ASSOCIATION OF GENETIC SUPPORT OF AUSTRALASIA INC. NEWSLETTER

Making the right connections since 1988

NOVEMBER 2012 ISSUE 98 ISSN1033-8624 Funded by NSW HEALTH & Ageing Disability and Home Care (ADHC)

The AGSA newsletter is kindly sponsored by Genea

GENEA
WORLD LEADERS IN FERTILITY

CONTENTS

MISSION STATEMENT

To facilitate support for those affected directly or indirectly by genetic conditions throughout Australasia.

EDITORIAL

Christmas is rushing towards us and already the shops are full of Christmas decorations. As I sit here writing this it is a very grey dull day and the rain is drizzling down. This weather certainly doesn't reflect the exciting couple of months AGSA has had.

On the weekend of 16th November AGSA ran a rural outreach seminar and Sibshop in Lismore. This was our third visit to the area and we had a good turn out with excellent speakers.

Recently we held our final Leadership meeting for the year where support group coordinators got together and shared stories and information. Although the meeting was small much information gleaned with lots of discussion and laughter. If you wish to be a part of the group please email Jennifer on projects@agsa-geneticsupport.org.au
Our next meeting will be in February before Rare Disease Day on 28th February. We will keep you posted as well as providing information on the format Rare Disease Day will take this year.

This year I am very pleased to announce I have been appointed to the National Health & Medical Research Council (NHMRC) Human Genetics Advisory Committee (HGAC) by The Hon Tanya Plibersek MP Minister for Health. The appointment runs to June 2015. HGAC is a Principal Committee of NHMRC.

Recently I attended their first meeting in Canberra Chaired by Professor Robyn Ward along with 15 other committee members. This is a challenging position, sitting five times a year for 3 years to discuss many major issues in the field of genetics.
http://www.nhmrc.gov.au/about/committees-nhmrc/human-geneticadvisory-committee-hgac

AGSA has attended two Genetics Testing Working Party meetings. Issues of concern to families were recorded with lively and healthy discussion which have been included in the report. The final paper will be completed in December and will be open for public consultation.

On 14th October, 80 people attended AGSA's 12th BRCA Information Day held at the Sebel Sydney, Surry Hills. 73% of attendees were newcomers. AGSA would like to thank the genetic counsellors for all their hard work in planning and attending the day which was a resounding success.

I am happy to announce AGSA has been successful in securing a Community Grant from nib Foundation to research the *Unmet needs of families living in the Hunter Region.* We are currently advertising for a research assistant to start work on this project next year.

AGSA's AGM will be on 12th December at 7pm.

AGSA is holding a fundraising sausage sizzle at Bunning's in Mascot (Cnr Bourke & Gardeners Rd) on Friday 22nd February 2013 so if you are in the area come along and say hello.

I look forward to seeing you either at our AGM, Bunnings or Rare Disease Day.

Until then, Best wishes to everyone for a safe and happy Christmas break.

DIANNE PETRIE OAM

CONTACT CORNER

AGSA will publish requests for contact and letters from people searching for families with similar experiences, from those seeking or contributing specific information as well as other resource information.

Anyone who wishes to reply to a request or a letter should write direct to the individual or group concerned where an address is provided. The AGSA office many be contacted for the information to be passed on in the case of anonymous requests. Privacy and anonymity will be ensured if requested.

While AGSA aims to facilitate contacts between families it is unable to assess the suitability of these in individual cases.

It should be remembered that a shared genetic condition does not mean an equally shared value system between families. Different degrees of acceptance and different mechanisms for coping will be encountered and a non-judgmental approach is recommended in establishing contact.

CONGENTIAL DISORDER OF GLYCOSLATION
A mother of two children aged 4 and 2 with this very rare genetic condition is trying to find someone else to talk with. Please contact AGSA for information.

BUSCHKE OLLENDORF SYNDROME
A mother of a six year old boy in the Netherlands is looking for information on this condition and contact with others. Please contact AGSA for information.

AUTOSOMAL RECESSIVE SPASTIC ATAXIA OF CHARLEVOIX SAGUERAY
A lady with this condition would like contact with someone else who has the same condition. Please contact AGSA for information.

TRPS TYPE III– TRICHORHINOPHALANGEAL CONNECTIVE TISSUE DYSPLASIA
A mother of a 10 year old girl would like contact with others who have similar issues. Please contact AGSA for information.

PENI-SHOKEIR TYPE I
A 23 year old lady would like contact with others. Please contact AGSA for information.

WERNER SYNDROME
www.het-werner-syndrome.com
I received an email from a young man in the Netherlands. He asked me for our support in finding people with the same disease condition. He is affected by a very rare genetic disease called 'Werner Syndrome', also known as Adult Progeria. The incidence is 1 in 1 million.

Right now this young man doesn't know anyone in our country and is looking for people in other countries. He is already registered in the rare disease database and he also created his own website on the disease: www.het-werner-syndroom.com

Please can you look among your contacts (patient organizations, physicians etc) to check whether you know someone in your country with the same diagnosis? It is very important to him.

I thank you already in advance for your support.

With kind regards,

Maryze Schoneveld van der Linde

Department of Families Housing Community Services& Indigenous Affairs

Better Start for Children with Disability expansion

Information for parents & carers
From 1 January 2013, children who are aged under 6 years and have been diagnosed with Prader-Willi, Williams, Angelman, Kabuki Make Up, Smith-Magenis, CHARGE, Cornelia de Lange or Cri du Chat syndromes or Microcephaly will be able to register to access early intervention funding of up to $12,000 (up to a maximum of $6,000 per financial year) under the Better Start for Children with Disability (Better Start) initiative.

Since 1 July 2011, children who are aged under 6 years and have been diagnosed with cerebral palsy, Down syndrome, Fragile X syndrome and moderate or greater hearing or vision impairments, including deaf blindness, have been able to register for access to the early intervention

funding under Better Start.

The funding can be used to pay for services such as speech pathology, audiology, occupational therapy, physiotherapy, optometry, psychology, orthoptics and services of teachers of the deaf. Families have until their child turns seven to use the early intervention funding.

This targeted early intervention during the pre-school years' aims to complement existing Commonwealth and State and Territory services and to assist these children to have the best possible preparation for their transition to school.

Support for Families with older children
In addition to the early intervention funding under Better Start, children with the previously listed disabilities (including those who are over the age of six) may be eligible to receive Medicare rebates for:-

- the development of a treatment and management plan by a general practitioner or by a specialist or consultant physician (for a child under 13 years of age)

- up to four diagnostic / assessment services delivered by psychologists, speech pathologists, occupational therapists, audiologists, optometrists, orthoptists or physiotherapists to assist the referring practitioner with diagnosis or to contribute to a child's treatment and management plan (for a child under 13 years of age)

- up to twenty treatment services delivered by psychologists, speech pathologists, occupational therapists, audiologists, optometrists, orthoptists or physiotherapists (for a child under 15 years of age, providing a treatment and management plan is in place before their 13th birthday)

Parents and carers of eligible children are not required to complete a formal application to access the Medicare component of the initiative, but need to be referred by their general practitioner or by a specialist or consultant physician.

The Medicare component of the Better Start initiative is administered by the Department of Health and Ageing. Further information about the Medicare component of Better Start is available at http://www.health.gov.au/internet/main/publishing.nsf/Content/children_disability

Support for families living in outer regional and remote areas
Families living in outer regional or remote areas may be eligible for a $2,000 one-off support payment. This payment recognises the difficulties that families living in remote and outer regional areas face in accessing services and will assist with expenses such as travel and home visits.

Registering for the Better Start initiative
Parents or carers of eligible children need to register their child for Better Start before they turn six in order to access the early intervention funding.

The Better Start Registration and Information Service (RIS) is operated by Carers Australia.

To register your child for the Better Start initiative, please call the Carers Australia RIS on **1800 242 636**.

In order to register for the Better Start initiative families will need to provide proof of:

- the child's age (e.g. the child's birth certificate or passport),
- the child's residential address (e.g. a recent utilities bill or rates notice),
- the child's diagnosis,
- residential status (if applicable), and
- the child's Centrelink generated Customer

Reference Number (CRN).

Further information
Please visit
www.fahcsia.gov.au/betterstart

Enquiries can also be directed to the Better Start Helpline on
1800 778 581 or emailed to
Better.Start@fahcsia.gov.au

SUPPORT GROUP NEWS

Association for Children with a Disability NSW (ACDNSW)
The updated, 4th Edition of ACD NSW's flagship publication for parents of children with a disability, Through the Maze, is now available at www.acdnsw.org.au

DAISI – Disability and Aged Information Service Inc.
Play Quest
Cage and Play Lismore Special Needs
At Playquest (SNAP) provides a comfortable environment where parents, carers, and children can come and unwind without any concern.

Every Wednesday afternoons from 3-6pm
Cost: Free for 0-12 months, $5.00 for under 2 years and $9.50 for 2-11 year olds

CONFERENCES

10th ACPHG- Asia Pacific Conference on Human Genetics
5-8th December 2012
Crowne Plaza Hotel, Kuala Lumpur
Malaysiahttp://www.apchg2012.org/

34th Annual Lorne Genome Conference
17-19th February 2013
Mantra Resort, Lorne, Victoria, Australia
http://www.genome-conf.net.au/

The 13th International Meeting on Psychosocial Aspects of Hereditary Cancer
7-8th March 2013, Sydney, Australia
http://www.impahc2013.com.au/

Joint Conference of HGM 2013 and 21st International Congress of Genetics
13-18th April 2013
The Sands Expo and Convention Center,
Marina Bay Sands, Singapore
http://www.hgm2013-icg.org

Porphyria Patient Day
Saturday, May 18th, 2013 as part of the International Congress of Porphyrins and Porphyrias at the conference center KKL in Lucerne, Switzerland. Information on the congress is available at
www.porphyrinsandporphyrias.org

Australian Addison's Disease Association Inc.
NSW/Sydney Chapter Annual Addison's Seminar, 2013
The Addison's Journey: Self Management
Saturday, 25th May 2013
Kerry Packer Auditorium, Prince Alfred Hospital, Camperdown, Bronwyn: 0427 601 795

Familial Aspects of Cancer 2013
25-28th August 2013
Cairns Convention Centre, Cairns, Australia
http://wired.ivvy.com/event/A1DYPE

PERSONAL STORIES

"All for the love of Kirsty"

Written by her parents, Robert and Debra.

Now

Kirsty is 25 years old, and has lived away from the family home for the past 9 years. Kirsty currently lives in a group home, along with four housemates, and 24 hour support staff. Kirsty is 190cm (6'2") tall, weighs 85kg; she has a diagnosis of Sotos Syndrome and Bi Polar Disorder, along with a moderate intellectual delay.

There have been endless bureaucratic hurdles for us to overcome along the way, to finally have her placed in an environment where her every day needs are met, where she has a quality of life and a bright future. She is gaining her full potential and independence to the best of her abilities.
Kirsty happily attends a day options program where she is very productive and popular amongst her peers. She loves dancing and karaoke, her favourite singer has always been John Farnham!

Then

Kirsty was diagnosed with Sotos Syndrome at age 3 years. Over the next few years, this diagnosis gave us explanations as to why Kirsty was growing rapidly, why she had low muscle tone, long hands

and feet, autistic tendencies, challenging behaviours, her struggles with numbers and maths,
even though she learned to read and write and speak at the appropriate ages.

Due to her low muscle tone and weight, Kirsty did not walk until 27months. Her birth weight was 4.81 kg and she continued to gain 1kg a week, at age 6 weeks she was almost 11kg. Finally, this rapid weight gain settled, but she continued to grow "upwards" at an accelerated rate, until the age of 15. Kirsty had no particular health problems during the early years.

At age 12, Kirsty started to exhibit more frequent aggressive behaviours. Behavioural management plans and strategies, both at home and school were clearly not working. We became exhausted anticipating her every move, her absconding from home and school, being physically abusive to both her parents, teachers and peers. She was in danger of self-harm and putting everyone around her in danger.

Finally, when Kirsty was 15, "the straw that broke the camel's back", came when we answered the umpteenth phone call from her school, asking us to pick her up as she was being verbally and physically abusive to both teachers and students. My husband told the principal *"we couldn't pick her up, we could no longer cope with her either, that if we took her home, one of us might snap under pressure, that we were all in danger of harm*". The principal was very supportive, and the next job was for him to notify the police, for the police to take Kirsty into "custody" until emergency respite could be organised.

That is where the heartache (for us) began. Our entire focus was always to provide the best possible support for Kirsty, for her individual needs to be met, and here we were, telling the authorities we were abandoning her. It was a bitter realisation for us, as it was the only way to get the support ***KIRSTY*** needed for long term care. It was never about us, it's always been "***All for the Love of Kirsty".***

We were to learn there was no "system" and there was no referral process for people with disabilities to be assessed in appropriate health care facilities, and the lack of facilities. We set precedents along the way, court orders and guardianship orders, advocating for Kirsty to numerous government bodies, encountering endless rounds of meetings, complying with all the rules, until we were heard. We were advocating on ***Kirsty's*** behalf, for ***Kirsty's*** rights, the same rights we are all entitled to, regardless of ability.

Our Thoughts

The diagnosis of Sotos Syndrome, at times, masked the fact that Kirsty may have had a second condition, apart from Sotos Syndrome. We had to insist the

health and community services look further than Sotos, as not everyone with Sotos is the same. They only share the same set of criteria that makes them "Sotos".

We have, and always will, advocate for Kirsty as an ***individual***, to make certain ***her*** needs and rights are met, so she can remain independent and the happy and healthy young woman that she is today.

AGSA would like to sincerely thank Robert and Debra for kindly sharing their experiences during a very difficult time in their lives. It is greatly appreciated.

From Google Alerts

Enzyme therapy allows child with rare genetic mutation comfort closer to home

by JANET ST. JAMES
WFAA
Posted on October 29, 2012 at 1:23 PM
Updated today at 1:23 PM

DALLAS -- Being poked and prodded is nothing for eight-year-old Tearrian Salois, who often feels like he's on fire -- from the inside out.

"Sometimes my feet burn and my hands burn because of the disease," he explained. "It hurts really bad."

Tearrian has Fabry disease; a rare, painful, and potentially-deadly genetic mutation that causes dangerous fats to build up in blood vessels, tissues and organs. For more than a year, Tearrian has travelled from Montana to Dallas every week for experimental enzyme therapy that

relieves the burn.

The treatment is part of a clinical trial at Baylor's Institute for Metabolic Disease. The institute specializes in cutting-edge research and treatment for genetic diseases so unusual; they affect only a handful of people on the planet.

"Some adults were actually committing suicide because they were suffering so much from the effects of this disease," said Dr. Raphael Schiffmann, a pediatric neurologist who has overseen Tearrian's case. "Yet, the tests were normal."

Schiffmann and Tearrian's family were frustrated when the manufacturer decided to discontinue the clinical trial that helped make Tearrian's life bearable. Often, when drugs for uncommon diseases go away, patients die.

"It would've hurt," said Tearrian's mother Tina, through tears. "I would've have to watch him suffer. Because sometimes the over-the-counter drugs don't help."

Instead, the institute and Tina Salois fought to get Tearrian on an adult drug that he could receive through an infusion closer to home.

Last Tuesday was his last treatment in Dallas. Everyone associated with his case hopes it also symbolized his first step toward being a healthy homebody.
E-mail jstjames@wfaa.com

CAUSES OF CANCER ARE NOT COMMON

LEADING cancer researcher has called for greater emphasis on personalised treatment after a landmark Australian study found the underlying genetic cause of pancreatic cancer differed among patients.
After sequencing the genome of 142 people and their pancreatic tumours, scientists found no two cancers were the same.

The findings, part of an international program to identify the genetic drivers behind 50 different cancers, have implications for the treatment of other types of tumours.

"The big thing to come out of this is that cancer is not one disease," said Andrew Biankin, from the Kinghorn Cancer Centre at the Garvan Institute of Medical Research and a lead author of the study. "While there are common genes in each tumour, based on this we think each cancer should be treated differently."

Pancreatic cancer is one of the most lethal cancers. The majority of patients die within a year of being diagnosed – a survival rate that has not improved in almost 50 years.

As part of the study, published in the journal *Nature*, the team identified more than 2000 genes involved in the cancer, adding to the half a dozen already linked to the disease.

While four genes were present in 50 per cent of pancreatic tumours, most mutations were found in less than 2 per cent of cancers.

Several study participants were placed on more targeted medication after their doctors were alerted to their genetic profile. "It changed the treatment of not just the individual patients but, because we've detected some inherited genes, it has also impacted on the wellbeing and the care of their families," he said.

One participant, Zenon Slotwinski, whose mother died of complications from pancreatic cancer in April 2010, learnt he and his two sisters had inherited a known cancer gene from her.

As a result, Mr Slotwinski, 54, is in a pancreatic and prostate cancer screening program, and his sisters have undergone preventative operations to remove susceptible organs. "For me knowledge is a good thing but everyone has a different view," he said.

Professor Biankin said standard cancer treatments were based on where the cancer originated, and doctors were required to first treat patients with drugs shown to be most effective on most patients before moving on to other medications.

"Unfortunately most patients with pancreatic cancer never get to the second drug because if they haven't responded to the first drug, the disease is so aggressive they end up dying before they get their second choice, [which] might have been the right one for [that patient]," he said.
To improve tumour treatments, researchers around the world are building a "genetic knowledge bank of cancers" that doctors could refer to when deciding the drug most suited to the genetic profile of their patient's cancer, Professor Biankin said.

Clinical trials are now conducted on a particular drug for a specific cancer, a process that can take years and require hundreds and sometimes thousands of patients. Personalised cancer

treatments are being used throughout health systems in the United States, Britain and Europe but are yet to take hold in Australia, he said.

From next month the Australian Pancreatic Cancer Genome Initiative will run a clinical trial in which the treatment for pancreatic cancer patients will be matched to their genetic profile.
From the Sydney Morning Herald, Thursday 25th October 2012

Scientists in the US have created embryos with genes from one man and two women.

By Nick Phillips Science
SMH Thursday 25th October 2012

Read more: http://www.smh.com.au/technology/sci-tech/scientists-grow-embryos-from-three-adults-20121025-286fv.html#ixzz2AGWsnfZv

SCIENTISTS have grown five-day-old human embryos with a new technique that uses genes from three adults. To create each embryo, genetic material from one woman's egg was inserted into another donor egg before being fertilised with male sperm.

Researchers hope the technique will eventually help women with diseases of the mitochondria - the energy powerhouses inside cells - avoid passing the disorder onto their children.

Illustration: Cathy Wilcox
But the controversial procedure, first demonstrated in human embryos in Britain in 2008, has raised ethical concerns among some people who believe it would be inappropriate to produce offspring with genetic material from three adults.

The technique new, developed by scientists at the Oregon Health and Science University, involved transplanting the nucleus, which contains most of an organism's DNA including the instructions for characteristics such as sex, height and eye colour, from one woman's egg into a healthy donor egg whose own nucleus had been removed.

The egg was then fertilised with donor sperm. If the technique, known as spindle transfer, was used in a woman with a mitochondrial disease, the new egg would contain the mother's chromosomes but the donor's healthy mitochondria.

''The technique works pretty well - technically mitochondrial DNA can be fully replaced in human oocytes,'' said lead researcher Shoukhrat Mitalipov.

Just under half of the eggs that underwent spindle transfer completed normal fertilisation and developed into embryos.

From those, the team was able to produce stem cell lines to demonstrate the gene replacement had been successful.

While the research shows the procedure is technically possible, it would be years before it could be offered as a fertility therapy for humans. The creation of embryos with more than two genetic parents is banned in Australia.

While mitochondrial DNA play no role in the development of a person's characteristics, defects in this type of DNA can cause a range of diseases including blindness, epilepsy and mental retardation.

According to the Australian Mitochondrial Disease Foundation the disorders likely affect more than 100,000 Australians. In 2009, Professor Mitalipov conducted spindle transfer in female macaque monkey eggs, which were then reinserted into the mother's wombs and grew into healthy babies.

Follow-up studies of the monkeys, now three years old, showed they were developing normally, he said.

In 2010, a group of British scientists published research on human embryos they had grown by transplanting the genetic material from a fertilised egg into a donor egg, a process known as pronuclear transfer.

''At this point it is hard to say which of these techniques is better,'' said Professor Mitalipov, whose findings are published in the journal *Nature*.

The British research prompted the government to launch a public consultation on the ethical issues associated with the technique. A report is due next year.