ASSOCIATION OF GENETIC SUPPORT OF AUSTRALASIA INC. NEWSLETTER

Making the right connections since 1988

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GENEA
WORLD LEADERS IN FERTILITY

CONTENTS

MISSION STATEMENT

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

EDITORIAL Happy Easter everyone.

With the closure of Darrell Lea where will I find chocolate treats!!?? I hope you have a good break.

Rare Disease Day was a collaborative event with the Steve Waugh Foundation, BEEHAC and AGSA at the Sydney Cricket Ground. It was an early start and we all wore our distinctive black Rare T-shirts and posed behind Stevie the weather man from Channel 9. We managed to get national coverage and a number of friends spotted us. A number of community service announcements went out which were well received.

Thank you to the AGSA people who participated.

nib *foundation* on Rare Disease Day issued a Press Release announcing their partnership with AGSA on the project to identify unmet needs in the rare disease community in the Hunter region resulting in media coverage by radio and ABC news. Catherine Spinks, a Genetic Counsellor, has been employed for a year to undertake this research which involves running a couple of seminars, sibling workshops and interviewing families. Not to mention all the necessary report and ethics approval submissions.

It is lovely to have Catherine on board.

AGSA also welcomes Ayesha our new Administration Officer. Ayesha is doing her Masters in Public Health at Sydney University. We are still looking for a Projects Manager to replaced Jennifer who has left to finish her training as a psychologist, an offer she just could not refuse.

I have attended a number of Human Genetic Advisory Committee meetings in Canberra which are very stimulating. I am pleased to say rare diseases are on the agenda.

Sue Hawkins presented on peer support training at our latest leadership meeting and the topic for the next meeting will be siblings.

AGSA's fundraising sausage sizzle at Bunnings Mascot raised $900. Thank you to our wonderful helpers, Scott & Marlene Brightwell, Claudia Bereny and Ayesha Wijesinghe.

Dates for the diary – IDEAS Expo Newcastle Entertainment Centre. AGSA will be holding an informative seminar at 2:15pm-3:30pm on 4th May. Please come along. Until the next newsletter, keep well.

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.

- ***Pristanic Acid Disorder***
- ***Isaacs Syndrome***
- ***Deletion 10q 23.1***
- ***4p Duplication 8p 23 Deletion***

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.

CENTRELINK

The difference between the Carer Payment and the Carer Allowance:

- The Carer Payment is an income support payment for people with no income. This payment is means tested.
- Carers Allowance is a supplement payment and is not means tested.
- People who are entitled to receive the carer payment are also entitled to receive the Carer Allowance.
- The same form is used for both the payment and the allowance. Forms however are no longer available online and you need to ring to have it delivered.
- If your care recipient enters a nursing home or other residential facility, the Carers Payment and Carers Allowance are cancelled.
- When applying for Carers Allowance or Carers Payment, the level of care provided is required as applications are processed by a scoring system.
- If you are a carer and work more than 25 hours per week you are not entitled to a Carer payment. However, if you have a family member who cares for your care recipient while you work, they may be entitled to the Carer Payment.
- If you receive a Carers Payment, a Health Care Card is automatically approved for you.
- You do not have to live with your care recipient in order to receive a Carers Allowance.

If you are receiving a compensation payment, money is usually taken out dollar for dollar from your pension.

Social work services are available at Centrelink for crisis payments.

If your care payment is rejected due to your assets, you can get in touch with Centrelink and make an appointment with a Financial Information Officer, or you can apply for 'Hardship provisions'. This will be assessed by a Complex Assessment Officer.

In order to qualify for a Disability Support Pension the person must be assessed as not being able to work for over 15 hours a week. However, once a person has a Disability Support Pension, they would be able to work for over 30 hours a week. You must be on a Disability Support Pension, Care Payment or other income support payment in order to receive Rent Assistance.

Questions and Answers:

Q1. Can you back pay the Carers Allowance?

A1: From 1 July 2006 backdating rules for Carer Allowance were standardised to a maximum of 12 weeks prior to the date of lodgement of the claim for customers who are caring for either a child or adult.

Care Receivers under 16 years of age - If the qualifying event occurred more than 12 weeks prior to the claim for Carers Allowance (Child) or Health Care Card (HCC) only, Carers Allowance/HCC only can be backdated up to 12 weeks. If the qualifying event occurred less than 12 weeks prior to the claim, CA can be backdated to the day of qualification. Backdating provisions do not apply to CA (auto).

Care Receivers 16 years and over - For a carers Allowance (adult) claim to be backdated, the qualifying event must include 'acute onset'. If the qualifying event occurred more than 12 weeks prior to the claim then Carers Allowance can be backdated up to 12 weeks. If the qualifying event occurred less than 12 weeks prior to the claim, Carers Allowance can be backdated to the day of qualification.

Q2: Can someone apply for a Carers Payment once they reach the aged pension age?

A2: YES. To qualify for Care payment the carer must:

- Satisfy the *residence requirements*
- Be personally providing constant care for an adult with a *qualifying ADAT score* in the home of the

care receiver. Note: The carer is not required to live with the care receiver.
- The carer may cease care for a maximum of 25 hours per week (including travel time) for the purposes of paid employment, self employment, voluntary work, attending an educational institution or training, and still qualify for CP. Hours where the carer continues to provide care for the care receiver *are not counted*.
- Have income and assets below the maximum allowed under the *pension's income and assets test*.

Q3: What is a Chronic Disease Management Plan?
A3: This question should be directed to Medicare as Centrelink knows nothing about it.

Q4. What elements of Centrelink assistance is available to help a DSP recipient with moving into their own home –i.e. help with rental, Telstra, gas, electricity, computer etc.
A4:
a – Rent
Rent assistance is an additional component to the payment to help paying rent.

b - Telephone
Telephone Allowance (TAL) is paid to assist customers with the cost of maintaining a telephone service. A higher rate may be paid to DSP recipients (under 21 with no dependent children) who also subscribe to a home internet service. Entitlement to TAL depends on whether or not the customer or their partner has a telephone service (with or without home internet) connected in their name and the type of payment they receive. The current rate is $25.20 quarterly.

c - Electricity
Clean Energy Advance (CEA) payments and Clean Energy Supplements are measures in the Clean Energy Future Household Assistance Package that provides financial assistance to help manage cost of living impacts from the introduction of a carbon price on 1 July 2012. These payments are non-taxable and are exempt income under social security and family assistance law.

Eligibility
The CEA and Clean Energy Supplements will be paid automatically to eligible customers who are in Australia and receiving a qualifying payment for the CEA and Clean Energy Supplements.

Income support customers who also receive Family Tax Benefit (FTB) may receive separate CEAs with their income support and FTB payments, and separate Clean Energy Supplements with their income support and FTB payments.

Rate
The amount of the CEA and the rate of the Clean Energy Supplement depend on the customer's qualifying payment type and individual circumstances.

d - Help to pay bills
Centrepay is a free and voluntary bill paying service available for customers to pay their bills directly from their Centrelink payments.

Customers receiving an eligible Centrelink administered payment can nominate to have amounts deducted from their payment for bills such as private rent, gas, electricity, water, council rates, ambulance services, telephone, education fees, funeral benefit schemes, legal fees, whitegoods and medical expenses.

Deductions are transferred directly to a Centrepay Third Party Organisation (TPO) for part or full payment for services. Centrepay was established in 1997 and is similar to, but does not replace the Rent Deduction Scheme (RDS) which allows the deduction of housing payments directly from the customer's Centrelink payment to pay State and Territory Housing Authorities.

Hereditary Hemorrhagic Telangiectasia (HHT) - WHAT IS A REGIONAL NETWORKING ALLIANCE? (USA)

Living with a rare disorder can be isolating and frightening. Whether you are newly diagnosed with HHT or have been living with the disorder for years, you don't have to face this illness alone.

A Regional Networking Alliance is a group of individuals united through a common purpose. It is an informal group of empowered volunteers that work closely with the HHT Foundation to facilitate local connections for individuals and families affected by HHT. A Regional Networking Alliances mission may vary according to the needs of the local community. The Regional Networking Alliance is meant to foster stronger links between the HHT Foundation, local communities, and individuals across the country.

The HHT Foundation has an ever-growing network of support groups called Regional Networking Alliances (RNA) operating across the United States.

Directory of Regional Networking Alliances can be found at www.hht.org

WHY JOIN A REGIONAL NETWORKING ALLIANCE
There are no limits to what can be accomplished when you enlist the power and potential of individuals working together for a common purpose. A Regional Networking Alliance can provide a forum for individuals to share their concerns and experiences. In a Regional Networking Alliance, you can discover the tools you need to utilize on a day to day basis, finding answers to your questions, and obtaining access to resources and services sharon.williams@hht.org. Our goal is to have one RNA for each state; therefore we have adopted a policy to name each RNA group based on the state in which the Regional Coordinator resides.

Regional Networking Alliances are critical to the mission of the HHT Foundation. We recognize and value the Regional Networking Alliances local expertise, their ability to reach out to others affected by this disorder in their area, their understanding of this disorder, and their ability to make a real difference. The HHT Foundation will work hand in hand with the Alliances to provide support and resources when and where needed.

CONFERENCES

International Congress of Porphyrins & Porphyrias - an International Porphyria Patient Day will be held on Saturday 18th May 2013 at the conference center KKL in Lucerne Switzerland. www.porphyrinsandporphyrias.org/patient-day.

For further information about the **Congress of Porphyrins and Porphyrias** and the latest scientific program, please consult the official congress website: www.porphyrinsandporphyrias.org

Ehlers Danlos Conference
Date: 19th-21st July 2013
Venue: To be advised
Contact AGSA for details.

Beckwith Wiedemann Syndrome (BWS) Conference 2013 Sydney Australia
Date: 30 October – 1 November, 2013
Venue: Novotel Sydney, Parramatta
350 Church Street
Parramatta NSW 2150
The conference is aimed at providing updates on the latest research into BWS, clinical procedures, the screening protocol and sharing experiences between families and medical professionals from across Australia, New Zealand and the USA.

PROFILE
A – Z OF GENETIC CONDITIONS

PROTEUS SYNDROME

(Summary of Lindhurst et al)
Researchers at the National Institutes of Health in Bethesda, MD, have recently identified the genetic alteration that causes Proteus syndrome, an overgrowth condition thought to affect fewer than 1000 people worldwide.

Proteus syndrome causes unequal overgrowth of various body structures. Scientists studying this condition believe the overgrowth seen in Proteus syndrome is caused by a genetic alteration in the parts of the body that show signs of the condition unaffected body areas have normal genes.

The NIH team, headed by Dr. Leslie Biesecker, used new gene sequencing technologies to look at the genes of many people with Proteus syndrome. They found a mutation in the AKT1 gene in tissue samples from affected areas of the body in 26 patients with Proteus syndrome.

The mutation in AKT1 was not found in people who do not have Proteus syndrome and in many tissue samples from unaffected areas of the body in patients with Proteus syndrome. The AKT1 gene produces a protein that plays an important role in regulating cell growth. The AKT1 mutation found in patients with Proteus syndrome results in an abnormal protein that causes increased cell growth and proliferation.

Researchers are hopeful that understanding the genetic cause of Proteus syndrome will lead to better diagnosis and improved treatment options for patients with this rare condition.

Questions and Answers about the Genetics of Proteus Syndrome

What is the genetic cause of Proteus syndrome?
Researchers at NIH have recently learned that Proteus syndrome is caused by a mosaic alteration, or mutation, in a gene called "AKT1." The NIH research group, led by Dr. Leslie Biesecker, recently published their findings in a journal article in the Name of Journal, and you can find a copy of the complete article here. This fact sheet will answer some questions people may have about this discovery.

What is a "mosaic" genetic alteration? Our bodies are made up of millions of different cells. Each cell has its own copy of the genetic code in it that serves as the cells' instruction manual. A mosaic gene alteration is a change in the genetic code that is present in some of the body's cells but not others.

Scientists studying Proteus syndrome have previously hypothesized, or guessed, that the genetic alteration responsible for causing the condition would be mosaic. This is because of the way that Proteus syndrome affects people – some parts of the body appear to be affected with the condition while other body parts appear normal. Also, no two people with Proteus syndrome are affected in exactly the same way.

How did researchers find this gene alteration?
The NIH team, with financial support from the Proteus Syndrome Foundations of the US and the UK, used new genetic sequencing technologies to look at all of the genes of a few patients with Proteus syndrome and some of their relatives. By reading through, or "sequencing," all of these individuals' genes, they found that one gene, AKT1, was altered in patients with Proteus syndrome but normal in unaffected relatives.

The NIH researchers tested the AKT1 gene in 29 patients with Proteus syndrome and they found an AKT1 mutation in 26 patients. The source of the sample, that is, where the DNA used for the test came from, turned out to be very important in this study. When the NIH researchers tested the AKT1 gene in cells that came from clearly affected parts of the body, such as an abnormal skin biopsy, they found an AKT1 mutation in about 75% of the samples. When the cells of clearly *unaffected parts of the body were tested, they found* the mutation in only about 30% of the samples. Only very specialized testing was able to detect this mutation in blood cells, and only two patients' blood cells were positive using this specialized technique.

These results match what scientists have believed about Proteus syndrome: the genetic alteration that causes the condition is not present in all of the cells of the body. This makes genetic testing for AKT1 mutations in people with Proteus syndrome complicated because the sample that used for testing may not actually have the gene change in it or it may be present in such low levels that are not able to be detected.

What is the AKT1 gene and how is it altered in people with Proteus syndrome? Genes are the body's instructions for growth and development because they provide the blueprint to make proteins, chemicals that are responsible for all the body's important functions.

AKT1 is a gene that makes a protein that acts like a switch that controls when cells should grow and die off. People with Proteus syndrome have an altered AKT1 gene in some of their cells. This altered gene makes an abnormal protein. The official name for this alteration is "c. 49G>A, p.Glu17Lys," and it is called an "activating mutation."

This means that the AKT1 gene has "spelling error" that causes an abnormally active protein to be made in the body. This active protein is thought to increase rates of cell growth and may prevent cells from dying off when they naturally would. Researchers think that this helps to explain why patients with Proteus syndrome experience overgrowth and are at an increased risk to develop tumors.

What does this discovery mean for people with Proteus syndrome? Understanding the genetic cause of Proteus syndrome could lead to significant advances in diagnosing and treating this rare condition, and there is reason for patients and their families to be excited and hopeful about this discovery. For researchers, this discovery will help focus efforts to develop animal models of Proteus syndrome, test new drugs and other therapies, and gain a better understanding of the complications seen in many patients.

Fertilized egg divides into many cells to form an embryo arise. Fertilized egg divides into many cells to form an embryo. As the cells continue to divide, the DNA in one of the cells become altered.

The AKT1 gene in one of the cells changes – where the DNA code should have a "G," it has an "A" instead.
As the cells of the growing embryo continue to divide, the number of both the cells with a changed AKT1 gene and the cells with an unchanged AKT1 gene expand and contribute to the formation of organs and tissues
The developing baby has two types of cells. Some have the normal AKT1 gene and some have the altered AKT1 gene.

The parts of the body that developed from the cells with the altered AKT1 gene grow differently than normal cells. This is why the body parts of people with Proteus syndrome are unevenly affected.

EVENTS

On the 28th of February all around the world people embraced **Rare Disease Day**, to create awareness for rare diseases and hopefully bring about public attention to the need of continuing research and funding in this area. To get involved, AGSA took part in a collaborative event with the Steve Waugh Foundation and BEEHAC at the Sydney Cricket Ground. Despite being a fairly early start (6am!), the event went of famously, and was featured on Channel 9's TODAY Show weather broadcast. Those with rare disease from all over NSW attended the event, and got to meet some famous faces such as Georgie Parker, Channel 9 weather man Stevie and Steve Waugh. Everyone had a marvellous time munching on their sausage sizzle with the stars, and taking part in the various activites that were planned. Overall it was a great day and hopefully raised awareness for rare diseases.

NEWS IN RARE DISEASES AROUND THE WORLD

Rare diseases not well treated in China

24/02/13 21:39

BEIJING - Nearly 10 million patients suffering from rare diseases in China are facing difficulties receiving proper diagnosis and treatment, a medical expert said on Sunday.
Speaking at a conference on rare diseases in Beijing, Ding Jie, vice director of the Peking University First Hospital, said many patients failed to get a correct diagnosis. This is because of doctors' limited medical knowledge about rare diseases, most of which are genetic and therefore difficult to confirm.

It is also difficult for diagnosed patients to receive timely treatment due to a shortage of effective drugs, most of which are imported from foreign countries. They usually take a long time to receive approval and get to market, Ding said.
Some patients gave up treatment even though imported medicine was available, Ding said, adding that costly drug prices as well as incomplete medical security are major barriers.

According to the World Health Organization, a rare disease is one that affects a small percentage of the population, ranging from 0.65 in 1,000 to 1 in 1,000. More than 6,000 rare diseases have been confirmed so far.

AGU is partner in international campaign of rare diseases

24/02/13 12:46

BAHRAIN - Under the care of HE the Bahraini Minister of Health, Dr. Sadiq Shihabi Yahya and HE Dr. Khalid Abdul Rahman AL -Ohaly, the President of AGU, Al Jawhara Center for Molecular Medicine / Arab Gulf University is celebrating the International Day of Rare Disorders.

Dr. Cristina Skrypynk, chair of the Rare Disease Day Bahrain 2013 Organizing Committee, said, "The Al Jawhara Center for Molecular Medicine and Genetic Disorders, in collaboration with Primary and Secondary Care Departments from the Bahrain Ministry of Health, will host on Saturday, February 26, 2013 a scientific symposium. Also, a public awareness campaign for rare diseases will take place on Monday, February 28, 2013 at Seef Mall, in Manama."

Dr Cristina mentioned, "Rare disorders are scientifically defined as diseases that affect one person out of every two thousand people, most of the diseases causes are genetic and ways of cure are unfortunately unknown. More than 100 million people are suffering from rare disorders all over the world. Studies have estimated that around 20 million people are suffering from rare disorders out of about 240 million Arab countries people from."

So far, more than 700 genetic phenotypes or diseases have been recorded in the Arab population. The center of Arab Genomic Studies (CAGS) estimates that Arab countries spent about $30bn a year on their patients suffering from hereditary diseases. Most rare diseases are genetic, and thus are present throughout the person's entire life, even if symptoms do not immediately appear.

Dr. Cristina added, "Spread of the phenomenon of consanguineous marriages in the Arab societies, aging parents and high birth rates in the same family, all contribute to the growing of rare disorders, and therefore the awareness of these consequences should be major goal of all interested people in health care."

Furthermore, the International Day for Rare Disorders was organized for first time in 2008 by the European Organization of Rare Diseases (EURORDIS) and since then more than 1000 events have been organized around the world with the participation of hundreds of thousands people from 63 countries participated in 2012 campaign.

These campaigns have contributed to the development of national strategies and policies for rare diseases in a number of countries around the world. From the GCC countries, only UAE participated to the 2012 Rare Disease day event.

This year, the International Day for Rare Diseases is celebrated in the Kingdom of Bahrain throughout a symposium on the 26 of February and a public awareness campaign on the 28 of February. The events are organized by a team from Al Jawhara Center/Arabian Gulf University and from Bahrain Ministry of Health, primary and secondary care units.

Dr. Cristina said, "The main objective of this campaign lies in raising awareness among the public and decision-makers, researchers, and involve them in contributing to alleviate the suffering of these people affected by rare disorders, and the abolition of the borders between the various parties local and global in order to exchange experiences and collaborate in developing effective solutions for these chronic and severe diseases."

As an action of celebration of the World Rare Disease Day 2013, LYSOGENE announces the launch of its new website-section entirely dedicated to Patients,

their Families and Patient Organizations (PO's)

28/02/13

PARIS – LYSOGENE today announces the launch of its website-section entirely dedicated to Patients and Patient Organizations (PO's) as a sign of its endeavour to value all those living with an MPS III condition and those who incarnate their voices. With this new action, LYSOGENE wishes to reaffirm its passionate commitment to fight against heavy-debilitating and life-threatening rare diseases with high unmet medical needs and to bring valuable treatments to s and families in high demand.

The European Commission will provide €144 million of new funding for 26 research projects, with the objective to deliver 200 new therapies for rare diseases by 2020, the EU's executive announced today

28/2/13

The projects, announced on Rare Disease Day 2013, are expected to help improve the lives of some of the 30 million Europeans suffering from a rare disease.

The goal is to pool resources and work beyond borders in order to get a better understanding of rare diseases and find adequate treatments.

The Commission is hoping that the projects, which will be conducted together with national and international partners, will deliver 200 new therapies for rare diseases and the means to diagnose most of them by 2020.

Over 300 participants from 29 countries in Europe and beyond will be brought together in the selected projects, including teams from leading academic institutions, smaller businesses and patients' groups.

"Most rare diseases affect children and most of them are devastating genetic disorders resulting in greatly reduced quality of life and premature death," said Máire Geoghegan-Quinn, the EU Commissioner for Research, Innovation and Science.

"We hope that these new research projects will bring patients, their families and health professionals closer to a cure and support them in their daily battle with disease," she said in a statement.

Better diagnoses to improve lives

The 26 new projects cover an array of rare diseases including cardiovascular, metabolic and immunological disorders. They will aim at developing substances that may become new or improved therapies for patients and understanding better the diseases' origins and mechanisms.

The projects will also try to help a better diagnosing and improve the management of rare diseases in hospital and healthcare settings.

Teams will work on varied challenges, including a new 'bioartificial' liver support system to treat acute liver failure, powerful data processing operations to develop novel diagnostic tools and biomarkers and screening strategies for therapeutic agents against rare kidney diseases.

They will also work on the clinical development of a drug to treat alkaptonuria, a genetic disorder which leads to a severe and early-onset form of arthritis, heart disease and disability for which there is currently no effective treatment.

Many of the new projects will contribute to the International Rare Diseases Research Consortium (IRDiRC) which is the biggest collective rare diseases research effort in the world. The new projects will bring the number of EU-funded collaborative research projects related to rare diseases to close to 100 over the last six years. Altogether, they represent an investment of almost €500 million.

EC promises $187M for ambitious effort to spawn 200 new rare disease drugs

5/3/13

A few days ago the European Commission mapped out a plan to help spur development of 200 new treatments for rare diseases by the year 2020. And the Commission put its money where its mouth is, committing $187 million for 26 research projects.

In full swing, the research initiative aims to bring together some 300 investigators--academics, biotech and pharma investigators--from 29 countries to work on new treatments related to cardiovascular disease, acute liver failure, rare kidney disease and more. And it adds to a series of initiatives that now account for 100 projects and about $650 million in EC financing.

"Most rare diseases affect children and most of them are devastating genetic disorders resulting in greatly reduced quality of life and premature death," said Máire Geoghegan-Quinn, the EU Commissioner for Research, Innovation and Science, in a statement. "We hope that these new research projects will bring patients, their families and health professionals closer to a cure and support them in their daily battle with disease."

Rare diseases have become one of the central focuses in the biopharma R&D world. Everyone from GlaxoSmithKline ($GSK) to a host of startups believe that

the promise of a more efficient development program combined with a much better understanding of the genetics of rare ailments make this a growth area capable of spawning dozens of new products. And while the patient populations are small, costs associated with these drugs can be among the highest in the industry.

Draft NDIS Rules released for comment

5/3/13

The Australian Government today released draft Rules which support the NDIS Bill 2012. These Rules provide more detail on how the NDIS will work. They cover:

- becoming a participant (eligibility);
- supports for participants (including the criteria for assessment and deciding which supports will or will not be funded);
- registration of providers;
- plan management;
- the appointment of nominees;
- how children's interests will be determined; and
- the protection and disclosure of information.

Feedback on the draft Rules can be made until 23 March. NDS urged the Government to release the draft Rules for consultation prior to their tabling in Parliament and welcomes their release. We are pleased to note that the draft Rules respond to some of the concerns we have been raising. For example, we have argued that people with disability should be able to choose a service provider to be their plan manager (providing that safeguards are in place). The draft Rules allow this to happen.

NDS will scrutinize the draft Rules and make a submission as well as meet with officials during the next fortnight. Members wishing to provide input to the submission are asked to contact Philippa Angley at philippa.angley@nds.org.au or 03 8341 4302, or Tessa Thompson at tessa.thompson@nds.org.au or 03 8341 4305 by 20 March.

Yesterday NDS appeared before the Senate Committee inquiring into the NDIS Bill. The Committee questioned NDS in detail on many aspects of the Bill (and the Rules) including: registered providers; workforce training; the need for capital funding for accommodation; services which should be block-funded; fundraising; and the eligibility age limit.

The NDS submission on the NDIS Rules Consultation Paper is now available. NDS thanks those members that provided input.

PERSONAL STORIES

Funicular Myelosis

I am a 65 year old male and my journey began in October 2003, although according to my Neurologist it was undiagnosed for about 10yrs. I was lying in bed and I was unable to move my legs, and had severe tremors so much so that I was hospitalised.

During my time in hospital I was found to be severely B12 deficient, and was suffering. They diagnosed me with ***Funicular Myelosis***. The way they found this out was by a myriad of tests and standing upright legs together and closed my eyes – I just fell over.

I was then put on a monthly regime of injections, all was good for about 3 months and then the stiff muscle ataxia cramps, loss of various movements, oedema etc started. So my specialist increased the injections to fortnightly and I was put on an exercise programme at the Rehab department of the hospital, oh and by the way I was told I was not allowed to drive.

So I started to get on with my life, bought myself a mobility scooter to get around on and then I started to have intermittent paralysis of both legs, arms and hands. I could not eat a meal without the arm paralysis happening so now and then I was unable to go anywhere without a carer, in most cases my wife taking me. I am unable to use my scooter any more.

I underwent further tests MRI, SSEP (evoked potentials) Nerve conduction tests, EMG, specialised Blood tests all normal, so of course what does the good DR say? It's all in your head. My own GP and the people who saw me at Rehab did not agree with his diagnosis, so I had further Nerve Conduction Tests. They too were normal. Also, to enable me to again become independent, I now have my motorised wheelchair using chin control, and it is so good to have regained my independence. I have nicknamed it THE BEAST, as it is very large.

My condition has improved in regards to my arms and hands but as soon as I do any exercise or walking my legs become very heavy and painful and the paralysis episodes occur, or my legs just collapse below me. I was having physio every 2 weeks at my local hospital, this has now been stopped. I then started having very severe upper body spasms. Anything I was carrying would go flying, you should see the roof in my lounge where the TV controller has hit it, and these were up to 6 times a day at one point then 5-6 times a week and now with medication I have to

this point reduced thanks to 2700 mg Gabapentin and 75mg Dantrolene daily.

My new neurologist thought I had a variation of Stiff Person Syndrome and at least two other neurological deficits. Blood tests were unremarkable. Have had further EMG testing done, needles in hand, thigh, eyelid and left of off spine. These results were also unremarkable. Just had a full spine and brain MRI. I then started having very severe leg spasms - whole leg and feet twist violently inward resulting episode pulling the muscles all the way to my hip, very painful. Had started hydrotherapy on weekly basis with constant supervision, during this time my condition became much worse, my legs hands arms and feet would invert to the point of agony.

Then in late June I presented to my local hospital in terrible agony, but I had the presence of mind to tell them to contact my Neurologist, and she arranged for me to be transferred to one of the top hospitals in Australia, under the Professor of Neurology. Then I was put through so many tests, Ultra Sounds, CT Scans etc. At this time I was in a very deep depression. I was shell shocked, did not know what was going every time I tried to walk my legs collapsed below me. Eventually through medication and my own ability to better control the spasms things started to get better, even the geniuses at the hospital would still not give me a definitive diagnosis. I lay between Stiff Person Syndrome and Isaacs' Syndrome or a syndrome called after me, Stiff Person being the most likely.

I was then transferred back to my local hospital to begin rehabilitation. They had to teach me to walk again, something we take for granted. This was incredibly hard as you have to retrain the brain again. They also gave me access to a psychologist. This along with the visits from the pastoral care worker, who actually was a friend from my Church brought me out of my deep depression. During these sessions I really opened up and laid all things bare. I was then allowed to return home but was still having to use my walker or wheelchair, and had to have services put in place to shower me etc. This was tough for me as I am a very independent person, and stubborn as mule.

Physiotherapy is ongoing and has helped me immensely to get fitter. It took me 6 months to be able to walk confidently without any aid and to walk up some stairs. I am not allowed to walk outside for fear of seizure or falling, and I still must have a carer with me at all times, my beloved wife usually fulfils that demanding role.

My disability does not define me and has been in many ways been a blessing. It has allowed me to be a very significant part of my grand children's lives and to volunteer in many capacities, and to advocate for others with disabilities and their carers so often the unsung heroes of our society.

Rare disease a challenge

By Kerrie Armstrong Feb. 26, 2013, 2:03 p.m.
Rare Disease Day February 28, 2013 Camden Advertiser

DAVID Napier is a rarity — so rare he is literally one in a million. But not in a good way. The Camden resident has stiff person syndrome, and it has taken him 10 years to find a doctor that could treat him.

"I was the first person to be diagnosed with it at St Vincent's Hospital in 12 years," Mr. Napier said. He hopes everyone will spare a thought tomorrow for people like him as part of Rare Disease Day. He said it was difficult for people with rare and unknown diseases to get media coverage, fund-raising for research or support.

"It's fantastic what we do to support different organisations and diseases, but you don't get that for people with rare diseases. There is no funding, no research." Mr Napier said he was finally able to track down the specialist he now sees with the help of the internet. He uses the internet to keep in touch with other people diagnosed with rare diseases through support group chat rooms. "Most of the support and knowledge we get is from each other," he said. "There are only 10 or 12 of us in Australia."

Treatment is also expensive, he said. "The medication you need is not covered by the Pharmaceutical Benefit Scheme. The medication I am on costs $138 a month. Because I am over 65 I don't qualify for rebated physio sessions in the hospital."

Tanya Jarvis - I also suffer from this rare disease but I live in the USA. My best friend that I met on the Internet on NORD lives in Australia we have been friends since 2007 when I was diagnosed. We really need so much more awareness all around the world. In 2005 there was very little or no help available, so in 2009 I created SPS - Australia - on face book. There are now 194 people who follow this page from all around the world. I agree with David regarding the lack of awareness for all these Rare Diseases.

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