for children and young people, enabling them to interact with one another, for siblings to gain support, and for parents and caregivers to access the professionals in the knowledge that their children and young people have professional support.

Another function of the BDFA is to support families through decisions about their young person's care needs and where they will be living once they reach 18 years. Many families choose for their young person to remain with them at home, although some young people go to live at Heather House, a care home which has provided care for many young people with JNCL over the past few years. The

BDFA also supports families to find a counselor who knows about JNCL, as many of the local counselors are not familiar with the disease and families can find this very unhelpful.

Collaborative working is the core to the way in which the BDFA delivers its services to children and families. The Association does not believe that it can or should try to support families on its own; the BDFA will continue to advocate on behalf of the families, working with other disability organizations to improve services and care for children and young people with all types of NCL diseases across the UK, because it is only together that stakeholders can make a difference.

Notes from an educator in Scotland

Children and young people with JNCL often build up what to parents is an alarming amount of documentation about the provision of education and therapy for their child. Pupils in Scotland, depending on their local authority, will have a Plan which should encompass all aspects pertinent to their situation (see also Chapter 11). In a School, such as The Royal Blind School, there will be review meetings regarding the education, health and care of the pupil. These reviews take place at least once a year and more frequently as required, for example in preparation for transitions.

For parents these reviews may be daunting owing, in part, to the number of team members present, which can exceed ten. Around the table may be a community pediatrician, an educational psychologist, local authority professionals in charge of placements for pupils with additional support needs, a social worker, a physiotherapist, a speech and language therapist, an occupational therapist, a habilitation specialist, a class or form teacher, a deputy head teacher and a principal teacher. At a time of transition, there will also be professionals from the service the pupil will be attending. At one such review, twenty professionals attended. The parent found this overwhelming and did not attend the next reviews, even after reassurance that the numbers would be fewer. This parent had been encouraged by the school to liaise with the BDFA but could not be persuaded. The potential for a difficult transition process for the adolescent increases when the family and the team do not share goals or strategies for attaining them.

The experience of other parents was very different because they engaged with the BDFA once they had a diagnosis for their child and before the child came to the school. The role of the BDFA cannot be overestimated. The BDFA worked with the parents to communicate with the school and placement authority and the professionals responsible for implementing placement. For any parent the BDFA is also a link to meeting other parents and to attend weekends where the

whole family is welcome and can share their concerns whether as a parent or a sibling. The school's link with the BDFA ensures that the most effective practice in education for children with JNCL is shared and any new ideas or innovative practice can be shared. This kind of professional dialogue is vital to maintaining the highest standards of learning for all children and young people with JNCL.

The Batten Disease Support and Research Association (BDSRA)

Since 1987, the BDSRA has been a guiding force in advocacy for all forms of NCL. Funding research, supporting parents and developing programs that go to the heart of families' needs have been keys to the organization's success.

BDSRA reaches families across the US and world in a variety of ways. Social media, through Facebook, Twitter and Instagram provide platforms to report news, respond to questions and provide educational content to families and loved ones. At this writing, there are 900 family members and caregivers from different parts of the world on the BDSRA private page, monitored by staff members.

Families also connect in person each year at the annual three-day family conference, which includes programming for parents, extended family members, young siblings and researchers. Workshops, celebrations and remembrance are all a part of this event. Because so many are in attendance, researchers are able to engage with families to ask questions, complete natural history information about the disease, and collect blood and saliva samples. These data are keys to scientific progress.

In addition to ongoing clinical referrals for parents, school consultations for teachers, and clinical trial patient support, the BDSRA is keenly involved in research development. Parents and supporters have funded research projects for all forms of the disease, leading to some of the human clinical trials moving forward now. Each of these families raised money for research they knew would never help their own child, but that might benefit children with NCL diseases of the future.

Each year the BDSRA performs a basic and translational research merit review process, requesting proposals from around the world. Each proposal is reviewed by experts in lysosomal disease, pharmacy, genetics, pediatric neurology and other fields. Those with the highest scores are funded by BDSRA and family and other foundation partners. Family foundations such as Noah's Hope/Hope for Bridget, Our Promise to Nicholas, Beyond Batten Disease Foundation, Drew's Hope and Taylor's Tale among others fund research for the type of NCL disease their children have had.

BDSRA and other family foundations provide consultation to researchers, industry, and regulatory agencies to help them understand the voice of individual persons with JNCL in drug development. The Association does this by organizing focus groups, recruiting families to speak at public hearings, and providing letters of support for legislative efforts important to the families, such as healthcare coverage, newborn screening and improved regulatory processes to further rare disease scientific exploration.

Support, research and advocacy are the pillars of BDSRA, driven by Batten family social action, funding and hope for a brighter future.

The Finnish NCL Family Association

The Finnish NCL Family Association was registered in 2007 but has worked actively since the 1980s as a group of families with children who have JNCL. The main aim of the Association is to share information about JNCL and provide peer support to the families. The Association has about 100 members, including 35 families with a member who is diagnosed with JNCL (CLN3), two with late infantile neuronal ceroid lipofuscinosis, LINCL (CLN2 and CLN5), 20 external members and two company members. There is a separate group for infantile neuronal ceroid lipofuscinosis, INCL (CLN1), with around 40 families with a child who has this diagnosis.

The Finnish NCL Family Association organizes four to six events each year, including one or two events for families, one event for parents only, and weekends for mothers and fathers. In those events, there are usually some lectures by professionals and official annual meetings, but the main aim is to be together and have valuable peer support. The Association shares information via their webpage, email and events. It organizes a Facebook group, which is also a good way to share information and provide peer support.

The Finnish NCL Family Association has a history of strong relations with the Finnish Federation of the Visually Impaired (FFVI), which provided an NCL specialist for families until 2015. In 2017, the Association started working with the Norio Centre of rare and genetic disorders, including funding for an NCL specialist in one year. The continuation of this position depends on future funding. The Association is not getting regular funding and all activities and events are organized and managed by members of the Association. The lack of publicity and keeping a low profile are hindering the recruitment of sponsors, but raising the profile may have a negative effect for some families.

Links to the associations

Batten Disease Family Association	http://www.bdfa-uk.org.uk/
Batten Disease Support and Research Association	https://bdsra.org
Bildungszentrum für Blinde und Sehbehinderte in Hamburg	https://bzbs.hamburg.de
NCL Danmark	http://dsvf.dk
Norsk Spielmeyer-Vogt Forening	https://www.nsvf.org/
Oppimis-ja ohjauskeskus Valteri, Onerva	http://www.valteri.fi
Royal Blind School	https://www.royalblind.org/education
Statped	http://www.statped.no/
The Finnish JNCL Family Association	https://www.jncl.fi
The German NCL Family Association	http://www.ncl-deutschland.de/
Vision Centre Refsnæs	http://synref.dk/
WESC Foundation	https://www.wescfoundation.ac.uk

27

Behavioral and Emotional Reactions to a Difficult Life Situation

Stephen von Tetzchner, Jochen Lippe Holstein and Per Kristian Haugen

Juvenile neuronal ceroid lipofuscinosis (JNCL) is a serious and complex disease. Children and young people with JNCL experience a sequence of challenges, each of which may have a pervasive influence on their lives. First comes the onset of visual problems and gradual complete loss of vision, then academic problems in school, followed by motor and language problems. These impairments often lead to reduced participation in activities with peers and friends. Over time, children and young people with JNCL will realize that their own development is following a different course from that of others. Their achievements are falling behind, resulting in an increasing gap between their abilities and those of their peers. They may experience frustration and feelings of loss and insecurity related to what is happening. It is important that such feelings and the reactions of the children and young people are met with special attention and understanding. The present chapter discusses these reactions and how children and young people with JNCL can be helped to cope with their life situation in the best possible way.

Emotional and behavioral reactions and JNCL

Children and young people with JNCL will react in different ways to their difficult life situation, both emotionally and behaviorally, and with internalizing or externalizing reactions. Some of them withdraw from some or many situations, rejecting suggestions for activities that they might otherwise find very attractive. Others react strongly (i.e., overreact) to the slightest demands, possibly because of fear of failure or being an outsider, and may show aggression towards other people or objects. Sometimes such behaviors occur without an apparent cause. There are however individuals who show resilience by adapting and coping in spite of many challenges (Goldstein & Brooks, 2013; Rutter, 2013), and expressions of resilience may also be found among children, adolescents and adults with JNCL.

Although there are few studies addressing the emotional and behavioral reactions in individuals with JNCL, those studies are consistent in the finding that there is a higher level of emotional and behavioral problems in this group than in peers with typical development (Adams et al., 2006, 2013; Marshall et al., 2005; Santavuori, Linnankivi, Jaeken, Vanhanen, Telakivi, & Heiskala, 1993). Adams and associates (2006) asked parents of 25 individuals with JNCL, aged 6 to 18 years (average 12.3 years), to complete *Child Behavior Checklist* (CBCL, Achenbach & Rescorla, 2001) and *Scales of Independent Behavior* (SIB, Bruininks, Woodcock, Weatherman, & Hill, 1996). On the CBCL, the highest scores (indicating more severe problems) were on the subscales «Thought problems», «Social problems», «Somatic complaints» and «Aggressive behavior». On the aggression subscale, three participants were given scores in the clinical range and seven in borderline clinical range. The average scores for «Anxious/depressed» and «Withdrawn/depressed» were close to the general population mean of the same age. On these two subscales, one individual had a score in the clinical range and five others had scores in the borderline clinical range. Among the 20 most frequent items that were «somewhat true» or «often true» were «Can't get his/her mind off certain thoughts, obsessions», «Demands a lot of attention», «Argues a lot», «Talks too much», «Temper tantrums or hot temper», «Clings to adults or too dependent», «Sudden changes in mood or feelings», and «Impulsive or acts without thinking». On the SIB, one was given a score in the serious range and nine in the moderately serious range on the «Asocial» subscale (two items: a) Socially offensive behavior and b) Uncooperative behavior). Two individuals scored in the moderately serious range on the «Externalizing» subscale (three items: a) Hurtful to others, b) Destruction of properties and c) Disruptive behavior). None scored in the moderately or more serious range of internalizing behavior (three items: a) Hurtful to self, b) Unusual repetitive behaviors, and c) Withdrawn or inattentive behavior). The scores on the two checklists were a little above the average, indicating somewhat more problems than in the general population. However, it is important to emphasize that checklists do not represent a diagnosis. They were completed by the parents and the items on CBCL were not adapted to individuals with severe visual impairment.

In a Finnish study with 27 slightly older children and adolescents (mean age 15 years, range 9–19), the most prominent behavioral features were problems of social interaction and aggressive behavior (Bäckman, Santavuori, Åberg, & Aronen, 2006). Similar to the study of Adams and associates (2006), symptoms of depression were reported to be rare. A study with 42 children and adolescents (mean age 10.2, range 5-15 years) found that restlessness, aggressive behavior and fear were common features (Santavuori et al., 1993). Difficulties with sleep were especially addressed by Santavuori and found to be a common problem. Depression was reported for four of the 42 individuals but Santavuori considered

unrest, aggressive outbursts, anxiety and self-injury as symptoms of depression. Combining several studies comprising 70 children, Adams and associates (2013) found a small increase in raw scores on the subscales «Social problems» and «Aggressive behavior» on the CBCL from early to later childhood, and a decline in scores on these two subscales from late adolescence into adulthood. However, these averages hide considerable variation.

In the present study (Appendix A), parents were asked about various emotional and behavioral problems. About 75 percent of the participants with JNCL had shown some kind of emotional symptom (anxiety, depression, fear, etc.). These reactions tended to appear for the first time in childhood, but some appeared first in adolescence and early adulthood (Figure 27.1). Common emotional reactions were anxiety, depression, passivity and excessive sadness. As one could expect, there was sadness due to changing social settings, loss of interaction with peers and loss of friends – but still relatively high contentment despite the disease.

It is understandable that many children and young people with JNCL react with anxiety, depression, and fear. Anxiety is a typical reaction to the unknown. For example, individuals with autism spectrum disorder have difficulties understanding other people and why they act as they do, and anxiety is very common in this group (Kerns & Kendall, 2014). Depression may reflect a state of lack of power, of having little influence on decisions about one's own life, and low mood and passivity are frequent reactions in this group. Anger may be an expression of fighting back and not accepting what is happening. Depression can also express itself in agitated and disruptive behavior (American Psychiatric Association,

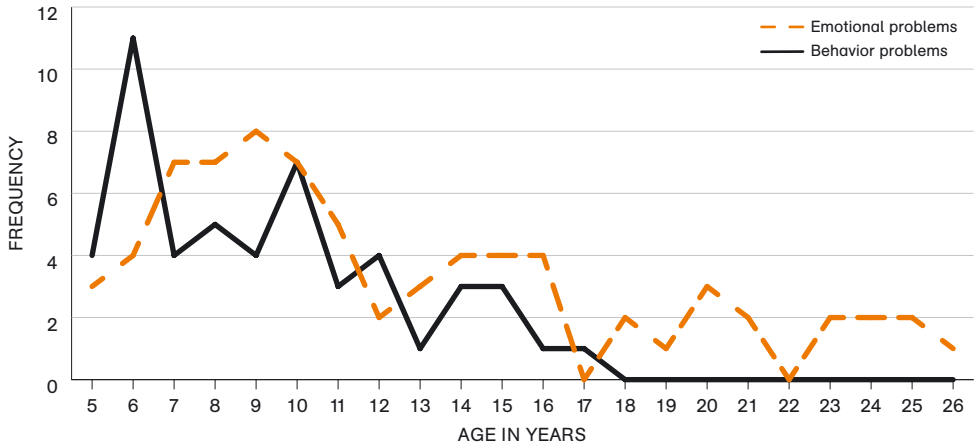

Figure 27.1 Frequency of subjects at each age when emotional reactions (anxiety, depression, fear etc.) and behavior reactions were noticed for the first time

2013), and Santavuori and associates (1993) point to unrest, aggressive outbursts and self-injury as symptoms of depression.

Behavioral reactions were observed less frequently than emotional reactions, in about 56 percent of the participants, but the distribution of first observation followed a similar pattern (Figure 27.1). The most common behavioral reactions were verbal and physical aggression, while spitting, swearing, object destruction and self-injury were less common (Figure 27.2).

The results from the present study with a relatively large sample of individuals with JNCL demonstrate that the disease often leads to emotional and behaviour reactions but also the significant variation in such reactions within this group.

Many research studies measure negative reactions to life events without providing parallel measures obtained from rating scales for positive responses. A positive attitude to life is indicated or assumed by a lack of emotional and behavioral problems. However, everyday life is usually a balance between positive and negative elements, and this situation is also true for individuals with visual impairment and cognitive challenges. Only focusing on negative elements will not sufficiently represent the overall situation of the child or young person with JNCL. This biased profile is well illustrated in the quotation below.

> *Our daughter was never aggressive or ungrateful. She accepted her disease. She was always friendly, said «please», «thank you» and «no thank you» and is very well liked at school, in her residence and in her leisure time activities. She likes many things. Eating is most important for her; she loves*

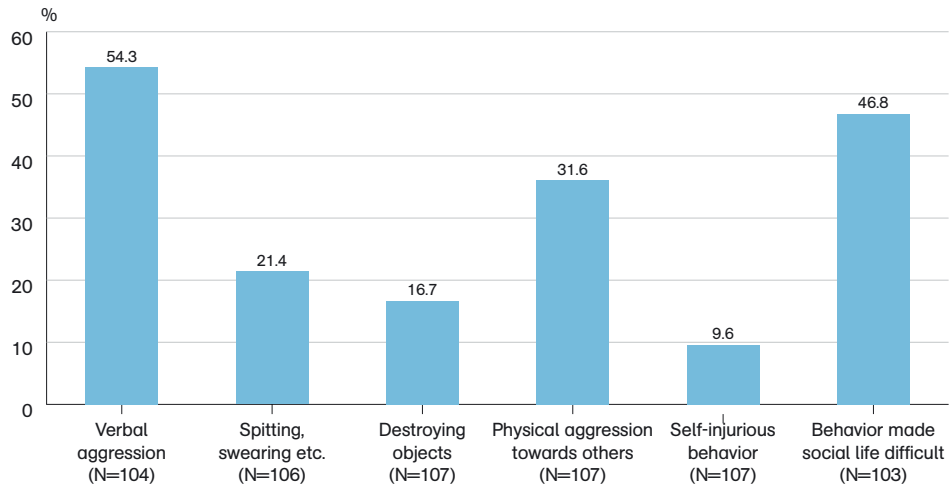

Figure 27.2 Percentage of participants who had shown different forms of behavior problems

her gherkins (58 kg weight, 162 cm height). She loves to watch her Harry Potter and Walt Disney DVDs, and she knows everything by heart, speaks along and laughs about the same passages. She is a cheerful and kind-hearted person. Her laughing comes from her heart. She is grateful for every little bit, and answers this with «thank you», «no, thank you» or «please».

In the present study, parents were asked about the frequency of four major moods in their child at different age levels. Figure 27.3 shows that it was a positive and enthusiastic mood that characterized the individuals with JNCL in almost all ages, although the scores of troubled, anxious and irritated and annoyed moods increased with age.

Children and young people with JNCL have more challenges than others. The small number of earlier studies and the present study (Appendix A) have found that many in this group react to their difficult life situation in different ways, emotionally and behaviorally. The large variation suggests that the brain disease in itself is not responsible for all the reactions and so it is necessary to investigate the influence of other factors, such as the quality of the physical environment and the responsiveness of people in the environment. These will influence the well-being and the emotional and behavioral reactions of children and young people with JNCL, as well as how these reactions are met by people in the environment. This complex interaction can best be described as a transactional process (Sameroff, 2010). In the present survey, parents reported one common compensatory strategy they favored was to try to reduce stress, as illustrated in this quotation.

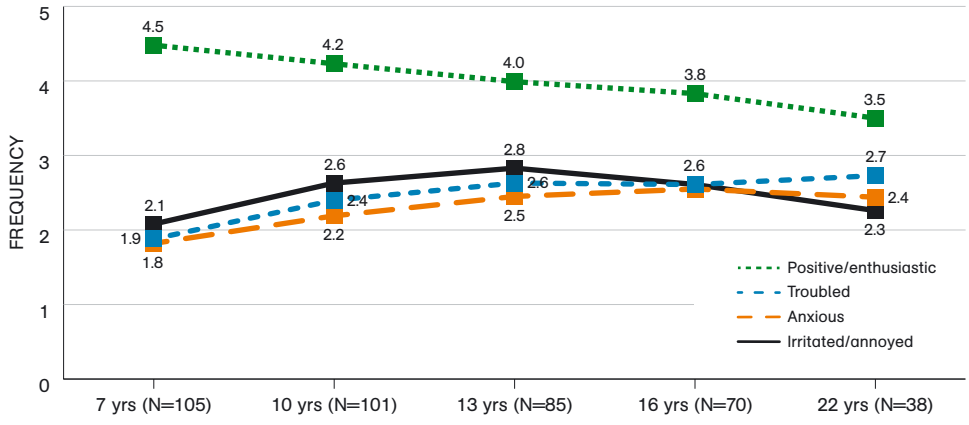

Figure 27.3 Frequency of different moods across age

Scale used: 1=Never, 2=Seldom, 3=Sometimes, 4=Often, 5=Very often

> *The environment was extremely shielded, all wishes were fulfilled. Therefore the behavioral disorders had no or only sometimes a negative impact.*

> *We avoided situations which meant a lot of stress to her. So we stayed at home, in the garden, cared for pets, long walks (later in the wheelchair). With the dog in the woods and along the lake. We did not make any plans in advance but we did the things that she wanted to do, like handicrafts, meeting people of her age, or caring for animals.*

This finding emphasizes the importance of three key needs: assessing the various declines, ascertaining how to influence the everyday functioning of children and adolescents with JNCL, and establishing how support may prevent or reduce secondary effects of the various features of the disease.

Vision loss, dementia and behavioral and emotional reactions

Blindness is a core symptom of JNCL which leads to uncertainty and a lack of situational overview. The consequences may be further aggravated by another core symptom, cognitive decline, which makes it difficult to explain to the individual in meaningful ways what is happening to them and how they can cope. People and objects alike lose their habitual appearance and orientation, rendering mobility more difficult (see Chapter 16). Information about what is going on in the wider surroundings must now be obtained through the use of hearing. People typically rely on visual cues to know who is in the environment, what they are doing, and what activities might be possible. These factors influence their choice of topic and patterns of social interaction. Without the visual cues, many participation opportunities go unnoticed, with severe consequences to the child's play, collaboration and communication, and school performance.

Frustration and misunderstanding

It is a general finding that frustration may lead to aggression, hopelessness and despair (Breuer & Elson, 2017). The assumption that the reported behavioral reactions were a result of a difficult life situation was supported by comments from parents in the present project (Appendix A), who pointed to frustration and misunderstanding as common causes of anger and aggression.

> *When reality turns out to be different from what she had imagined.*

> *He is an amazingly well-balanced young man who only reacts in an aggressive way when he feels misunderstood. This is often due to his massive loss of speech. He still is full of beans. It seems as if he wants to fill every minute with important stuff without any breaks (whether during the day or the night).*

Transition situations with potential loss of control were also mentioned as difficult situations which might lead to emotional or behavioral reactions.

A young person with JNCL may show anger and desperation due to writing problems, and, for example, throw the braille machine off the table or insult fellow students. In such situations, a soothing distraction can be that together, the teacher and student leave the situation, take a break or change to tasks over which the student feels more mastery. Singing a familiar song or recounting a fondly remembered event were often mentoned as useful strategies in the present study, which may change both the general atmosphere and the student's mood.

Anxiety and depression

The life situation of children and young people with JNCL also implies a vulnerability for emotional problems. Losing vision may be experienced as dramatic: «Fear is a logical symptom, because loss of vision elicits both anxiety and insecurity and children experience the environment as dangerous» (Santavuori et al., 1993, p. 247). The results in the present project (Appendix A) indicate almost the same prevalence of behavior problems among children and adolescents with JNCL as with congenital blindness, around 50 percent. Whereas behavior problems among children with congenital blindness are related to missing or delayed social skills (Alon, Ophir, Cohen, & Tirosh, 2010; Runjic, Prcic, & Alimovic, 2015) early behavioral difficulties in children with JNCL (e.g., disruption or verbal aggression) are reactions to frustration. In the present study, few participants with JNCL showed any physical aggression towards others and very few children showed self-injury.

In adult dementia, emotional and behavioral disorders are related to how the individual person experiences changes in competence that accompany cognitive decline (Cerejeira, Lagarto, & Mukaetova-Ladinska, 2012). The disorders are not a direct result of the changes in the brain; instead anxiety and depression reflect reactions to changes in personal competence in routing occupations of life. Depression is a frequently cited symptom throughout the course of dementia and relates to both psychological and biological factors (Lyketsos & Olin, 2002). Key factors contributing to depression include failure to master new ways of accomplishing familiar tasks and inability to adapt to changes (Enache et al., 2011).

In the earlier stages of dementia, people are aware of their difficulties remembering everyday things and decreasing ability to complete the basic activities of daily life. This awareness phase is accompanied by growing reactions of frustration. However, with increasing severity of the disease, they may become less aware of the changes they experience and the source of their frustration possibly shifts from an internal to an external focus (see Chapter 5).

Another population that experiences anxiety, depression, and behavioral disorders includes high-functioning children with autism spectrum disorder (Joshi et al., 2010). Within this population, these disorders may be related to confusion originating in the children's problems with mind understanding and general cognitive problems. By contrast, the visual impairment of children and young people with JNCL leads to less control and more uncertainty, and the cognitive decline makes it difficult to find new ways of doing things and coping with issues. As in adult dementia and autism spectrum disorder, reactions may be observed as forms of anger and aggression, as well as passivity and withdrawal. This implies a need to ascertain the factors underlying behavioral reactions.

In the present study (Appendix A), parents were not asked to distinguish between anxiety and depression because the distinction may not be discernable to a lay person. However, the fact remains that anxiety and depression are different problems, despite sharing symptoms, and frequently presenting as comorbid states: while children with anxiety are wary of potential threats, children with depression ruminate on hopelessness and loss (Hankin, 2012; Hankin, Gibb, Abela, & Flory, 2010). Children and adolescents who are depressed show less pleasure, interest and concentration than they normally would, and easily become tired. They sleep poorly and have little appetite, low self-esteem and little confidence. In the case of JNCL, having little confidence may be both an expected reaction to loss of functional competence in everyday tasks, and a symptom of a resulting depression. Depression can become deeper when it leads to social isolation and having fewer friends. The inability of adolescents, as a group, to solve the problems they meet may contribute to typical symptoms, such as stress, a poor self-image and dysphoria (Hankin, Young, Gallop, & Garber, 2018); however, atypical symptoms include restlessness, anger, irritation, weight gain and increased sleep (Ballard, Day, Sharp, Wing, & Sorensen, 2008). A similar atypical pattern of symptoms may be observed in young people with JNCL.

> *She oftentimes argues and just has her way of thinking about things and is not receptive to other ways of thinking about it at this point. So there are times where she sits in the breakfast room and has to be moved to a different table, you know. We have put her on a table with girls, eighth grade girls sometimes, in breakfasting and that seems to help because she doesn't seem as confrontational with those kids.*

Later, as the disease progresses, adults with dementia tend to become increasingly passive, not taking initiative and are seemingly not interested in participating in activities that they previously liked to engage in. Passivity may also function as a way of ensuring that nothing bad happens, even if it also means that nothing good happens. The person with dementia keeps control by being passive. Some become more impatient and irritable. It can be threatening and very frightening to experience not understanding or having control of one's own situation. Some people experience panic-like anxiety, typically in situations where they notice that they come out short on tasks that they previously have managed without difficulties (Badrakalimuthu & Tarbuck, 2012; Goyal, Engedal, & Eriksen, 2019). In young people with JNCL, anger over legs that can no longer be used for safe walking can lead to vehement pounding on the thighs. Word-finding or articulation problems can lead to aggressive rage or desperate crying – or to a total lack of communication. Individuals with JNCL may have to adapt to a new life situation due to further physical and mental changes, so they can feel safe and comfortable again. But then there is another change in the situation: young people with JNCL are repeatedly being confronted with something happening to them which they cannot control.

> *It was always important to her to be independent. She always got very angry when she was not able any more to manage things on her own. It was difficult for her to accept help.*

The result may be insecurity, anger, fear and grief, all of which could be accompanied by externalizing reactions.

Children and adolescents spend a lot of time at school, where their problems often become more apparent; and where their depression is typically aggravated by an awareness of increasing sense of social isolation and having fewer friends than they had previously. When adolescents are unable to solve the problems they face, they experience even more stress, low self-esteem and depression (Roberts, 2015). In severe adult dementia, there may be behavioral changes in the form of suspicion, delusions, socially inappropriate behavior, impulsivity or emotionally indifference (Steinberg et al., 2008). Such reactions may sometimes be due to misinterpretation or misunderstanding due to the cognitive impairment and a reduced overall grasp of what is going on. Similar reactions are recognizable in adolescents and young adults with JNCL.

> *The special behavioral issues mostly occurred in early adolescence. They were not permanent and did not affect life in a special way.*

Parents and teachers may experience situations and behaviors that are not easy for anyone involved to handle. The children and adolescents with JNCL can suddenly behave aggressively towards others or against themselves, as well as become extremely withdrawn, sad or closed. The reason for the behavioral reactions is not always apparent.

Attachment

Attachment represents another perspective to understanding the emotional and behavioral reactions of children and young people with JNCL. They may sense not only their own lack of control but also their parents' lack of control: the parents are no longer omnipotent. The attachment system may be relevant for their reactions. According to Bowlby (1982), attachment is an innate behavioral system for ensuring protection. He defines attachment behavior as «any behavior that results in a person attaining or maintaining proximity to some other clearly identified individual who is conceived as better able to cope with the world» (Bowlby, 1982, p. 669). Attachment behavior may be activated by unfamiliar people, places and routines, pain, fear and stress. Illness and fatigue increase the need for protection and will heighten children's need for an attachment figure – a parent or another close person (Bowlby, 1969, 1982). Events occur in which children or young people with JNCL feel a need for protection but feel that help is not available and that there is no secure person to turn to. Experiences with events such as these may reflect that the persons with JNCL do not have the psychological protection that attachment implies. One parent described this in her daughter:

> *When she was six years old she started to develop separation anxiety.*

Central to separation anxiety is the fear of being separated from attachment figures. Children may cling to caregivers and react with angry outbursts, kicking, screaming and crying when someone tries to separate them. Children may also experience physical symptoms when leaving for kindergarten or school, or in other ways refuse to go to school. Adolescents can be anxious to be left at home alone.

Anger may also be a reaction to an experienced loss of protection, to the unavailability of protection from an attachment figure. The anger has two functions: first, to empower the child to overcome the obstacles on the way to reunion with the attachment figure and security, and second, to try to ensure that the attachment figure will be available in the future. This type of anger does not destroy the emotional bond between child and attachment figure, but instead strengthens it. Since fear and anger are activated under similar conditions, they often occur simultaneously, and an increase in the intensity of one emotion can

at times strengthen the other (von Tetzchner, 2019). It has been suggested that challenging behavior in children and adults with intellectual disability and autism spectrum disorders sometimes reflects attachment reactions (Janssen, Schuengel & Stolz, 2002; Perry & Flood, 2016; von Tetzchner, 2004).

Exploration is necessary for learning about the world, and exploration is activated by unfamiliar or complex objects and locations. The usual driving forces for exploration (i.e., novelty, unfamiliarity) are identical to the forces driving children to seek an attachment figure (the unknown and uncertainty). Exploration and attachment seeking are thus activated under almost the same conditions, but attachment behavior is elicited mostly by fear and insecurity, while exploration takes place when children feel relatively secure. When the attachment figure moves further away from the child, attachment behavior is activated, while exploratory behavior is more easily activated when the attachment figure approaches, for example in situations involving fascinating objects such as toys or animals. By means of attachment, familiar adults become a secure base for controlled exploration of objects and locations that otherwise can elicit insecurity or fear, a base the child alternately leaves and returns to (Ainsworth, 1963). Thus, attachment and exploration are to some extent complementary ways of meeting new situations, meaning that exploratory behavior is inhibited in situations in which children show attachment behavior.

The attachment and exploratory systems emphasize the need to develop security-giving structures that allow children and young people with JNCL access to social and societal participation. Self-directed exploration may be independent or interdependent (see Chapter 16), and supporting exploration may include activity-based education and adapted tasks that allow students to learn through their own discoveries. Exploration may also imply combining established knowledge with new knowledge. For students with JNCL, exploration may function as an activation of prior knowledge and thereby refresh their memories of earlier experiences. This may range from identifying simple tactual or auditory sensations to investigating whole or parts of complex events, which the teacher may help to integrate (von Tetzchner, Fosse, & Elmerskog, 2013). When the student with JNCL has been guided to explore the whole school and has become better acquainted with peers and staff, the school environment will be experienced as secure, which in turn may prevent emotional and behavioral reactions in children and adolescents with JNCL.

Reactions to loss of friends

As the disease progresses, interaction with peers tends to be reduced (see Chapter 22). Friends are central to young people's lives, and the breakup of a close friendship can be a difficult experience and lead to depression, guilt and anger

(Rubin, Bowker, McDonald, & Menzer, 2013). Furthermore, depression is often accompanied by other impairments, including emotion regulation disorders. Psychological problems such as anxiety, depression, and poor self-esteem are all related to experienced loneliness. Since emotion regulation has a social basis, regulation disorders can have a serious impact on children's social development. Some years into school age, children begin to compare their skills, attitudes, possessions and families with those of their peers; however, low self-esteem based on such comparisons can lead to depression (Harter, 1987). This in turn may lead to cascading effects of decreased interest and motivation to participate in peer activities, lower activity levels, and little support and attention from the environment. This emphasizes the need for structure, control and positive challenges within a recognizable and familiar framework.

When social contacts are lost and new ones are difficult to establish and build, changing school may sometimes help.

> *His emotional and behavioral problems got better when he changed from regular school to the LBZB (school for students with learning difficulties).*

If loss of friends becomes absolute, this may be experienced as dramatic and traumatic, and the result may be comparable to post-traumatic stress disorder (PTSD), a reaction to a stressful or traumatic event a child has experienced or learned about. Children with PTSD can have intrusive memories and recurrent disturbing dreams with fear. The distress is persistent and intense, and the child finds it difficult to calm down. They can feel a great deal of shame, guilt, fear and confusion, and express few positive emotions. They tend to be irritable and angry, are easily startled, and may have difficulties with sleeping and concentrating. They also participate less in play and other positive activities. The symptoms of PTSD involve unregulated emotions and can be reminiscent of behavioral problems. Because some of the reactions are the same, PTSD can be confounded with depression.

Concluding comments

Studies show that the prevalence of emotional and behavioral problems is higher in individuals with JNCL than in the general population. These reactions are related to their declines and the consequences these declines may have for coping in everyday activities and participation in social life. Many factors contribute to these reactions which may be understood from different perspectives, including insecurity, confusion, loss of personal power, control, self-efficacy, and loss of

experience of protection. However, the studies also show there is considerable variation in the scope and severity of these reactions; and that support and adaptation of the physical and social environment may prevent or reduce emotional and behavioral reactions.

It is therefore of critical importance to seek to implement preventive measures. In the present study, behavioral reactions were reduced for some students with JNCL after adaptations were made in the environment to meet the student's social and learning needs; for instance, efforts to compensate for the loss of vision helped the situation, suggesting that the reactions were related to the loss of mastery. Both educational strategies and the social environment of the school are important in this connection, not only in early childhood but throughout school and after the individual has left school. For example, during adolescence, mood fluctuations are quite common in the general population (Laugesen, Dugas, & Bukowski, 2003; Zeman, Cassano, Perry-Parrish, & Stegall, 2006). Thus, it may also be a period where young people with JNCL need special consideration.

An important aim is to provide measures that may help persons with JNCL create meaning and coherence in their lives. One approach is to create a biography with important contacts, themes and experiences, which can enable the person with JNCL, family and staff to remember and use information of special significance to the person with JNCL, and thereby constitute a foundation for security, best possible life flow and meaning-in-activity (see Chapters 11 and 23). Particularly when memory problems are emerging, events that are significant for the individual can be used to call forth pleasant memories and access to familiar structures of action. The following quotation illustrates both the challenges contributing to emotional and behavioral reactions, and a solution to cope with the challenges.

> *In retrospect the psychological stress during the process of blindness (from 7 to 14 years until adolescence) reached its peak when she had to experience consciously that she was not able to keep up with the others any more although she still wanted to be part of them. At first she became disappointed, sad and angry, then she felt resignation and a kind of acceptance in the end. At this stage she wrote a diary and invented stories which focused on herself and the desperate plight because of the increasing blindness. Writing seemed to help her cope with the disease. That is why she did not suffer from very big psychological problems. She even regained her zest for life. At school and in the sheltered workshop she was known for her cutting remarks or her derisive laughter.*

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Afterword

The aim of the project and this book has been to collect information about the developmental course of juvenile neuronal ceroid lipofuscinosis (JNCL) and the experiences of parents and professionals related to the education and support for children, adolescents and adults with this disease. An important motivation was to make readers aware of educational tools and strategies that may be useful but also to inspire new thoughts and insights in the readers. If that is the case, our work in this project and in editing the book has filled its purpose.

Working with individuals with JNCL is challenging, but for most of us, it is also a rewarding experience. We believe that drawing on the ideas and competence summarized in this book, readers will have a more solid basis for their work and guidance in choosing strategies which have been found to produce positive results in the past.

The project and this book would not have been possible without the formidable contributions from members of this project, parents, teachers and care givers, who have given their ideas and knowledge to the project. In addition, valuable contributions have been received from professionals with many years of experience working within the fields of education, communication and language, habilitation, blindness, dementia, music and physical therapy.

For us, the Editors, the project period (2014–2018) has been a time of learning. Many of the views expressed in this book result from long and fruitful discussions and integrate quite different points of view. No doubt, this process has added considerable value to the final product.

The main idea that JNCL must be understood from the perspectives of blindness and childhood dementia underlies the survey and interviews in the project and is very visible in the book. These two features are characteristics of the developmental course of JNCL, but many of the disease's developmental consequences are also found in people with other diseases. We believe the book will be valuable for these groups as well.

Finally, we have chosen to examine education and learning in individuals with JNCL from a lifetime perspective. This approach implies that the capacity for learning extends beyond the school years. Early intervention and the acquisition of skills, the maintenance of these skills, and ongoing learning thus constitute a continuous process, which will contribute to the quality of life also for adults with JNCL.

The Editors

Appendix A
Methods

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

The aim of the present study was to investigate the experiences of parents, educational staff and care staff related to education and support services for children, adolescents and adults with juvenile neuronal ceroid lipofuscinosis (JNCL). It is a survey and interview study (see Robson & McCartan, 2016) with informants from six countries: Denmark, Finland, Germany, Norway, the UK (England and Scotland only), and the USA.

The study is a part of the international project, «Juvenile Neuronal Ceroid Lipofuscinosis (JNCL) and Education» (2014–2017) which was funded by the European Commission through Erasmus+ grant No. 2014-1-NO01-KA200-000388. In addition, the project was supported by participating organizations and volunteer work. Preparatory activities in 2013 and 2014 were funded by The Directorate for Children, Youth and Family Affairs and the Directorate for Education and Training in Norway, the Norwegian NCL Family Association and Statped, Norway. Statped Midt in Norway coordinated the project.

Participants

Recruitment of informants was made through parent associations, educational institutions and medical institutions participating in the project. They distributed a letter with information about the project and a consent form to families with a member with JNCL. Parents who had given consent were asked to nominate teachers or other non-medical staff as potential informants (hereafter named "professionals"). The information letter and consent form were sent to the nominated professionals, including the information that they had been nominated by parents. Many of the parents informed the teachers or staff members that they would receive a letter of information asking them to take part in the survey. Some parents did not want to involve a teacher or another professional. Other parents did

not want to take part in the survey but did nominate a teacher or staff member who consented to take part in the project. This means that the group of individuals with JNCL described in the surveys by parents was not identical to the group described by the professionals. It is not possible to compare responses from parents and professionals, because many participants were only represented either by parents or by professionals. The reason for including information from both parents and professionals was to obtain as much information as possible and from more than one source: both a parent perspective and a professional perspective. However, it was not an aim to compare the experiences and views of parents and professionals.

The parents and professionals who completed the questionnaires and took part in interviews are here called *informants*, while the individuals with JNCL who were described are called *participants*. To protect personal information, the consent form was returned to the national coordinator of each country who de-identified the informants and participants (removed names and identifying information) before the results were sent to the researchers. The researchers thus do not know the names of those who participated.

A few teachers and other staff members completed surveys describing the same individual with JNCL, and a few teachers and staff included more than one individual in their questionnaire responses. These were not included in the data analyses presented in this book because the surveys were about individuals and not about groups.

Figure 1 shows the numbers of parents and professionals who were informants in the surveys and interviews. The number of informants in each country is not proportional to the country's population. This may reflect national differences in

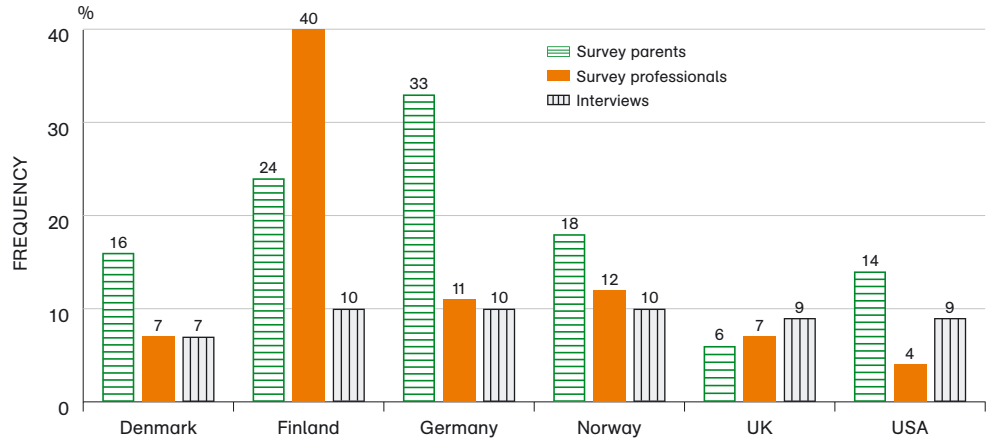

Figure A.1 The number of individuals from each country who completed questionnaires and participated in the interviews

Table A.1 Age (years) and sex distribution among participants with JNCL represented in the three surveys

Survey	N	Mean age (SD)	Range	Males/ Females
1 Parents	78	17.1 (6,5)	6–34	37/41
2 Bereaved parents	33	24.9 (5,3)	17–34	12/21
3 Professionals	81	13.0 (8.1)	8–27	41/40

Note: A few informants did not provide information about the age of the participant. In Survey 2, the mean age refers to the age when the participants passed away. The total number of participants are 135. Some of these participants appear in two different Surveys; in Survey 1 and 3 or in Survey 2 and 3.

prevalence but is mainly due to differences in the way informants were recruited. In Denmark and Norway, all families with a family member with JNCL were invited to participate in the survey through the national family organization and the educational centers responsible for the follow up of individuals with JNCL. Together they knew the names of all or nearly all families in the country. Information letters were sent to all the families, also the bereaved families. In Finland, Germany, the UK and the USA, the associations sent the information letter to families who were receiving services from the association or who had received such services in the past, including some parents who were bereaved. In Finland, most relevant families were known to the parent association. The German sample was mainly recruited from the northern part of Germany, where most families were known to the institutions participating in the project, and the sample was thus representative for this geographical part of Germany. There was a relatively low number of participants from the UK and USA. Recruitment of professionals working with individuals with JNCL depended on a nomination from parents, and thus reflects the parent group. It is not clear how these recruitment procedures have influenced the results. It is a strength that the study has a relatively large sample, but the sample is more representative of individuals with JNCL and educational systems and support services in Northern Europe than in the UK and USA.

Three different surveys were organized. Survey 1 was for parents, Survey 2 was for bereaved parents and Survey 3 was for professionals (see below). A total of 192 questionnaires were completed in the three surveys, for 135 participants, 63 males and 72 females with JNCL. Survey 1 includes information about 78 participants with an average age of 17.1 years (Table A.1). Survey 2 comprises 33 individuals who had passed away and their mean length of life was 24.9 years. Survey 3 comprises 81 participants with a mean age of 16.4 years. All participants had a diagnosis of JNCL. The youngest participant in the surveys was six and the oldest was 34 years old. All participants attended or had attended school but there

were rather few students aged between six and nine years. In surveys 1 and 3, thirty-nine participants were past school age, 20 years or older.

The plan was to complete ten interviews in each country, five with parents and five with professionals. Twenty-five interviews with parents and 30 interviews with professionals were completed, resulting in more than 2000 transcribed pages. The transcriptions of non-English interviews were translated to English. All personal names used in the interviews were de-identified in the transcriptions.

One reason for including several countries in the study was the possibility of getting a sample that was large enough to uncover the variation that may exist in development, learning and mastering among individuals with JNCL, as well as in educational systems and support services. The variation in results might reflect both biological differences and cultural difference in education and support services. The present study presents new information because it is the first large study addressing issues related to education and support for individuals with JNCL. However, this study should be supplemented with new studies that have samples that are representative for countries with other educational and support services than those in Northern Europe, which constitute the majority of the participants presented in this study.

Surveys and interviews

There were three different surveys in five different languages (Danish, English, Finish, German and Norwegian).

- Survey 1 – For parents (with focus on the history and the present)
- Survey 2 – For bereaved parents (with focus on the history)
- Survey 3 – For teachers and other professionals (with focus on the present)

The surveys include demographic information, and information about the present status of the participants with JNCL. They include many of the same themes, but the information was somewhat different. The main parent survey includes both retrospective information and information about the present situation of the individual with JNCL, while the bereaved parents were asked to give retrospective information. The professionals were mainly asked about the present situation of the participants.

The surveys include information about when different problems became noticeable, the diagnostic process, abilities and challenges, school attendance and educational strategies, support services, and the participant's social interaction with peers. For many issues, the parents were asked to use a five-point scale to rate their

child's skills and achievements at different age levels: 1) no competence, 2) low, 3) moderate, 4) high, and 5) very high competence. Satisfaction with education was evaluated on a similar scale: 1) no satisfaction, 2) low, 3) moderate, 4) high, and 5) very high satisfaction. The participants' interests in different activities compared to peers were indicated on this scale: 1) much lower interest, 2) lower, 3) same, 4) greater, and 5) much greater interest than peers. Social interaction with peers was indicated on the scale: 1) no interaction, 2) minimal, 3) reasonable, 4) pretty good, and 5) very good interaction. For some activities and behaviors, informants were asked about frequency: 1) never, 2) monthly, 3) weekly, 4) daily, and 5) several times per day. Different moods were indicated as being present: 1) never, 2) seldom, 3) sometimes, 4) often, or 5) very often. The contributions of diverse services were evaluated on the scale: 1) no contribution, 2) some contribution, 3) moderate degree of contribution, 4) high degree of contribution, and 5) very high degree of contribution. Negative effects were indicated on a similar scale: 1) no negative effect, 2) some, 3) moderate, 4) high, and 5) very high negative effect. Informants were asked to evaluate the impact of education and services on the scale: 1) no impact, 2) low, 3) some, 4) high, and 5) very high impact. Parents indicated level of cooperation with different services on the scale: 1) no cooperation, 2) low degree of cooperation, 3) moderate, 4) high, and 5) very high degree of cooperation. Parents were asked to evaluate the usefulness of different sources of information, including from school, courses and associations on the scale: 1) not useful, 2) useful to a low degree, 3) useful to a moderate degree, 4) very useful, and 5) extremely useful.

For most issues, the informants could add comments to their evaluation scores, and many informants used this opportunity to give supplementary information and clarifying examples and views.

The interviews were semi-structured. There were two different interview guides, one for interviews with parents and one for interviews with professionals, and they were available in five languages (Danish, English, Finnish, German and Norwegian). The interviews were mostly made face-to-face but due to geographical distances and difficulties finding an appropriate time, telephone and Skype were used for a few interviews. The interviews followed the interview guide but with flexibility, and informants were free to convey any information they thought would be relevant. The interviews were conducted by local project partners and it was ensured that the interviewer was not affiliated with the informant and was not responsible for educational or other support services for the participant with JNCL. The local interviewer transcribed the interview, translated it into English when needed, and sent it to the researchers.

Informants from the surveys and interviews were de-identified and given a project identification number. Both quantitative and qualitative data were mainly analyzed in Norway.

Procedures

The questionnaire with factual information and evaluation scales was sent to all parents who had given consent. The parents were always recruited first, and professionals were only approached if parents had given consent and nominated a professional. The information letters and consent forms for teachers and other professionals were sent to the professionals indicated by the parents. Parents and professionals could complete the questionnaire on paper and send it by mail, or electronically on Internet.

The participants were asked to indicate on the questionnaire whether they were willing to participate in an in-depth interview. The parents and professionals who were interviewed were drawn among those who had expressed willingness to take part in an interview. After completion of the interview, the informants were contacted on telephone to inquire if there was anything they would like to add (they had been informed about this during the interview).

Analysis

Quantitative analyses included mainly descriptive statistics, with frequencies, mean, standard deviation and range, and a small number of correlations. All statistics were performed with IBM SPSS Version 25. Qualitative data were analyzed using a thematic approach (see Guest, MacQueen, & Namey, 2012). Direct quotations from parents' and professionals' contributions were used to illustrate trends in comments as well as trends in quantitative data.

Ethics

The study was approved by the Regional Committees for Medical and Health Research Ethics in 2015 (No. 2015/1464). The parents had control over the recruitment of professionals, as professionals were only approached if parents had given their consent and nominated a person.

References

Guest, G., MacQueen, K. M., & Namey, E. E. (2012). *Applied thematic analysis*. London, UK: Sage.
Robson, C., & McCartan, K. (2016). *Real world research, Fourth edition*. Chichester, UK: Wiley.

Appendix B
Resources

Batten Disease Family Association (BDFA)
http://www.bdfa-uk.org.uk/
Batten Disease Support and Research Association (BDSRA)
https://bdsra.org
Beyond Batten Disease Foundation
https://beyondbatten.org/research/state-of-science/
Bildungzentrum für Blinde und Sehbehinderte in Hamburg
(Educational Center for the Blind and Visually Impaired in Hamburg)
http://bzbs.hamburg.de
NCL Danmark (The Danish NCL Family Association)
http://dsvf.dk
NCL Gruppe Deutschland e. V. (The German NCL Family Association)
http://www.ncl-deutschland.de
NCL Net
http://ncl-netz.de/en/specialclinic.htm
NCL Resource
http://www.ucl.ac.uk/ncl/
Norsk Spielmeyer-Vogt Forening (The Norwegian NCL Family Association)
https://www.nsvf.org/
Oppimis-ja ohjauskeskus Valteri Onerva
(Learning and Consulting Center Valteri Onerva)
https://www.valteri.fi/onerva
Royal Blind School
https://www.royalblind.org/education
Statped
http://www.statped.no/
Suomen JNCL-perheiden tukiydistys ry (The Finnish JNCL Association)
https://www.jncl.fi

Synscenter Refsnæs (Vision Center Refnæs)
http://synref.dk/
University of Rochester Batten Center (URBC)
https://www.urmc.rochester.edu/neurology/batten-disease-center.aspx
WESC Foundation – The Specialist Centre for Visual Impairment
https://www.wescfoundation.ac.uk

Juvenile neuronal ceroid lipofuscinosis (JNCL) is a rare neurological condition characterized by the onset of blindness and dementia in childhood, but with considerable individual differences. This book is concerned with the developmental course of the disease and educational and non-medical intervention for children and young people with this condition. The book is based on an international project on JNCL, dementia and education, and presents evidence-based practices in various areas. It gives the reader insight into educational strategies and tools which may support learning and maintenance of knowledge and skills in children and young people with JNCL, as well as the experiences of parents and staff. The text is illustrated with many small case stories.

The chapters are written by professionals and parents from different countries and give a broad knowledge foundation for planning education for students with JNCL and contributing to their learning and a meaningful life. The book is intended as a knowledge base and source of practice for parents, educators and support staff. The book focuses on JNCL and its many manifestations and symptoms, but may be useful also for professionals working with other young people with early blindness or dementia.

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