Gateway

News from Huntington's New South Wales

Volume 18 No 2 **Winter 2015**

Save the Date

WALK4 HOPE

HUNTINGTON'S AWARENESS MONTH
SEPTEMBER

PARRAMATTA PARK
Sunday 27 September

SPEER'S POINT
Saturday 19 September

Our 40th Anniversary celebrations continue with our 2nd Walk 4 Hope.
To date there are two walks scheduled for Huntington's Awareness Month in September.
Keep in touch by visiting our website www.huntingtonsnsw.org.au and Facebook page
We look forward to you, your family and friends joining us.

Huntington's NSW 1975—2015

blue illusion®

Thursday 2nd July
6pm-8pm
Fashion Parade and
Shopping Night
at
Blue Illusion
258 Darling St Balmain

Bubbles, nibbles and goodie bags
Come and join us there and support Huntington's NSW

For more information contact Pauline at paulinekeyvar@fams.net.au

Bike Riding 'Round Australia for HD - Update

Since our last newsletter, Madison has raised in excess of $22,000. It's not too late to support him on the leg home by making a donation at https://give.everydayhero.com/au/mad-1

We'll be cheering him on when we arrives back home in Terrigal on 20th June.

Mad Hatters T Party raises over $20,000

Wow, what a night. The Mad Hatters T Party was so much fun with over 130 people getting into the mood at the Intercontinental Hotel as part of our 40th anniversary celebrations.

Our President, Brian Rumbold with his wife, Margaret, Alex Greenwich and Pauline Keyvar

Guests arrived wearing a magnificent array of hats and were welcomed with a glass of champagne. We were entertained by the Mad Hatter and Charisma Belle and we were delighted that Alex Greenwich, the Member for Sydney, was able to join us.

Holly Faulkner and Dr Clement Loy

There was frenzied bidding during the live auction and a fashion parade of amazing hats by Philadelphia Philpot. Overall we raised over $20,000 and all funds raised will be going to the establishment of a Youth Support Program.

Special thanks must go to the fundraising committee, Penny Bachell, Glenette Jeffrey-Konig, Kairen White, Anna Falkinder and Sally McKay who were so ably led by our Fundraising & Marketing Consultant, Pauline Keyvar. We are most grateful for all their hard work, their enthusiasm and commitment.

Beautiful hats from Philadelphia Philpot

It's time to renew your membership!!

Yes it's that time of year again. Membership renewals for 2015/2016 are now due. For $22 per year (including GST) you will continue to receive research news, practical advice and updates in *Gateway* and have unlimited access to new publications, special events and formal and informal support networks offered by the HD community.

A strong membership will ensure that the Association continues to be representative of, and relevant to, people affected by HD in NSW and the ACT. So why not encourage another family member or friend to join?

A membership form is enclosed for you to complete and return with your cheque/money order/credit card details.

As the 30th June is fast approaching, it's also a good time to make a tax deductible donation to further the work of the Association. We would be most grateful!

"I refuse to let this disease get the better of me"

Wellington woman leads the fight against Huntington's disease

A 25 year old local woman Amy George has tested positive to the Huntington gene. A killer brain disease which has no cure but she's fighting not for herself but the many others who are fighting the rare disease.

The young mother knows one day the disease may take her .

'I refuse to let this disease get the best of me. Being an advocate takes all my frustrations, angers, and fears about Huntington's and turns it into a positive. I have hope and believe in a cure of Huntington's Disease. In order to find better treatments and a cure there needs to be more awareness and money raised. So here I am vowing to wear something Blue until the end of the year to make some noise about HD. I'm hoping to raise $6000, but I'd love to raise much, much more."

"In 2009 I tested positive for the Huntington gene, which mean's that at some stage in my life I will develop Huntington's Disease.

Huntington's disease is an incurable, hereditary brain disorder. It is a devastating brain disorder for which there is no currently 'effective' treatment. Nerve cells become damaged, causing various parts of the brain to deteriorate. The disease affects movement, behaviour and cognition - the affected individual's abilities to walk, think, reason and talk are gradually degraded to such a point that they eventually become entirely reliant on other people for their care. Huntington's disease has a major emotional, mental, social and economic impact on the lives of patients, as well as their families. I want to raise more awareness for HD.

It was said that HD is not a rare disease but a rarely known disease.

Sad isn't it. Such a devastating disease and it's probably something you've never even heard of."

She is asking locals and the business community to donate money and also wear blue to raise awareness.

https://give.everydayhero.com/au/hope-for-huntingtons

Acknowledgement: Wellington Times, 20th May 2015

Thank you Amy for sharing your story with your local community. Good luck with your fundraising efforts.

Some happy news to share!

In March this year our son, Cameron, was married (finally). We will always be grateful to our newest daughter-in-law, Marlisa, for suggesting that they be married at the nursing home where Bill now lives.

Bill is in the later stages of Huntington's disease, now bedridden and this was the only way he could have the pleasure of being part of the day.

At first we thought to keep it a secret and surprise him, but then decided to tell him and give him something to look forward to.

Boy, did he look forward to it, he was so very excited, for months. He may lack expression but he managed to let us know just what he wanted… the waistcoat I ordered him was slow arriving and each time I saw him he would hit at his chest and say 'where' 'where'…he was overjoyed when it finally arrived. I might add it was there weeks ahead, hanging in his wardrobe with the rest of his party gear.

Cameron didn't want a bucks night, instead 'the boys' spent the Saturday afternoon prior to the wedding with Grampa, together with a bottle of scotch … Grampa slept well that night.
The nursing home is "Jemalong" in Forbes NSW, we have to give them a plug … they turned over the chapel and the activity room and allowed us to do anything we wanted in order to make the venue as special as possible. We should also add that this is without a doubt the most brilliant facility you will ever find. The level of care and concern in every area is amazing.

On the wedding morning Bill was calling his nurses at 6am, wanting them to get him up so he would be ready and waiting, they were wonderful with him and joined in his excitement. They even took photos and had them up on his wall before the service was over.

The boys arrived early (with another bottle of scotch) to spend some time together, poured Grampa several drinks … he didn't spill a drop. He was a very proud and somewhat pickled, father of the groom.

The service was wonderful and even now if someone asks him how it all went, he points to the pictures and says only three words……'magic, got drunk'.

Thank you Susie, Bill, Cameron and Marlisa for sharing your story with us.

Do you have a Story to Share?

If you have a contribution that you wish to make to the Newsletter please send it to us at the Association office (see details on the back page) or by email to Robyn at robyn.kapp@huntingtonsnsw.org.au

Research Project: An Evaluation of Pre-Symptomatic Testing in Huntington Disease

What is this study about?
If you undertook pre-symptomatic genetic testing or supported someone who went through pre-symptomatic testing for Huntington disease between January 2009 and December 2013, you are invited to take part in this research study. The aim of the study is to evaluate the current practice in genetic services across Australia. This study will help guide the genetics services that provide pre-symptomatic testing.

Participation in this research study is voluntary. So it's up to you whether you wish to take part or not.

Who is running the study?
The study is being carried out by the following researchers:

- Tenielle Clinch, Master of Genetic Counselling Student at the University of Sydney.
- Associate Professor Kristine Barlow-Stewart, Director, Master of Genetic Counselling Program, University of Sydney.
- Fiona Richards, Social Worker, The Children's Hospital at Westmead, Sydney.
- Robyn Kapp, Executive Officer, Huntington's NSW.
- John Conaghan, Social Worker, Hunter Genetics, Waratah, Newcastle.

Tenielle is conducting this study as part of a Master of Genetic Counselling at the University of Sydney and it will take place under the supervision of the researchers listed above.
The study is being funded by the Master of Genetic Counselling program at the University of Sydney.

What will the study involve for me?
This study will involve the completion of an anonymous questionnaire. The questionnaire can be answered online, or you can request a paper copy of the questionnaire and a reply-paid envelope.
You can complete the study online at https://www.surveymonkey.com/r/HDPSTsurveyPIS
The questionnaire will start with introductory questions including age, gender, and level of education etc., followed by questions about your experience as an individual who has undertaken pre-symptomatic testing for Huntington disease. There will be a number of specific questions asking you about factual things that happened. There will also be questions that will ask you to rate whether or not you agree with a certain statement; these questions are designed to assess your opinions/views of the testing process. The questionnaire once completed can be either submitted online or posted in the reply-paid envelope supplied.
You can request a paper copy of the study by contacting Robyn Kapp on (02)9874 9777 or robyn.kapp@huntingtonsnsw.org.au.

How much of my time will the study take?
It is estimated that the questionnaire should take about 20 minutes of your time.

Can I tell other people about the study?
Yes, you are welcome and encouraged to tell other people about the study. Please feel free to send the online questionnaire link to anyone else you know who might fit the criteria of the study.

What if I would like further information about the study?
When you have read this information, Tenielle Clinch will be available to discuss any aspect of the study with you further and answer any questions you may have. If you would like to know more at any stage during the study, please feel free to contact Tenielle at tcli9579@uni.sydney.edu.au

Book Review: Inside the O'Briens

Lisa Genova

Twenty-two years ago Lisa Genova was a first-year neuroscientist when she witnessed a key moment in medical history. She was working metres from a team of doctors, including neurologists Dr Anne Young and Dr Allan Tobin, who isolated the genetic mutation that causes Huntington's disease, in her home town of Boston, in the USA.
"It was an exciting time to be witnessing this historic moment but 22 years later there is still no treatment," says Genova who has a PhD in neuroscience from Harvard.
"Huntington's disease is an orphan disease ... there is no celebrity who has it and many people don't know about it, so there's not as much incentive for drug companies to put money into developing treatment."

Lisa Genova is well known for her compelling books about the human brain. The Oscar-winning movie Still Alice, about a brilliant professor struggling with early onset Alzheimer's disease, is based on her novel of the same name. In her other novels, she has also tackled brain injury in Left Neglected and autism in Love Anthony.

Genova wanted to ensure her next novel, Inside The O'Briens, shone a spotlight on another worthy cause.

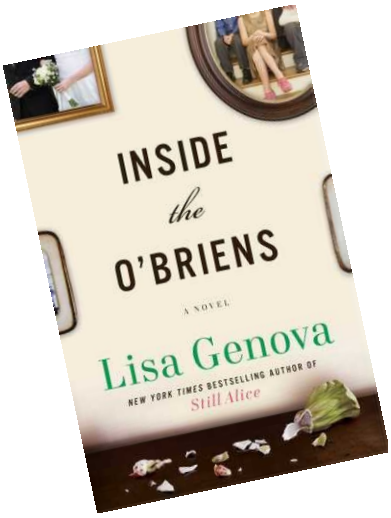
The story centres around a family that carries the gene for Huntington's disease. Joe O'Brien, a Boston police officer and father of four adult children, is 44 years old when his wife first notices that something is different about her husband.

He drops things, he fidgets and he has sudden temper outbursts, which are not part of his normal personality.

He has noticed himself that he takes longer to fill out reports at work and that he sometimes falls down, but he puts those problems down to stress and a knee problem he has been having.

His wife Rosie insists on taking him to the doctor and after some preliminary testing he ends up at the neurologist where he is given the diagnosis of Huntington's disease.

Joe's mother died when Joe was 12, but he never understood the circumstances surrounding her death. He remembers her wasting away in the hospital, but he always thought it was because she was a drunk. History is revised when he realizes she had Huntington's disease and that she had passed the gene down to him.



Now, each of Joe and Rosie's four children has a 50-per-cent chance of getting the disease. The children struggle with the knowledge that they could have the predictive test and find out if they too have the gene-— is it better to know or to live life wondering? In particular, Katie, 22, isn't sure she wants to know. She wrestles with the idea of telling her boyfriend, or not, and how the knowledge or lack thereof, will affect their relationship.

While Genova hopes her novels inform readers, her priority is to create a gripping human story. She interviewed several neurologists including Young and Tobin and she met many families including the Sullivans, whose daughter Meghan, 26, died last year.

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Genova dedicated the novel to Meghan. "For everything to work it has to be a story first," she says.

It's not all about Huntington's — Inside the O'Briens includes interesting extra themes like baseball, yoga and professional dance.

As she has in all of her previous books, Genova brings a complicated medical diagnosis to real life with sensitivity and thoughtfulness. She gives readers an insider's view of what it's like to live with something very wrong with a person's brain.

Her compassionate storytelling is full of human emotion, all the way from boiling rage to love and gratitude, to despair.

Adapted from reviews by Tracy Sherlock, Vancouver Sun and by Fiona Purdon, The Daily Telegraph (Sydney)

Inside the O'Briens is published by Simon and Schuster and the RRP is $26.00
However, it has been seen in Big W for $16 and is available from Angus and Robertson for $20.45
You can also check out your local library.

Carers' Support Group

Huntington's New South Wales and Huntington Disease Service – Westmead Hospital

invite you to
come along to our

CARERS' SUPPORT GROUP

for a get together with other carers who, like yourself, are caring for a partner, a family member or a friend with HD.
Come along and join us as we share our chatter, laughter, tears and experiences.

Sessions will be held at
Huntington's NSW Offices
21 Chatham Road, West Ryde NSW 2114
2015 Sessions

01 July, Wednesday, 10.30am*
19 August, Wednesday, 10.30am
23 September, Wednesday, 10.30am*
28 October, Wednesday, 10.30am
27 Nov or 04 Dec, Fri, Year-end Get-Together (TBC)

** Denotes School Holidays*

To RSVP and for further information, please contact either: Jet Aserios or Cecelia Lincoln, SW Department, Westmead Hospital on 9845 6699 or Robyn Kapp, Huntington's NSW on 9874 9777

Providing Hope for the Future

Recently we received a wonderful donation of $20,000 in the form of a bequest for our regional support and research. This gift was unexpected and we are so grateful to the Late Edith Ross for remembering Huntington's NSW in her will.

A gift in a will some years ago from the Late Elsie Court meant that we were able to purchase our office here in West Ryde. Gifts like these mean that we can put more funds into the support we are able to provide for families impacted by Huntington's Disease across NSW and the ACT, particularly for people in regional areas.

Once your family is provided for, Huntington's NSW would be most grateful for your gift. Leaving a gift to the Association in your Will is easy and if you are considering changing or making your Will please contact Pauline Keyvar or Robyn Kapp (02) 9874 9777 for a confidential discussion. For further information our 'will wording' is on our website www.huntingtonsnsw.org.au.

Lighting the Way: A new Biomarker for HD

A new biomarker reveals brain changes in early Huntington's disease.
By Melissa Christianson, Edited by Dr Jeff Carroll

In Huntington's disease, brain cells begin dying long before disease symptoms arise. Unfortunately, good tools for monitoring early brain changes – and testing whether new therapies slow or stop them – have not previously been available. However, a newly developed tool aiming to overcome this problem may mean big changes for the way we track Huntington's disease.

Symptoms in Huntington's disease are caused by the death of brain cells in specific parts of the brain. However, one of the curious features of the disease is that outward symptoms don't typically appear until a good many brain cells have already died. Thus, there is a big disconnect between the timing of the brain changes and the outward symptoms of Huntington's.

This disconnect makes early, proactive treatment of Huntington's disease really important. To understand why, imagine Huntington's as a fire burning inside a building. If no one calls the fire department until half the building is charred, so much of the building will be lost by the time the fire brigade arrives that no amount of effort will save it. In much the same way, waiting to treat Huntington's disease until outward symptoms appear means allowing important brain cells to die – and once these cells die, they can't be replaced.

Therefore, effectively treating Huntington's disease will almost certainly require intervening *before* brain cell loss causes outward symptoms. Unfortunately, good tools for tracking brain changes early in Huntington's don't yet exist. This lack of tools is a problem, because it means that doctors and scientists are essentially blind to what's going on in the brain early on in the disease.

Returning to our fire analogy, trying to treat Huntington's disease while blind to early brain changes is like trying to fight a fire with your eyes closed. If you can't see the fire, you don't know if you are containing it – or if instead you have your water hose pointed in the wrong direction entirely. Similarly, if doctors are blind to early brain changes in Huntington's disease, they can't accurately tell if new therapies prevent or slow down these changes. This means that they must spend valuable time – time that individuals with the disease don't have – waiting for outward symptoms to develop before determining if a treatment has any hope of working.

Trying to treat HD while blind to early brain changes is like trying to fight a fire with your eyes closed.

To avoid this waiting game, we are in urgent need of tools for tracking early brain changes in Huntington's disease. Just like tools for monitoring the temperature of a fire provide important information about the status of the fire, tools for monitoring early brain changes in Huntington's provide important information about the status of the disease.

Sign, Sign, Everywhere a Sign

In the clinic, such tracking tools are known as "biomarkers". Biomarkers give **signs or signals** of what's going on in diseases like Huntington's. They can be any kind of test - running the gamut from blood tests to thinking tests to brain scans and everything in between - but they all have one thing in common: they measure something concrete about the disease. A good biomarker lets us monitor the status of the disease, which is important for predicting progression or telling if a treatment is working.

In a practical sense, therefore, biomarkers are really important for the Huntington's community because they can make clinical trials of new drugs quicker and more reliable. Having good biomarkers would be a powerful weapon in the fight against Huntington's disease.

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A Biomarker for PDE10

Fortunately, a good biomarker of early brain changes in Huntington's disease may be just over the horizon.

The new biomarker is focused on PDE10 – a brain protein that has already made a splash in the Huntington's research community. Scientists think that PDE10 helps brain cells communicate with one another and that it might be a good drug target for the disease. In animals, PDE10-targeting drugs improve brain cell survival and delay the onset of Huntington's-like symptoms. In the clinic, an ongoing trial currently recruiting subjects is now testing whether PDE10-targeting drugs improve symptoms in humans with HD.

Two features make PDE10 particularly well suited as a biomarker for Huntington's disease. First, it is found almost exclusively in parts of the brain where brain cells die in Huntington's disease. As a biomarker, therefore, it would give information specifically about problem areas in the disease. Second, although these brain cells normally make a lot of PDE10, they start making less and less long before they die in Huntington's. Thus, a PDE10 biomarker would provide information on brain cells that are sick but not yet dead.

Together, these two pieces of information would give PDE10 the potential to be a really powerful biomarker – one that would allow doctors to specifically monitor the health of at-risk brain cells in Huntington's disease *before* outward symptoms develop.

Glow, Little Biomarker, Glow

With this idea in mind, scientists at Pfizer created a PDE10 biomarker for tracking early brain changes in Huntington's disease.

In essence, the new biomarker is a picky, sticky substance that attaches tightly PDE10 but not other proteins in the body. Importantly, this picky, sticky substance has tiny glowing bits attached to it. Though tiny, these glowing bits are a big addition: they let scientists using a special camera track the substance wherever it goes. The substance itself is safe, so scientists can administer it to individuals with Huntington's disease – and then watch where it travels inside the human body.

Most of the new biomarker ends up stuck to PDE10 in brain cells, which means that it accumulates in exactly the brain areas that we want to watch in Huntington's disease. In these areas, healthy cells (with lots of PDE10) glow brighter than sick cells (with only a little PDE10) in danger of dying. By measuring the brightness of the glowing biomarker with a special camera, therefore, scientists can monitor the health of these at-risk brain cells over time.

Putting It to the Test

Pfizer scientists put this new tool to a practical test in a study published in the Journal of the American Medical Association.

In their study, the scientists took pictures of Huntington's-vulnerable brain regions lit up by their new glowing biomarker, and then they looked closely at the pictures to see if the regions looked different in individuals with and without early Huntington's disease. Importantly, the Huntington's individuals in this study had **very early disease**: they either only mild symptoms or no symptoms at all.

The new PDE10 biomarker may directly improve and speed up the search for HD therapies.

When the scientists analyzed the pictures, they found that the brain regions from the different groups of subjects in fact looked very different – even though there were not obvious differences in outward symptoms between subjects with and without Huntington's disease. Specifically, for important brain regions, the glow coming from the PDE10 biomarker was brighter in healthy volunteers than individuals with Huntington's disease. Further, even among Huntington's individuals, the glow in those with no outward symptoms was brighter than that in those with mildly symptomatic disease.

Thus, there was a strong relationship between the brightness of the new PDE10 biomarker and the extent of Huntington's disease. This relationship was much stronger than what scientists could detect using existing tools.

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Based on these results, the scientists believe that their new biomarker is sensitive to early brain changes in Huntington's disease.

How Does This Help Us?

The PDE10 biomarker could be a really powerful tool for the Huntington's community, because it offers an easy, accurate way to monitor brain changes early in the disease. Better disease monitoring makes assessing if a potential treatment is working both easier and more accurate, which speeds up clinical trials. Thus, even though the biomarker itself is not a treatment, it may directly improve and accelerate the search for new Huntington's therapies.

Further, this biomarker could allow doctors to predict when an individual with pre-symptomatic Huntington's will begin to experience symptomatic disease. This prediction would be invaluable both for clinical studies (where it is important to test therapies in patients whose exact disease status is known) and for individuals living in the shadow of looming symptomatic disease.

The Bottom Line

It is encouraging to see a large, well-resourced pharmaceutical company invested in running efficient, effective clinical trials for Huntington's disease. Although the Pfizer scientists must do more work to verify their new biomarker's sensitivity, the potential benefits of this new tool in the search for Huntington's therapies are clear. Across the board, the development and validation of good biomarkers will speed the search for effective treatment.

Acknowledgement: www.hdbuzz.net

Did you know that ...?

In April 1975, nine people met at the Toongabbie Hotel, Toongabbie and formed a steering committee with a view to establishing the New South Wales Huntington's Disease Association. They contributed $75 to cover immediate expenses and on 27th May 1975, the inaugural meeting of the Association was held at Lidcombe Hospital.

Who would have imagined that four decades later we would be holding a fundraising event at the Intercontinental Hotel in the heart of Sydney to celebrate the 40th anniversary of their vision? Discussion at the inaugural meeting included the social aspects of the disease, the inadequate facilities in Sydney, the lack of counselling and the fact that general practitioners were unable to make a diagnosis.

At the first Annual General Meeting, held in May 1976, a constitution was adopted, and the Association commenced activity as a purely voluntary body.

In those early days, we didn't have our own newsletter so we paid the Victorians 10 cents each for copies of theirs. (Imagine doing that today!)

Over the next few years various fundraising activities were conducted, a newsletter was started and the first information pamphlet was produced.

In 1980 we commenced advocating for a nursing home unit at Lidcombe Hospital for people with HD; and although this did not eventuate until 1988, an HD clinic was established in 1982. Also around this time the first seminar for families and professionals was held as well as the first of what has proved to be a successful and continuing holiday camp program.

Of course, that first committee would have been wishing and hoping that by 2015 a cure would have been found but as we are aware, that was not to be.

Huntington's NSW is still needed to support families impacted by HD throughout NSW and the ACT. You can be assured of our ongoing commitment to this cause.

In friendship
Robyn Kapp
Executive Officer

Huntington's New South Wales

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Email: info@huntingtonsnsw.org.au
Web Site: www.huntingtonsnsw.org.au

AHDA (NSW) Inc
The Australian Huntington's Disease Association (NSW) Inc is a not-for-profit organisation established in 1975.

Our Mission
The energies and resources of the Australian Huntington's Disease Association (NSW) Inc are directed towards satisfying the needs of people with or at risk for Huntington's Disease and their families in NSW and the ACT by providing and/or facilitating delivery of a range of quality services.

Our Philosophy
People with Huntington's Disease and their families are individuals with equal value to all other members of Australian society, with the right to treatment and care by knowledgeable professionals and care givers, the right to appropriate support services and the right to have the best quality of life possible.

Our Services
These include education and information; advocacy; counselling and referral; holiday programs; family support; rural outreach and client services.

Our Board
President: Brian Rumbold
Vice President: Deb Cockrell
Treasurer: Richard Bobbitt
Secretary: Amanda Dickey
Member: Felicity O'Neil

Association and Other Useful Contacts

Huntington's NSW

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