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DIRECTIONS

FAMILIES OF SPINAL MUSCULAR ATROPHY

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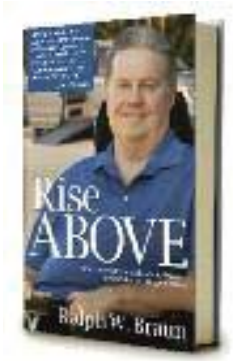

Dr. Mary Schroth, Chair of the FSMA Medical Advisory Council, and Charlie Sykora, SMA Type I

A Childhood Worth Fighting for

The Story of Cariad Howat

Written by Her Father, Jez Howat

As a family we don't spend lots of time on the past, or telling 'our story'. We don't have a website, Facebook or Myspace pages or anything like that. There are a number of reasons; we don't fit in to a 'support group' mould…basically, we have never been good at 'comparing notes' with others, or finding solutions to other peoples' problems. Being honest, we have enough emotional and physical baggage to carry on our own. It was hard enough coping at the time without worrying about 'being there' for others.

For the general public, if they have read or heard anything about SMA Type I, it's usually in the context of people struggling to work out the rest of their lives after its impact. Or it is reflective, with parents and family remembering their angels and still trying to make sense of it. These things tend to help others empathise with the destructive nature of SMA, but for us they are hard to read – and my guess is that for many families living with SMA, this is probably the case.

Now, we have a daughter with SMA Type I, and she is six years old. We have no idea what the ratios and survival rates are but we know we are very lucky that Cariad is not only with us, but that she is living as normal a life as she can. She attends school, is reading and (with help) writing. She is also speaking…a lot! We play games on the Wii, she loves to cook and…well, she is a normal kid trapped in a disabled body! Yes, she is a little angel – but she can be a little devil at times too.

And in truth that's one of the most important things to understand - underneath all that 'stuff', our little girl is a typical, stroppy, funny, loving, normal kid (depending on where you are reading this, you might like to cut this bit out for the medics).

Our journey here has been eventful with many difficult times, difficult conversations and difficult choices. However, what has occurred to me is that there is something that we could hope to offer others, and that is what we (medics, parents and carers) **need** to understand what we need to deal with children who live with this condition, and equally as important, what we need to **forget**.

We had the usual prognosis of a very short lifespan with not much air space to breath. Its no wonder that all many people see of SMA Type I is the aftermath. The worst part was that it feels so 'medical' and matter of fact, with little time for humanity outside of 'medical ethics' and palliative care.

Cariad was diagnosed at four months old. At six months she had respiratory collapse. During a 12 week drama we were asked to let her die several times. We also went through the 'what do you want to do?' question pretty much every day for three months - you know, the one where you get the feeling that they know the answer already, but are waiting for you to come round to it. During that time, we knew little about the condition, and the situation seemed to move so fast. We weren't offered any real choices of care and we didn't know enough to make a difference anyway. This changed very quickly!

The first thing that we found essential is **UNDERSTANDING**. Please don't read that as 'knowledge' of the condition – there is a mile of difference between knowing something and understanding it. Packaged in 'knowledge' is the prognosis – one of those things that parents, medics, etc need to learn to forget! Its grip on how a child will live their lives, what others expect of them and what services will be offered (or denied) is very much overstating its true place in things. Understanding the prognosis means knowing when it's served its purpose, and disposing of it accordingly!

I remember being told, shortly after Cariads diagnosis, that "there really isn't much point in putting a physio plan together - it's not going to do her any good is it?" I wonder how many parents could tell similar stories. Prognosis is useful. But it is not the law. Let's get it into perspective. Prognosis is an easy option – funding can be put into categories, care plans be made more generic, expectations put into safe boxes. It is also (to some degree) driven by a fear of being wrong (or sued) and also because it's harder to treat each person as an individual with an ever increasing workload.

It was interesting how things changed once Cariad reached two years old, and then three. We are still asked occasionally what the prognosis is…we just retell the official prognosis and watch the reaction, before shrugging our shoulders.

OK, this is simplifying things a bit. There are hard decisions to be made

based on what is predicted for a child's life. Some will not make the same choices we made. Some will go further than we want to; others may think we have gone too far. That's fine. However, it's a parent's responsibility to make those choices for their children with a truthful understanding provided by those who should have an understanding of the condition – grounded in the prognosis but knowing its limitations and accepting the child as an individual.

Rant about prognosis over, this brings me to the second thing we feel is needed - **CHOICES**.

We wanted Cariad to have the opportunity for life, and believed that she could experience this in a productive way, with the pleasure/pain/etc. that goes with that (which was the medics' main criteria for 'quality of life' 5½ years ago). The treating hospital disagreed with us that Cariad could achieve this. We asked that, if Cariad had an infection, she should be intubated for 72 hours for antibiotics to have an effect (the issue is setting a timescale). However, we didn't (don't) want her to have a tracheotomy or permanent hospitalisation.

I am happy to say we ARE right…and based upon that simple evidence, I would expect medics to take a different attitude now. It is a good question though; what is an 'acceptable quality of life' in the real world? This was a lot harder than deciding whether to allow her to die. We don't want 'life at any cost' but believe there must be a reasonable measurement. In some ways we are fortunate because she can now decide many things for herself.

Moving on to other choices, how about technology? We have embraced technology (Bi-pap, cough assist, suction, etc.). Technology has helped and sustained Cariad in good health, and we couldn't be without it. You also have to choose where this stops. This brings you into funding debates; as technology is not cheap… and someone has to pay. There is a very healthy debate about technology and funding and it does help to try to see this difficult dilemma from both sides. However, better people than I are unsure about this, so it's best left there! One thing I would like to say is that prognosis often has a major impact on the level of technology available (unless you are willing to buy it yourself). I am happy to be corrected on this, but as I have a healthy disregard for the P word, I do not think that is the most appropriate solution.

And the final tool we have discovered is **TENACITY**. When you have reached the right decision for you, stick to it.

I said that we chose to give Cariad a chance – even though we were being told there was none. We found that Cariad was receiving the wrong kind of care, although the hospital would not give her a Bi-PAP ventilator until she was three. We found evidence the best treatment was probably a Bi-PAP, and that children with her condition could survive longer. The hospital dismissed this evidence as it came from outside the UK. As a result of this disagreement our hospital launched us on a date for a judicial review (in the UK, if parents and medics disagree, the medics can get a court to decide…and thereby overrule).

With the help of the UK charity for SMA Type I, the Jennifer Trust, we began to stop 'following' and started to understand our choices, and the limits of those choices! With an offer of help from the Royal Brompton Hospital in London, we were saved from having to fight in court. They knew SMA and **understood** the condition. They hadn't much experience of introducing (effectively) a baby to Bi-PAP, but gave it a go with huge success. They were then able to treat the underlying respiratory infection.

There is no point in suggesting we made all our choices without their help – we needed lots of it and were allowed to ask dumb questions, throw tantrums occasionally and also to be real people – and Cariad a real child instead of an SMA Type I sufferer. We are grateful for their ongoing support and encouragement – even though Cariad has only been back into hospital once in the last three to four years (apart from visits and check ups). It's important to also say that we have built some significant bridges with our local Care Trust – especially post prognosis. They have implemented a comprehensive 'emergency arrangement' and have put a lot of effort into making sure she gets the best of care…though under the Brompton's guidance and specialist advice.

Tenacity is extremely important in the long term survival of an SMA Type I child. Find out who needs to be involved; nutritionists, OT's, speech and language, physiotherapists, portage, educational specialists, community team. Get as high up the tree as you can and build a relationship. It is only with the support of all these people that you can be sure you are getting everything you can.

What about the future? What about the family's life? One medic - who I would love to name but have actually forgotten his name - suggested that having a disabled child would place an unbearable strain on the family. I couldn't possibly write my response here, but don't all kids put unbearable strain on a family from time to time? We are actually incredibly lucky to have a family life where there is so much love, attention and effort is made to ensure that each and every day is normal and packed with family activities.

I really hope that more and more children with SMA Type I get an opportunity for longer life with non-invasive means of management. I am convinced that NIV via Bi-pap, together with a well managed package of care, is granting children with SMA Type I a better outlook, and I am very proud of the people at the Royal Brompton (Prof. Andrew Bush, Dr. Michelle Chatwin, Dr. Anita Symmonds, etc.) who are spearheading this in the UK. Most of all, I am proud of my little fighter, Cariad, who puts a great big smile on the face of every nurse, teacher and friend that she meets – I was never that sociable at her age!