DIAGNOSING RETT SYNDROME – MY EXPERIENCE

My name is Marisa Knott and I'm going to tell you about my experience as the mother of a young child recently diagnosed with Rett Syndrome.

My daughter's name is Isabella and she is almost 4 years old. Isabella was initially a very placid baby and she seemed to progress well in that she began speaking and clapping hands on demand from when she was just 3 months old, however by the time she was 6 months we noticed she was physically delayed. Isabella didn't sit up competently on her own until she was 12 months. She never crawled conventionally, instead she bottom shuffled to move around. She didn't bear weight on her legs until she was 13 months and then there was the constant hand mouthing that has been present throughout her life.

We'd had several appointments with paediatricians and health nurses who kept assuring us how it's most common for larger babies to be physically delayed.

Then, one day she wouldn't talk or repeat words as she had done every other day. At first I thought she was being stubborn but when weeks passed I became more concerned. Then, months later the aggression began, head banging, biting, pinching, pulling and screaming for no reason other than what appeared to be frustration. The hand mouthing became so bad she'd often bite through the skin on her wrists. To those who are familiar with Rett Syndrome, all this may sound familiar as being common behaviour of a child going through the first to second stage however, at the time no one could tell us what was happening.

When Isabella was 22 months, we took her to see another paediatrician. She was concerned Isabella may have had epilepsy due to the aggressive behaviour, so she arranged for an EEG to be performed. She also arranged for blood tests for a chromosomal screening, Prada-Willi / Angelmans and Fragile X Syndrome and also a sugar level and thyroid check.

A few weeks later, the paediatrician called me to say the results of the EEG indicated that Isabella was experiencing myoclonic seizures. I was told she needed anticonvulsants immediately. I asked her of the side effects and she said "drowsiness at first that will ease as Isabella gets used to the drug". She also told me she had arranged for us to see a paediatric neurologist. The next day we commenced Isabella on the drug Rivotril (clonazepam).

Unfortunately, Isabella was far from drowsy, in fact her head banging and aggression worsened. When we saw the neurologist, I asked him whether they were absolutely certain Isabella had epilepsy and asked him if that could be the reason for her delay. He said, "Isabella definitely has epilepsy but it's more likely she has something that's causing the epilepsy and developmental delay". I then told him that since commencing the anticonvulsants her aggression seemed to worsen. He said, as it had only been two weeks to keep increasing the dose by one drop per week until we saw results.

So, as advised we kept increasing Isabella's dosage and subsequently her behaviour and aggression became so difficult, her carers at the time could not cope so I was forced to temporarily stop working to care for her fulltime. At that point I became **determined to find out what was wrong with my daughter.**
I began reading about so many different disorders that cause developmental delay on the Internet. I read about the three disorders I knew she'd been tested for. None of these conditions really fit Isabella's profile and wondered why she'd been tested for them. So then I began entering words, such as, regression of speech, repetitive movements, hand mouthing and each time Rett Syndrome would appear on my screen. Isabella met all but one of the necessary criteria and other than the fact she was unusually tall and solid for her age, everything else seemed to fit. I recall after intensively reading the official website saying to my family**, "I think I know what's wrong with Isabella".**

I then called the paediatrician and asked if she could organise Isabella to have the MECP2 test. She agreed Isabella had characteristics similar to Rett Syndrome but also a range of many other conditions and I should be prepared that I may never know what was wrong.
I told her I'd read about some of the disorders she'd already been tested for and how Isabella didn't really fit the profile at all, yet the tests had been performed. Why? She said when a child has significant global developmental delay it is "routine" to test for these more common disorders for process of elimination. Routine? So even though she doesn't fit the profile these disorders are still to be eliminated, then why can't Rett Syndrome be eliminated? She said I should discuss it with the neurologist at my next visit and she'd also arrange for me to see a genetic consultant.

As Isabella's behaviour reached crisis point, my GP decided to contact the specialists as she was concerned it may have been the anticonvulsants causing it. The neurologist called me at home to say that excessive aggression is a common side effect of clonazepam. He told me to begin reducing the dose and at the next appointment we'd speak of introducing a new drug. As soon as I began reducing the dosage, Isabella actually began to show improvements. She seemed happier and I recall this time being when she began cruising the furniture. She also discovered a new word "more" and said it fairly often and always appropriately, however she has since lost all speech.

At the next appointment, the neurologist told me he wanted Isabella to undergo a video EEG, which may provide us with information on whether she perhaps had a type of epilepsy syndrome. I asked him if we could wait until after the video EEG before we introduced any other drugs to which he agreed. Then I told him I'd been reading about Rett Syndrome and how girls with the condition were often misdiagnosed with epilepsy, cerebral palsy or autism and how Isabella certainly fit the profile. He then replied with, I quote, "no she doesn't have Rett Syndrome, her head would be shrinking", he also said as the prognosis for some of the conditions we were discussing was not good, sometimes it's often better for the child if we don't know and I was "torturing" myself by browsing the internet but if I was that concerned I should wait to speak to the geneticist.

So, I took Isabella to see the geneticist. I asked him about Rett Syndrome and he said he didn't believe Isabella had it. He said her head circumference and **the fact Isabella had always been physically delayed didn't support a Rett Syndrome diagnosis**. **He said Rett's girls didn't only regress verbally but also physically**. He then said it would cost around $500.00 for a test should I be the one to request it. He told me to come back in a month during which he would have spoken to his colleagues about what to do next.

Isabella underwent her video EEG and when we next saw the neurologist he said he had good news, Isabella's reading was completely normal, no seizures at all and it seemed her head banging was most likely from frustration. In hindsight we now know Isabella was most likely also going through the period of regression when she began losing some of her skills and the clonazepam obviously enhanced her aggravation.

So on went my search for a diagnosis. When I next saw the geneticist, he told me he wanted to perform more tests for some rare metabolic disorders that can cause aggressive behaviour. I again asked him about Rett Syndrome and he again dismissed it by telling me he didn't think it would be worth my money for the MECP2 test. At that point he asked,
"How exactly is a diagnosis going to help you deal with Isabella? It's not going to change anything for her and you should just focus on obtaining assistance in helping you cope with her behaviour and disabilities instead."

It felt as though I kept hitting brick walls. No one was listening to me and the feeling of being ignored was so frustrating. The appointment ended with me conceding to have Isabella tested further for a range of other genetic and metabolic disorders and if all tests were negative he would reassess her in 12 months.

In the following months, I can recall Isabella squeezing and manipulating her fingers. She began dropping her cup and eventually couldn't pick it up as easily. Then she began punching her chin constantly so we had to splint her arms. When we next took her to the paediatrician for a follow up appointment, I again told her of my concerns about Rett Syndrome. She told me that perhaps I should see yet another paediatric neurologist that had experience in Rett Syndrome to "allay my fears".

I stayed awake each night, thinking about the possibility of my daughter having Rett Syndrome or worse, never knowing what was wrong at all. I wondered how many other parents were in my shoes and the thought of not knowing was something I personally could never deal with. We wanted to know as soon as possible, so I called the paediatrician and I must admit I was probably very aggressive when I demanded to know whether or not she herself could request the MECP2 test as we decided against waiting to see **yet another** specialist and we were fully aware that a negative result is not always conclusive.
So, she finally succumbed and the blood test was requested.

Five agonizing months later, my husband and I were told what we'd suspected for over 14 months. Isabella's blood test results confirmed there was a mutation type consistent with Rett Syndrome, and there it was.

So, now that we know;
Does anything change as far as Isabella is concerned? No.

Do I change the way I've been caring for her since she began losing her skills? No.

Does my love for Isabella change in any way? Absolutely not.

However on the flip side;
Do my husband and I want to extend our family now we know the risk of recurrence is less than 1%?

Do I have more tolerance and patience with Isabella when she's frustrated and has tantrums when we can't understand her?

Do we give Isabella more time to accomplish tasks because we know she has the will but finds it more difficult than most other children?

Do we have a name to write on forms and to tell others when they ask what's wrong?

Can we now plan ahead with where we're going to live, what type of equipment we should order in advance and which school or program to enroll Isabella in?

Do we have the support of an organisation we can join to be in contact with other affected families?

I can finally answer yes to all of those questions now that we have a diagnosis.

Had the MECP2 test been performed when I first requested it or had the specialists information been updated, we would not have subjected Isabella to so many unnecessary tests or anticonvulsants and going through that period of regression may have been a little easier to deal with had we have known **what** we were dealing with.

I understand years ago before the responsible gene was found, many girls were diagnosed much older when the hand wringing began or the decline in head circumference or lack of physical growth was prominent. But you know, my daughter is unusually tall and solid for her age, her head circumference is tracking just under the 50 percentile and up to now there hasn't been constant hand wringing. These are some of the reasons why the specialists initially couldn't or wouldn't consider eliminating the possibility of Rett Syndrome.
It just doesn't make sense to me that they were requesting tests for so many other genetic and metabolic disorders that are just as expensive and they were confident she didn't have.
I understand there is a funding and time issue. It has also recently come to my attention that some families have waited up to 19 months for their test results, which, as a parent of an affected child seems just so long to wait.

I have since been to see the geneticist who admitted to me, and I quote "we have learned a lot from this experience". The current method of diagnosis is based on a clinical one and the blood test is there to support it, but only after all other conditions are eliminated. Surely, considering the behavioral differences among affected girls, this procedure and government funding for genetic testing needs to be reviewed. Isabella had been subjected to well over $5000.00 worth of EEG's, MRI's, x-rays, ultrasounds, blood and urine tests which seems such a waste of valuable government funding, not to mention my family's out of pocket expenses.

The lack of knowledge and understanding of Rett Syndrome amongst hospital staff is also of great concern. This is the 2nd most common cause of disability in girls and yet when Isabella was recently taken to hospital, not one person in that emergency ward had even heard of Rett Syndrome - I must ask - how then, can they properly care for my daughter?

Some days I still ask myself, why me, why my daughter? The fact it takes a half hour to dress her, or the amount of physical energy it takes to go on a holiday or even just to go shopping and knowing the challenges we **still** have ahead of us overwhelms me. However, my daughter is the light of my life, she has taught me patience and an unconditional love I never knew was possible. She may not be able to tell me in words she loves me, but I can see it in her eyes and with her cuddles.

I now pray that one day soon, **professionals here or around the globe** may find a preventative treatment or perhaps even a cure before it's too late for my Isabella. My aim now is to not only raise my daughter with as much love and normality as possible but to also educate others about Rett Syndrome.

I'd just like to close by congratulating and thanking Dr Helen Leonard and her team. Because of the amazing amount of research that has been done over the years, we now have an indication of what to expect in the future with our precious Silent Angels.

Marisa Knott – 21/11/2005