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The Story of Our Life with Eleonora
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In May 2018, a little over a year ago, Emma and I were here in NY with our daughter Eleonora to meet Dr. Wendy Chung and her team. We wanted Dr. Chung and her team to meet Eleonora in person, we wanted to contribute experiences and knowledge to the research that is going on if possible.

We were warmly welcomed and it was a very rewarding meeting - for the first time since Eleonora received the KIF1A diagnosis, healthcare professionals showed genuine and sincere interest in her as a person and how the diagnosis affected her. We were seen, we were confirmed. We were met with care, curiosity and interest - we were offered help and support.

Although Eleonora had a world-unique diagnosis, she was actually the only known with her specific KIF1A mutation, Swedish health care showed no major interest in the diagnosis and Eleonora. It was surprising, frustrating and sad.

In order to give Eleonora the attention she deserved in this context, we had to travel 6200 km of bird path - from Sweden to the United States.

We demanded nothing big, we understood that our meeting with the researchers would not lead to Eleonora's recovery and it would not save her from an early death - we just wished Eleonora to be seen for the little unique individual she was and is.

Thank you Wendy, Lia, Joanne and to the others in the team for giving us what we wanted!

Our trip to NY last year required extensive preparation, logistics and large costs - traveling with a child as seriously ill as Eleonora is a huge arrangement to implement. The trip and stay in NY were filled with stress and worry - we were way beyond our comfort zone. We are happy and proud that we completed the trip, we are grateful that we were given the opportunity but at the same time we note that we would never do the same trip again. We achieved our goal with the trip - to show ourselves and the surroundings that nothing is impossible!

Now we are here again, we have traveled from Sweden to NY - this time without Eleonora. Not because we chose to travel without her, because she is no longer with us.

We are here to support the KIF1A community with our presence, we are here to honor our beloved daughter and to pay tribute to her memory.

I am Eleonora's dad. Eleonora was born on October 18th, 2015 and passed on February 14th, 2019 - Valentine's Day. Eleonora had KIF1A or KAND, as it is also referred to.

It is a great honor to stand here in front of you all and to be able to speak to you today. Emma and I have traveled here for the purpose of participating in this conference and we would like to thank the board of KIF1A.ORG, who has made it possible for us to be here.

"To sit in the waiting room of death with a blank ticket number"… that is how I sometimes describe the situation of being a parent to a seriously ill child. We knew all along that death would come prematurely but we did not know when.

Our child's diagnosis was neurodegenerative and progressive, it was extremely rare. We learned that there wasn´t any cure – nothing could be done for the actual diagnosis, only treatment for the symptoms and complications arising from it.

As the diagnosis was so rare, even the slightest glimmer of hope felt like it was out of reach – there was limited knowledge, limited research, and next to no financial means. We received an honest but disheartening message – there were no financial means to do research about a diagnosis that affects such a small minority of the population. A small group of seriously ill people, consisting exclusively of children and youths, were not going to get help, purely because of financial constraints. This message is extremely difficult to accept in the role of a parent. My child was going to die and no one was actively going to try and do anything to prevent it.

There was nothing else to do but to wait for the inevitable end, to say the least, a different existence and reality. To experience the joy of becoming a parent but at the same time realize the fact, that the time together with our child would be exceptionally limited. This felt unreal, sad, frustrating, stressful and lonely. It was us against death – we were confined to that waiting room against our will, and once there, we wished for nothing more than that it would never become our turn.

It was very welcome news when we learned that research about KIF1A was about to start. There was no question that we would do everything in our power to help and to use our influence where possible. We were aware of the fact that research would not save Eleonora´s life, we were beyond that hope. It would have been a race

against time, time we did not have. But KIF1A was not only about Eleonora, we wanted and still want to be part of the work towards giving hope to other children and families.

When parenthood turns into everything you never imagined – how do you handle such an overwhelming existence? Where do you find strength? Where do you find joy? Where do you find hope?

This is my story…

The day Eleonora arrived, will forever remain the happiest moment of our lives. The day she passed is the toughest we have ever lived through, however, that day will forever have a special meaning for us. Valentine's day… the day we celebrate love, well suited for our little Princess.

Eleonora reached the age of three years and four months.

About two days after the delivery, the doctors grew suspicious that Eleonora had ongoing seizures. We were about to go home. All that remained was the final routine health check before discharge. But rather than being discharged, we were rushed to the neonatal intensive care unit. Eleonora had strange movement, she appeared very still and tired, she was muscle hypotonic. Despite

comprehensive examinations and tests, no explanation to her condition was found. After two weeks stay in the hospital, we were allowed home with the promise of continuous follow up.

During the next five months we frequently visited a paediatric neurologist, on each occasion we had no answers. We could all see that Eleonora did not meet any of the expected milestones - she did not make any progress in her motor skills development. Eleonora appeared to be completely uninterested in her surroundings – there was no eye contact, she did not respond with a smile and her movements were very constrained.

Eleonora's condition was discussed and various explanations were presented – the paediatric neurologist was shifting between "immature movement patterns", "delayed development" and "likely severe brain damage". We were thrown between hope and despair. Was Eleonora going to recover and catch up in her development? Was she going to have a disability? Would she face a life full of big challenges and difficulties ahead?

At that point we did not even suspect just how awful it actually was…

At home we tried to live as normal as possible – we had a beloved daughter and we were together as a little family. We had our first

child and were overjoyed at being parents, however, the happiness was at the same time stained by fear and uncertainty.

Emma has no background or education within healthcare but she immersed herself and then filled herself with knowledge of rare diagnoses – she was determined to find an answer. I myself have experience within healthcare and have, to a certain extent, some education within that area but I did not want to, or rather did not dare to, immerse myself as Emma did. I realized it was pointing in the direction of something very negative and very disturbing. I just completely avoided reading some things.

Emma studied, read and debated. She described and compared different rare diagnosis – it scared me, it stressed me. Each diagnosis was worse than the other – I did not want to hear, I did not want to lose hope.

During spring 2016, when Eleonora was a little more than 5 months old, we attended emergency care. Eleonora showed signs of epilepsy. EEG showed a chaotic brain activity, with the whole brain involved. Eleonora turned out to have a severe type of epilepsy and was in a fatal condition.

The suspicion of severe brain damage was confirmed. It felt like the earth had opened beneath our feet and our dreams of a “normal” parenthood were brutally crushed.

This was the time when frequent and sometimes long lasting stays in hospital began. A genetic screening and evaluation were initiated. Blood and tissue samples were taken, both from Eleonora and from us, the parents. A genetic aberration was suspected to be the cause of Eleonora´s difficulties.

About three months later, we received the results of the test – Eleonora had a mutation on the KIF1A gene. The specific mutation had never before been registered in another individual and its placement would have devastating consequences. At this time, in 2016, the number of known cases with an aberration on KIF1A was approximately 50-100 people.

The physician explained that Eleonora would have complex disabilities and her life was expected to be short. The biggest threat to her life would be airway related complications such as pneumonia.

Shortly after Eleonora`s KIF1A diagnosis she got another additional diagnosis – PEHO syndrome. PEHO stands for Progressive encephalopathy with Edema, Hypsarrhythmia and Optic Atrophy. Simply put - this is a progressive brain disease with peripheral fluid accumulation, a very unusual and severe form of epilepsy and an

optic nerve that thins away. In addition to these symptoms, PEHO syndrome is characterized by atrophy of the cerebellum and the brainstem.

PEHO syndrome is often associated with mutations on another specific gene but in some research reports it is connected to KIF1A. The syndrome is a rare neurodegenerating illness, belonging to the family of infantile progressive encephalopathies.

As mentioned before, Eleonora did not reach any milestones in her development. Her psychomotor skills could be compared to a newborn baby and some of her physical functions deteriorated with time or just stopped functioning completely – a progressive pattern.

Initially the epilepsy was Eleonora's biggest difficulty. She had a few episodes of infantile spasm, or as it also called, hypsarrhythmia - a very rare, serious and tremendously difficult to treat form of epilepsy. During her whole life, Eleonora suffered from frequent and generalized seizures - the seizures involved the entire brain. In the beginning the epilepsy was the most prominent and dominating sign that Eleonora suffered from severe brain damage. We knew that every single seizure was potentially fatal and that scared us. The epilepsy was considered as resistant, however, it could be considerably controlled with medicines. After a while we learned

how to deal with the seizures and they no longer seemed as frightening as they did at the start.

As time passed, the respiratory problems would become the most prominent and dominant condition to deal with. Eleonora had enormous problems with stagnation of mucus in her airways and lungs. The chest muscles were weak and she sustained from frequent and long apneas, she often suffered with difficulties in breathing. During her life, Eleonora experienced ten episodes of pneumonia, of which the last one, indirectly caused her death.

The severe muscle hypotonia in combination with atrophy on the brainstem, where the brain respiratory center is located (latin: truncus encephali), caused major breathing problems. The muscles surrounding the breathing mechanisms were weak and the signals and impulses from the brainstem did not properly work. Eleonora had longer and more frequent apneas – the oxygenating was in imbalance and became a regular demand on the respiratory- and circulatory system.

We spent a lot of time on physiotherapy with a focus on respiratory care. Each morning and as needed, we did a program that lasted for approximately 90 minutes – inhalations consisting of medication for widening the airways, anti-inflammatory and sodium chloride of different strengths. The positioning and physiotherapy mobilized the

mucus and eased removal of mucus from the upper airway. The removal of mucus was aided by a vacuum suction unit. This treatment was very demanding for Eleonora but vital – It assisted her breathing and prolonged her life.

We actively tried to affect her life in a positive way through our determination and increasing knowledge. We were extremely dedicated to our task when it came to the survival of our beloved child. We developed our own methods for Eleonora's respiratory care – we were inventive and found effective solutions and methods. At all times, of course, we consulted with the healthcare professionals.

Eleonora was completely muscle hypotonic, she did not have the ability to perform any movement by will. She could not hold her head, nor body, upright by herself. Her body was like a rag doll.

Eleonora also suffered from reflux and sometimes vomiting, caused by the weak muscles around the stomach entrance - that progressively deteriorated.

Eleonora could initially eat, she was nursed and bottle fed for a while but then she had difficulty swallowing food and fluid - dysphagia. Food and liquid consumption by mouth became something that could be a danger to her life. A G-tube became essential and Eleonora was continuously fed with food, liquid and medication through this tube.

The dysphagia, difficulty swallowing due to neuromuscular disorder, led to serious risk of suffocation by her own saliva, reflux or vomit. There was also a risk that the reflux or vomit would end up in her lungs (aspiration), resulting in chemical pneumonia.

Eleonora had impaired vision however, she reacted very positively to strong colours.

Eleonora was a very quiet child – she never cried, she had no speech and she was very rarely upset. Occasionally we saw her smile but never laugh. In spite of this - she developed her own way to express herself through sounds, mimicking and body language. For us it was very clear when she was unhappy or when she was satisfied.

Eleonora had extensive disabilities with enormous difficulties and challenges but she rarely complained. She was satisfied as long as she had love and companionship.

Our main common factor is KIF1A or KAND, but our different challenges still differ. For example; depending on which country we live in, there is a significant difference in the opportunities to get help and funding.

There are major differences in how the diagnosis affects the individual and we find that Eleonora, along with a few others, was one of the absolute worst affected.

Because of these differences, our experience is that it was often difficult to discuss and highlight the individual challenges in a larger forum. What is seen as a problem in Sweden, may not be seen as a problem in another country - for example, the United States.

I mention the following in order to illustrate the situation of a seriously ill child with extensive disability from a Swedish perspective.

In Sweden, the healthcare system is in general very good and of a high standard. The healthcare is run by the government and is available to everyone. The physicians are competent and practice well researched medical knowledge.

Eleonora was connected to a wide range of experts within various different fields, neurology included. She was always well looked after and cared for in emergency situations but only a handful healthcare professionals had knowledge of her diagnosis and special needs. When we went for care, often we were the ones who had to tell and instruct the healthcare professionals – we were her ultimate lifeline, we were always there for her.

In Sweden we also have a well-established social security system. Children and youth have free healthcare, free medications and

physical aids as needed - all paid for by taxes. For some disabilities the individual can have a personal assistant (comparable to a nurse or caregiver) and there are many ways that offer a good standard of living with very fair terms and conditions for the individual. However, the application process is complicated and lengthy.

When Eleonora was around two years of age and after more than a fair amount of struggle, she got personal assistance. This extra help made it possible for Emma and myself to focus more on parenthood. The responsibility for the very complicated care, that was required for Eleonora, could at times be placed on someone else other than ourselves. This support gave us well needed breaks that increased our resilience, endurance and also our ability to be able to handle our life situation with a very ill child.

In this company, I am quite sure there is no need to say how much is required of a parent to a child with special needs. I am sure we all agree it is a very demanding existence indeed.

This is how it was for us – our entire life was painted by Eleonora's diagnosis – we struggled everyday 24/7 for Eleonora to live, for her to survive.

We absolutely hated what the illness did to Eleonora. We hated what the illness did to our life as a family.

We grieved that Eleonora was deprived of things that would otherwise be normal for a child to experience. We grieved that we were deprived of things that would otherwise be normal and joyful to experience.

We never got to see her crawl, pull herself up, or walk. We never heard her pronouncing her first words. Never saw her dance. We never experienced her first day in school, never saw her fall in love. Never saw her growing up to be a young woman.

But… we were parents, we had a child – we loved and love Eleonora and we were prepared to do anything for her. We tried to appreciate what we had, not grieve for what we did not have.

For me it took a long time to do just that, to appreciate what I had.

I had a hard time bonding with my daughter, I had to learn about a fatherhood I did not know – a fatherhood that was different from the norm. Each day I came closer to Eleonora, every day my love for her grew – she taught me to be a father on her conditions.

We tried to live in the present without thinking about what was sooner or later to come – Eleonora's death. We knew, but we chose not to touch on the subject. If we had lived our lives thinking about death and what could be around the corner every single day, we

would most probably have broken down and given up hope. We wanted to appreciate every single day together with Eleonora.

During 2018, Eleonora's breathing deteriorated. The apneas incidents increased in number and lasted for longer. She suffered from mucus building up, that resulted in an inability to breathe. The reason for the deteriorating condition was an increasing weakness of her chest muscles along with a degeneration of the brainstem.

Eleonora's condition made her eligible for palliative care and a special care plan was established. In agreement with physicians, we decided that no resuscitation was to be carried out if Eleonora came into a life threatening condition – no CPR was to be performed and no respiratory aid would be given.

Through this palliative care plan, Eleonora would receive the appropriate care right up to the end - giving her optimum comfort. Eleonora would not suffer!

Eleonora had around six months left to live - something we were not aware of at this point in time. She was not immediately dying, but was in the "early palliative stage".

Late at night, on February 6th this year, Eleonora suffered from a mucus clot - which caused acute breathing difficulties. She was admitted to hospital. Eleonora suffered from pneumonia, possibly caused by a pulmonary aspiration.

Initially the condition was treated successfully with antibiotics, but as she in addition contracted influenza, her condition worsened. During the course of the week, several acute situations arose. Eleonora had major difficulty oxygenating herself, she had very shallow breathing and her general condition deteriorated.

Wednesday, February 13th, a week after she was admitted to hospital, her condition was extremely serious and late at night she was in a critical condition. Eleonora's breathing was very shallow and she had increasingly high levels of carbon dioxide, her body could not deal with it by itself. The only thing that possibly would help, was to intubate and use respiratory aid. But this, we had previously decided against. Eleonora would not suffer!

The physician explained that she had exhausted all her medical knowledge and she had no more alternatives left – there was nothing more at this point that could be done to save Eleonora's life.

It is a devastating decision to face, it is inhumane – we decided to let Eleonora pass. The decision was without comparison the worst we

have ever faced but then and there it was obvious – Eleonora was not to suffer, she was just to receive love and tenderness.

Once the decision was made, February 13th had turned into the February 14th – Valentine's day.

Fifteen hours later, at exactly 4 PM/16.00, Eleonora took her last breath and her heart stopped beating.

The cause of death was ARDS – Acute respiratory distress syndrome. In other words, a respiratory and circulation collapse.

Eleonora´s last moment in life she spent embraced by Emma and myself – we gave her love and a feeling of safety, by not being alone.

When Eleonora died, a piece of me died with her. Eleonora did not suffer during her last 15 hours – her journey towards death was calm and silent, but to follow her fighting for breath and see her weakened breathing was filled with painful agony.

It was surreal and definitely cruel – our little girl left and we would never ever see her alive again.

To live with a seriously ill child, whose time is limited, is a very psychologically straining existence. 24/7 of making sure that your

child will have the opportunity to survive and live. We struggled for 3 years and 4 months for our child. The pressure was continuously high and the respite for this was very rare.

Life with Eleonora was a paradox. It was a life of many parts that I would not wish upon anyone else but it was also a life of many parts that I would not want to be without. It was incredibly beautiful. We lived hand in hand with sorrow and joy, guided by the love Eleonora gave us.

When we received Eleonora's diagnosis the very fabric of our existence was changed, however we learnt or rather were forced to learn how to live with this difficulty, we learnt how to appreciate the small things. When Eleonora passed, we did not only lose our child - we also lost our direction in life. Yet again the fabric of our existence was changed. Every single day we struggle to find happiness again.

Just a couple of days ago it was the 6 months anniversary of Eleonora's passing. The grief and sorrow is still difficult and agonizing.

Eleonora has made an enormous imprint, not only on us as parents, but also on people she met in her life and on people around the world – people who never met her. She taught people some important things – she gave love, joy and hope.

Eleonora showed the spark of life, strength and patience. She handled huge challenges and stress without complaint. People who met Eleonora used to say that she had a special aura and charisma of calm and serenity. She spread knowledge and love. She made us wiser, more humble and more grateful.

Eleonora became our own Superhero.

In Sweden Eleonora has become a figure head and a symbol for seriously ill children and people with disabilities. She created public opinion and politically influenced issues related to disability rights. She contributed to changing and improved better living conditions for other children with disabilities. This makes us enormously proud.

Malmoe, the city where we live, is the 3rd biggest city in Sweden. Eleonora will give her name to a prize that is to be awarded by the municipality on December 3rd – the United Nation 'International day of disabled persons'. The prize will be awarded to someone who has made an important difference to people with disabilities – for equality, self-worth, freedom and life quality.

It is incredibly honorable and beautiful, that Eleonora's name and memory can live on in this way.

The littlest feet make the biggest footprint! We will never forget Eleonora and the world will never forget her.

Thank you for your attention and thank you for letting me share our story with you.