The Undiagnosed

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Wilhelm Foundation

What's the name of the disease?

Almost everyone who has a disease has an answer


But not Lukas or his parents!
He has a disease that still is undiagnosed!

Freya is also undiagnosed

Freya and Lukas
are not alone!

Approximately
6 children per 10.000
children suffered
from an undiagnosed
disease

The Undiagnosed

	Known rare disease	Disease is not yet discovered
Not yet diagnosed (but should be)	X	
Undiagnosed		X
Misdiagnosed	X	X


Kajsa is 20 years old
and still undiagnosed!

The journey to a diagnosis

Not yet diagnosed

- 25 % of patients waited from 5 to 30 years for a diagnosis (EURORDIS survey)*
- 30 % (Australian survey)

Misdiagnosed - wrong diagnosis

- During that time 40 % received an incorrect diagnosis (EURORDIS)
- 50 % (Australian survey)

In the worst case senario a late diagnosis could lead to irreversible consequences and even be lifethreatening

**of eight relatively common rare diseases*

The Undiagnosed

Patients for whom medicine has failed to provide a diagnosis

Dr Gahl, National Institutes of Health (NIH) USA

Perhaps you wonder why I’m talking about the Undiagnosed?

I’m not a specialist,

not a doctor


I’m a mother of four children
Our three youngest children suffered from an undiagnosed disease
Emma
Wilhelm
Hugo

Wilhelm was our second child and healthy from birth

as his elder sister

- "Child epilepsy"
- Stomach problems
- When he was infected he got asthma and false croup at the same time

Wilhelm

Provocation

- Lactose

The result

- Normal as every other tests

Home again. But Wilhelm didn’t feel so good

The same night..

he got a fever (40, 5^{o} C) and difficult to breath

Back to the hospital

No one could explain why it happend

as it had many times before

Since they couldn’t find anything unusual

They told us that we could give him ordinary food

We did as we were told

But only for a short time

Because Wilhelm got really bad

- His cough was so bad that the his eyes were bloodshot
- He became very swollen in his face
- He got a terrible stomach pain

- When they X-rayed his bowels they couldn’t see anything because it was “air” all over

This examination didn’t lead to anything




Otherwise he was almost like an ordinary guy









Then I didn't know that there was a thing like undiagnosed diseases

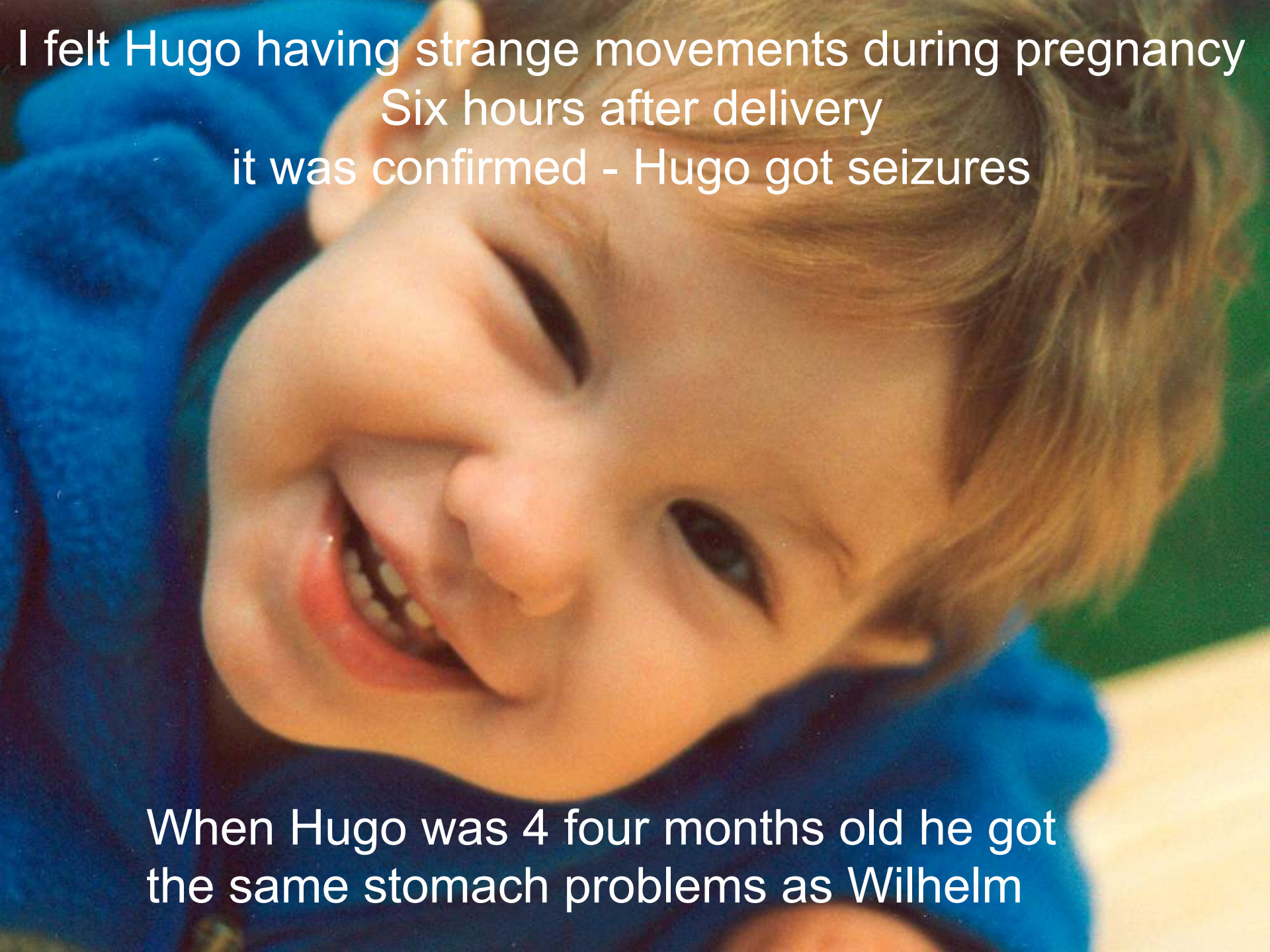
All tests were normal in Wilhelm's case

or

In Felica's case the test results don't match a known disease

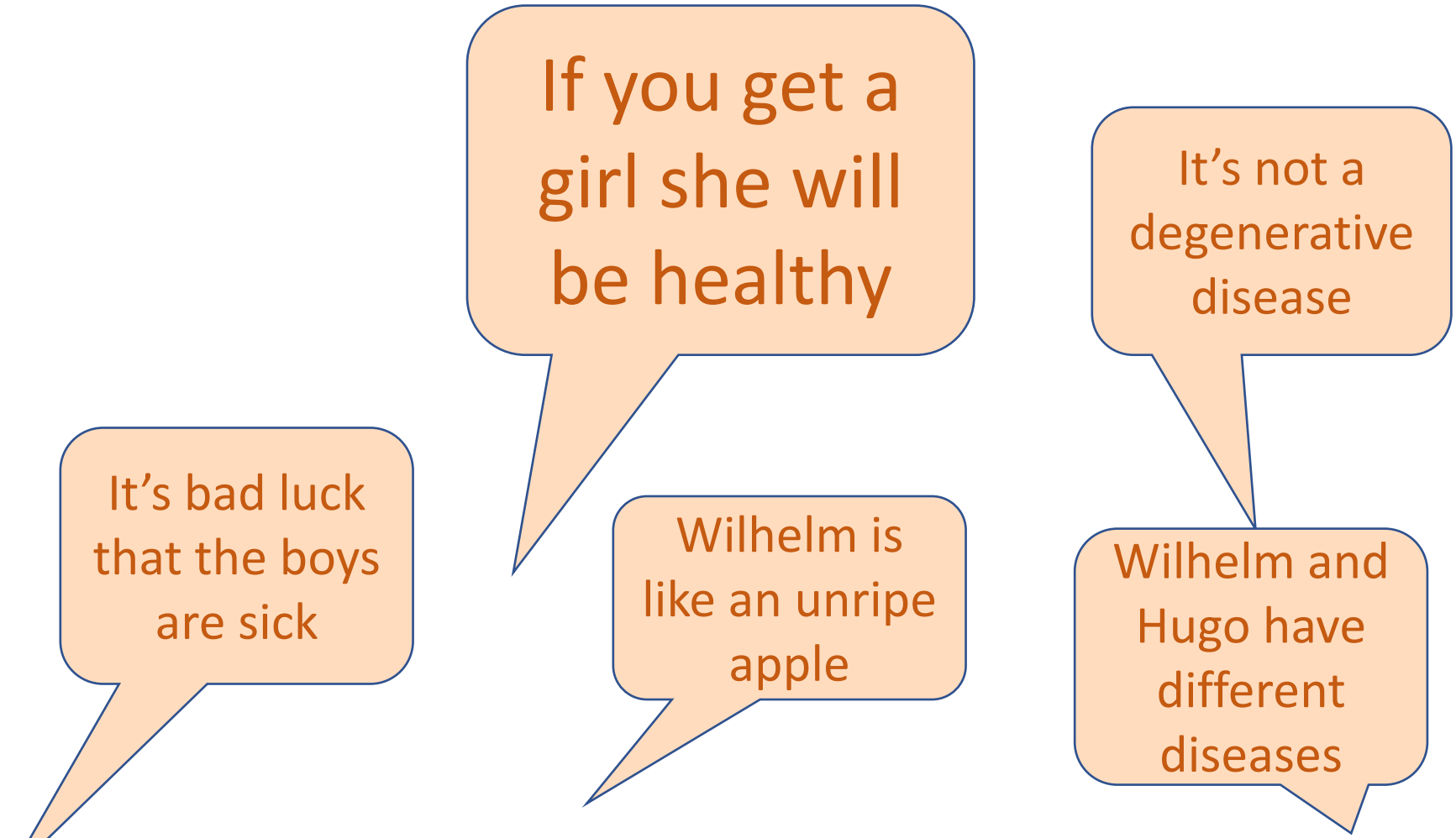
In both cases no one can give a diagnosis


We longed for siblings
The doctors told us that nothing was wrong even the genetic evaluation was normal
Our beloved Hugo was born


I felt Hugo having strange movements during pregnancy
Six hours after delivery
it was confirmed - Hugo got seizures
When Hugo was 4 four months old he got
the same stomach problems as Wilhelm

Still every test were normal

Some comments from doctors:



Emma

Once again I felt the same strange movements in the last trimester

She got confirmed seizures
30 minutes old

Tried to live a normal life

We concentrated on living and searched for diagnosis when they slept

Phenotypes

Wilhelm

- Epilepsy
- Special formula feeding
- Sensitive to infections

Hugo

- Epilepsy
- Special formula feeding
- Atypical autism
- Mental retardation
- Emma and Hugo had their own language that the closest understood
- Sensitive to infections

Emma

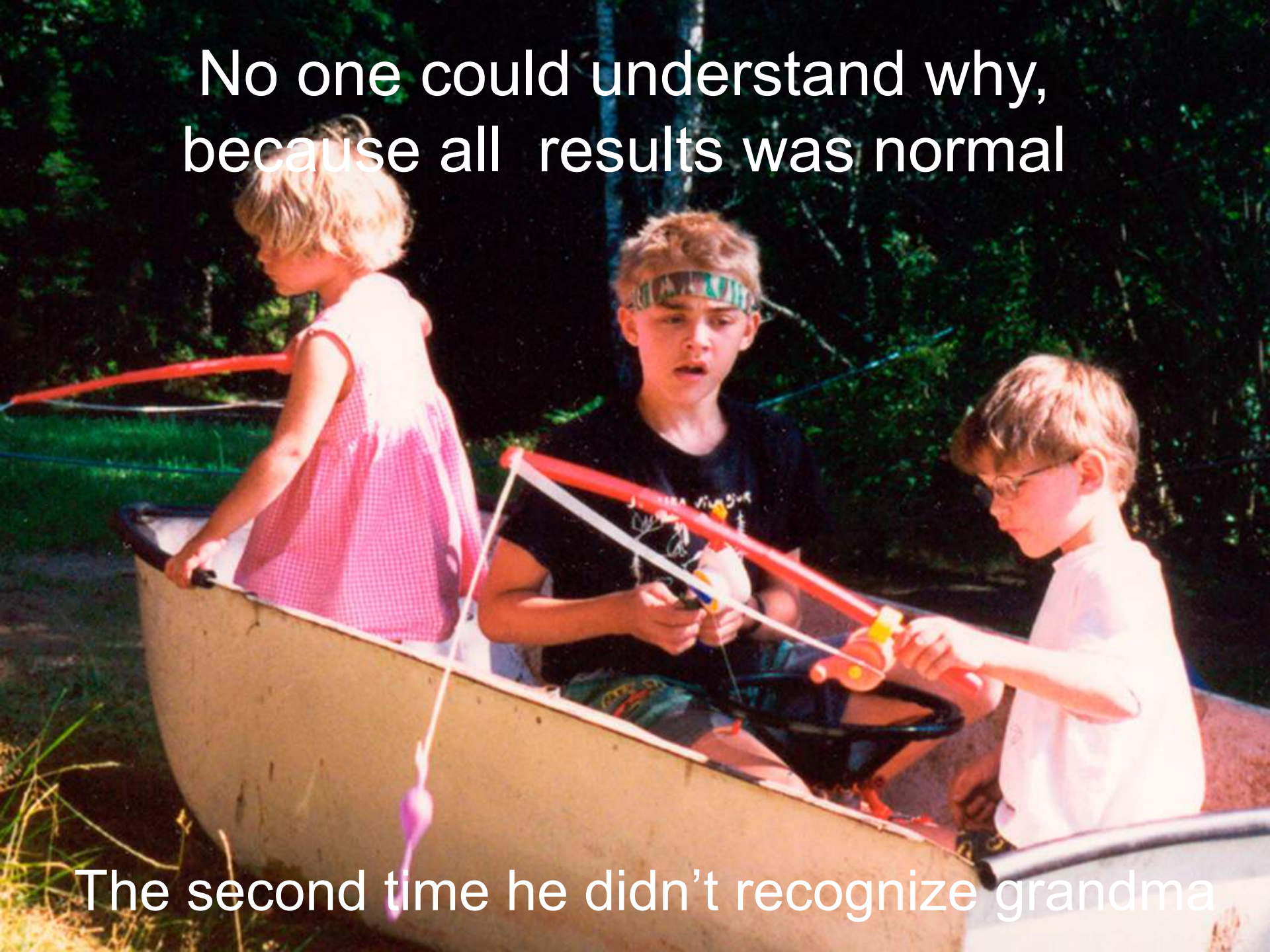
- Epilepsy
- Special formula feeding
- Apnéa
- Atypical autism
- Emma and Hugo had their own language that the closest understood
- Sensitive to infections





At 12 years old Wilhelm changed

- He couldn’t ride a bike anymore
- He run into 1 meter high stinging nettles
- Impossible to understand the home work
- Lost skills
- Didn’t want to play with his friends
- Didn’t want to read books
- Prefer to play with the his friends younger siblings
- Played with Hugo och Emma and their toys
- Want us to read books for younger children


No one could understand why,
because all results was normal
The second time he didn’t recognize grandma


Wilhelm was diagnosed with dementia
Carpe diem
We tried to do the best of the situation

In search for a diagnosis

One more visit to specialists this time to Great Ormond Street Hospital, London

Once again all the test results were normal and no diagnosis

Next stop Johns Hopkins Hospital USA

After 2 weeks at Johns Hopkins Hospital USA

Home again, without a diagnosis!

I said to my husband Mikk that we have to do something!

"We must try to collect the specialists!"

"And try to get them to talk to each other!"

"And collaborate!"

A world congress!

Our dream began…


Wilhelm passed away, 16 years old

Just before Wilhelm's funeral we funded Wilhelm Foundation to collect money

We won’t be bitter – we have to do something

Undiagnosed - consequences

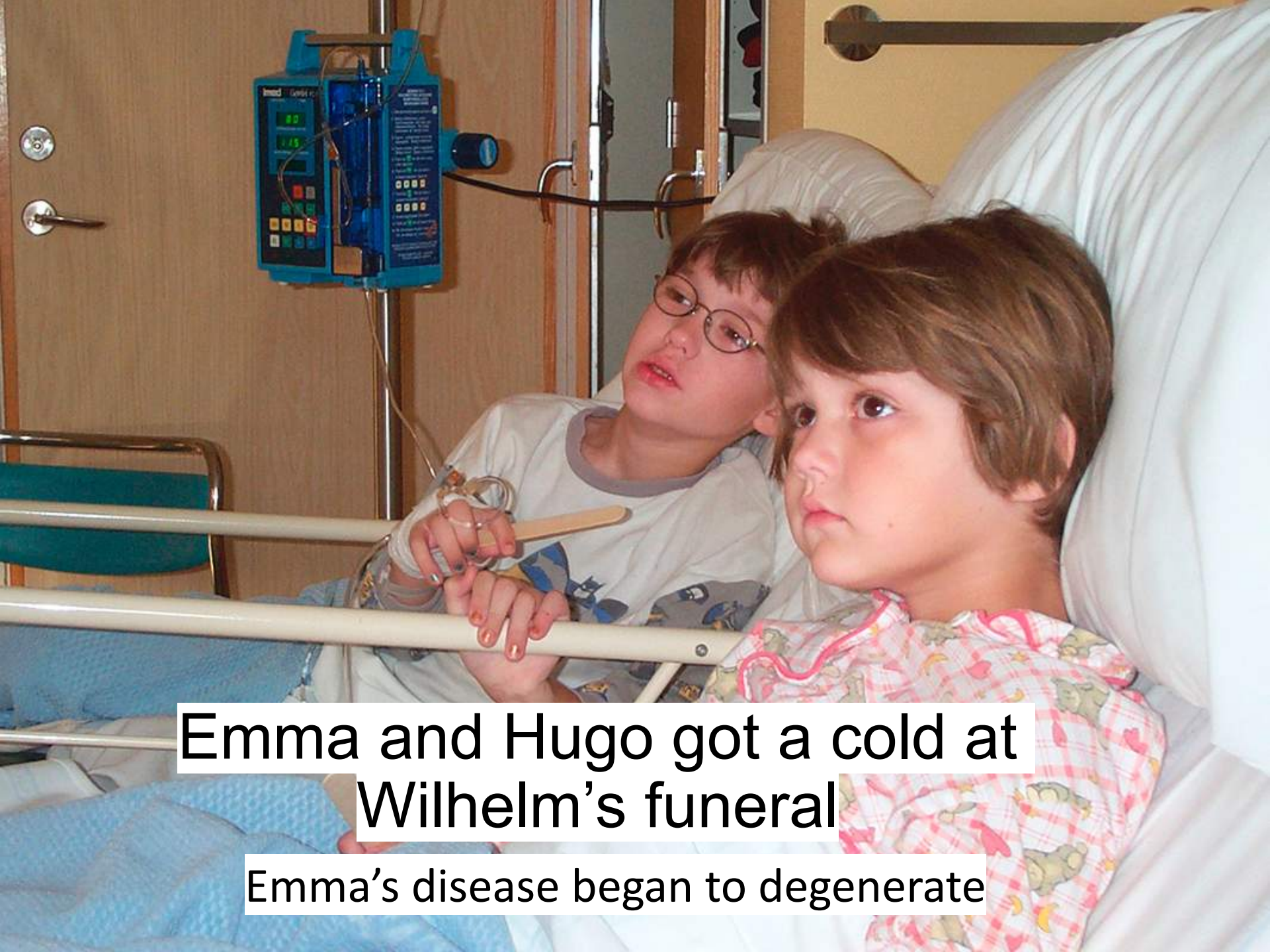
- No treatment
- Misunderstood
- Difficulties for the family and the siblings
- Limited or no help from the society
- No tests
- No prognosis
- Dies much to young

How to help the families

- World congress – we wanted to collect famous specialists around the world to make them cooperate
- Support
- Raise awareness






Emma and Hugo got a cold at Wilhelm's funeral
Emma's disease began to degenerate


Emma had frequent periods of coma


Wilhelm’s home nursing team told us; It was too difficult to be in our home once again – they loved our children too much

Carpe diem

The following year Emma's periods of coma got longer and longer

One year after Wilhelm past away
Emma died only six years old

Hugo alone…

"What should we do?"

Hugo got bad

Whitout a diagnosis – no prognosis

– no wheel chair

Two years later
Hugo passed away
at an age of 10

In three years we lost our three beloved children in an Undiagnosed disease

Wilhelm Foundation

For all the children with an undiagnosed diseases and their families

Goal:

All the Undiagnosed diseases should be solved

How?

International Conferences - World congresses

Difficulties without a diagnosis?

Medical

- There is no treatment
- Lack of understanding
- No prognosis
- No one knows if it's an genetic disease

Psychological

- Can be difficult to comprehend for the child's surroundings, family, and school.
- Parents asked them themself ”Did I do something wrong?”
- ”Is it my fault?”

Rights

- Can be difficult to get a aproperty help
- Difficult to receive the help that it should from society.
- Refferals to other doctors
- Second opinion
- Special dental care
- Assistive equipments

2013 - outside of Sweden

Our first meeting with Dr Gahl NIH USA

We told him about our dream - a world congress

Dr Gahl said YES! Dr Groft also said YES!

They wanted to collaborate with us

First International Congress 2014 Rome

Thanks to Dr Gahl, Dr Groft and Dr Taruscio

Undiagnosed Diseases Network International was formed at the first and second conference

www.udninternational.org


The laste conference was in Sweden.
This is the Secret room at Nobel Forum, where the Nobel Committee decide who gets the Nobel Prize for Physiology or Medicine
The Fifth Conference was a collaboration between:
Dr. Gahl NIH USA, Professor Nordgren, Professor Wedell chair of the Nobel Committee for Physiology or Medicine, and Wilhelm Foundation

Finally a Swedish UDP - Karolinska UDP

Prof. Ann Nordgren
Karolinska UDP

Prof. Anna Wedell
Karolinska sjukhuset

At the congress in Stockholm
The Undiagnosed

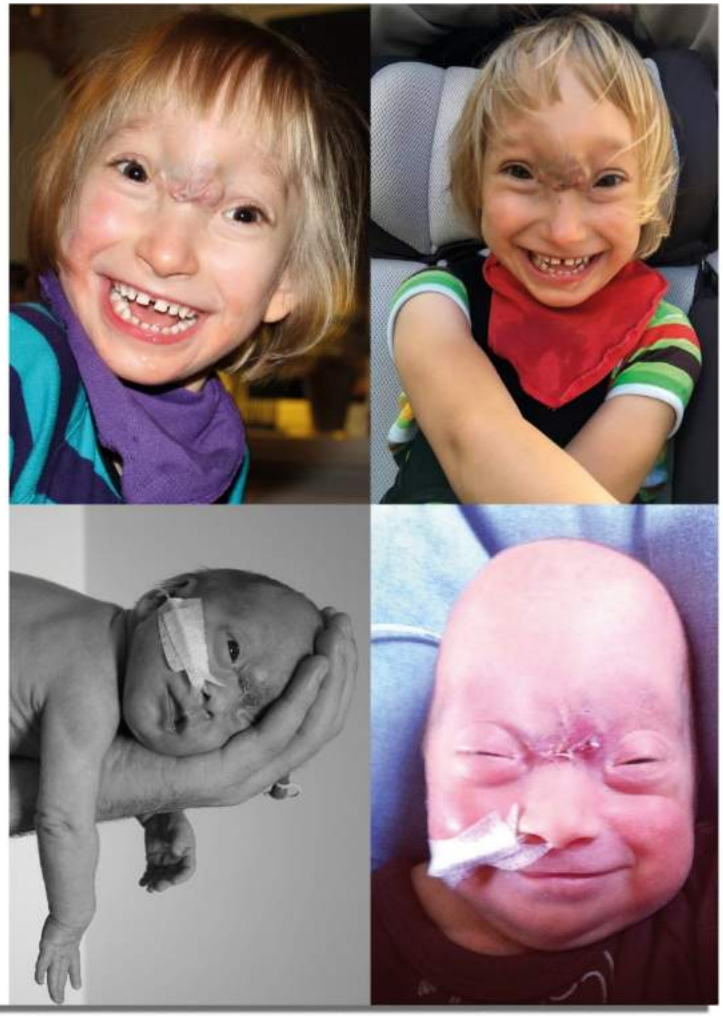
A photo exhibition and booklet with
Undiagnosed diseases

Freya
one undiagnosed child

The Undiagnosed

Children in Sweden

Name	Freya
Year	2011
Description	Freya is a six year old with lots and lots of energy, she's curious, stubborn and she seems pretty content with life. A very happy girl that is an ivf and donor (sperm) baby. On the routine ultrasound they noticed Freya has corpus callosum agnesis and her head was 20% smaller than normal. Nothing showed on the amniocentesis. Freya was born 8 weeks early, my water broke and the doctors couldn't stop the labour from starting. When she was born we found out she had a encephalocele above her nose. She had surgery on that the next morning. Freya needs help with all everyday things: getting dressed, feeding (eats mashed food), wears nappies (started toilet training), can walk with help (holding someones hand or in her walker), just started crawling a little, very movable on the floor, can pull herself up to standing. Freya has foot orthoses to keep her feet in the right position. Her left side is weaker and harder for her to control because of CP. She's got bad eyesight especially on her left eye, where she also squints, and wears glasses. But she seems to be able to grab things within her reach very well without her glasses anyway. Her eyes sometimes "jumps" when she tries to focus on something. Her hearing is more than good, hears everything and she loves music. A psychiatrist has done an evaluation on Freyas general development level and it's result was about 17 months old. This and that she seems to have a high pain threshold means we have to watch her all the time so she doesn't hurts herself playing. She also have a thing for pulling her hair and like everything else she puts it in her mouth. She drools a great deal. Freya doesn't pull her hand or toys out of her mouth when she gags which makes her throw up if you don't stop her in time. She also bites if you're not careful, can bite herself as well. The evaluation the psychiatrist did also showed that she has ADHD and a very short span of concentration. Freya doesn't speak but makes a lot of noises, high squeals and sounds. We can hear her say "mama" and "papa". She's has problems sleeping at night but it has got better with help of medicine. Doctors think it's her epilepsy that disturbs her sleep, she got her diagnose at 9 months of age. Her medicine keeps it in control but gets the odd seizures for which if needed we give her diazepam. Usually it's if she's coming down with a cold, slept bad or if her stomach isn't working as it should.
Symptoms / Signs	Corpus callosum agnesi, Chiari malformation type 1,Encephalocele, Microcephaly, Severe developmental disorder, Epilepsy, Cerebral palsy, ADHD, Bad eyesight, No speech
Current Treatments	For her epilepsy Keppra and Ergenyl Retard. Diazepam if needed to stop seizures. Melatonin and Frisium to sleep. Botox in both calves and inside thighs for her spasms every 4-5 months.
Prior Treatments	Ergenyl fluid, Both legs in casts knee high, twice (last year and this year) for 4 weeks after a botox treatment.
Previously Considered Diagnoses	Morning glory syndrome, Joubert syndrome
Contact	Helene Cederroth, helene@wilhelmfoundation.org



The Sixth International Conference on Undiagnosed Diseases, Naples Italy in June 20-21

Thank you for listening!

Helene Cederroth

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www. wilhelmfoundation.org

EURORDIS Black Pearl Volunteer Award 2018