The Undiagnosed

Helene Cederroth

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 scenario a late diagnosis could lead to irreversible consequences and even be life-threatening

*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° C) and difficult to breathe

Back to the hospital

No one could explain why it happened

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.
If you get a girl she will be healthy

Still every test were normal

It’s not a degenerative disease

It’s bad luck that the boys are sick

Wilhelm is like an unripe apple

Wilhelm and Hugo have different diseases

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 property 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 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.

The last 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.
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

Freya is a one-year-old child with lots and lots of energy, she cannot stop being active and she seems to enjoy pretty much life. A very happy girl that is on her own and loves dancing (openly) baby. On the other hand, they noticed Freya has some difficulties with co-ordination and her head was 30% smaller than normal. Walking around the playground, Freya was born 8 weeks early, my water broke and the doctors couldnt stop the labour from starting. When she was born we found out she had a cephalohematoma above her nose. She had surgery on the following morning, Freya needs help with all everyday things getting dressed, feeding (not that much food), using the toilet, and she needs help to tailor the right size of the clothes. We cant pull her up to help, Freya has foot orthotics to keep her feet (right position). She is fragile and hard to control because of CF that gets back right especially on her left side where she also suffers, and wears glasses. She seems to be able to grab things within her reach very well without her glasses anyway. Her eyes sometimes "jump" when she tries to focus on something. Her hearing is more than good, her vision is good and she loves music.

A psychiatrist has done an evaluation on Freya general development level and it’s much was about 17 months old. This can be that she seems to have a high pain threshold means we have to watch her all the time on she doesn’t hurt herself playing. She also has a thing for pulling her hair and if she needs help she puts it in her mouth. She has a great deal, Freya doesn’t feel her hand or leg or out of her mouth when she writes, Freya doesn’t feel her hand or leg or out of her mouth when she writes, Freya doesn’t feel her hand or leg or out of her mouth when she writes.

The evaluation the psychiatrist did also showed that she has ADHD and a very short span of concentration. Freya doesn’t speak yet comes a lot of noises, laugh sounds and words. We can hear her say “mama” and “papa” when she has problems sleeping at night but it has got better, with help of medicine. Doctors think it’s her curiosity that disturbs her sleep. She got her diagnosis at around 4 months old. Her medicine helps it control but gets the cold symptoms for which we need to give her asprin. Usually it’s coming down with a cold, sleep better or her stomach isn’t working as it should.

Symptoms / Signs
Canada-colonism agent, Clinch median-type.李先生, Other symptoms, ADHD, very short PSP, Sleep.

Current Treatments
Freya is on the waiting list for a medical procedure that is needed to stop sleeping. She is on Enfamil and Enfamil in to adult formula for her meals every 4-6 months.

Prior Treatments
Enfamil milk, both in 1st year, twice, last year and this year.

Previously Considered Diagnoses
Enfamil milk, twice in 1st year, twice last year and this year.

Contact
Helene Cedersund, helene@thefoundation.org

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<th>Name</th>
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Symptoms / Signs
Canada-colonism agent, Clinch median-type, E. histolytica, Severe developmental disorder, Enfamil, central sleep, ADHD, very short PSP, Sleep.

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The Sixth International Conference on Undiagnosed Diseases, Naples Italy in June 20-21
Thank you for listening!

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www.wilhelmfoundation.org

EURORDIS Black Pearl Volunteer Award 2018