Cure SMA Conference
June 14-17 2018
Dallas, Texas

Report by Fiona Tolich

I cannot start with anything other than praise for the wonderful people who pull this conference together – it really was phenomenal!

The impact this has had on my life is immeasurable. It has sparked a fire in me I thought I already had, or perhaps it has now created an inferno that I hope never goes out. The feeling of community was immense – researchers, clinicians, families, individuals… all coming together with no judgement, just complete respect for one another and an understanding of what each other was going through.

There were times where I felt confronted with what this disease does to people, including a moment where I was face to face with a child unable to breathe, but there were also so many more moments of hope from those already on a treatment path and from those who have achieved so much with this condition… in some cases, really beating the odds.

With well over 1600 attendees, the saying that everything is big in Texas was not wrong! I know that Anna Sutherland and I had travelled the furthest from New Zealand but we were not alone in our travels, with people from 30 different countries coming together for this one annual event.

To help describe the magnitude of the conference here are some other numbers:

- 450 researchers and health care professionals in attendance
- 55 family workshop sessions
- 185 researcher and care presentations

When you take into account that SMA is an ‘orphan disease’ (rare disease), it is simply amazing that an event like this can attract so many people and that a community like this can drive for such significant change. This is a broad and large community united in its mission to find a cure.

Newly Diagnosed Session

The Cure SMA Conference began for me with attendance at the Newly Diagnosed session (a session for those newly diagnosed or those who had never attended a conference before). It was here that we were introduced to Kenneth Hobby, the President of Cure SMA.

18 months ago, the community was still in a state of hope but that has started to morph into practical reality for many that help is here. Many in the room had access to Spinraza… this is what I want for our community at home – a chance to get our own piece of practical reality.

The exciting news is that more developments are in the pipeline and more treatments will be released. The one thing about SMA is that we know the cause of it – that those who have it are missing the SMN1 gene. Other diseases have different versions and mutations, but we are the same (something that will undoubtedly help to make significant breakthroughs). It is the SMN2 gene that has given the drugs/treatments a target. There is some hope with this back up gene as everyone with SMA has it, it is just the number of copies that they might have that are different.

The SMN2 gene makes a small amount of survival motor neuron protein. If an individual has 3 or more copies of SMN2, research shows us that they are more mildly affected, if they have just 1-2 copies then they have enough to keep them alive through birth but not enough to see them through life.

The mission for Cure SMA is a simple one. First slow the disease (current management strategies are doing this), then stop the disease and finally cure it. As much as there is a lot of positive work going on right now, there is still so much more to be done.

The key message is that we should not wait – do not wait for the next trial, do not wait for a treatment that is easier to administer… what you can get now is more important. We need to stop the deterioration – it is too important to wait. Every day you wait could be the loss of a motor neurone or more. New Zealand needs access to treatment now and I am on a mission to help drive this alongside Anna Sutherland

and our New Zealand SMA Reference Group! Our people and our community are important and we simply cannot be patient and wait!

It was exciting to also travel with Julie Cini from SMA Australia – what she has achieved across the Tasman with access to Spinraza for under 18's is so exciting. We wish them well with their next task of getting access to adults and hope we too can achieve in New Zealand.

Thomas Crawford MD, an SMA expert from John Hopkins Hospital in Maryland was amazing to listen to. He described DNA as being like a string of pearls on stretches of string. DNA takes the necklace and makes a copy where the pearls have to be put together. The pearls represent exons. In SMA, one of those pearls is missing – exon 7 of SMN1.

He also explained that everyone has mutations and 1/50 people have this mutation. Further to this, 1/2500 couples both have the same mutation. If you are unlucky, then SMN1 is missing from the child (think the pearl necklace).

The reality with SMA is that the weakness comes from the loss of motor neurones, everything else is completely fine. The consequences of that are what we see with SMA – skeletal deformity, weakness, no strength to cough, respiratory difficulty and so much more. As more motor neurones die, the impact becomes greater on the individual.

Until there is a cure, SMA is described as being like the Mafia – once you are in, there is no getting out. I, for one, plan on getting us all out! A cure is possible and I believe it will happen in my lifetime! "Without faith, nothing is possible. With it, nothing is impossible" Mary McLeod Bethune

Researcher Sessions

Although I was not in attendance at the Researcher Sessions as I'm not a researcher but someone with adult-onset SMA, I thought it warranted a mention. The information below has been taken from the Cure SMA page.

The SMA Researcher Meeting is the largest research meeting in the world specifically focused on SMA. The goal is to create open communication of early, unpublished scientific data, accelerating the pace of research. The meeting also furthers research by building productive collaborations—including cross-disciplinary

dialogue, partnerships, integration of new researchers and drug companies, and educational opportunities for junior researchers.

Finally, since the meeting is part of our Annual SMA Conference, researchers have the chance to interact with, learn from, and educate families affected by SMA.

At this year's conference, several companies released new data from their programs. The data released covers the entire SMA population – from infants to adults – and includes different therapeutic approaches, including those that target the underlying genetic cause of SMA and those that work on the muscles affected by SMA.

Final Word

This is just the start, those families who travelled from New Zealand (Anna and I) and Australia (Julie Cini and Zoe Watson) made a pact to go to different sessions to get as much information as possible. It is our commitment to get this information to you as soon as possible. This information will include nutritional recommendations, yoga therapy, aquatic physical therapy, orthopaedic management and so much more.

It is also important to note that on the final day of conference, it was announced that next year's Annual SMA Conference will take place between June 28th - July 1st at Disneyland in Anaheim, California. I for one am hooked – what I have taken from this conference and what I hope to share was well worth all of the efforts, sacrifice and tears when I left my kids behind. I highly recommend it if you can make it!

As a final word though, the work is just beginning here in New Zealand, and I want to treat it like a sprint relay race – the quicker we get out of the starting blocks, the better chance we have of achievement, but we all need to work together to get to the finish line. We need to work together for the sake of our community.