ATR-x Syndrome

information for families

The Dutch ATR-x SYNDROME Foundation

Index

General information on the ATR-x Syndrome

What is ATR-x Syndrome?

In the Netherlands there are about 25 boys known with this syndrome. ATR-x is one of the many genetic abnormalities that cause developmental delay.

ATR-x Syndrome stands for x-linked Alpha Thalassaemia Mental Retardation Syndrome.

The three main features of the ATR-x Syndrome are:

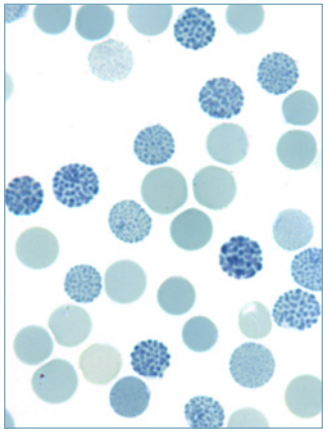
- *severe developmental delay*
- *characteristic facial features*
- *some form of anaemia (called a -thalassaemia)*

The syndrome occurs only in males. Females are generally (in about 75% of cases) carrier of the gene with the mutation, but there are also known families where the mutation was a 'mistake at conception' and the mother is not a carrier.

Diagnosis

Medical examination of the ATR-X Syndrome

Searching for mutations in the ATR-x gene and making the diagnosis is nowadays relatively quick and easy. DNA testing via *Whole Exome Sequencing* is the recommended method in the Netherlands.



The characteristic facial expression in a child with severe developmental delay is the most recognisable feature to test for ATR-x Syndrome.

Features

Learning difficulties

Most of the affected boys (96%) have learning disabilities which can be classified as serious.

Speech is usually absent, although some children learn a few words and a small repertoire of sign language. Some are barely aware of the family and the environment. Others understand where to find a biscuit tin or how to turn on the TV and how to perform simple tasks.

About half of the children learn to walk; 45% learn to walk before they are nine years of age.

Characteristic facial features (90%)

Many syndromes are recognisable by characteristic facial features (like Downs syndrome). This applies also for ATR-x Syndrome, although the characteristic features are more clearly recognisable at an early age. The size of the head is often small (microcephaly, 75%), the eyes are wide apart, the bridge of the nose is broad and flat, the nose itself is small, triangular and turned up at the end. The upper lip is narrower and the lower lip is full and turned outwards.

Anaemia (α thalassaemia) (85%)

Anaemia can occur in children with ATR-x Syndrome. This is due to a reduction of the manufacture of one of the proteins that produces haemoglobin. This form of anaemia is called α-thalassaemia. It can be diagnosed with a simple blood test. It is a mild kind of anaemia which does not require treatment.

Neonatal hypotonia (85%)

At birth, the children are usually muscularly weak and during the first 6-9 months their development is delayed. Milestones such as sitting or crawling, are delayed.

Genital abnormalities (80%)

Genital abnormalities occur in 80% of the cases. This can occur in mild forms. The testicles are not descended, the penis may be small and underdeveloped. The foreskin may be poorly developed or partially missing. With some children it is unclear what gender they are. These children have no uterus or ovaries, but small malformed testicles that can only be found with keyhole surgery. The problems of genital development is likely due to insufficient production of male hormones and this may also be the reason that a number of the children have problems going through the normal development of puberty.

Abnormalities of the skeleton (90%)

These are quite diverse and can be seen as the children grow. Occasionally a child is born with a clubfoot. Joints may be in a fixed flexed position, especially in the fingers. During the aging process a curvature of the spine can occur.

Heart problems (20%)

Only some of the boys have control of bowel and bladder. One of the most common problems is that urine during emptying of the bladder can again flow upward towards the kidneys. This can increase the chance of causing urinary tract infections.

Kidney problems (15%)

Slechts een deel van de jongens heeft controle over de darmen en de blaas. Eén van de meest voorkomende problemen is dat urine bij het legen van de blaas weer omhoog stroomt naar de nieren. Dit kan leiden tot een grotere vatbaarheid voor urineweginfecties.

Digestive System

After birth, there may be issues with feeding due to the limited presence of the sucking reflex. Frequent spitting up of food or vomiting are common and will often become less as the child grows older. Swallowing is rather uncoordinated for children with ATR-x Syndrome - some children will show a reluctance to eat and swallow and will choke regularly. The profuse drooling and burping, which is so characteristic, are likely to be associated with uncontrolled swallowing. Many mothers will indicate that their son uses multiple bibs in the course of the day. One or two cases are known in which the gut has gone "on strike" because the normal contraction of the gut stops. Almost all children suffer from a form of constipation.

Growth

Many of the children are small in stature (65%), their growth is not consistent with that of others of the same age. Some children grow normally during childhood but their growth can become stunted during adolescence (between 10 and 15 years).

Behaviour

The children are usually described as cheerful and affectionate. They exhibit a wide range of emotions: joy, enthusiasm, excitement, grumpiness, sadness - depending on the circumstances. There are reports of sudden emotional outbursts with persistent laughing or crying for no apparent reason. Like most children they love attention and to be played with, they like especially noisy games, frolicking, playing with water and musical toys. Some children exhibit quite repetitive behaviour. They bite their hands or put their hands in their mouth. Most children rest easily in the evening and generally fall asleep without any problems. However, there are known cases where parents say that their son often stays awake all night long.

Epilepsy (30%)

About a third of the boys suffer from seizures. Generally they can be treated with medication. Children who suffered from attacks when they were very young, seem to have less problems when they grow older. Some boys get seizures into puberty.

Life Expectancy

Because this syndrome has not been recognised very long, we don't have a clear picture of life expectancy. Some adults are healthy in their thirties. There is not much information available about boys deceased and causes of death. Some children have died before the age of 5. Pneumonia happens regularly because infections in the bronchial tubes occur due to vomiting and choking on food.

The characteristic facial expression in a child with severe developmental delay is the most common reason to test for ATR-x Syndrome.

Max

Name: Max Vis

Age: 2,5 years (07-05-2012)

This is Max. Max is 2.5 years old. He is a very social, cheerful and happy child. He is very easy and very people-oriented, the more people the better. He has a lot of perseverance. He is easily pleased. He is very positive in what he does, and he likes to see happy people around him. He loves music, singing and eating. He has recently started exploring, he pushes himself forward on his buttocks. Annoying behaviour is that he pulls peoples' hair and pinches.

Process to diagnosis

In July 2013 Max was diagnosed with ATR-x Syndrome.
The DNA test was conducted when he was then about 2 months old because Max did not grow well. When DNA testing the parents did not know that he had ATR-x. When they told about their son Thomas, who is now deceased, they looked at what similarities there were between the syndromes of having a healthy daughter and a 'not healthy' son. *"Our intuition was that there was something wrong with Max,"* his mother said. For example he couldn't eat and drink properly.It took three months before the diagnosis was known. The ATR-x Syndrome was determined by means of DNA. The mother is a carrier of the ATR-x gene, which was determined by blood tests.

How did you experience the process?

"Well, of course, quite emotionally. You hope for a healthy baby. Especially at the ATR-x weekend we saw many older children with ATR-x who were in different stages of development and that came as quite a shock. It was difficult to combine the care for our son with our work, so my wife now has stopped working. In the beginning it was a rollercoaster but now we're getting more rest. In the beginning he was in hospital every 6 weeks but now he is much calmer because he doesn't have to go there so often anymore" his father says.

Symptoms and development

Max has some characteristic features of ATR-x. He drools a lot. He also has a developmental delay. He will learn certain skills, but he learns slower. In terms of developmental age he is now around the age of 1. He's ahead with some skills and with others he is lagging behind. Further he gets physiotherapy to learn to walk and speech therapy which helps with his eating. Max uses a special walker so he can get around. He also has a wheelchair.

Daily life of Max

He goes to school twice a week, from 9 to 3. A physiotherapist, an occupational therapist and a speech therapist come here and they have developed a special plan for him. *"It is pretty heavy for a child of 2 but he thinks it's fun. School is his second home. I am amazed at what he is learning there "*, his mother said.

On Wednesday afternoon they get family support. This person does special activities with Max.

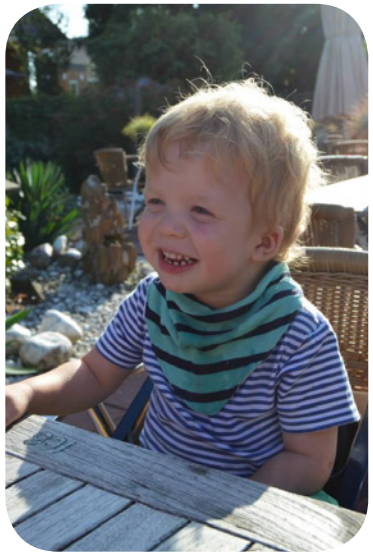
Daily life of parents

Mother stays at home and father goes to work. There must always be someone present with Max. *"We now have a cot where we can put him, so we can leave him alone for just a moment. It is restricting for our daughter. She has to stay indoors more often, because we cannot go outside so easily with Max. He is very susceptible to colds. And when that occurs, he spits, which we want to avoid, so we are very careful. We only go out if the weather is good"*, the parents say.

Great moments

"There are quite a lot of them! It's often little things that make it worthwhile like in his response. For example, he greets everyone and that's sometimes really funny. We love that he is trying to stand up. Also his sister can play really well with him. You measure your happiness to a different extent. You notice that little things are very beautiful."

Flint



Name: Flint Brophy

Age: 3 years (20-03-2012)

This is Flint. Flint is 3 years old. He is very cheerful and finds almost everything interesting. He is curious but also vulnerable. He used to get overstimulated very quickly but now he is super friendly and social. He is enthusiastic, but easily distracted. His parents say, 'he is a happy little guy.' His pain threshold is very high and he has no sense of danger.

Process to diagnosis

In September 2014 Flint was diagnosed with ATR-x Syndrome. Flint was born prematurely, in the 33rd week of pregnancy. When he was about one year old, it became clear that he had a developmental delay. In retrospect, there were many signs of ATR-X Syndrome in those first weeks. He was very weak, had low muscle tone, and he had regular problems with his intestines. He also had difficulty eating and drinking. But it was not yet clear that this was ATR-x. After one year, the parents went to the hospital because since his premature birth his development had become worse. Doctors began investigate to find out what was the reason for the delay. There was a lot of testing and this lasted from March to August. In August, the latest results were known and the family was told *"We cannot find anything."* After that the family was referred to the Academic Medical Centre. Ultimately the clinical geneticist decided to test all genes for a mistake, both in the father, the mother and Flint himself. Here, they have found that it was ATR-x. Mother is not a carrier of the gene that is causing the syndrome.

Symptoms and development

Flint has a number of external features of ATR-x. Additionally, squinting his eyes, he suffers from muscle weakness and genital abnormality (his testicles are not descended). Flint has a developmental delay along with a developmental age of about 12 months. It takes longer for him to react to things. In terms of behaviour, he really likes hitting things because of the sound it makes, reading books and playing in the bath. He also likes to cuddle animals and other children. At around 1.5 years old Flint could sit up unaided, and he can crawl since he was 2 years old. Now he has started to stand up by himself and can walk holding on with one hand, or with a walker. He wears splints. In terms of language development we do not see much progress although we do see the progress in understanding the vocabulary and non-verbal communication. *"Furthermore, it is becoming more visible that Flint is different from other children. In the beginning it is normal that he cannot speak and wears a diaper and now it will become more visible the older he gets, "* his mother says.

Daily life

Flint goes to his grandmother's every Monday and Wednesday, and Friday he goes to the nursery. On Tuesday he's home with his mother and on Thursday he is at home with his father. On weekends the parents are both at home. He goes to a normal / regular nursery. He gets help from an institution to join in with the other children. *"Usually we take him to grandma on Sunday, so that we as parents also have an evening together. We try to go swimming once a week and we go to a rehabilitation centre once a week where he has physiotherapy and speech therapy. Sometimes we think, it is very tough for him, but then we look at him and see him smile and then we think, it's not so bad after all",* his mother says.

Great moments

"It's truly inspiring when you see how cheerful he is. It is a challenge to think; I have to go shopping, or something else, but you just have to put everything on hold and go along in his world. Because his world is actually quite nice. And if you also adapt to his pace then you also get a lot more contact, which is beautiful. Also beautiful are the smaller milestones that are achieved. Our goals / milestones are much smaller than the ones lots of parents have. The first time sitting up straight, or the first time grabbing a piece of bread by himself. That's really a great moment. It may sound less obvious to other parents, but the first time that your child really looks at you is great. That normally happens quite quickly but with Flint it took a long time before he would also respond to us. It is best when we can leave our concerns behind and just look at him, he's really happy. Sometimes he grumbles but actually he is always very cheerful. He really enjoys life."

Ricardo

Name: Ricardo van Gurp

Age: 12 years (08-05-2002)

This is Ricardo. Ricardo is 12 years old. He is a sweet and cheerful little boy. He laughs a lot. He loves to cuddle and is, therefore, everyone's friend. However, he can be very tense.

Process to diagnosis

In May 2005 Ricardo received the diagnosis ATR-x Syndrome. Ricardo was admitted in hospital when he was 6 months old, because he hardly ate or drank. In hospital they saw that the structure of the brain was not right, and he was referred to the Academic Hospital, where this was confirmed after the necessary studies. Looking for the cause, they were referred to the clinical geneticist. "*It will be like looking for a needle in a haystack, there are more than 1,001 syndromes, we were told*", his mother said. Based on the external features the clinical geneticists had a presumption of ATR-x Syndrome, this could not be confirmed by DNA alone. After 9 months of investigation, again blood samples were taken which were sent to Dr. Gibbons in Oxford. There, ATR-x Syndrome was confirmed. Research showed that not only mother and grandmother are carriers of the ATR-x gene, but several family members: his cousin Jorn also was examined again and tested positive for ATR-x Syndrome, after confirmation in Ricardo. Jorn is now almost 27 years old.

How did you experience the process?

"It just happens to you, you have to cope with it and you must go on, you want to get the most out of life. We try to be open towards others and try as much as possible to function like a normal family. Just after we had heard that Ricardo would be disabled for the rest of his life, our doctor came to visit us at home. He gave us good advice and he was very firm in that: "you are a family of four (Jarno was not born yet) and all four of you are equally important in the family, keep this firmly in your mind. It should not be that your life only revolves around Ricardo. "We try to do that as much as possible. That often means making choices, which is not always easy. "

Symptoms and development

Ricardo has a number of external features of ATR-x. He has high muscle tone, suffers from sleeping problems and possibly there is epilepsy present. In terms of developmental age Ricardo is around 9 to 12 months. Ricardo cannot sit independently, cannot talk and walk (never crawled). Ricardo seeks contact and recognises your voice. You do have a social bond with him. He expresses himself by sounds. He is currently receiving physiotherapy and a special therapy called 'Veronica Sherborne'. He also has a wheelchair, a special playpen, an adapted and adjustable bed, he has a feeding tube in his stomach and splints instead of shoes. He has had speech therapy for a long time for eating and mouth movements.

Home outplacement

At the age of 9, Ricardo was placed into a special institution. Previously he used to stay there some nights in the same group. When he was smaller and younger, a babysitter or day care was not an issue. As he became older and bigger and he was bound to his wheelchair, you noticed it became increasingly difficult to find a nanny. *"Then it is nice that the residential facility is close to our home, where everything is well organised. This enables us to live fairly normally as a family. This way we can give more attention to his sister Jennifer and brother Jarno. Especially now we know that he really likes it at the institution where he gets all the rest and attention he needs, "* the parents say.

Daily life

Ricardo lives 7 days a week at the institution and during the day he goes to the day care that's on site. *"We have no fixed times when we get him, we can come and go whenever we please. Wednesday is the morning I (mother) spend with him. Every week I attend a parent-child group with him, where we do fun activities with other moms and kids. We close the activity while enjoying a drink with something sweet and we share experiences with each other. Then I take him home on a regular base and I will bring him back the next day before the day care begins. If there is time in the weekends, he also comes home and then we bring him back on Sunday night. If we are unable to take him home we certainly visit him during the day. When he is at home, we're very busy with caring for him, "* the parents say.

Great moments

"We enjoy it when Ricardo laughs. Sometimes the whole family is in tears of laughter as Ricardo is laughing and having fun. We enjoy the moments and Ricardo himself enjoys everything. If he is comfortable and happy, we all enjoy his warmth and the attention he gives back to us. And also if we hear and see others who enjoy him and who say he is so cuddly and cute. That's very nice to hear!"

Tom

Name: Tom Hartmann

Age: 13 years (01-10-2001)

This is Tom. Tom is 13 years old. He is very enthusiastic, busy and very cheerful. He is everybody's friend and also a bit of a tyrant. Tom is someone who finds it difficult to limit his enthusiasm. He is always in good spirit and can easily persuade everyone to play with him.

Process to diagnosis

In September 2004, Tom was diagnosed with ATR-x syndrome. From birth it was not clear that Tom was different. However, he suffered from muscle weakness, but this was not disturbing. After three months his parents noticed that something was wrong. He reacted differently than his older brother. He made no eye contact. He looked past you. It was as if he did not see you. Furthermore, he stayed behind in his development. He also did not want to eat. In his first year, he would only drink infant milk with a few tablespoons of porridge. When Tom was about 2.5 years, he became ill and stopped drinking. He ended up in hospital, where he got a nasogastric tube. He then arrived at the Academic Hospital and he was 7 months in an institution, where he received intensive eating-therapy. When he was discharged from the institution, he ate and drank everything. Soon after he could attend the day care centre where he goes to with great pleasure till this day. When he was in hospital he was viewed by many specialists. A clinical geneticist thought she had seen this before. They recognised the facial features and the hands. After further DNA study, ATR-x Syndrome was diagnosed.

How did you experience the process?

"When Tom got sick and ended up in hospital, the doctor asked if we had help. When we asked why, he looked surprised at us and said, "Your child has a disability". This was the first person who told us. In the beginning you collapse and have to find your way. You need to adjust your dreams and future expectations. Now after so many years we found our way to cope with it. We have all accepted it and one single word or one look says it all. We're also glad that his older brother, Daan, is not ashamed of Tom. He holds talks in school about Tom and shows the educational film. It is very important to be honest with each other and keep talking to each other about Tom. This way we really grew closer together as a family."

Symptoms and development

Tom has some characteristics of ATR-x. He has a curved back and crooked little fingers and his testicles are not fully descended. He is hypermobile, he has a high pain threshold and he suffered from muscle tension. Tom does not have the symptoms to the worst extent. He can walk and communicate in his own way. He can say some words and uses sign language. His developmental age is around 2 years and 3 months. His development goes very slowly.

His gross motor skills are good. His fine motor skills are less. Tom has physiotherapy. There he learned to crawl and walk. Tom also has speech therapy once a week. The speech therapist teaches him mainly new gestures. In addition, Tom wears sturdy shoes for walking (he tends to walk on his toes), a buggy for going long distances, a special tandem, a special bed, custom seat, computer with touchscreen for eye-hand coordination and an iPad with the app of the Dutch sign language centre. Finally, Tom gets help with specific requests.

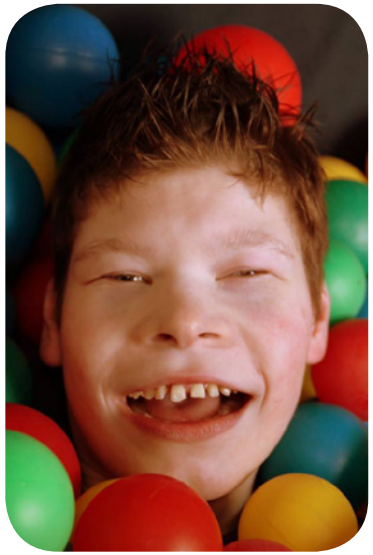
Daily life

Tom needs a steady rhythm. This means going to bed at fixed times and eat at regular times. For him it's best to have the same activities every day, but at home that is not always possible. *"We sometimes try to do something else on purpose, and further we don't have the freedom to go where we want whenever we want. We need to plan everything."* the parents say. Tom goes to the day care centre every day and every other week on Saturday he attends a special club. The other Saturdays there is a personal mentor who does all sorts of activities with him.

Great moments

"Tom is very affectionate, you get a lot of love from him and he is also open to that love. You never know in advance what awaits you. We are experiencing some really beautiful moments together. It's often little things but the fun of experiencing these together has only increased. The nice thing about Tom is that he is so pure in what he does. He's just himself. "

Marien



Name: Marien Verhoek

Age: 15 years (18-05-1999)

This is Marien. Marien is 15 years old. He is a cheerful boy when his physical well-being is good. He has a captivating smile and greatly enjoys physical contact (hugging or just frolicking games). He amuses himself with a toy that makes noise or which he can chew on. His cheerfulness can change quickly into restless behaviour. Marien then bites and beats himself.

Process to diagnosis

Marien was diagnosed with ATR-x Syndrome when he was 6 months old. During the maternity days Marien was not quite awake, he was lethargic and there was minimal eye contact. It was decided to have an additional check-up carried out by the doctor of the clinic and the midwives, but they could find nothing special and thought the parents were over anxious. After 3 to 4 months, the parents themselves contacted the general practitioner. The GP made an appointment with the eye doctor, as there might be something wrong with his sight because of the lack of eye contact. Meanwhile, the parents made an appointment with the clinical geneticist of the children's hospital and they made an appointment for January. However, in December Marien was struck by a coma, the cause was unknown, and was admitted to the ICU at the Academical Medical Centre of the Emma's children's hospital. From there, everything went fast and within 2 weeks the diagnosis ATR-x Syndrome and Periodic Central Hypoventilation were made via the clinical geneticist (and Dr. Gibbons).

How did you experience the process?

"The diagnosis itself was a confirmation of our suspicion. It was a relief that we now knew for sure. We could reasonably picture the future, because two of my brothers (the mother) also have the syndrome. In the beginning when our son was small, we had intensive mail contact with a family who had a boy of the same age with this syndrome. We saw many similarities. Now contact with other peers is very useful too. They sometimes have a broader view than if you remain within family relations. Furthermore, we had to give up many things. Because Marien is very susceptible, we cannot just go to the village, the pool, or the beach when we want to. The weather has to be really hot and windless. Our world has become smaller because of that. But on the other hand, you also meet helpful people who will always support you."

Symptoms and development

Marien has problems sleeping and since the last 3 years additional epilepsy. He has problems with his digestion so we can't feed him solid foods. He has obstructive and central hypoventilation. For this Marien has a monitor and uses oxygen at home. During the nights he is attached to this, and also when his saturation is below 90. The developmental age of Marien is somewhere around 6 to 9 months. His social development is about the same age. Furthermore Marien cannot talk, and spoken language also seems incomprehensible. He does recognise his own name though. When he sees things, he puts them in his mouth. Marien could sit at the age of 2.5 and propelled himself at the age of 6. He can pull himself up to stand, however, he can only stand independently for just a very short while. He has had physiotherapy from about 6 months and still has it now.

Daily life

"Marien is mostly cared for by us, his parents. Some days we get help by hiring someone from his personal budget. Since October 2014 Marien stays in a guest house about once a month. This relieves us, and then we can do activities with our other three children, which we cannot do with Marien. Marien goes to the children's centre 5 days a week. They treat him with respect and with humour. They provide for the intensive care that he needs and the guidance he gets is aimed at offering sensory experiences and contacts. Stimulation of his development in the areas of motor skills, communication, socialization and self-reliance comes in small steps. As we and Marien get older, you notice that it is becoming more physically demanding", the parents say.

Great moments

"We enjoy if Marien enjoys a romp game. Or if he is lying satisfied against you when you're reading to him. If you lie in the bed box with him, and he comes to you, and holds you.

His hearty laughter if he likes something, or rather his mischievous smile when he does something 'naughty'.

It's the little things that make it fun. Nothing is taken for granted, everything is special. And those are the moments we enjoy!

Cees

Name: Cees ten Katen

Age: 23 years (26-08-1991)

This is Cees. Cees is 23 years old. Cees is cheerful but also compulsively present. He cannot coop with any changes. Life costs him a lot of energy. Furthermore, he loves music.

Process to diagnosis

Cees was diagnosed with ATR-x Syndrome when he was about 12 or 13 years old. Only a few months after birth the parents noticed that Cees was behind in his development. He had starting problems with drinking. Because they thought that Cees had hearing problems, he went to the audio logical centre. There was nothing wrong with his hearing, but they wanted to further investigate his behaviour. There they came to the conclusion that Cees has severe multiple disabilities. There was an appointment made with a paediatrician. The paediatrician observed Cees and indicated that there was something wrong. Cees had a lot of medical examinations. First they thought of the Angelman Syndrome. But finally DNA tests revealed that Cees has the ATR-x Syndrome. Also was revealed that his mother carries the gene that causes the mutation.

How did you experience the process?

"At the audio logical centre they wanted to do another test because they thought something else was wrong with Cees. This test showed that Cees had a developmental age of 6 months, and that it would probably not change. When Cees was 3 years old, he went to a children's day care centre, but he was constantly sick, so he stopped going there. When Cees was 9 years old he started again at the children's day care centre."

Symptoms and development

Cees has problems sleeping and since the last 3.5 years he additionally has epilepsy. Cees choked quite often and suffered from reflux, but he now has medication for this. His developmental age is around 6 months. Furthermore, Cees cannot speak. He can walk a few paces and he has learned to crawl at physiotherapy. He has poor eyesight and sleeps with a special tie so he cannot get out of bed.

Daily life

"Cees needs care and guidance 24 hours a day, 7 days a week. Everything is a big struggle. Cees was placed out of house for half a year, unfortunately it failed. He now lives back home. Up to his 18th birthday he went to a day care centre. Now he goes to the adult day care in Hoogeveen. We take him there every morning and in the afternoon they will bring him back home again, " the parents say.

Great moments

"Great moments are when we see Cees rocking and enjoying his music, especially his favourite singers. And at bedtime as Cees puts his arms around you and wants to hug ... It sounds weird, but we learn the truth of life through Cees because he is so pure and real! "

Photography: Miranda Wolf

Information on the Dutch Foundation, the Policlinic for Rare Disorders and the Weatherall Institute of Molecular Medicine

The Dutch ATR-x Foundation

As a foundation we can pursue a number of goals:

- *establish contacts with peers and experts to share experiences and information in order to gain more insight into the life of a person with the ATR-x Syndrome*
- *maintaining direct contacts with practitioners and experts on care needs and the latest studies / developments of the ATR-x Syndrome*
- *increasing awareness of the ATR-x Syndrome in general and among physicians,*
- *paramedics and other health care providers in particular so that timely and accurate diagnoses can be made and a resulting treatment can take place in the best possible way*
- *represent the interests of individuals with ATR-X Syndrome and their parents /*
- *care givers and family members to increase the welfare and opportunities in the community*

The Foundation is committed to giving information about the syndrome. In addition, the Foundation organises family weekends and other activities. Thus, contacts can be made with peers and experiences can be exchanged. Finally, the foundation wants to create more clarity about the medical and behavioural characteristics that often occur in the ATR-x Syndrome through specific medical examinations.

Policlinic for Rare Disorders

The clinic for Rare Disorders at the Radboud University Medical Centre in Nijmegen focuses on the diagnosis and monitoring of children and adults with rare forms of mental retardation. The clinic is part of the outpatient clinical genetics at the Radboud UMC.

This Policlinic is for patients with unknown causes of intellectual disability and for patients whose rare form of mental restriction already has a known genetic cause. The clinic has particular expertise in various rare syndromes.

The clinic will carry out research into some symptoms associated with ATR-x Syndrome. Parents can come here with questions and it is a centre of expertise for the practitioners of our children.

A partnership will be formed with the research team from Oxford and with the Dutch ATR-x Foundation, so that there eventually will be more clarity about the life of individuals with ATR-x Syndrome.

The Weatherall Institute of Molecular Medicine

The research team of the Weatherall Institute of Molecular Medicine at the John Radcliff Hospital in Oxford is led by Dr. Richard Gibbons and specializes in DNA research related to the ATR-x Syndrome.

Doug Higgs and Richard Gibbons have worked together on this condition for over 20 years. The group has been involved in describing the clinical features, identifying the gene involved and the nature of the defect in affected individuals.

Their present work is focusing on the following areas:

- *helping with the diagnosis of ATR-x Syndrome*
- *identifying the underlying genetic lesion in affected children*
- *carrier and prenatal testing for families*
- *developing new diagnostic tests for the condition*
- *discovering how common ATR-x Syndrome is*
- *finding out about the different ways it affects children*
- *how the clinical features in ATR-x Syndrome arise*
- *determining the function of the ATR-x gene product and how it controls the activity of other genes*

Fund Raising

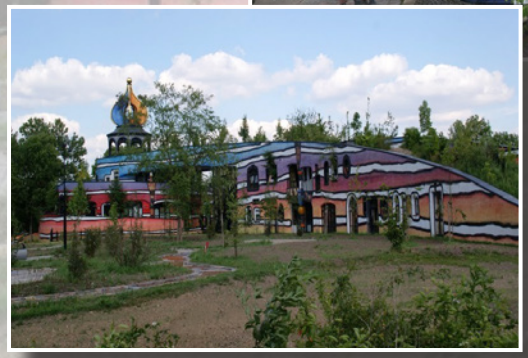
March 2011

ATR-x Holland Tour

May 2014

Peer Weekend

May 2011



Educational Film

September 2012

Music Activity

May 2013

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We want to thank Dr. Gibbons and Dr. Higgs for sharing their information in our leaflets

The Dutch ATR-x SYNDROME Foundation

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