

Supporting Australians with Paroxysmal Nocturnal Haemoglobinuria

# RARE BUT REAL

NEWSLETTER ISSUE NO4 MARCH 2014

## Event Spotlight

# Rare Disease Day, 28 February 2014

**O**n 28 February 2014 the Genetic Support Network of Victoria (GSNV) hosted 'Rare Disease Day' at Murdoch Childrens Research Institute in Melbourne. The invitation was open to medical professionals, rare disease support groups and charities, and any other person with interest in rare disease. The aim was to encourage professional awareness around this important day.

The topic for discussion was 'responsibility' and what responsibility means to those suffering from, or involved with, a rare disease. The event featured four key speakers each with different views on 'responsibility'. Guest speakers for the morning included Dr. Sue White, Clinical Geneticist at Victorian Clinical Genetics Services, Nicole Millus, Director at Mucopolysaccharide & Related Diseases Society Aust. Ltd, Mandy Jacobs at Australian NPC Disease Foundation Inc, Flora Pearce, Metabolic Social Worker at Victorian Clinical Genetics Services.

Mandy Jacobs and Nicole Millus shared their personal stories from the point of view of mother with children who had inherited a genetic rare disease as well as from the support association role of their respective foundations. Each discussed their experiences of guilt and responsibility for their child's diagnosis, which can be, for the most part, overwhelming. Further, both discussed the struggles with the lack of material, research and support in the respective genetic rare diseases and how they both became empowered to either establish or join support groups to promote awareness of the rare disease, information and ongoing support at various levels.

Dr. Sue White and Flora Pearce spoke in turn about the responsibilities Clinicians have in aiding families coming to

"Rare Disease Day – Murdoch Childrens Research Institute in Melbourne"



terms with a rare disease diagnosis, addressing issues of guilt one may feel who may either be the carrier or have a special relationship with the person who has been diagnosed. Both explained the struggle people face admitting they have these feelings and how different clinicians work with families and supporters to assist them in reaching a level of acceptance.

Before conclusion of the morning each speaker formed a panel for Q&A from the guests. While many issues were raised, none more prevalent than that of the lack of information provided on diagnosis whether that is because the disease is rare or that some medical staff are not aware of new information or support groups and networks formed. This point highlighted the importance of individual support groups for rare disease and the desperate need for a National Plan and rare disease policy, similar to that in Europe, to end the ad-hoc treatment by Government to those suffering a rare disease, end the tiresome lobbying, and obtain the medical care needed to improve the quality of life.

## A few words

# A message from the President

I would like to wish all our members and supporters a very Happy New Year and I hope you all enjoyed the holiday period including watching the Australian Cricket team's great win in the Ashes.

2014 is shaping up to be a big year for the PNHSAA and our members with the Committee's focus in the first half of the year is increasing the awareness of PNH and the PNHSAA in conjunction with other rare diseases with Rare Voices Australia.

For the second half we are calling for member and supporter interest for a proposed 2014 National Conference to be held in Melbourne in August. Further details can be found in this newsletter. Given the success

of the National Conference in 2012 all the Committee members are encouraging participation so planning may commence for another informative and rewarding day for our all our members.

Finally the PNHSAA website has been up and running for nearly one year now and we are continuing to add more information and links every couple of weeks so I urge you to please keep looking and write to us with any information requests.

As always a big thanks to the Committee for their efforts last year and thank you to all those members who continue to financially support us as without you there would be no PNHSAA.



Matt Dean, President

## Forming alliances

# Rare Voices Australia

Rare Voices Australia ('RVA') is a non-for-profit organisation that was established in February 2012 to be the united voice for all Australians living with a rare disease. The aim of RVA is not to replicate the services that are already being provided by individual disease groups (such as PNHSAA) but rather to act on a national scale improving big picture issues such as identifying policies that need to be implemented or modified to support patients with a rare disease and then lobby government to bring about the required change. One of the big projects RVA is working on is implementing a national plan on how our health system manages rare

diseases in Australia. This National Plan aims to improve diagnostic techniques, patient access to treatment or supportive therapies as well as regulate how data is collected so that research opportunities in the space can

be improved and allow collaboration with international bodies. Our PNHSAA Secretary, Jenny Sturrock has recently joined the Board of RVA and hopes to bring change to the LSDP's process of funding orphan drugs. Stay tuned!

For more information on RVA contact [info@pnhsaa.org.au](mailto:info@pnhsaa.org.au) or RVA direct at [admin@rarevoices.com.au](mailto:admin@rarevoices.com.au) or [www.rarevoices.com.au](http://www.rarevoices.com.au).

## Calling all members

# National Conference 2014

Following the success of the 2012 National Conference held in Melbourne and post-conference survey results indicating that members of the PNH Community wish to meet frequently, we are calling for Expressions of

Interest from Members for attendance to the proposed 2014 National Conference to be held in Melbourne on Saturday, 30 August. To register your interest, contact us at [info@pnhsaa.org.au](mailto:info@pnhsaa.org.au) or 0432 321 207.

**Share your story**

**Linda Charlton, former PNHSAA President**

*In 2010 Linda Charlton formed the PNHSAA with four others, to bring the PNH sufferers and supporters together as a community and to lobby government to approve funding of Soliris (eculizumab) on the Life Savings Drug Program. Since her departure as president and committee member from the Association we caught up with Linda to find out what she has been up to.*

I moved to Melbourne four years ago and was warmly welcomed by everyone, particularly Grace Dean and her family. I got a part-time job as a receptionist at an osteopathic clinic in Moonee Ponds, but after a couple of months most of my time ended up being taken by the Soliris campaign.

After the funding was secured, I found myself completely tapped of energy, physically and emotionally. I was really sick of being a PNH patient, telling people how dangerous and debilitating a disease it is and I was angry at the politicians who had kept back treatment funding to balance the budget. I felt I had done all I could for the support group at that time and chose to step down to recover and return to study.

In 2005 I was studying remedial massage but had to stop while the doctors figured out my diagnosis. In 2011 I was finally ready to re-enrol and went a step further to qualify as a Myotherapist in December 2012. It was an easy transition from behind the desk to treating clients at the clinic and I've been lucky enough to work with the athletes of Melbourne Victory FC.

In October 2011 I enjoyed a month in the US visiting my father and have had a chance to explore Melbourne's restaurants, cafes, art galleries and festivals with my partner Jeff and friends. For the past 6 months I have struggled with some unrelated health issues, but the PNH is stable, although I still need the odd transfusion. It's been a busy four years which has seen me transition from Brisbane Lions to a North Melbourne fan and become a master at pub trivia. I enjoy keeping fit with yoga and my latest challenge is studying classical ballet.

*The Committee thanks Linda for her continued support and sharing her story. To share your story for our next newsletter, email us at [info@pnhsaa.org.au](mailto:info@pnhsaa.org.au).*



**Share your story**

**Antony Gottlieb, Member**

*Antony Gottlieb has been a dedicated member of the PNHSAA and we were all too pleased to share his story on the 60th anniversary of his visit to his treating hospital.*

It's been nearly 3 years since I was diagnosed with PNH. So how has my life changed in this time, well thanks to the amazing results from the drug I'm treated with I feel healthy and full of energy to take on most of what daily life throws up. I'm really into fitness training and love that I'm still able to train on a regular basis 4 times a week with other training partners. I am also a huge foodie and really enjoy going out trying new cafes and socialising with friends on a regular basis without any hindrance from my condition. While my treatment for PNH gives me no side effects and no pain, what it does provide me is a constant reminder of how lucky I am to be able to carry on the way I am, even with a chronic disease. In a few days time I will be making my 60th visit to the Day oncology center at the hospital I attend every fortnight (not that I'm keeping count, but hospitals keep lots of

records). Yes by now I'm very accustomed to the whole procedure of the regular infusion I get, and to be honest it's no big deal, especially given when I look around at all the other patients at the hospital who have their own fight to stay healthy and for some unfortunately the fight to stay alive. It is extremely sobering and humbling for me to have this constant reminder and it certainly puts most problems I have in life well into perspective. Visiting my specialist haematologist every couple of months in itself is more to me than just a catch up to see how I'm tracking, I know I'm exceptionally lucky to hear a good report from him as we discuss over my test results as I know over the last couple of months he has had to deliver plenty of terrible diagnosis results to many patients. So me walking into my oncology appointments and walking out an hour/hour and a half later and being able to go visit friends, go back to work or even go to the gym and of course the best bit eat whatever I like, is a huge blessing and one I will never take for granted ever.

## Show your support

## Campaigning to save Australians living with aHUS

The aPSGA (aHUS Patient Support Group Association) represents people living with, or caring for someone with the ultra-rare, life-threatening disease, aHUS, which can damage vital organs, including the kidneys, heart and brain. An estimated two-in-one million Australians are living with aHUS. Ten per cent of all people with this devastating disease die within the first year of diagnosis.

There is only one clinically proven, life-saving and life-transforming treatment available for aHUS. Soliris (eculizumab) stops disease progression and prevents premature death and vital organ damage in patients with aHUS. The treatment is the same for that which was funded PNH, through the Government's Life Saving Drugs Program (LSDP) in January, 2012.

There are no alternatives to Soliris. Tragically, without this treatment, one-in-two aHUS patients will die, require dialysis or develop permanent kidney damage within a year of diagnosis.

In March 2013, the Pharmaceutical Benefits Advisory Committee (PBAC)

rejected the manufacturer's submission to fund Soliris through the LSDP. The treatment will again be considered for funding at the PBAC's next meeting, scheduled for between March 11-14, 2014.

In the lead-up to this decision, the aPSGA will be mounting a national media awareness campaign, coordinating ongoing MP outreach and producing an online submission to urge the Government to fund Soliris through the LSDP for those patients who require urgent access to this life-saving treatment.

To help save the lives of the two-in-one million Australians living with aHUS, sign our petition at [www.ahus.com.au](http://www.ahus.com.au). To lend further support to the aPSGA, submit an online submission to the PBAC on behalf of the small group of Australians living with aHUS requiring urgent access to Soliris.

To learn more about the aPSGA petition, online PBAC submission, or to lend your voice or support to this worthy cause, contact aPSGA on [info@apsga.org.au](mailto:info@apsga.org.au).

## Event spotlight

## Annual General Meeting 2013

The Annual General Meeting (AGM) for 2013 was held in August in Melbourne and received good attendance including several people phoning in from interstate. The AGM provided the opportunity to reflect on the 'year that was' including the 2013 re-branding of PNHSAA and the new website, growing membership and feedback received. It also provided a forum to discuss the Committee's plans for 2014, particularly the proposed 2014 National Conference.

The Committee Members voted in for 2014 are:

**President:** Mathew Dean

**Treasurer:** Michael Patterson

**Secretary:** Jenny Sturrock

**Committee Representatives:** Grace Dean & Chhean Khoun

The Committee looks forward to another successful year in 2014.

## Committee spotlight

## Meet Mathew Dean, President

In my third year as president of the PNHSAA, I thought it was about time I shared a little about myself. I am a property Development Manager in Melbourne within the commercial sector which is a new role for me commencing only this year as previously I worked as a Project Manager in construction for over 10 years. The change has been an exciting and challenging one.

I have been happily married for three years to Grace Dean (Committee Member) and my passion is travelling, recently spending 3 months in Europe (eating) and Nepal (hiking to lose the weight) where I completed the Annapurna Base Camp Trek. My other interests are photography, music, exploring Melbourne's fantastic restaurant scene, and sport

(spectating mostly rather than participating) with my favourite being cricket, you can't beat Day 1 of the Boxing Day test at the MCG.

Lastly I would like to touch on how personally being involved in the PNHSAA is for me. After Grace was diagnosed with PNHSAA in 2008 there was no immediate support for her and no readily available answers or information for either of us. Usually what was available was out-dated or overseas based. Bringing about a change as to how information is shared to the PNH Community is something that I am privileged to be a part of and grateful to the Committee and the broader PNH community for all the support and encouragement.

## Get involved Calling all volunteers

At times information cannot be readily available or updated and events may not be organised as timely as the Association would like. We appreciate the support and assistance of volunteers to aid us in bringing to our members the information, material and events you have asked for. Your support can be as minimal or as 'hands on' as you would like. Please contact us at [info@pnhsaa.org.au](mailto:info@pnhsaa.org.au) to become a volunteer.

## Team support Become a member

Become a member and support the Association which aims to help connect Australians affected by Paroxysmal Nocturnal Haemoglobinuria, increase awareness and understanding of the disease, and advocate best possible care for patients in order to reduce the confusion and isolation that comes with a PNH diagnosis. To become a member, email us at [info@pnhsaa.org.au](mailto:info@pnhsaa.org.au).

## Hello We'd like to hear from you

The PNHSAA would like to hear from its members with suggestions or fundraising ideas, newsletter stories or other charity events the Association can get involved in. Email your suggestions at [info@pnhsaa.org.au](mailto:info@pnhsaa.org.au).

## Power of money Donations are welcome

As we are a small Association we have very limited funding to assist us in providing support services, information and hosting educational and social events for our members around Australia. We appreciate your support and donations to PNH Support Association of Australia Inc. BSB 013-423 Account No:2545-47305

## The big T: Thank you

The Committee wishes to thank all of its members which are made up PNH patients and supporters for all the support, encouragement and donations made to the Association this financial year. We look forward to evolving and growing our community.