Noonan Syndrome Seminar

This issue

ISSUE
1
August
2016

Thank you for kindly completing our evaluation forms. Your feedback was encouraging and will guide us in our planning of future events.

Genetic Alliance Australia's primary role is to provide peer support and information to people affected directly or indirectly by a genetic condition. This support is through organising genetic seminars and getting people together.

Sunday 21st August 2016, 10.00am-3.00pm

Kinghorn Cancer Centre, 370 Victoria Street, Darlinghurst NSW 2010

Dear Families,

Thank you very much for attending the Noonan Syndrome seminar. It was nice to meet all of you and put names to a face. Fifty two people attended the seminar on Sunday. We really appreciate the time you took to travel and spend the day with us. A few of you came from quite far: Queensland, Australian Capital Territory, Victoria and even Western Australia.

Conversations were lively during networking time and it was nice to witness a friendly atmosphere that brought everyone together. I am glad that a lot of you made it to the picnic at the Sydney Royal Botanic Garden on Saturday: these opportunities to network are invaluable. I hope you all got the chance to meet new people and exchange contact.

I would like to thank all our speakers once again for giving very informative and engaging presentations. I hope it wasn't too overwhelming or confronting at times. Many thanks to Janelle Aronsten and Miranda Laird for sharing their personal story: they were very inspirational and many parents could relate to your experience.

I hope to see some of you at the 23rd Genetic Disorders Awareness Week Launch on 7th September 2016 (details p.3). If you would like to attend, please contact Dianne Petrie on 02 9295 8359.

We are also organising our next Telephone Support Group starting Tuesday, 11th October 2016 (See details p.2). For more information, call Dianne on 02 9295 8359 or email at dianne@geneticalliance.org.au .

Attached to this newsletter are the presentations. I have also included resources from the Noonan Syndrome conference that happened last July in Minnesota, USA. Please let me know if there are any other resources you would like access to.

Thank you once more to all attendees. We look forward to catching up with you soon. Until then - Remember we are only a phone call away.

Best wishes from Genetic Alliance Australia.

Doriane Ranaivoharison, Projects Manager

Dane's story

Remi and Dane on their way to school

After five years of IVF Dane and his twin sister Remi arrived in August 2009. A C-section was required at thirty-six weeks as Dane was tracking above a singleton pregnancy leaving no room for his sister to grow. Remi was born first with no complication but Dane followed two minutes later requiring bag and mask ventilation. He was losing his lower body pulse indicating a cardiac issue and had a nasal gastric tube inserted in the first twenty-four hours. Dane looked very different to his twin. He had silver white hair, nystagmus and his only obvious dysmorphic feature was his tiny low set rotated ears that I would tug at, thinking I could fix them - Yes Mother of the year!
Dane was transferred to Sydney. Cardiac surgery followed and genetic testing returned with a normal Karyotype. Dane also had a duplex collecting system on his left kidney. Post-cardiac surgery, Dane lost a lot of weight.

His delays became obvious as his twin sister reached each milestone. Little did I know he would be fifteen months before he could support his own head. The nose tube remained as his reflux was extreme and he would prefer to starve than drink and deal with the gagging, retching and vomiting. The sight of a bottle would result in him screaming in fear. Dane's reflux cycle was torturous and relentless. Due to his food aversion, also known as Post Traumatic Feeding Disorder, he was connected to a feeding pump. His stomach could not tolerate bolus feeds. The nightly retching episodes varied from thirty minutes to four hours. He was termed "failure to thrive" and had bowel surgery, fundoplication and gastrostomy surgery. The latter procedure stopped his ability to physically vomit and he could finally get blocks of sleep. He needed to eat to cease the reflux caused by liquid tube feeding but his fear of food was extreme.
We continued with our hospital feeding team for two years using the food play strategy alongside his twin sister for every meal and snack with no progress despite stopping feeds periodically.
Researching for experts in severe food aversion I came across the pioneers based in Austria. Support sites had also mentioned a specialist trained at Austria who came to families across the globe. It seemed for the first time in Dane's life that the stars aligned as we secured the services of this Doctor who came to us funded by a huge campaign that raised awareness of food aversion. No words can truly describe watching what happened during Dane's rapid tube wean therapy: this approach was exactly what he needed. The first two days were about creating a bond of trust as the psychologist won him over with endless fun.
By the end of day two, we had our breakthrough. Dane willingly engaged in a game involving a reverse psychology strategy, resulting in him gobbling McDonald's fries. The two-week treatment introduced a small safe menu and Dane was never tube fed again. He could now join the world and be with his twin at Pre-school. It was also wonderful to bring the psychologist back the next year with his colleague to treat some more children with complex issues.

In July 2014, a new dermatologist met with us as Dane's eczema had resulted in hospitalisations due to infections. He was extremely intrigued by Dane's appearance. He felt Dane fitted the condition Oculocutaneous Albinism Type-2 that created lack of pigmentation in the skin, white hair and piercing blue eyes that have involuntary side to side movement. After a meeting with an ophthalmologist Dane was sent for electrical activity testing that returned inconclusive.

Later that year, Dane was diagnosed with sleep apnoea and was scheduled for tonsil and adenoid surgery. Following the operation the surgeon informed me that Dane had a sub-mucous cleft palate. He looked at his long list of issues that had unfolded over the years and strongly suggested the possibility of a syndrome. Genetics was contacted and we waited for our appointment.

It was time to find a school for the twins and pre-school had advised us that Dane could do what his peers could if he had one-on-one support. He struggled with concentration, attention and had extremely poor fine motor skills. I was permitted to apply for three schools. Our local school suggested a 'multi-categorical' class for Dane but the teacher warned that it could be confronting for new parents. She was correct. I was concerned that if Dane copied his twin sister, he would also copy the children with extreme needs. It wasn't the right place for him. Another school advised that they could not keep him safe as there was no funding. I was very lucky with the third school. It was a private school with very few stairs and a learning centre to slow down the curriculum for Maths and English. The school saw the need to cater for Dane with a full time teacher's aide in and out of the classroom regardless of funding. The twins could be together and Dane could join mainstream classes after lunch. They transitioned to school in February 2015.

In July 2015, our appointment with genetics arrived. Noonan Syndrome (NS) was the immediate suggestion and test results returned confirming Noonan like Syndrome with loose anagen hair - SHOC2 mutation. A diagnosis provided an answer to everything but created a very paradoxical feeling as 'global delay' was replaced with Noonan Syndrome. The words can be hard to process - 'delay' meant temporary to us, 'syndrome' means permanent. At least the school could now apply for funding. I was to learn that Noonan Syndrome is not on any government disability lists. The limited funded school disability categories make our NS kids fall through the cracks. The signalling issue in the RAS-MAPK pathway can create learning disabilities across behaviour, attention, concentration and social communication. Slower speed of processing information is also common in NS and many children may need a full time teacher's aide. This has become my next mission: to enlighten the government and hope to bring about change. With support from my federal MP and an extensive letter from an expert Neuropsychologist in the field, I took Dane's story and that of NS kids to the ministers. A response is yet to return on a federal level.

Dane is progressing at his own pace at school and is feeling very included by the school community. Whenever he receives a merit award the school shows its incredible support. At age seven, Dane is exactly half of his twin sister's weight and stands on hundred and one centimetres. Regardless, his infectious 'cocktail party' personality is so large it outweighs his tiny stature.

OTHER RESOURCES:

An Overview of NS by Dr. Bruce Gelb
http://www.youtube.com/watch?v=pjfBf7IJnO0

NS & Cardiology Issues by Dr. Bruce Gelb
http://www.youtube.com/watch?v=g6mJ9nnD2pg

Genetics 101 by Beth Hopkins Denenberg, Pediatric Genetic Counselor
http://www.youtube.com/watch?v=z4XvzrIWnNQ

GEMSS: Genetic education material for school success.
www.gemssforschools.org

TELEPHONE SUPPORT GROUP:

Are you a parent, family member, or carer of someone with a genetic condition?

Do you feel isolated in your family/caring role?

Would you like to talk with other people in a similar situation?

Our next Telephone Support Group will start on Tuesday, 11th October 2016.

How our telephone support groups work:

Our telephone support groups are essentially a conference call with up to six parents, siblings or carers on the line with two qualified Genetic Alliance facilitators. The calls last for one hour, once per week for six week or less, at a time that suit participants. This enables participants to take part in group discussions in the comfort of their own home at no cost to them.

During each conference call, we discuss a range of topics such as accessing services, communication, behaviour, sibling issues, schooling, grief & loss, and any issues raised by the group. People share experiences and support each other - many stay in contact well after the group has finished.

How do I participate?

If you would like more information or to register, please contact **Dianne Petrie**:
Phone: **+61 2 9295 8359**
Email: **info@geneticalliance.org.au**

EVENTS:

Genetic Disorder Awareness Week
7th September 2016, 6pm-8pm
Parliament House NSW, Strangers Dining Room
Theme: "Australian patients and families' perspectives on Genome Sequencing: Survey results, translation and implications"
Contact Dianne Petrie (02 9295 8359) if you would like to attend.

Telephone Support Group
11th October 2016
Contact Dianne Petrie (02 9295 8359)

5th International RASopathies Symposium
28th -31st July, 2017
Chairs: Frank McCormick and Katherine A. Rauen
Renaissance Orlando at SeaWorld

CONTACTS:

Noonan Syndrome Awareness-Australia Community
https://www.facebook.com/noonansyndromeawareness

Noonan Syndrome Foundation
http://www.teamnoonan.org

RASopathiesNET
http://www.rasopathiesnet.org

Genetic Alliance Australia:
Contact details below.

If you would like the contact details of any of the other speakers please contact Genetic Alliance Australia

C/- Garvan Institute of Medical Research
Level 6, 384 Victoria Street
Darlinghurst NSW 2010
Australia

Email address: info@geneticalliance.org.au

Website: www.geneticalliance.org.au

Tel: + 61 2 9295 8359
Fax: + 61 2 9295 8181

Office Hours:
Monday: 10 – 4pm
Tuesday to Friday: 9am – 5pm

Genetic Alliance Australia aims to:-

Provide a contact point for families who are affected by genetic conditions so rare that they do not have their own support group.

Facilitate access to individual support groups for those families with a particular genetic disorder.

Provide a forum for the exchange of information between support groups regarding available community services.

Educate the medical and allied health professionals and the community about genetic disorders.

Consult with government bodies, both Federal and State, for appropriate funding for genetic services.

Funded by: NSW GOVERNMENT