Rett Syndrome

RTT

Nordic Conference on Rare Diseases

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- This is a picture of our girl a few weeks after she was born. We always knew she was an angel, but we never expected her to be a very special Silent Rett Angel.
The beginning ...

• We are parents of three children, 7, 5 and a newborn
  • Our girl is 5 years old and she has RTT
• She was referred to the SDCC in the fall of 2009 and was diagnosed with typical autism in June 2010
• SDCC also requested a DNA test for Fragile X and RTT,
  • Results came from a lab in Canada in April 2011 with confirmation of RTT (MECP2/c880)
• These were not the results that we were hoping for
• It was a huge blow for us,
  • Our girl has a rare disease with currently no cure!
    • but at the same time it’s a relief to know were you stand, because then you know what path you have to go ...
What is RTT?

- Rett syndrome, RTT, is a complex neurological disorder.
- It affects almost only girls.
- People with RTT are profoundly and multiply disabled and totally dependent on others for their needs throughout their lives.
- RTT affects about 1 out of 10,000 to 15,000 female births.
- Most cases of RTT are caused by a mutation in the MECP2 gene, which is located on the X chromosome.
- RTT is characterized by normal development during the first few months of life, followed by a loss (regression) of language and motor skills.
- RTT can resemble autism, cerebral palsy, epilepsy or developmental delay.
What to expect ...

• RTT is a progressive disease, which means that the symptoms will likely get worse over time.
• The speed at which a RTT progresses in a given child and the severity of symptoms she experiences varies from child to child.
• They may experience improvement in their behavioral symptoms between ages of 2 and 10. Many girls with RTT remain in this “plateau stage” for the rest of their lives.
  • Communication and motor skills usually don’t decline past this point.
• On average, most girls survive into their 40s or 50s.
Is there a cure?

• We knew nothing about RTT in April 2011, but then my wife began to Google,
  • It is not an option to quit!
• Researchers know that a mutation in the MECP2 gene prevents the MeCP2 protein from doing its job, and disrupts the normal functioning of nerve cells.
  • Adrian’s Bird mouse model revelation in 2007
• She found a clinical trial with a drug called IGF-1 which is seeking to reverse the RTT symptoms at the Boston Children’s Hospital
  • [http://www.childrenshospital.org/clinicalservices/Site1886/mainpageS1886P0.html](http://www.childrenshospital.org/clinicalservices/Site1886/mainpageS1886P0.html)
  • We enrolled our girl in June 2011, in the hope that she will be selected to participate in the study (Phase II)
• This gave us hope to carry on
RTT and the Icelandic system

- RTT is a rare disease. Little or no experience is at hand in the Icelandic health system to treat RTT. There is also a limited knowledge in the system about RTT to advise appropriate medical treatment and counsel.
- This is quite understandable, RTT is a rare disease and it is unreasonable to expect that Icelandic doctors and other specialists are capable in this small nation to be specialists in rare diseases and keep their knowledge competitive and up to date with recent research in the field.
- In our daughter’s case, her “team” is comprised of very good people with broad expertise and knowledge, willing and doing their best for our daughter, but that is unfortunately not enough.
  - We need, like all who work with children to know for sure if we are doing all that we can for our daughter.
Limitations

- Our „team“ could not convince us that the methods and treatment they discussed were the best one’s possible
  - This was a condition all of them knew about, i.e. our concern on their limitation on RTT
- We then decided to try to get a second opinion and a back up for our „team“
  - We wanted to go with our girl to the Rett Syndrome Programme at the Children’s Hospital in Boston to get a second opinion
- In our case, there was no „trigger“ in the healthcare system which went off to facilitate getting a second opinion and counseling on RTT from abroad
  - We had to have the initiative ourselves
Special committee

• We applied in August 2011 to the Special Committee on Hospitalization Abroad to cover costs for a medical consultation for our daughter
  • The substance of the law is that, if the insured person urgently requires hospitalization abroad because necessary assistance cannot be provided in an Icelandic hospital the expenses are covered by Icelandic Health Insurance
• Our doctor and the SCDD sent in their reviews concerning our application
  • They both supported our application and pointed out that it was necessary to ensure right and trustworthy medical treatment that she would be able to get a medical consultation from the RSP at the Children´s Hospital in Boston
  • This support was very important
• The Committee approved our application in October 2011, after gathering further information directly from the RSP in Boston and other questions, on the request of the Committee.
RSP, Children’s Hospital Boston

- We went with our girl to the Rett Syndrome Programme at the Children’s Hospital in Boston in December 2011.
- This visit met our every expectations, we got plenty of time with the doctor and she answered all our questions thoroughly
  - We prepared our visit carefully and sent before us a long list with questions
  - We asked all our members in the „team“ to come with their own questions so we could get them answered, many of the used the opportunity
- Generally speaking we could say that the counsel of the „team“ was in line with the recommendation from the RSP
  - We have changed some of our treatment methods and we got clear answers concerning medical issues of RTT, such as medication etc.
- Last, but not least we established a formal link between the professionals at RSP with our Icelandic counterparts.
  - This is invaluable to us to know of a strong research oriented and experienced people who are willing to give us and our people counsel and guidance in the years to come for our daughter.
... cure around the corner?

- The theory,
  - ... if we can find a way to reactivate the normal \textit{MECP2} gene on the inactive X chromosome, we may cure the disease 😊

- Jennie Lee, MD, PhD of Harvard University, is one of many funded investigators on RTT pursuing this line of inquiry:
  “There is the beautiful mouse model work of Adrian Bird that shows us that you can be born with this deficiency and be cured through gene therapy or reactivation of the normal copy of \textit{MECP2}. That is profound. How many congenital diseases can we say that about? Rett is one of those congenital genetic diseases for which a cure could actually happen.”
IRSF - RSRT

• International Rett Syndrome Foundation (IRSF) and Rett Syndrome Research Trust (RSRT) are the two largest RTT associations worldwide who finance research and give support to RTT communities.

• The story behind Rett Syndrome is complicated. It involves a devastating genetic affliction that starts with young girls and includes incredible family dynamics, groundbreaking treatment, care and science.
  
  • The film focuses on three Rett girls and their families, the RSRT efforts to find a cure and the optimism surrounding treatments and forward thinking scientific breakthroughs.
    
Outcome

• This link with RSP in Boston is invaluable to our daughter
• This process has convinced us that we are doing all that we can for her
• This work has made it possible for our „team“ to collaborate with their colleagues at RSP about RTT
• We´ve made our small contribution to making more information about RTT available in the Icelandic Health Care system

• The „system“ is often ready and able to help, but it needs the drive and initiative of parents and associations to make things move, because everybody is willing to do his part, they just need a little push
I thought I would have to teach my daughter about the world, it turns out I have to teach the world about my daughter.
They see a girl who cannot talk.
I see a miracle who doesn't need words.