



Nordic Welfare
Centre

Re-CHARGE

Voices about living with
CHARGE syndrome



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Foreword

A rare diagnosis is defined when the affected population is less than 1 in 2.000 persons, while an ultra-rare diagnosis is defined when fewer than 1 in 50.000 persons are affected. The prevalence of persons born with CHARGE syndrome in the Nordic region is approximately 1 of 10.000 born each year, making the population very small in each country. In some regions there might be only one person with the syndrome. This means that the support system around the individual has no prior knowledge of the syndrome.

For this reason, there are great benefits in the Nordic region from sharing knowledge and examples of best practice.

The purpose of this publication is to raise awareness about the diversity and complexity of living with CHARGE syndrome. The publication is aimed at both professionals who might meet persons with CHARGE syndrome in their work, and relatives and the close network who want to know more to be able to give support.

Medical facts often highlight the negative aspects of the syndrome, and there is a lack of literature on what living with CHARGE syndrome may entail. This is why we have gathered voices of persons with CHARGE syndrome and relatives and professionals from the Nordic region. The focus is on functionality rather than medical descriptions. We want to give these persons a voice and fill a gap when it comes to describing all the things that may work, given the right support.

The Nordic Council of Ministers have a vision of a socially sustainable Nordic region – for all. In this publication, we focus on creating the conditions for achieving these goals for persons with CHARGE syndrome through increased knowledge.

This publication has been produced by members of one of the Nordic Welfare Centre's networks, the Nordic Network on CHARGE syndrome.

The Nordic Welfare Centre would like to thank all the authors and the editorial team of the Nordic CHARGE network who have met and built trust with the participants in the book, often more than once.

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Introduction

In this publication, we want to emphasise possibilities and show how persons with CHARGE syndrome can lead good lives with proper support.

Living with CHARGE syndrome can entail different aspects and challenges. Often, the focus revolves around the medical aspects, and less on how individuals can function in their day-to-day lives. Persons with this syndrome represent a small group which means that when professionals encounter someone from this population they may have limited knowledge about the syndrome. And even though there is lots of knowledge regarding the medical aspects of CHARGE syndrome, there is a lack of case descriptions and interviews, telling the story from the perspective of the persons living with the syndrome themselves.

We hope that this publication can show the diversity and give a voice to a group that seldom is heard. Hopefully, parents, legal guardians, and relatives can get a more positive picture of what life with CHARGE syndrome might look like.

We have spent the last few years collecting case studies from everyday practice and interviewing people with CHARGE syndrome and their families. We want to share the experiences of the participants in this publication.

Due to the small population of people with CHARGE syndrome in the Nordic region, we have anonymised the texts and chosen fictitious names.

Meet Thomas's mother who gives a very personal account of how it was to learn that her newborn baby had CHARGE syndrome. Read about Dominic who is in primary school, knows three languages but also struggles with balance and sight, which makes him exhausted at the end of the day. Karl recently got his drivers' license and wants to move to the capital to get to know more people who knows sign language. Together with Charlie, Anna, Inga, and Linda they have all let us into their lives, displaying challenges, personal growth, and development.

We hope that you, as the reader, can relate and find similarities with your own life.

/The Nordic network on CHARGE syndrome

About the Nordic CHARGE Network

Established in 2002, The Nordic Network on CHARGE syndrome has more than 20 years of experience in networking across the Nordic region.

Limited knowledge about CHARGE syndrome in various professional environments and among parents affects individuals living with the syndrome. By focusing on educational, and behavioural aspects, as well as the psychological consequences of CHARGE syndrome, the participants in the network strive to increase knowledge and understanding.

The opportunity to meet professionals from other Nordic countries has made a significant difference so far. The network has shared experiences, supported each other in forming national networks and projects, and found ways to apply the knowledge in the daily practices.

The network produces information and spreads knowledge about CHARGE syndrome to families and professionals in the Nordic countries. We also collaborate with European networks, Deafblind International (DbI), and other international networks when relevant.

[Read more about The Nordic Network on CHARGE Syndrome.](#)

What is CHARGE?

CHARGE syndrome is considered a rare diagnosis, caused by a congenital genetic mutation. The syndrome affects several organs, often involving hearing loss, visual impairment, and challenges with balance, all which require a great need for medical treatment and special educational efforts.

The name CHARGE syndrome is an acronym, created in 1981, with six criteria that initially described the syndrome, with the diagnosis requiring the presence of four out of six features. However, this acronym is no longer used for diagnosing CHARGE syndrome.

The acronym:

C: Coloboma (a congenital malformation of the eye causing defects in the lens, iris, or retina).

H: Heart anomalies (different types of congenital heart conditions).

A: Choanal atresia (a congenital narrowing of the back of the nasal cavity that causes difficulty breathing).

R: Retardation of growth and/or development.

G: Genital anomalies (including hormonal anomalies).

E: Ear anomalies and/or deafness (physical deformations and/or deafness/reduced hearing).

Today, we know that at least 60 percent of the persons with the clinical diagnosis have a mutation of the CHD7 gene. This means that a blood sample can determine whether the syndrome is present.

CHARGE syndrome has other features not included in the acronym, such as an affected cranial nerve that can lead to facial palsy. Different definitions of the diagnostic criteria have been made over the years.

Children with CHARGE syndrome often face life-threatening conditions from birth, such as heart problems and breathing

difficulties. Frequent and lengthy hospital stays, surgery, and other treatments are common. Problems with swallowing and breathing can also cause difficulties when the child returns home from the hospital, and these problems sometimes persist throughout their lives.

The senses of smell and taste, hearing, vision, balance, and the sense of movement are also frequently affected. The impairment of multiple senses results in delayed development in children. At the same time, intellectual capabilities often fall within the normal range.

Those are the medical facts. However, a person with CHARGE syndrome is so much more, and that is why we have written this publication.

This is a different acronym borrowed from the [Dbl CHARGE Networks](#) book [You taught me:](#)

C: Creative

H: Humorous

A: Active

R: Regulative

G: Goal directed

E: Explorative



Thomas's mother's journey to acceptance

When Thomas's mother first learned of her son's diagnosis, she would not believe it. "There can't be so many things wrong with you!" But with time she learned to accept and to discover more positive sides with the help of an online group. "Thomas is so much more than the syndrome, and he makes me very happy and proud."

We are visiting Thomas and his mum at their home to learn about their experiences at the hospital following Thomas's birth. Thomas, now 18 months old, plays on the floor, pulling himself up to a standing position by the sofa. He is eager to say hello to us as his mother speaks.

– I didn't realize anything was wrong, apart from a heart defect discovered during the pregnancy. I was supposed to give birth at a different hospital but was referred to this one due to his heart condition. They did numerous exams and tests, including amniotic fluid and blood tests, but everything appeared fine. The tests did not reveal CHARGE syndrome, and I don't believe they specifically checked for it.

No straight answers

Thomas's birth was normal, and he was born full-term without complications. However, he was immediately brought to the intensive care unit because of the heart defect. They had done ultrasounds monthly at the hospital and everything else seemed fine.

The day after Thomas's birth, his mother visited the intensive care unit where Thomas was lying in a respirator. When asked, the doctors didn't give any straight answers, only mentioning that Thomas was struggling to breathe.

– I assumed it was because of his heart condition, she says. I thought it would be a matter of a few days before he recovered. But then there was more... They said that he couldn't swallow. He was doing very poorly at the beginning with breathing and swallowing. He had acid reflux, and all the food just came back up again. They inserted a PEG tube, and he was no longer fed orally. They did try, but it just didn't work.

Learning about CHARGE

After a couple of weeks at the hospital, Thomas was diagnosed with CHARGE syndrome.

– I had never heard of CHARGE before, Thomas's mother remembers. I thought: "What on earth is that?" I didn't want to believe it. It was very hard. I just thought: "It's not possible! There can't be so many things wrong with you."

"I didn't want to believe it. It was very hard. I just thought: 'It's not possible! There can't be so many things wrong with you!'"

Thomas' parents didn't know anyone who had a child with special needs or suffering from severe illness. The doctors explained the diagnosis, that he might not be able to see, hear, or swallow, and he had problems breathing.

– This was too much for me, explains Thomas's mother. It was like a surreal, painful dream. I heard what they said, but deep inside I didn't want to believe it. I just hoped that they had made a mistake.

A frustrating time

They learned that Thomas might not be able to do as much as a normal, healthy child. Maybe he would struggle with speech, vision, and hearing and his brain could be affected, and maybe it would take years for him to learn to walk and to develop strong legs.

– The only thing I heard was, “He can’t do this, he can’t do that, he’s never going to be normal, like other children. He will be dependent on help for the rest of his life”. That’s not what I wanted. I wanted him to manage fine, right?

Thomas’s mother was in a state of shock, and the doctors only focused on the negative aspects.

“Thomas’s mother was in a state of shock, and the doctors only focused on the negative aspects.”

– I couldn’t believe any of it.

A bomb of information

Thomas’s mother wished that the doctors had told her everything gradually, bit by bit.

– It felt like an explosion, a bomb of information! I don’t know if they could have handled it differently. They had an important message, of course, but for me, it was just too much at once.

She took some time to think for herself, to call her sister, and to cry. She just hoped that Thomas would get well. That he would not be in pain or discomfort. But when she looked at him, lying in intensive care for a long time already, struggling to breathe, she realised that it was all true.

From that point, she began to accept that this was a part of him, and it was not something he would recover from. He will have to live with the syndrome for the rest of his life.

– In the beginning, time moved very slowly, but gradually I began to accept that he for real was quite ill.

“In the beginning, time moved very slowly, but gradually I began to accept that he for real was quite ill.”

Facebook group for support

Thomas's mother wanted to find a place where she could talk with

someone who would understand her and who knew what CHARGE syndrome was. There wasn't much information online, but then she found a Facebook group where members share their experiences, write, and publish pictures and videos. This has been a great support for her. People also gave positive information; they showed how some children with CHARGE syndrome thrive in education and work. In a global group, there are members from many different countries all over the world. Many of them have children born around the same year as Thomas.

– At the hospital, as soon as they made the diagnosis, they put Thomas in a category, you know. I don't think that's right because he is so much more than the syndrome.

Acceptance is crucial

Knowing more, Thomas's mother reflects on the advice she would give herself if she could go back in time to right after birth.

– I would say, don't panic! Back then, I really didn't believe that I would ever see all the things he is capable of doing now. He does do so many things, small things that count a lot! Even though he has serious difficulties, his personality, and the things he does make me very happy and proud, she says.

– Acceptance is crucial. I would advise myself to accept it early; then everything will be all right.

Wishes for the future

A constant worry is that children with CHARGE syndrome, aside from motoric problems, often are ill, and they frequently end up in hospital.

– If Thomas catches a cold he might have to go to the hospital. I hope this can change, that his immune system gets stronger, and that he won't have to be so ill.

Thomas once got very ill with a new infection. Seeing him lying there with such a high fever, wasn't easy, Thomas's mother recalls. Managing such a high fever is challenging for anyone, but especially for babies.

– Why are they supposed to go through this? What have they done? Why can't they be allowed to be like all the others, enjoy themselves, and just be babies? asks Thomas's mother.

Now she sincerely hopes that the future brings Thomas a lot of joy.

– All we can do now is wait and see what the future holds.

– Eventually, he will learn to walk and maybe pick up sign language, she says while Thomas is playing on the floor, smiling, and babbling just like other babies of his age.



Strategies helped Dominic adjust to primary school

Starting a new school, and meeting new teachers, assistants, and friends is a big adjustment for any child, and it takes time to adapt. Every family wants their kids to feel good, thrive, and enjoy school. Meet Dominic, a seven-year-old boy who loves numbers and lego.

Dominic lives with his parents and brother. He loves numbers, letters, watches, clocks, and to build with lego. He has a unique memory for names, people, places, and dates, speaks two languages, and can understand English as well. He can read in all three languages proficiently.

"Dominic is kind and loving and has a great sense of humour."

Dominic is kind and loving and has a great sense of humour. He enjoys spending time with adults and talking with them about life and interests. Also, he likes to learn about other people and their family lives.

Among his friends at his inclusive primary school, he is popular. After the first weeks of school, he began to show them a little more interest and now he spends more time with them. Dominic likes the school subjects and finishes his work quickly, so he gets extra tasks.

Speech- and physical therapy is regular on a weekly basis. When he was younger, he had occupational therapy as well every week. Dominic gets support from teachers, special education teachers, a social educator, and teaching assistants at school.

He doesn't participate in any sports or hobbies after school at the moment and the family has been waiting for a support family or after school support for two years now. It is difficult to find someone for this job, and many families experience the same problem. His parents try to give each other some time for themselves, but both of their extended families live in another country, so it is not easy to find support.

Challenges with CHARGE syndrome combined with talents

Dominic has a facial nerve paralysis which mainly affects his facial appearance. He cannot close his right eye and needs eye drops several times a day. His vision is 6/12 in both eyes, which means that he can read or see things at a six-meter distance that a person with normal vision could see at a twelve-meter distance.

Dominic is sensitive to light, but he does not have coloboma. However, he was born with a heart condition called mitral regurgitation, which means a leak in the mitral valve in the heart. The family also recently discovered that he lacks a sense of smell but can still taste food. CHARGE syndrome has impacted his speech and both fine and gross motor skills. He is also diagnosed with autism and has hearing loss in his right ear due to the absence of a cochlea, so he uses hearing aids. This affects his balance.

"Despite these challenges, Dominic has exceptional talents for numbers, people, and things that interest him."

Despite these challenges, Dominic has exceptional talents for numbers, people, and things that interest him. He speaks two languages and in addition can read in English. When using his first language, his vocabulary and understanding are better, and he uses 5–6-word sentences.

Dominic likes to repeat things a lot, and his language comprehension and communication are in constant progress. He used sign

language till he was five years old, and it helped him a lot, but now he no longer uses signs.

Assistance in everyday life

Due to his hyperopia (farsightedness), Dominic wears glasses. When outdoors he also has filter glasses with a protective frame against the wind. For the time being, he does not need enlarged text or pictures, but the school knows what to do if this changes. The school has access to advisors at a national institute for blind and visually impaired and deafblind, who provide guidance on Dominic's sensory functioning.

It is important to stay close to Dominic when speaking with him, ensuring eye contact and good acoustics. This facilitates the communication.

He has a designated contact person at the National Hearing and Speech Institute for adjustment and assistance at school.

Dominic's balance is weak due to the lack of his cochlea. Earlier he used to wear a soft helmet outside for protection, but he does not need it anymore. He tends to stumble and fall easily and tires quickly. The use of extra time combined with good quality shoes are important for his comfort. For longer walks, he needs a pram or a bike.

Daily routine is important

Dominic is sensitive to both loud noises and light. He prefers routines and may find sudden changes challenging, such as when the teaching assistants cannot be with him due to illness or if the daily schedule changes. For a period of time, in the after-school program he enjoyed reading the same book every day, sitting on the same sofa.

"Dominic has excellent skills and memory on topics of interest and can do his daily activities independently."

Dominic has excellent skills and memory on topics of interest and can do his daily activities independently. However, he prefers to do things his way. When he is in distress, the teachers may not always understand why this occurs.

Sometimes, too many changes coincide, creating a lack of control. One semester Dominic got both a new teacher, new teaching assistants, and new support staff. He needs time to get to know everyone and build trust with them.

Keeping him well-informed through spoken language, written information, or both, is a good strategy so that he knows in advance what to expect during the day. In kindergarten he and his class had a visual schedule and that has been used in primary school as well.

After school, Dominic is often tired and can have challenging behaviours. Sometimes he struggles to sleep which also affects his energy and well-being.

Different support systems

The support systems differ between kindergarten and primary school. Kindergartens often assign one teacher or teaching assistant to have the main responsibility for supporting a child during the day, which includes different types of therapy, organising meetings, sharing knowledge, and working with advisors. The team of teachers works together during the day with the children.

In primary school, a team of people provides support and works with the children during the day, including teachers, special education teachers, developmental therapists, and teaching assistants. In addition to this the children meet a lot of other persons during the week. Both systems have their advantages and disadvantages.

In situations where Dominic needs to relax, the teachers found it helpful to go to a quiet room with him before returning to class. They have also noticed differences in his energy levels during the day, and the week. He has more energy in the morning, and at the beginning of the week but is tired at the end of the day and the end of the week. This is taken into consideration when planning the week's schedule.

Relationships developed with time

Dominic has good relationships with the teachers, although he can become dependent on specific individuals and get upset when they are not around.

In the beginning, when his classmates wanted to play with him, he

was not very interested. After the first semester this changed, he was more responsive and began to interact with them. He still struggles with group work sometimes which may be related to CHARGE syndrome or his autism diagnosis.

The teachers have tools to assist with social learning, but those are not always suitable due to communication challenges. Understanding Dominic's feelings and triggers is crucial for the teachers if he gets upset, and that understanding takes time.

During the first year, various situations would upset Dominic, which showed some potential triggers such as loud noises, group size, proximity, room acoustics, or if he was tired or hungry. Sometimes a class, that he usually enjoyed, could upset him but the teachers did not understand why.

Strategies to help Dominic adjust to a new school

- **Independence:** Encouraging Dominic to get his books, papers, snacks, etc, and setting reasonable demands. This includes setting limits, being firm but kind, giving him time, assisting him at the beginning of a class, giving him space when needed, and then again refocusing on his interests, and finding more new and interesting things.
- **Visual aids:** Using a visual schedule helps Dominic to understand the agenda for the day or month.
- **Time:** Allowing time for adjustment, such as progressing from having lunch in a quiet room to joining the entire class in a larger setting.
- **Breaks and quiet time:** To provide breaks or quiet time when needed. Introducing breaks during the day to reduce anxiety and increase routine and predictability.
- **Adjustment:** Organising the environment/classroom and making changes to enhance interaction. For example, removing a couch he always sat in during circle time encourages him to sit with his classmates and interact with them.
- **Group size:** Organising small groups when necessary, ensuring more attention and opportunities for interaction.
- **Introducing a variety of activities:** Encouraging a variety of activities, like clubs in the after-school program, to help Dominic participate more actively.

- **Sensory overload and stress:** When Dominic feels overwhelmed, it's important to listen, give him room, and provide quiet time for him to regain his balance.

Strategies that gave results

With time, adjustment, and good assistance Dominic is now happy in school. He is more independent, social, and relaxed. Now, he gets less upset, interacts more with his classmates, and participates in all activities during the day. He handles unexpected events much better. Knowing the routine and schedule makes him safe and calm and helps him enjoy school.

The first year in school is always an adjustment period. Getting to know the teachers, classmates and the school requires effort and patience. To build trust takes time, but a positive attitude, passion, teamwork, and solution-oriented mindset will give results.

Every family wants their children to participate in school and society with joy, understanding, and the freedom to be themselves, to belong, and to be accepted. It is essential to maintain good communication and cooperation with families, providing support in their everyday lives.

For Dominic, the CHARGE acronym means

C: Challenges with vision: sensitivity to light, with visual acuity at 6/12, but without coloboma.

H: Heart condition, mitral regurgitation.

A: Anosmia, a loss of smell.

R: Remarkable talent for things he is interested in. General impact on speech, and fine and gross motor skills.

G: Not applicable.

E: Ear/hearing-problems, loss of hearing in the right ear and the absence of a cochlea.



Karl wants to live on his own

Karl recently learned how to drive. "I am generally a calm person but can get a little bit stressed when I am driving a car." Karl is a resilient young man navigating through life both with CHARGE syndrome and the challenges of driving a car. He faced many challenges from the very beginning, but with support, he is now building a life of his own.

Karl speaks sign language and lives with his family in a small town where there was little or no knowledge about CHARGE syndrome when he was born.

Due to breathing problems, Karl was rushed to intensive care right after birth. He had surgery the next day, and for the first four and a half months of his life, he spent time in the hospital with his parents.

– It was a tough experience, Karl's mother states.

Karl was almost three weeks old when he got his diagnosis. A doctor who had worked at a children's hospital in Texas recognised the symptoms.

– We were told not to look it up on the internet, but we did. That was hard, and we never did it again, says Karl's mother.

Instead, the parents talked with the doctors to get answers to their questions and decided to see what would happen. What would the prognosis be and how could they help him improve in every way?

They got support from doctors and social workers. As no one spoke sign language in their small town, they had to seek help from an institution that teaches sign language. But they wish that there would have been better support regarding his education throughout the school years.

Karl wants to move to the capital

Now they just wish that Karl, in the future, can live by himself, be independent, get a job, and hopefully have a family. To achieve this, his parents wish to support him as much as possible.

– Karl wants to move to the capital to be more in contact with deaf people. He would like to get an apartment on his own and be independent, says his mother.

If she could go back in time she would change a lot of things, especially to stand up more for herself and for her son's needs regarding education.

"If she could go back in time she would change a lot of things, especially to stand up more for herself and for her son's needs regarding education."

– In my opinion, children with special needs, such as deafness, should have the opportunity to attend special schools where they can meet kids with similar problems. Deaf children have the right to the same opportunities as other children, but this is not the reality, and that needs to change.

"Karl lives at home with his family and enjoys computer games and sports."

Cheers for Man United

Karl lives at home with his family and enjoys computer games and sports. His sister is playing basketball for a college team in the USA, and Karl watches every basketball game with his hometown team.

His favourite football team in the Premier League is Manchester United.

He has an assistant in school, and a sign language interpreter, who translates to him what the teacher is saying.

– She is very good and a really good friend of mine, Karl says.

Karl also needs support when he goes shopping to be able to communicate with people who don't know sign language. If he doesn't have an interpreter at hand his parents or sister some times help him. However, he does not need guided mobility.

Karl uses Signwiki, Messenger, and Snapchat to communicate. And he has a system at home that lights up when somebody rings the doorbell and a special alarm clock with a light that wakes him up in the mornings.

"I am generally a calm person but can get a little bit stressed when I am driving a car."

Karl got his driver's license

– I am generally a calm person but can get a little bit stressed when I am driving a car.

Karl hopes that this stress will disappear when he becomes a more experienced driver, but sometimes this anxiety means that he drives less.

– I prefer having one of my parents with me when I drive. They are really helpful, he says.



Charlie participating in new activities and environments

We have followed Charlie and his assistants for over a year as they worked on adapting to a dance activity. To support persons with CHARGE syndrome, for them to actively partake in new activities, we need knowledge on how to adapt both the physical and social environment.

Charlie is a smart and persistent 30-year-old with a strong personality. He has a great sense of humour, appreciates practical jokes, and likes to play around. For the past ten years, Charlie has lived in an apartment in a residential facility adapted for persons with congenital deafblindness. Most of Charlie's neighbours have known him since his school years.

"Charlie has a great sense of humour, appreciates practical jokes, and likes to play around."

Charlie has assistants present both day and night to provide company or support when needed. He sleeps between 4-6 hours a night. Despite this, he remains alert and active during the day, but due to his sleeping pattern, he needs his assistants present during the night as well.

Charlie likes being occupied. He enjoys the outdoors, going on car trips and exploring his surroundings. He also enjoys turn-taking

games and engaging in cooking and taking care of his apartment together with his assistants. He prefers close interaction in a One-to-one setting, where the other person can focus exclusively on Charlie. In larger groups, he often observes, while in smaller groups, he interacts more directly.

Before taking part in an activity with another person, Charlie wants to sit and look into the other person's eyes, very close, for a long time. He holds the other person's hands and plays a rhythmic, swinging game to settle down and find a common tempo. After this he is ready for adventure.

Living with CHARGE

Charlie's sensory integration and perception of the world around him are affected by his CHARGE syndrome. He has a narrowed visual field and possibly a cerebral vision impairment. He can observe the smallest changes in his environment one day, whilst the next he may walk straight into a parked car. He needs a guide-interpreter to be safe when he is in traffic, and, by choice, also when he is exploring new places. He visually understands simple, adapted, and concrete sign language but often prefers sign language in a tactile modality. Bright colours attract him, and pictures and photos are easier for him to see on a screen with high brightness.

Further, Charlie has functional deafness. He neither responds to sound nor use hearing as a channel of communication. When tested in childhood, he had a hearing loss of 60 decibel in his right ear and is considered totally deaf in the left ear. Charlie has cup ears, which are very common for CHARGE syndrome, making it difficult to try out hearing aids. After primary school, he has completely stopped using his hearing aids.

"When it comes to how clothes feel against his body, Charlie is very sensitive; they must fit tightly."

When it comes to how clothes feel against his body, Charlie is very sensitive; they must fit tightly. He also appreciates tactile stimulation such as massage, showers, and baths.

Charlie does not seem at all affected by temperature changes. He likes to wear many layers of clothing even if they make him very

warm. This is most likely linked to a need for pressure on the body to enhance his body perception.

He enjoys pressure on joints and muscles. It can be very soothing for him if someone taps on his body rhythmically from top to bottom putting extra pressure on his joints. Charlie often sits with his legs folded beneath him. This increases his awareness of his body. Charlie also enjoys sitting in confined spaces. He appreciates pressure on his body, by means of a weighted blanket, a bean bag, or another person leaning on him.

In new places, he needs to lie on furniture or other things, on his back with his legs bent. Sometimes he puts one foot against the knee of the other leg and places his head lower than his body. This position puts pressure on large joints and helps him perceive his surroundings. This is his strategy for calming down, to self-regulate. Charlie appreciates if the assistant is imitating his position and gives him plenty of time to prepare for exploring the new environment. He needs to observe his surroundings in peace without demands. He also needs long time after activities to process what's been happening.

Charlie has scoliosis, which, along with narrow airways, asthma, and allergies, is challenging. He needs to control his weight to avoid too much pressure on his lungs.

Self-regulation – ways to stay in control

Charlie stresses easily. He needs to know where he is going, and why, to be willing and able to participate in activities. Changes in physical or social environment is hard for him. The same is true when many people move around and worse if they are moving things around. Charlie then has an increased need, compared to when he is in his own home, to check that furniture and things are in order. He likes things to be in a straight line and persons around him can help make things easier for him by supporting his need for structure.

Charlie has certain behaviours that are his way of dealing with these situations. They are strategic and conscious ways to self-regulate, calm down, and maintain self-control:

- First, he connects his hands and bite on one of them.
- He then may need to stand up and spin around a few times, with one foot planted firmly in one place and the other making the circle.
- If this doesn't help, the next way to regulate is to run in small

circles. Often Charlie is making sounds while running, it is easy to hear that he is worried.

- Lastly, if none of these strategies help him, Charlie chooses to leave the activity and/or the room.

Different communication strategies

Charlie uses body language, tactile gestures, and signs to communicate. He also uses concrete objects to show what he wants to do.

Charlie understands adapted sign language through both visual and tactile means. This includes using short sentences, adjusting the pace of communication, and considering distance. The needed adaptation depends on Charlie's state of mind, motivation, and the conversation topic. Concrete objects combined with signs are used as augmentative and alternative communication (AAC). For example, to clarify what to do outdoors, the sign for car is used in combination with the actual car key.

Charlie's year with a dance activity

One winter a few years ago, Charlie was introduced to a new daily activity: a dance session focused on body awareness, body image, and social skills. This should suit his interests perfectly. Despite taking place in a group setting, it involved one-to-one interaction with an assistant close to Charlie throughout the session. Together they followed the dance instructor's teachings.

The activity would enhance the rhythm of the music with joint movements such as rocking, back-to-back. In the dance, the assistant used fabrics, feathers, balls, pea bags, and hands to create breezes, beats, pressures, stretches, and strokes on the participant's body.

To avoid a new environment, the activity began in a familiar setting, the residential facility's common room.

Curiosity

Before the dance activity was started, the furniture in the common room was rearranged. Beanbags and yoga mats were laid out to mark the location of the dance area. Charlie was very curious about what was happening. After it was explained to him that the furniture would be placed differently for the duration of the activity, Charlie laid down on a beanbag but found it difficult to get fully settled. There were a lot of things he needed to keep track

of. He wanted the furniture back in place and asked for this, several times. The assistant tried to help Charlie endure and calm down by, among other things, laying down next to him.

Charlie participated in the beginning of the session when rhythms were tapped on the floor near his body. He did bite his hand but remained lying down. In the next exercise, Charlie got up and tried to correct other participants and move them around. He wanted to have the assistant close to him but could not calm down. After an exercise, when a large piece of silk fabric was raised and lowered over Charlie, he got up and left the room.

When the dance activity was over and the furniture was back in its usual place, Charlie wanted to check several times that everything was in order and found it difficult to do other activities that day.

A new environment

For the rest of the term, Charlie could not settle into this activity. He joined in, rearranging the furniture before the dance activity, and participated at the beginning. But often, after ten minutes, went home to his apartment.

After the summer break, the dance activity moved to the daily activity centre. In a large room, the dance area was defined by curtains, screens, and a ring of yoga mats with the equipment in the middle. There were also some beanbags in different colours for those who wanted to use them instead of lying or sitting on the yoga mats. The room has good sound equipment, and the walls and floor are made of wood, which provides good vibration amplification of the music.

Charlie quickly chose an orange beanbag as his place and the assistant sat next to him. Again, he found it difficult to stay in the activity with the group. After a few exercises, he decided to move to a sofa behind the curtain at one end of the room and lay down with his head in the assistant's lap. In this position he could continue to do some of the exercises, though separated from the group in a smaller, confined space. This repeated itself over the coming sessions.

Changes became a turning point

After two months, some further changes were made.

Charlie especially appreciated the pea bags as they provided weight and pressure on the body and could be stacked in many fun

ways. The pea bags became the symbol of the dance activity and were used as AAC before, during and after the activity.

Charlie's beanbag was moved towards one wall to reduce distractions and other colourful beanbag were placed around him.

"We made sure that Charlie always had a familiar assistant with him who is used to interpret his expressions and interacting with him."

We made sure that Charlie always had a familiar assistant with him who is used to interpret his expressions and interacting with him. It was not possible to have the same assistant each time, but a small group was selected and rotated between sessions. They also received guidance in the dance itself with clarifying explanations on how the different exercises in the session focus on strengthening interaction and body perception.

Video was used to show the progress of the interaction between Charlie and his assistants, to help analyse which exercises and materials seemed to attract Charlie the most. Then, these were expanded and used more in the activity.

After these changes, Charlie became more resilient and partook longer in the activity.

Yet, another two months and further adaptations were made in the room, to reduce sensory impressions and create a cozy and safe atmosphere. Charlie's place in the activity was moved to a corner marked with two large fabrics on the walls, one blue and one orange. His orange beanbag was placed in the corner and the ring of yoga mats was moved closer making it easier for Charlie to be part of the group.

Charlie fully participates

In the early spring, more than a year after the activity first was introduced, Charlie began to stay in the corner for the entire 40-minute session. Initially, he focused more on himself and his assistant, skipping some parts of the dance but Charlie now fully engages in the group.

Throughout the spring term, he actively participates in all

elements of the dance, often smiling, laughing, and showing clear appreciation of the different parts of the activity.

Checklist for introducing a new activity

- Discuss with the assistant why the activity is relevant.
- Create clear paths to the activity/location using colours and materials.
- Designate a clear place for the participant using colours, pictures, and symbols.
- Employ clarifying pedagogy in preparation as pictures, physical symbols, and signs.
- Use the same symbol and sign during the activity as in the preparation.
- Facilitate positioning that strengthens the possibility of participation.
- Give plenty of time for the individual to get used to the place/location of the activity.
- Adapt the pace of the situation to the individual.
- Wait for and follow the individual's initiative without making demands.
- Repeat the activity several times before changing anything.
- Allow plenty of time for evaluation and counselling.
- Use video analysis to reinforce interactions.
- Use video analysis to show progress, identify the next steps, and extension of the activity.

For Charlie, the CHARGE acronym means

C: He has visual field loss with no coloboma.

H: He was born with a hole between the heart chambers.

A: Charlie has narrowed breathing canal.

R: He is short in stature.

G: He has arrested puberty.

E: He has functional deafness and cup ears.



Linda's next chapter in a new city

Linda lives an independent life in her apartment in a residential facility. However, moving to a new city wasn't easy to begin with. She had to learn to focus more on herself and to understand other people's emotions. Now she feels capable of doing almost anything. "I like to learn new things", Linda explains when she tells her story of personal growth, adaptation, and the joy of overcoming challenges.

"In my job, it's good that the supervisors explain how to do things so that I can follow and understand. They use good visual support."

Linda who is 24 years old, moved to a new city a year ago, where she resides in her apartment in a building where there is staff available night and day. There are also common rooms for the residents to socialise. We meet Linda at her workplace, accompanied by a trusted interpreter, to talk about her life and personal development.

– After I had moved into the apartment, my mum noticed how tidy and clean it was; she was very impressed. Earlier, things could be messy, and I often ate carelessly. But after moving out, when I came home to visit my mum, she observed how I was eating more properly, leaning forward, and sipping carefully, Linda explains.

"After I had moved into the apartment, my mum noticed how tidy and clean it was; she was very impressed."

Living with CHARGE

Linda is blind in her left eye, and she is having difficulties seeing in the dark. Distinguishing patterns and processing strong colours give her problems when she reads manual signs.

– Sometimes my eyes flicker and I need to close them and rest until it stops. The flickering disturbs my vision so much that I need a guide-interpreter take me to a place where I can rest. In these instances, I rely on tactile sign language. I also need this when it is dark, Linda explains.

As an infant, Linda had a heart defect but after surgery, her heart was functioning well again. Her breathing was also affected, and she got a cannula in the throat when she was two years old which made eating difficult.

– At 14 years of age, the cannula was removed and after that, I could eat everything as usual, and breathe well, she recounts.

Linda is deaf in her left ear, but with the help of a bone-anchored hearing aid (BaHa), she can hear quite well with her right ear. Understanding lower-pitched voices is harder, while higher-pitched voices are easier. But she prefers sign language over spoken language.

– I can understand simple and clear words, but if there's a lot of talking around me it's hard to follow and I need an interpreter.

"I can understand simple and clear words, but if there's a lot of talking around me it's hard to follow and I need an interpreter."

A busy life

Linda enjoys walking, exercising, baking, cooking, painting, playing games, going to the cinema, dining at restaurants, and having

coffee with friends. Most of all, she likes the theatre. She feels capable of doing almost anything.

– I have a job now too, at a daily activity centre. I like to learn new things, it is fun. The supervisors explain how we should do things so that I can follow and understand, she says.

At work, there is a good visual job description with pictures, such as how to do handicrafts. They also have a day planner with pictures.

Linda learns to focus

Moving to a new city that she didn't know made it difficult for Linda to focus; as an example, she had to get used to new sounds. Linda gets easily stressed due to a hormonal imbalance. If other people argue, it affects her a lot. She worries easily, but the staff can help and support her.

– They tell me to try to focus on myself, to go up to the flat or another room and do something else, something funny, and to think about other things. If I have difficulty with sounds, the staff can remind me to take my hearing aid off.

– When I was younger, it was easier to focus on myself. But moving to a new city has been a good exercise for me to learn to focus and understand that others have their ways of doing things.

Linda teaches new staff sign language

A guide-interpreter assists Linda outdoors. She also needs adaptations for the optimisation of sound and light, computerised schedules, and sometimes she needs to be reminded of meetings and doctor's appointments.

– I get pictorial support showing what time to leave and what time we should be there, Linda explains.

Linda appreciates when the staff reminds her of things:

– For instance, if I start repeating myself, they remind me to focus on myself and not think about what others are doing. But they can also help me cook, and we have fun together.

Sign language is essential for Linda. The staff, comprising deaf, hearing-impaired, and hearing members, creates a diverse mix. Linda is curious to know and often asks about the hearing status of new staff.

– It's good to know, Linda notes. We help hearing staff, who are new to sign language, by teaching them new signs so they can adapt slowly to avoid misunderstandings. We spell slowly so that they eventually understand. The staff say it's okay.

Understands emotions better

As a child Linda often questioned her emotions, wondering why she felt angry or upset. Her mother explained to her that it's okay to feel angry and grumpy sometimes. As a teenager Linda became more aware of emotions and that everyone feels differently.

– Around 12–13 years old, I realised that it's okay, to have different feelings. Earlier, it was hard to understand why a person was sad, but now I'm aware that it's normal, she says.

When she was a teenager, she sometimes said inappropriate things, however, she has matured over time.

– In my twenties, I became more conscious of my words. Now I can even say to others "No, don't say that, think before you speak!", Linda adds.

She is grateful to both primary and secondary school for teaching her about right and wrong.

Linda recognises her personal development and so has her mother. She tells her that she has become more mature, developed, and grown.

– Sometimes, mummy wants me to be a little girl again, but I remind her: "No, no, I'm an adult now, mummy!"

"Sometimes, mummy wants me to be a little girl again, but I remind her: 'No, no, I'm an adult now, mummy!'"



Understanding and supporting Anna: A case study

This case study explores the life of Anna, a 38-year-old woman with CHARGE syndrome. Her interactions and communication methods give us insights into the challenges of living with CHARGE syndrome and the significance of personalised care and understanding.

Anna is 38 years old and has lived in various residential facilities since she was four. Each year, she receives 50 hours of special education for adults and has a contact person for ten hours a month. From 9 to 18 years of age, Anna spent weekends and holidays at a support family.

Anna lives in an apartment of her own in a residential centre. Her apartment consists of a bathroom, living room, sleeping area, and a small kitchen. She enjoys ballgames, watching children's programs like Teletubbies, doing jigsaw puzzles, and watching videos made by herself on her Ipad during her everyday activities.

The Ipad is also her communication tool. She films herself talking about daily experiences.

She spends time taking photos, writing words on the computer, organising laundry, and goes for long walks with an accompanying assistant.

"For Anna to thrive, she relies on support from people who know her personality, her interests, and who have a good knowledge of her body language and communication methods."

Reflecting on her day

For Anna to thrive, she relies on support from people who know her personality, her interests, and who have a good knowledge of her body language and communication methods.

Every afternoon and evening, Anna writes nine words in a calendar book functioning as her diary, with the help of an assistant. These words reflect her day, including things that are important to her, her experiences, and future appointments. She uses pictures on her Ipad as reference objects.

Anna also enjoys spending time in the communal kitchen and living room checking what her fellow residents are doing. In the kitchen, Anna has a weekly task of loading the dishwasher. She also helps her fellow residents and encourages them to choose a dessert for the weekend among the dessert pictures in a special box.

Living with CHARGE

The first four years of her life, Anna was hospitalised several times. She underwent surgery for a congenital heart defect and for cleft lip and palate. She lacks a passage between her nose and throat and had a tracheostomy implant.

Anna has colobomas in both eyes, horizontal undulating nystagmus (repetitive, uncontrolled eye movements) and, microcornea (small cornea, less than 10 millimetres in diameter), is nearsighted, and has had surgery for cataracts. She is visually impaired with a limited field of vision. Her ears are malformed, and her hearing is impaired. She functions as deaf. Her developmental profile ranges from two to eight years.

Overcoming limited vision

Anna reads and writes by holding her face close to the material; she uses her left eye for reading and her right eye for orientation.

She can't see upwards and can only see gestures from the top of her head downwards. Anna lacks depth perception and can't determine height differences. She finds it difficult when the light is low.

Block magnifiers and spectacle correction haven't been effective.

It is important to communicate within Anna's narrow field of vision. She is very dependent on her residual vision and on the adaptation to conditions to see better. Non-manual components of sign language, such as mimics, are hard for her, but she communicates using a combination of some signs, writing, pictures, and text.

Occasionally, Anna uses tactile sign language and relies heavily on her tactile sense. She can use her fingertips or pick up things to examine surfaces and objects tactilely and visually.

A walker increased her mobility

Her balance problem and limited vision make it challenging to navigate in unfamiliar terrain. However, this does not stop her from visiting new places.

Adapted lighting, and flooring inside and around the accommodation centre make it easier for Anna to move around. The use of a walker increases her mobility.

Anna's sensory loss is affecting her motor skills. She has shortened collarbones which has led to increased thoracic kyphosis^[1], and she has a slight scoliosis (a back misalignment). She compensates for balance issues with a wide-tracked gait and has begun to use a tall, rear-facing walker with forearm support in her daily long walks. This has helped her to increase her walking distance.

Anna walks in a wide-track gait with knees bent to lower the centre of gravity and her body swaying from side to side with each step. She often finds better balance by sitting with one foot on the other knee or with crossed legs.

Anna probably has no sense of smell and taste. A lack of olfactory bulbs is a common symptom in persons with CHARGE syndrome. Anna enjoys mixing liquid food, experimenting, for example, mixing ketchup, caramel, whipped cream, and fruit colouring.

1. Curvature of the upper part of the spine

Relying on assistants

Other people often guide Anna's use of language. She can restrain impulses only for a short time. She adapts her behaviour a bit with others and tries to calm herself rather than allowing others to comfort her. Anna does not express her feelings verbally; instead, she relies on assistants to understand and fulfil her wishes and needs, accepting their decisions.

Anna can wait a while for things she enjoys and relies on the assistant's suggestions and guidance in how things should be done. Anna has a sense of time and needs many repetitions to the daily routine, especially if there are changes to the plan.

Limited sign language

Anna communicates by visual sign language – but not at an age-appropriate level. She uses from one to up to eight signs, depending on the situation, and she likes to use reference objects such as pictures, books, and games.

One of her communication strategies involves asking questions using one to two characters, while simultaneously expressing a questioning attitude. The person she's communicating with needs to figure out what Anna is asking and try to get more details to understand.

Once Anna is motivated and has received information, she can participate in a conversation.

"Once Anna is motivated and has received information, she can participate in a conversation."

In-depth knowledge

Anna likes to hand over messages from one assistant to another, ensuring that everyone is informed. She can start a conversation with one word and then look at the assistants with the expectation that they know what she is thinking.

The assistants use triparty conversations to support her, limiting the sign space, supporting the signs tactilely, and using clear body language.

Anna understands more signs than she uses. She is a "data

collector" and wants information about fellow residents and staff regarding absence, illnesses, holidays, and shift schedules.

The communication topics Anna selects are often very personalised, person- and context-dependent, which requires an in-depth knowledge of her.

Pictures for communication

Numbers are a favourite: how many different things they had for dinner, how many buns were baked, how many presents she got for her birthday, etcetera. She also takes notes of time, such as when someone needs to go home or when she is going for a walk.

Anna expresses wishes for her social pedagogical schedule for the day and if making purchases, what to buy. She films her everyday experiences on the Ipad and takes pictures. Afterwards, she enlarges them to study them closer.

Interaction with Anna – a video analysis

We have included a video analysis here to give an example of Annas communication methods. In the video, Anna and an assistant are sitting at a table with a calendar in front of them. Anna grabs the assistant's right wrist and points with her other hand to another assistant who is sitting at the same table knitting. They both laugh before the conversation begins.

Anna: (Name) – holiday home.

Assistant: You say (name) and holiday home, what do you think?

Anna: (Pointing to her diary) Search – holiday – home – date – search.

The two assistants communicate verbally about the dates.

Anna lightly places her hand on the assistant's wrist to establish contact, subsequently pulling the assistant's arm towards herself. She tries to turn the assistant's head while maintaining hand contact.

Anna: (Changing subject pointing to her tracheal cannula and then to her calendar) Summer house – go again.

Assistant: We should also look at when to go to the hospital.

Anna: Summer house.

Assistant: On 30 March we can look at the summer house.

Anna: (Pointing at the assistant and then down at the table) Is (name) present?

Assistant: (not answering the question).

Analysis

The video shows several topics of conversation at the same time. From Anna's perspective, they are talking about holidays, the coming visit to the hospital, and who is coming and when.

The assistants also discuss the organisation of staff holidays. They should have discussed this before the conversation with Anna because she gets confused, and Anna and the assistant talk past each other.

Initially, Anna does not realise that they are talking about a colleague's holiday and not her own. A couple of times, Anna tries to return to and point to the calendar to confirm that they are talking about her holiday, while the assistant is still talking about when a colleague might be going on holiday.

A future action strategy must be for staff to coordinate dates for hospital visits, staff holidays, and possibly the exact date of the Annas holiday before involving her.

Better sign language knowledge

Another strategy is higher partner competence. The partners must be at a higher level of sign language than Anna so that they can challenge and refine her sign language level at the right times and with empathy. Enhancing Anna's ability to express herself in a more nuanced way will provide her with greater opportunities to influence her own life.

In the conversation, she is the one who points out that at the time of a possible holiday trip, there is also time scheduled for a hospital visit. Anna shows a great and long-term overview of time and a good memory. The video sequence also shows that Anna can focus on more than one topic at a time, as she brings several topics into play unprompted and on her initiative.

Support, but avoid confusion

Anna often mixes various topics into a single conversation, and the partner needs to help maintain focus and structure preventing unnecessary confusion for Anna. The ability to include related

topics in the main conversation is a cognitive skill that should be supported and developed. However, it is equally important for the partner to assist Anna in avoiding having too many different topics in one conversation, to prevent misunderstandings.

It might be tempting to conclude that the confusion is caused by the assistant in the video. This occurs when the assistant, without informing Anna, shifts the topic and starts to talk about the colleague's holiday while Anna is still talking about her own holiday.

When developing future strategies for Anna, it is important to consider the assistant's role in the conversation and Anna's overview and energy levels.

For Anna, the CHARGE acronym means

C: Coloboma in both eyes.

H: Heart defect; Anna has had surgery for a congenital heart defect.

A: Airway anomaly; Anna lacks a passage between the nose and throat, and had a tracheostomy implanted.

R: Restricted growth. Anna is physically not very tall and her developmental profile ranges from two to eight years.

G: Genitalia might be underdeveloped but there is no information if Anna has ever been examined for this.

E: Ears are malformed and the hearing impaired. Anna functions as deaf having a hearing loss of 35–40 decibel, possibly due to a cortical (brain-induced) hearing loss or her hearing opt-out.



Inga is finding balance in life

Inga struggles to balance strong emotions with a need for calm. "The emotions control me, and it easily becomes too much", she says, sharing her world affected by CHARGE syndrome. Communication is the key, and for Inga, it is important to feel safe and to trust the other person.

"Communication is the key, and for Inga, it is important to feel safe and to trust the other person."

We meet Inga in a café together with a staff member whom she trusts, to talk about her life with CHARGE syndrome.

Inga lives in an apartment of her own. She is content managing her own time and enjoys living alone. She has neighbours and staff to spend time with, in communal spaces, and the staff is always present in the building. They all know sign language, even the hearing staff. Inga says that the relationship with the staff works well.

– Feeling confident at home took time for me. I needed to understand that this is my home, Inga reflects.

Living with CHARGE

Inga can see well with her right eye, less so with her left, and relies on glasses. While she can perceive objects at long distances, this is

tiring and affects her vision.

– Bright light can be challenging. I can get confused when I've been in strong light and then go into a house, then my vision needs to adapt, Inga says.

Inga underwent heart surgery as a baby and now her heart functions well, though sudden events can cause rapid heartbeats. She has no problems with her throat when eating, but her mouth tends to get dry.

If it is hot outside, the temperature can affect her breathing. Emotions also have an impact. When feeling sad, she needs to take a nasal spray. Inga has a bone-anchored hearing aid (BaHa) that helps her hear all the sounds around her. She can't perceive speech, but she can hear people talking. When people speak calmly and use one word at a time, she can hear words too.

– When I remove the hearing aid, I barely hear, just sounds like a door closing or metal rattling.

Noise is hard

Coping with noise is hard for Inga, it disturbs her focus and affects her communication. Even after removing the hearing aid following a disturbing sound, the echoes persist in her mind, causing stress. She can get migraines triggered by light and sound.

Pressing situations, a lot of people, and busy environments affect her. The way other people use sign language, and where they are positioned, can make her tired trying to read signs.

– It can be easier or harder depending on how they sign, I must feel safe and trust the other person, she explains and emphasises that communication is the key.

Working on relationships

Inga enjoys social activities, walks, and watching films and sports on tv. She is very social and uses social media a lot.

"I also like being in nature and feel good when I am close to the sea, she says."

– I also like being in nature and feel good when I am close to the sea, she says.

Inga thinks that CHARGE syndrome primarily affects her relationships. There are a lot of communication misunderstandings, and it can be hard to handle these situations.

– The emotions are in control of me, and it easily becomes too much when I have to deal with things.

Routines and support

Inga receives assistance with cleaning and interpreting everyday situations, beyond just language.

– A sign language interpreter often works well, but not always. Sometimes there are misunderstandings due to differences in sign language. I might be a little unclear or too fast sometimes. I try to be flexible in how I sign.

– But it also depends on who is interpreting, so it's about relationships as well, she adds.

Inga needs accompaniment when it's dark outside and when walking on uneven surfaces. She also appreciates help when traveling, like taking a train from the station. Her home has a flashing doorbell, which also works as a fire alarm.

Routines make her feel good. Changes can affect her negatively sometimes, for example, if an activity is cancelled.

– But if it was something uninteresting, I don't mind, Inga remarks.

She always writes down reminders on her mobile phone.

High expectations

Inga has high expectations regarding how people should treat her. It's a question about how one is met and relationships rather than knowing routines.

– I want to be able to trust the staff, but it depends on the relationship and what we can talk about.

In her view, being met in a good way means that the staff should be warm, understanding, and provide a sense of safety. They should not be jumpy, angry, or grumpy, and they should avoid getting over-excited too easily. They need to know what distance to keep that allows her to clearly see the sign language. Sign language is of course important and even hearing people can be good at it.

Coping with stress

Different situations can cause Inga stress: time, the weather, and even money.

– I find it hard to deal with money sometimes. How much should there be? Is it enough or not? Small things like that.

In advance of special excursions that she looks forward to, Inga worries: will it be good or not?

– It's all about my own expectations and need for everything to be perfect, which stresses me, Inga explains. This also applies to her job and interactions with people in different situations.

– I have a lot of expectations for everything, absolutely everything!

Inga needs to actively manage the thoughts that control her and persist even when she tries to slow herself down. She finds it helpful to get reminders from the staff which aid in calming her thoughts.

– I try to take it easy and think that everything will be fine. I focus on here and now and take everything step by step.

"I try to take it easy and think that everything will be fine. I focus on here and now and take everything step by step."

Getting good sleep and eating well have a big impact on her ability to get through the day and understand what's happening.

– I used to sleep badly, but it's better today. Sleep and energy levels affect a lot.

Better understanding needed

Inga wishes that everyone who meets people with CHARGE syndrome would learn more, not be afraid, and gain a better understanding of what it is like to live with CHARGE syndrome.

- I want others to understand me, and I want to tell them how I function, but it's difficult since relationships and trust affect me.

She would like to advise parents who have children with CHARGE syndrome not to worry, as this will have negative effects.

- Don't worry, there are many opportunities ahead in the future!

Summary

Delving deep into the study of CHARGE syndrome and its diagnoses can be daunting. When reading, a picture of illness emerges, telling us everything about what is wrong, and what is afflicting the person, who might be someone's loved one.

Frequently we find ourselves on pages filled with information that is challenging to comprehend. When only viewing the medical aspects, a one-sided picture is created. But who is the person behind the diagnosis? In discussions within the realm of disability issues, the consensus is that we are not defined by our diagnosis; rather, we are individuals who happen to have a diagnosis.

Of course, diagnosing is important, for the surrounding to give the right treatment and make adaptations for the person. But we must not forget that there's always a person behind the diagnosis. With this publication, we aim to provide insight into what it means to live with CHARGE syndrome, uncovering the potential within these individuals. Or at the very least, we hope that we have been able to share some of the successes experienced by individuals with CHARGE syndrome.

Thank you for reading these personal stories. We hope that all of you may dance into the future like Charlie, or be the driver, choosing your own road, like Karl.

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