ISSUE 4

QUEENSLAND ANGELMAN ASSOCIATION

AUGUST 2010

INSIDE THIS ISSUE:

PRODUCTIVITY COMMISSION SUBMISSION	2
QLD FAMILY STORY	3
QLD DRAFT PLAN FOR DISABLITIES	4
INTERESTING LINKS	4

Workshop—Challenging Behaviours in Angelman Syndrome

Need help understanding some of the challenging behaviour in children and adults with Angelman Syndrome?

Professor Chris Oliver will soon be here to help!

Registratons are now open for the workshop with Professor Oliver on Sunday 26th September at the Mater Hospital, South Brisbane, commencing at 10.00 a.m.

This presentation will explore the more common forms of challenging behaviour in children and adults with Angelman syndrome. There will be a description of how genetic disorder can influence behaviour and information on the forms of the behaviours and changes with age, followed by a description of the main causes of behaviours such as aggression to others. Methods for assessing causes will be described and a range of intervention techniques will be presented with advice on evaluating how effective different techniques are. Throughout the presentation there will be an emphasis on early intervention, the importance of consistency across environments and linking cause to intervention.

This workshop will be beneficial not only for parents, but also therapists, educators and carers. Please feel free to share this information to anyone you feel may benefit from this session.

Registrants will have the opportunity to ask questions prior to the workshop that will be addressed on the day.

To register your interest in attending the workshop, please visit our website www.angelmansyndromeqld.org/index.php?id=event

Please note: The official launch for the Foundation for Angelman Syndrome Therapeutics will be held early evening on September 26.
Details to follow.

BIRTHDAY CANDLES

July

Tara

Molly

August

Grace

September

Lucia

Bridge to Brisbane Fun Run—August 29

Help us raise funds for Angelman Syndrome and get fit at the same time!

In a salute to our angels and how hard they work every day, we are putting together a team to walk, run or shuffle the Bridge to Brisbane this year.

Donations can be made through out Team Page at Everyday Hero www.everydayhero.com.au/team_angelman with all the proceeds going to the newly established FAST Australia! If you are interested in joining us, please email jen@angelmansyndromeqld.org
More details on the event can be found here http://www.bridgetobrisbane.com.au/

The Developing Foundation

The Developing Foundation Inc. was founded in 1978 as a support group by families seeking appropriate treatment for a family member with a brain injury or a developmental disability. The organisation is approved as a service provider by Disability Services Queensland.

The Developing Foundation supports families caring for a child or adult with a brain injury or developmental disability. The Foundation believes that the potentially adverse outcomes of brain injury and developmental delays can be minimised or decreased through education and intervention.

The Foundation offers families a chance to fundraise for their own individual needs through their annual Riverwalk on 12th September at West End, Brisbane. Riverwalk is a sponsored 5km walk along the Brisbane River.

This annual fundraising event helps families raise funds to provide support and care for their loved ones. 90% of the money raised by the family, goes to the family, so every donation makes a difference.

If you are interested in particpating in the Riverwalk and raising funds for your child, please see http://www.developingfoundation.org.au/news/item/id/4

"The heart of a volunteer is not measured in size, but by the depth of the commitment to make a difference in the lives of others. "

Teamwork divides the task and doubles the success

A big thanks to QAA Member, Emily McInnes, for sharing her family's story on (Page 3), and helping us with information and links for our newsletter, website & Facebook site.

This is your association, the more hands on deck the more successful we are! If you have a story to contribute to the newsletter (perhaps a profile or therapy story), Facebook or our website please contact us!

FAST Australia are also looking for volunteers to assist with the Foundation Launch in Brisbane in September, if you can help please contact admin@cureangelman.org.au

Epilepsy Qld are now on Facebook!

Join up to get all the latest news and events such as;

Cairns - Wednesday August 11—Understanding Epilepsy Workshop
- Thurs August 12 Morning Tea
Call 1300 852 853 to register your interest

Productivity Commission Submission

The Queensland Angelman Association are in the process of finalising a submission to the Australian Productivity Commission's Inquiry into Disability Care and Support. http://www.pc.gov.au/projects/inquiry/disability-support

The Commission are currently calling for both personal and organisational submissions detailing the current service gaps applicable, together with suggestions for an improved disability sector within Australia. Closing date for submissions is 16th August.

This is a real opportunity for our families to have a say and make a difference in future disability policy. If you would like to assist by sharing your circumstances and suggestions with us for inclusion in our submission, please email jen@angelmansyndromeqld.org

«This is a real opportunity for our families to have a say and make a difference in future disability policy "

Our Angel Family

In April we received the test results that both of our children, Alex and Hannah, have Angelman Syndrome. They have a mutation in one copy of their UBE3A gene (on Chromosome 15). It turns out that I am a carrier of this mutation, which does not affect me, but causes AS when inherited as it is on their maternal copy.

Alex is now 4, Hannah is 2.5 years old. Although they have identical mutations, they have very different personalities, and different histories. It was only Alex that was suspected of having AS, although we were told this was unlikely. On her own, Hannah might not have been diagnosed until much later, as it wasn't considered a possibility.

Alex had reflux, was slow to gain weight, and was difficult to transition to solids. He was capable of eating the food once it was in his mouth, but was very much against letting us get it in there. He also had low muscle tone, and a tremor/ataxia. Out in public he was a charmer, we had strangers stopping us to comment on how smiley and happy he was, and people who already knew him still remarked on it every time they saw him. As he was our first child, we just thought these were the usual challenges of child rearing, and that was his personality. When I returned to work, his daycare informed me that his development wasn't right. It wasn't just the delay in motor and language skills that concerned them, but also his lack of emotional range. I would often be called to collect him due to high temperatures, and on arrival would find him happily playing. We began the diagnosis journey, with no idea just how long a road it would be, and certainly no idea that it wouldn't be something we could fix.

When Hannah was born, we were so happy, realising this must be what a typical child is meant to be like. She did have low muscle tone, but we were told it was mild. She came along to her big brother's appointments, watching quietly from her pram, while sucking her thumb or sleeping. By the time she was nine months, I finally had to admit that she also needed help. She couldn't sit, wasn't gaining weight, and although she didn't mind food being put in her mouth, she couldn't eat it. She was given the global development delay label (same as Alex), and also failure to thrive.

Angelman Syndrome had been considered and dismissed (negative FISH and methylation tests) for Alex when he was 2, before Hannah's delays became apparent. Now that we had two children with global development delay, the doctors seemed baffled. The kids didn't have any obvious physical abnormalities, or medical conditions to suggest something to test for. Also, they seemed very different, so the doctors weren't sure if they had the same condition or something different. The general conclusion was that it most likely was a recessive genetic condition that both parents must carry. We weren't given much hope of getting a diagnosis, and were discouraged from pursuing it.

Since originally finding out about AS, I had continued to read about it every now and then. A lot of what I read sounded like Alex, especially the behaviours that were talked about, like hair pulling and love of water. On the other hand a lot of the diagnostic information and videos I watched showed more severe cases, and I would again put it aside.

I really owe a lot of gratitude to the ladies of the Queensland Angelman Association. The workshop with Dr Weeber last year gave me the opportunity to meet a boy that had a UBE3A gene mutation. He reminded me so much of my son, and I knew that we had to have the gene sequencing test. The QAA also started the process that led to the test being available in Brisbane (previously only overseas), and finally we were able to convince the doctors to test Alex. Once Alex's test came back abnormal, they did further testing that confirmed Hannah also had the mutation, with myself as the carrier.

Although my children don't meet the diagnostic / clinical criteria, they do have Angelman Syndrome. The geneticist was the most surprised person in the room the day they gave us the results. Since then I have learned that children with a UBE3A mutation are less likely to fit the pattern, which suggests to me it is time for the criteria to be reviewed, or at least to reflect that it is a spectrum, not absolute. I think about how many undiagnosed children must be out there, with their parents still stuck on the diagnosis rollercoaster, and how easily AS could've been missed in our case, especially for Hannah who was only tested after the fact.

Alex started walking after he turned three, and is able to use some makaton signs. He still tries to charm everyone while out and about, but at home when it is just mum he likes to be the boss and have things his way. Hannah is cruising furniture, and can walk with AFOs and a walker.

> ***«Although my children don't meet the diagnostic / clinical criteria, they do have Angelman Syndrome»***

She has recently stopped sucking her thumb, which has seen an amazing turn around - she eats like a champion and is much more interested in activities.

A big thank you to Emily for sharing her family's story. It is an example to us all how different our angels all are, and that diagnosis can be a rough road for many families.

QUEENSLAND ANGELMAN ASSOCIATION

www.angelmansyndromeqld.org

enquiries@angelmansyndromeqld.org

Interesting Links

Ageing & AS - Is there a link?

Exceptional Family TV - A Rare Angel

Clinical Guideline for the management of AS

Tap to Talk - iPad Application

Communication Device &/or free workshop on Literacy Development & Difficulties

Nursery Rhymes to See, Sing and Sign

Free Printable Visual Aids

Auslan Signbank

*We have so many links this edition we could not include the actual text of the link! If you are interested in the actual web address please contact us

Queensland Angelman Association is an unincorporated group of parents & carers focused on providing support & information to Queensland families who have been touched by Angelman Syndrome.

Through our experiences we have discovered the comfort in being surrounded by a loving & compassionate Angelman Syndrome community. For whatever challenges that may be faced during this journey, there will be others to support & provide advice. It is our goal to open a channel of communication between Queensland families.

We believe that information is power. It is important that our Angelman Syndrome families are provided the most up to date information on research, studies & the achievements of our angels around the world.

Queensland Draft Plan for Disabilities

From 2005-2009 a series of Shared Vision Conferences and Public Forums were held around Queensland to explore the future of disability services in Queensland.

It is reported the sector responded enthusiastically to this opportunity to share best practice, form networks and be part of a process for identifying strategies to create a positive future. As a result of this work, a Draft 10-year Plan for Supporting Queenslanders with a disability has been created.

Disability Services Queensland are calling for public feedback on this plan, and a copy of the consultation paper and questions can be found at http://www.disability.qld.gov.au/community/10-year-plan/index.html

Furthermore, there will be Regional Forms held to enable families to share their visions for a better future for people with a disability in Queensland. This forum is open to families, carers as well as interested service providers.

Information on forum dates and locatons can be found at http://www.disability.qld.gov.au/community/shared-visions/forum-2010/forum-dates-locations.html. This plan, and subseqent consultation, has been unveiled together with a boost in funding for 2010-11 to the tune of $1.054 billion for disability services in Queensland, and $528.7 million for Home and Community Care.

It is reported that the extra $12.5 million in 2010-11 will provide an extra:

- $4.4 million to support young people with a disability leaving school;
- $3.2 million to support people with spinal cord injuries;
- $2.9 million for early intervention and support services for the families of children with a disability;
- $1 million for the Community Living Initiative, to help people with a disability to live independently in the community;
- $1 million to create new autism early intervention centres in Bundaberg and Mackay.