Gateway

News from Huntington's New South Wales
Volume 14 No 2 **Winter 2011**

The World Congress on Huntington's Disease is coming Down under again in September! It was great to have it here in 1997 and it's fantastic that Australia is hosting it for a second time.

The Congress will include the Marjorie Guthrie Day – an IHA Family Day on Sunday 11th September (12 noon till 5pm). Attendance will be free of charge to delegates representing IHA organisations and anyone associated with an HD organisation.

Join others from around the world to share and learn more about youth needs, helping families stay strong through HD, coping strategies, starting a family, IVF, support groups, residential care facilities and caring – share the care!

You may like to organise a group of family and friends to attend the World Congress in Melbourne. Go to the official website **www.worldcongress-hd2011.org** for information on Speakers, Program, Accommodation, Travel Information and Bursaries, Tours and Partners Program, Satellite and Related Meetings, Exhibitors and more.

From the President

After rejoining the Board two and a half years ago I'm now assisting Huntington's NSW through a new period of transition. Due to unforeseen circumstances, the Executive Officer position is now vacant and the Board is considering the best way to take the organisation into its next phase of development.

My sincere thanks and gratitude go to Lily, Ramona, Toni and Mark for the work they have done to keep the office and our social club, carer support and outreach programs running so smoothly.

Of course, we would not exist without the ongoing support of the HD Community. Please take a moment to renew your membership using the form enclosed in this issue of Gateway. You will also notice that Huntington's NSW has taken a big leap into the 21st century by adding credit cards to its methods of payment!

We look forward to bringing you news of more positive changes in the coming months.

In friendship,
Robyn Kapp

Getting The Data Out – A New Scientific Journal Just For HD

The search for better treatments for HD requires a lot of effort by researchers across the globe. Time is of the essence: the ideal time for a treatment for HD is yesterday. Data produced by all these researchers need to be made available sooner rather than later, so that others can build on what is already known. PloS is a new journal launched to speed up the process of scientific discovery in HD.

The traditional way to publish new findings is for researchers to send data with a 'story' to a scientific journal. A panel of experts then reviews that story, to judge whether the data and story are solid, and also whether the story is important enough to be interesting to the journal's audience. This process is known as 'peer review'.

This approach has advantages: it ensures that what is published is scientifically sound. Once published, articles can be retrieved via the internet sites like PubMed, and used by other scientists to guide and advance their own research.

However, there are several drawbacks, too. Firstly, the time between doing the research and the story being published can be very long. Sometimes the story has to be offered to several journals, one after the other, before it is eventually published. Several years can pass like this.

Secondly, most journals survive by selling copies, so they have to rate the importance of each story. Anything that might not be interesting to that journal's audience is likely to be rejected, even if the story is scientifically sound.

That introduces bias to what's available in the scientific literature. It favours exciting stories, but makes it difficult to publish solid, well-conducted scientific research if the results are less glamorous – for instance, if they show that a particular approach, idea or experiment has not helped. These are known as 'negative results'.

Another problem is that a complete scientific 'story' might take five or ten years to research from start to finish. Along the way, interesting data might be produced, but because they don't tell a complete story, they are unlikely to be published and seen by other researchers.

"Negative data", or data that don't make a complete story, can still be really useful to other researchers. For science to make progress, knowing what doesn't work can be as useful as knowing what does.

Imaging ten researchers in different places, working on similar scientific projects, that could have been shown to be pointless already, if someone had published a single negative result. All that effort, time and money could have been put to much better use.

A new platform for publishing was launched in September 2010, aiming to make HD research more efficient by publishing the results that would otherwise never be seen, and shortening the time it takes to get data published.

This platform is called Public Library of Science (PloS) Currents Huntington Disease and is supported by the CHDI Foundation.

Renew Your Membership!

Your membership of the Association are due for renewal on 1 July 2011, unless you have pre-paid for the 2011/12 membership year, and a membership form is enclosed with the newsletter.

If you're unsure of whether you have pre-paid, please call Lily at the Association office and she can advise you.

It is important for lobbying and advocacy for the Association to have a membership that is representative of all Huntington's families and there is a "no fee" option for those who are on a pension or otherwise unable to pay the $22 fee. If you are a carer and take out a paid membership please encourage the person you care for to also join under the "no fee" option – there is strength in numbers!

Great news!

You can now pay your annual membership fee by credit card. If you'd like to use you Visa card or MasterCard to pay your fees, please complete the appropriate section on the enclosed form.

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PloS Currents HD aims to keep what's good about conventional publishing, crediting researchers for their work, and making the research available online.

Its emphasis, though, is on getting scientific data into the public domain for everyone to access, not within years but within days. As a bonus, this way of publishing is cheap for researchers and offers free open-access for anyone wanting to use it – whether they are scientists or not.

PloS Huntington Currents Huntington Disease is edited by eminent HD researcher Gillian Bates.

A Board of expert moderators reviews new submissions, just like a conventional journal would. However, the reviewers make a decision based purely on whether the science is sound, and not whether the story is 'important' or 'exciting'. After this quick check, the research is published immediately online. Because the process is so quick, findings may be available a few days after submission.

To improve interaction between researchers, readers can post comments, and the authors of articles can update their publications with new findings, so that various versions are available. Repeat submissions go through a review process to check they are sound.
Hopefully, all this will make PloS Currents HD a dynamic way to get good-quality data out into the public domain.

Here at HDBuzz, we believe that making scientific research available to everyone, quickly and reliably, is crucial to making progress. We are confident that PloS Currents will be a major step forward in the global search for treatments for HD.:

Taken from the http://en.hdbuzz.net/10 website

Camp Breakaway 2011

Camp Breakaway 2011 began in style - with sunshine and anticipation. Monday March 11th dawned warm and sunny as our excited campers and staff piled onto the excursion bus waiting at Elsie Cottage in West Ryde to take us to our familiar "home" for the next week.

Several of our staff members and volunteers had already made their way to Camp Breakaway, a facility set in 25 acres of lush bushland in San Remo on the Central Coast, and welcomed us warmly with the first of our 24 meals/ snacks of the week. Yes, the Huntington's Association Camp is renowned not just for its fun and frivolity, games, activities and "party night", it has, in the last few years since acquiring our new "chef", become a place of degustation and gastronomic delight. Robyn, Chef Extraordinaire, kitchen Nazi and Association President, took her kitchen duties with the seriousness of a surgeon, and tantalized our tastebuds 6 times a day with moist home baked cakes, fluffy pancakes, hearty casseroles and stroganoffs, curries, desserts...the list goes on...

Camp is not all about food however. We were once again very lucky this year as the skies were blue and sunny every single day, allowing us the opportunity to enjoy our leisurely morning walks to the lake after breakfast. The more active of our group enjoyed a competition of sorts, seeing who would make it up the hill first before breaking out in a sweat, while the rest of the group took in the scenery and sheer beauty of the lake in the morning light, stopping to pet dogs, watch toddlers on their bikes, taking photos of each other when the opportunity arose.

After lunch, Karen and Lily kept us busy in the afternoons with some very creative craftwork; painting, picture frame designs, birthday cards, and many other wonderful mementos to take home, and we spent the remainder of the day playing mini golf, bowls, skittles and a host of other games with interludes for coffee and a chat interspersed throughout the day.

It was difficult once again to leave the comfort of a familiar place, surrounded by friends - some new, some old - who understood what each other was going through.

The friendship, sharing and camaraderie that developed over the 5 days is testimony to what an open heart and mind can achieve, and achieve that we did.

Wonderful friendships and loyalties were forged, and there were handshakes, hugs and tears of farewell as we said goodbye to some of our friends for another year.

Social/Lunch Club

Good Times Keep Rolling at Social Club

We have had a lovely busy time with social club these last few months, whilst welcoming a few new members to our family. Lily and I have continued to don our aprons in the kitchen to cook up a storm when we have lunch at Elsie Cottage, and have been practising many new and wholesome recipes we learned from Robyn while at camp. No complaints so far – in fact we are all continuing to put on weight with requests for more home baked puddings and creamy sauces!

We have also continued with our outings every month. One of the highlights of our excursions have included a big day out to the Koala Wildlife Sanctuary, where we patted "Eric", a cuddly and tame (albeit a bit smelly) alpha male koala, and fed kangaroos and their joeys while they were hopping freely all around us. It was a wonderful experience for all our members to be so close to nature, walking along the canopied paths of the sanctuary and experiencing the delights of touching and feeding our native animals.

If you know of anyone with HD who might be interested in joining our group for social activities, a good chat with friends, and just time out with people who know what you're going through, please call us at the Association on 9874 9777 or email Ramona@ahdansw.asn.au




Do you have a story to share?

We want to hear from you. If you have a personal story or a contribution that you wish to make to the Newsletter please send it to us at the Association offices (see details on the back page) or by email to hdassoc@ahdansw.asn.au.

HD Research Update

IMAGE-HD Project Progresses

Following is an update on using neuroimaging methods to further the understanding of brain mechanisms in Huntington's Disease and working towards identifying an imaging biomarker of disease onset and progression for use in clinical drug trials. From the Image-HD Team: A/Prof Nellie Georgiou-Karistianis, Prof Gary Egan, Prof Julie Stout, Dr Andrew Churchyard, Dr Phyllis Chua, Dr Marcus Gray, Dr Juan Dominguez and Ms Rebecca Langmaid.

Using neuroimaging methods to further understand brain mechanisms in Huntington's disease and working towards identifying an imaging biomarker of disease onset and progression for use in clinical drug trials.

The Project: Neuroimaging methods have gained significant momentum in Huntington's disease (HD) research in recent years with high resolution techniques, like new developments in positron emission tomography (PET) and functional magnetic resonance imaging (fMRI), providing a dynamic representation of brain function that enables insight into underlying cellular dysfunction. Although structural imaging (MRI) has been included in various large scale multi-site studies in HD (i.e., PREDICT-HD, TRACK-HD), given its sensitivity in reliably detecting progressive volumetric changes pre clinically up to 10 years prior to symptom onset, functional and other types of imaging methods may offer a more complete understanding of the complex web of interactions underlying the neuropathology of HD. IMAGE-HD is a "multi-modal" neuroimaging study based in Melbourne, incorporating a range of neuroimaging techniques in the one large-scale longitudinal study. Baseline data from 35 early symptomatic, 35 pre-symptomatic and 35 healthy controls was collected during 2008-2009 and re-scanning of subjects is currently in progress. Neuroimaging results from the baseline study (structural, microstructural, functional) will demonstrate how neuroimaging methods can sensitively detect brain changes in pre-symptomatic HD up to 15 years prior to disease onset. Neuroimaging methods enable a more complete and comprehensive understanding of the mechanisms involved in neural breakdown that characterizes this devastating disease, and longitudinal studies will determine the sensitivity and reliability of potential imaging biomarkers in tracking disease progression, as well as to evaluate their links to clinical outcomes.

Our progress to date: As of April 2011 we have completed the second stage of longitudinal data collection, 18 months from initial baseline testing. We have one further time-point remaining due to commence May 2011. Since baseline we have highlighted subtle changes in brain tissue size (grey and white matter volume) associated with Huntington's disease. We have also observed very small differences in the way this tissue is structured (via diffusion tensor microstructural analysis). Based on these results we have proposed a biomarker which describes how the brain changes during the early stages of this disease which considers both volume and microstructure. We are currently preparing a paper on this data for publication.

Further examination of how the specific circuits in the brain function during set-response shifting (numbers and letters task) have found that during this task, the function of specific parts of the brain (ie frontal cortex) also reflect general cognitive and emotional difficulties that symptomatic HD patients may experience. This suggests this brain region is important in coping effectively with HD. Both of these findings have been accepted for presentation at the *International Human Brain Mapping* conference in Québec City, Canada in June 2011. Analysis of neuroimaging data during a working memory task (remembering number locations) reveals differences in working memory circuits in the brain in HD. This data has been submitted for publication in the journal *Cerebral Cortex*.

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Finally, we are also examining how volume, microstructure and brain function change over time. These findings will be reported by A/Prof Nellie Georgiou-Karistianis as part of a key note presentation at the *World Congress on Huntington's Disease* in Melbourne 11-14th September, 2011, Convention & Exhibition Centre. We encourage as many family members to register and attend this exciting event. The IHA Family day will be held on 10 September, 2011 (for more information go to www.worldcongress-hd2011.org).

Funding Sources: We would like to acknowledge and thank all participants for their invaluable contribution to this study. We would like to also thank the Cure Huntington's Disease Initiative (CHDI), Ltd, USA, and the National Health & Medical Research Council (NH&MRC) for funding this study.

Image HD project leader: Associate Professor Nellie Georgiou-Karistianis
Experimental Neuropsychology Research Unit, School of Psychology and Psychiatry, Monash University, Clayton, VIC,3800.

The HD Outreach Service Has Moved

The HD Outreach Service has now moved from Lottie Stewart Hospital at Dundas to Westmead Hospital. Please note their new contact details below:

Westmead Hospital HD Service,
Westmead Hospital,
Hawkesbury Road,
Westmead 2145

Phone: 02 98459960

Is Huntington's Disease Twice As Common As We Thought?

In an article in the medical journal The Lancet, Sir Michael Rawlins claims that traditional estimates of how common Huntington's disease is, might be dramatic underestimates. Why might this be, and what does it mean for the HD community and the search for effective treatments.

There are probably twice as many people with symptoms of HD – or even more- than previously thought.

Scientists and statisticians use the word 'prevalence' to describe how many people there are with a particular disease at a given time.

For a long time, the prevalence of 'symptomatic' HD has been quoted as 4 – 10 per 100,000. That means that in England and Wales, for example, where the combined population is 53 million, there should be between 2,120 and 5,300 people with HD.

But Rawlins reveals figures from the Huntington's Disease Association of England and Wales (HDA) showing that they provide care for precisely 6,702 people with symptoms of HD. Even if that were all the patients in England and Wales, it still translates to a prevalence of 12.4 per 100,000 – higher than the upper limit of the previously accepted range.

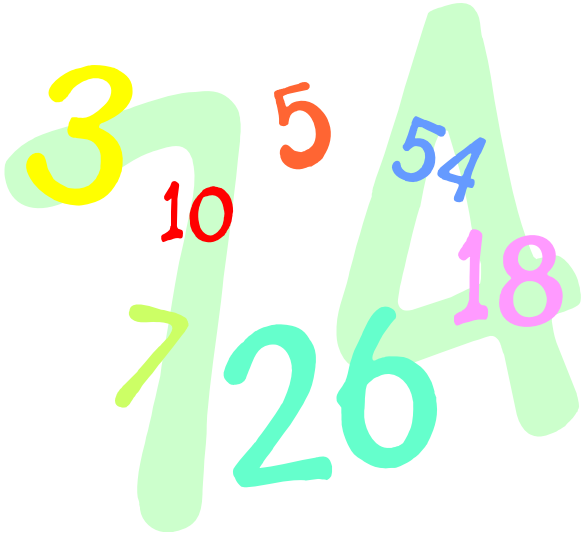
In reality, it's likely that even this new higher figure is an underestimate. There are areas that aren't covered by the HDA's services, and lots of people with HD who have never been referred to the HDA.

What's more, there is every reason to believe that these underestimates have occurred throughout the world, not just in the United Kingdom.

Why are the old figures wrong?
The traditional prevalence figures are wrong, Rawlins suggests, for a number of reasons.

First, most of the studies that led to the figures were done a long time ago, most before the genetic test for HD became available in 1993. In those days there was often uncertainty or a delay in reaching a definite diagnosis. Nowadays the genetic test often enables doctors to make a diagnosis of HD earlier, and in people with unusual forms of the disease that would previously have been missed.

Second, and perhaps more importantly, is the stigma that has long surrounded Huntington's disease. Many HD family members will be familiar with stories of relatives whose illness was not to be discussed, or who were sent to psychiatric institutions and never spoken of again. Many patients receiving a diagnosis of HD have kept it a secret from relatives, doctors and insurers, or prevented HD from being mentioned as an official cause of death.



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There are many reasons, of course, why such secrecy surrounded HD, but over the years it has contributed to a stigma that has prevented one of the most important basic tasks facing HD researchers – counting the number of people affected by HD – from being possible.

If accurate figures are to be reached, urgent new research is needed with new methods capable of overcoming some of these problems. This won't be easy but Rawlins is working with expert statisticians and HD clinicians to set up such a study.

Rawlins' article triggered a number of responses in the scientific literature. A group of Australian researchers led by Clement Loy, pointed out another possible reason for the underestimate: the 'baby boomer' population surge, means that right now there are more people than ever before in their 50's and 60's, a time of life HD often becomes apparent.

Patrick Morrison from Belfast, who helped to produce some of the original prevalence figures, pointed out that, thanks to improvement in symptom control and clinical care, HD patients are living longer than they used to, which has probably contributed to the increase in prevalence. His analysis of more recent data suggests a prevalence in the UK of 14 – 16 per 100,000 – about twice the current figures.

Why does prevalence matter?

This article and the more accurate numbers that will hopefully stem from it, matters because prevalence figures are among the things used by governments, health care organisations and research funding bodies, in deciding how much to spend on different conditions. If the prevalence of HD is much higher than previously thought, it is likely that these organisations have been devoting less money to HD than ought to have been received.

But as Rawlins points out, HD does not just affect those with symptoms. It is estimated that for every person with symptoms of HD, there are 5 more at risk – not to mention all those with no genetic risk – friends, partners, wives, husbands and those who've had a negative genetic test. If the prevalence of HD is higher than thought, there are a lot more of those people too.

A call to action

The timing of the article coincided with the launch of an All-Party Parliamentary Group on HD – a group of UK politicians, advised by HD experts, who are pledged to eradicate stigma and promote HD research and care. The launch was accompanied by a mass-rally at the Houses of Parliament of HD family members under the banner "Hidden No More" : http://hiddennomore.co.uk – reflecting a new desire in the HD community to work together to remove the stigma of HD – beginning with being accurately counted.

Note: This article was taken from the new website: http://en.hdbuzz.net which features HD research news in plain language written by scientists for the global HD community. Keep yourself up-to-date on the latest happenings on the world stage of HD research by logging on to this great site. We will be bringing you articles from this site in future newsletters for those who have no access to the internet.

Programs Reach More Rural and Remote Areas

We are already into the 2nd half of the year and have accomplished much in terms of our programs this year. Social club has been busy and, as the name implies, incredibly social. Our members have enjoyed gastronomic feasts at Lily's capable hands, as well as fun-filled outings to different areas in Sydney each month.

Last month I spoke at a Conference in Penrith for the Referral and Assessment Centre who are responsible for receiving referrals and assessing eligibility and service needs of people referred to the Home Care Service of NSW (HCS). It was an informative and animated session as many of the assessors had had dealings with people affected with HD but did not have the necessary skills and knowledge about the disease.

In the last month, both Mark and I have had several successful visits to the more rural and remote areas in NSW, meeting with isolated families affected by Huntington's Disease and talking to staff at Nursing Homes which care for HD patients. We have been astounded by the fortitude and tenacity of these families, who struggle with so much, yet never give up, even in the face of much adversity. We have heard gut-wrenching accounts of isolation from friends and other family members, life-changing questions unanswered, requests for help unheeded, and we would like to assure you that we will do our utmost to provide a listening ear to you and help you and your family to the best of our abilities.

We have continued to host the Carers Support Group in conjunction with Huntington Disease Service at Elsie Cottage. Our carers have found some solace in the fact that there are others in similar positions, and share experiences and impart words of advice and friendship to one another. It is a lovely, close-knit group of wonderful people who give so much of themselves, and we would love to welcome other carers to join us in this experience of sharing.

Canberra Support Group

Canberra and Wagga Outreach

It was delightful once again to visit our friends in Canberra last month. The support group for families with Huntington's Disease is thriving into a social network of exceptionally wonderful and resilient individuals - carers and people with HD alike who take time out every two months to meet for dinner and drinks, talk about the latest happenings, enjoy one another's other's company and provide a listening ear. The Canberra Support Group meets on the first Wednesday every alternate month, and welcomes new members to join them in their quest for the perfect schnitzel and lamb's fry...

I met a few other families in Canberra and Wagga who were unable to join the support group, but who benefited from meeting other families and who shared their stories with me. These courageous families battle with so much, yet continue to try to remain positive in all that they do, and it is humbling to be a small part of their lives.

If you have a family member with Huntington's Disease, or if you are affected by Huntington's and would like to talk or discuss options, please feel free to call us at the Association on 9874 9777 or email me at Ramona@ahdansw.asn.au. We are always here for you.

Canberra Lunch

Huntington's Disease Service

invites you to
come along to our

FAMILY AND FRIENDS 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.

2011 Sessions

13 July, Wednesday, 10.30am
24 August, Wednesday, 10.30am
28 September, Wednesday, 10.30am
19 October, Wednesday, 10.30am
16 November, Wednesday, 10.30am
Dec - Christmas Get-Together - TBC

Sessions will be held
at
Huntington's NSW Offices
Elsie Court Cottage
21 Chatham Road
West Ryde NSW 2114

For RSVP and further information, please contact:

Jet Aserios: (02) 9845 7528
Social Work Department
Westmead Hospital

Huntington's New South Wales

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Facsimile: (02) 9874 9177
STD Free Call: 1800 244 735 (Country NSW only)
Web Site: www.ahdansw.asn.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 10/11
President: Robyn Kapp OAM
Vice President: Anne Low
Treasurer: Richard Bobbitt
Secretary: John Conaghan
Don Ayres
Karen Bevan
Keith Dingeldei
Jim Finn

Association and Other Useful Contacts

Association Staff

Vacant
Executive Officer

Toni Ling Zhang
Administration Officer

Lily Shu Yue Ma
Administration and Activities Assistant

Ramona Watts
Family Support Co-ordinator

Mark Bevan
Family Support Officer

Huntington Disease Service

Dr Clement Loy
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Jet Aserios
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