Gateway

News from Huntington's New South Wales

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Enroll-HD

Huntington's NSW is excited to announce that the Enroll-HD study is now up and running at Westmead Hospital. This international study presents an opportunity for all family members affected by Huntington's disease to be involved in research. The study is an international collaboration, with study sites in the USA, Europe and South America. As of October 2015, close to 8,000 participants had been recruited world-wide.

The study is 'observational'. It is not a drug trial, but a way of gathering a large database of information to prepare for the next phase of research. Study visits are similar to usual clinic visits, comprising questionnaires, a physical examination, memory testing and blood collection. The main focus is on gene carriers, either affected or not, but the study is also open to the whole family.

The study involves a baseline visit followed by one visit annually. The first visit is the longest, and takes about two and a half hours. Studies after this should be around an hour. The Enroll-HD website is a great source of information about the study. You can find more information at the website, www.enroll-hd.org

The study organisers hope to recruit 20,000 people at risk or with HD worldwide. We are told that the study is designed with privacy concerns being paramount, and all information collected is de-identified. It is also possible to withdraw at any time.

If you are interested in participating in this exciting new study which represents the next phase of research into HD, you can contact the study coordinator, Therese Alting, at the Westmead HD Service. Telephone 9845 6310 or 0438 604 719 or email her at therese.alting@health.nsw.gov.au

The Board & Staff of Huntington's NSW would like to wish you and your families are very Happy Christmas and a Peaceful 2016.

Life Memberships

At the Annual General Meeting held on 14th November 2015, the Association recognised the outstanding and compassionate service of Mark and Karen Bevan; John Conaghan and Dr Clement Loy. In appreciation of their significant contribution to the Huntington's community they were awarded Life Membership. Following are excerpts from the citations of their awards.

John Conaghan, Karen Bevan, Dr Clement Loy & Mark Bevan

Karen and Mark Bevan joined the Association after Karen's father was diagnosed with Huntington's disease in the late 1970s.

Mark was elected to the Committee in 1981 and over the next nineteen years served various terms on the Board, undertaking the roles of President, Vice-President, Secretary and Board Member. Mark also played a pivotal role in the establishment of the Holiday Program first held in 1981 at "Gilbulla", Menangle.

In May 2010 he was appointed as the Regional and Rural Family Support Worker and held this position until his retirement in October 2015. He travelled throughout NSW and the ACT assisting families impacted by HD and providing education and information sessions to health professionals.

Karen coordinated and volunteered for the Social Club and Holiday Programs from 2001 until her retirement in October 2015. Over the past fourteen years she has attended twenty three holiday camps. Her enthusiasm and commitment to these programs have contributed in no small way to their success and to the enjoyment of those who participated. She also served on the Board from 2009 until 2012.

John Conaghan graduated in 1983 from La Trobe University, Melbourne with a degree in Social Work. He occupied various social work positions in Newcastle before taking up his current position at Hunter Genetics in 1991 working with families affected by HD.

He gained postgraduate qualifications in social work, family therapy, law and genetic counselling and teaches professional counselling skills to genetic counselling and social work students. He has contributed to research and supervised student research projects through the University of Sydney.

John served on the HD Genetic Testing Working Party, NSW Department of Health which oversaw the introduction of direct gene testing in NSW in 1993 and he collaborated with Huntington's NSW in the planning and development of Camellia Cottage, the first community residential facility in NSW which opened in 1997.

John also served on the Board of Huntington's NSW and was known for his advocacy for those families living in regional and rural NSW.

Associate Professor Clement Loy graduated from the University of Sydney in 1996 with MBBS and a BA (Pure Mathematics). He was awarded M Med (Clinical Epidemiology) in 2000 and his PhD from the University of NSW in 2013.

He underwent subspecialty training in cognitive neurology at the Dementia Research Centre, National Hospital of Neurology & Neurosurgery, London, and laboratory training at the Garvan and Prince of Wales Medical Research Institutes. His clinical expertise includes Huntington Disease and Young Onset Dementia.

In September 2008, he commenced as Neurologist and Director of the Huntington Disease Service at Westmead Hospital which is one the five largest HD services worldwide. He was instrumental in obtaining Commonwealth funding for the purpose built unit at St Joseph's Hospital, Auburn and he is a committed advocate on behalf of families impacted by HD.

He is a member of the Huntington Study Group and has been the site investigator or rater in the majority of international HD observational studies and clinical trials since 2008. He has been invited to a number of international committees on HD and since 2009 has provided media interviews and commentaries on HD for various radio and television programs and a documentary film.

Dr Loy has worked very closely with Huntington's NSW and has supported and encouraged the Association in many ways.

Our 40th Anniversary Year

At the Annual General Meeting held in November, I reflected on the past four decades of the Huntington's Disease Association in NSW. In the Annual Report I wrote:

"This year we recognise that it is forty years since Huntington's NSW was established and it is only through the dedication and service by many people that we have been able to provide meaningful support and encouragement for families who are impacted by Huntington's disease throughout NSW and the ACT.

Robyn Kapp, Dr Clement Loy and Brian Rumbold

At the present time there is no cure but there is hope. We certainly know much more about Huntington's than what we did in 1975 and as I think back over the past four decades, I cannot help but remember what it was like in those early days.

We had no written information – in fact we initially purchased the Victorian newsletter for 10 cents per copy. We had no funds and there was no such thing as an HD Clinic or specialised residential unit.

Former President, Ann Low, at he AGM

However, we did have a dedicated group of people who were determined to make a difference, who had hope. They held small fundraising events such as raffles and trash and treasure stalls. They turned up at Family Support Meetings at Lidcombe Hospital every second month and they craved any new information that came their way. They are to be congratulated and not forgotten for their enthusiasm and commitment to improving the lives of those affected by Huntington's.

Over the past forty years, there has been a reluctance for young people to become involved with Association. This is completely understandable – they want to get on with their lives, they don't want to be reminded of Huntington's all the time, could there be repercussions in the work place if it was known they that were from a Huntington's family. All perfectly legitimate reasons.

Newly elected Secretary, Therese Alting

However in recent times there has been an explosion of young people keen to support the Association, particularly with its fundraising ventures. They are enthusiastic and committed; they are creative and imaginative. They are the future of Huntington's NSW and I'm sure that they will take up the cause in the same way as those who pioneered Huntington's NSW back in 1975."

We look forward to 2016 with excitement and increased enthusiasm. We are excited about the research that is being undertaken collaboratively around the world and we are enthusiastic about establishing new programs for families impacted by HD and increasing our fundraising activities so that we might continue to support and care for our members and families.

In friendship

Goodbye … Welcome …

In October we said good bye to Mark and Karen Bevan who resigned from their positions as our Rural Family Support Worker and Activities Co-ordinator respectively. Mark and Karen have been involved with Huntington's NSW since the late 1970s and were awarded Life Membership at our AGM in November (see page 2). We wish them both well and best wishes for a very happy retirement.

We welcome Amanda Dickey as our Programs Officer. Amanda was first elected to the Board in October 2012 and has been an active member of the HD Central Coast Support Group since 2010. For 25 years she has witnessed how HD has affected close family members and inspired by their strength and sacrifice Amanda is a committed HD advocate.
We are delighted that Amanda has made the transition from Board to staff and in the coming year she will be working on adding to, and improving, the activities and events we offer to the HD community as well as establishing a volunteer program.

Rural Family Support Worker—Update

The Rural Family Support Worker position has been advertised and we are now at the stage of interviewing in order to find the best applicant for the role. We shall let you know the outcome as soon as possible. In the meantime, if you need to talk to some one please contact the office on 9874 9777 or 1800 244 735 (Country NSW only).

Holly's Interview

As we go to print, an interview with our Youth Ambassador, Holly Faulkner sharing her family story, has reached 28,777 people and has been watched by 9,200. We are so proud of Holly, she is always willing to fly the flag for Huntington's families and to give hope for the future.
To watch the interview, visit our page on Facebook.

Holly (right) with Brigitte Salden

Office improvements continue

Once again we have been successful in securing funding through the Community Building Partnership initiative of the NSW Government. We have received $5,314 to cover the cost of our new fence and updated signage and to improve the garden areas. We are indebted to the Member for Ryde, Victor Dominello, for recommending that we receive this funding.
In the past few years, through this program, Elsie Court Cottage has been painted inside and out; we have installed new blinds; laid new carpet and vinyl and purchased new chairs—it's great receiving so many positive comments about how nice our property is looking.

Through the eyes of a friend: changes in mood and behavior in early HD

The family and friends of individuals with HD often tell doctors that they began to notice changes in behavior long before a diagnosis was made. To better understand these early signs, researchers analyzed a psychological questionnaire filled out yearly for a decade by thousands of HD mutation carriers and their companions. The companions were more likely to perceive worsening symptoms over time.

By Leora Fox, edited by Dr Jeff Carroll

Understanding the early symptoms of HD

Huntington's Disease is inherited at conception, but for most carriers of the mutation, symptoms don't begin until middle age. Even though the HD mutation is toxic to brain cells called neurons, most mutation carriers spend several decades symptom-free. This means that the brain has a remarkable ability to withstand many years of exposure to the mutation.

The period before major symptoms have developed is known as the prodromal phase of HD, when behaviors may begin to change gradually and subtly. Often, the very first symptoms noticed by HD patients or their families involve small alterations in thinking, mood, or disposition. These symptoms are real, but it's not possible for physicians to say that they're definitively due to someone carrying an HD mutation because many people who don't carry the mutation also experience these challenges.

What are these early symptoms like? Maybe a punctual person finds it more difficult to be on time for appointments, or a spouse notes that a good sleeper has become a bit restless. Since these early signs don't usually interfere with daily activities, medical research didn't focus there at first. Now, we are aware that investigating early changes is important, because they can inform when and how to begin treatment, especially when new drugs become available.

Recently, a group of researchers concentrated on understanding the psychiatric and behavioral difficulties that can occur in prodromal HD. The

The input and support of trusted companions can be a great advantage.

work is just one arm of a huge study that relies on thousands of HD-positive and unaffected volunteers. Over the course of a decade, participants and their companions filled out a questionnaire every year, evaluating the participant's psychological health. The study revealed some of the subtle psychological changes that can occur in pre-symptomatic HD, and showed that close companions were more likely to notice worsening symptoms than the HD mutation carriers themselves.

PREDICT-HD: studying prodromal HD

The story behind this research actually began more than ten years ago, when researchers started recruiting for a huge study called PREDICT-HD. The overall goal of the work, which is ongoing, is to identify and understand the earliest signs of HD. HD mutation carriers and their families frequently report early behavioral changes, but diagnosis is usually based on movement symptoms that are more specific to HD.

To create standards for assessing patients and treating them with current and future therapies, clinicians need a clearer picture of what occurs during the years prior to the development of involuntary movements. This way, doctors can

make decisions based on documented history from HD carriers around the world, rather than isolated anecdotes from just their own experience with patients.

Volunteers participating in PREDICT-HD came from all over the world, at 33 medical sites in six countries. Each person generously agreed to visit a study site for a whole day or two once a year, for up to 10 years. Participants were examined by clinicians, received brain scans, completed written evaluations, and donated blood samples.

Importantly, participants in the PREDICT-HD study had to have already undergone testing for the HD mutation – a person at risk for HD could only enroll if they knew their gene status. As a comparison, the researchers also included a group of control individuals from HD families who did not inherit the HD mutation. From the test tube to the clinic, the findings from PREDICT-HD are helping us to better understand the earliest changes experienced by HD mutation carriers.

A yearly psychological pop-quiz

In the last decade there have been hundreds of publications about early HD based on data from PREDICT-HD volunteers. We'll zoom in on one study, which focused on prodromal psychiatric symptoms. Jane Paulsen, a clinical psychologist at the helm of the PREDICT-HD project, led the research team.

Every year, participants in the study completed a questionnaire about their psychological health. The test is used worldwide for many disorders, and it consists of 90 fill-in-the-bubble questions designed to measure a broad range of psychological problems. For example, a question might ask "In the past week, how much were you bothered by trouble concentrating?"

Respondents would rate each question on a scale of 0 (not at all) to 4 (extremely). Questions are designed to ask about feelings related to anxiety, depression, compulsions, interpersonal interactions, and many other categories.

Around 1300 participants took part in the study, both mutation carriers and controls, and most brought along a close companion to help assess their mental health using the same questionnaire. The companion was usually a live-in partner or spouse, but sometimes another family member or friend. The researchers were especially interested to see how mutation carriers' psychological scoring compared to individuals without HD, how their evaluations changed over an entire decade, and whether their companions' ratings matched their own.

Assessing mental health in HD: at the beginning, over time, and via a friend

The authors of the study used different types of mathematical analyses to answer three main questions about prodromal HD:

At the beginning of their participation in the study, were there already psychological differences between HD mutation carriers and unaffected individuals?

Yes. When they enrolled in PREDICT-HD, participants with the HD mutation rated themselves higher than control subjects on almost all aspects of the psychiatric questionnaire, including symptoms like anxiety, obsessive-compulsiveness, hostility, hyperawareness of physical illness or injury, and paranoia. Their companions also noticed these types of mental and mood changes, especially when their participating loved ones were closer to developing movement symptoms (such as those who were older, or had more severe mutations).

Over time, from the beginning to the end of an HD mutation carrier's participation in the study, was there a noticeable change in their psychological health?

Well, their companions noticed a change – but the mutation carriers didn't always agree. The majority of HD mutation carriers did not perceive their mental health to be getting worse over the years they participated in the study. However, their companions reported that certain psychological signs got worse, like anxiety, paranoia, and interpersonal distress.

Was there an overall difference in how participants rated their own symptoms, versus how their companions rated their symptoms?

Yes. The difference between the companions and the participants' ratings was especially striking in those predicted to have a higher likelihood of experiencing motor symptoms within a few years. Companions usually noticed more psychological distress in their loved ones than the HD mutation carriers did in their self-reports.

The message

What is the meaning of these results? First, analyzing participants at baseline (the very beginning of the study) showed that early on in symptom progression, HD mutation carriers and their companions noticed subtle changes in their behavior and personalities compared to unaffected individuals.

This is important because it confirms on a much larger scale that mood and behavior symptoms are apparent early on to patients and their loved ones. These types of symptoms can increase in severity over time before movement symptoms occur, to an extent that was not previously appreciated. Gaining a better handle on the psychological health of people with presymptomatic HD could help shape how and when people receive a diagnosis, and when might be a good time to begin treating symptoms such as anxiety, depression, compulsions, or difficulty sleeping.

Second, HD mutation carriers and their loved ones may perceive *longitudinal* changes in behavior (those that occur over time) in different ways. While many participants with the HD mutation did not believe that their symptoms were getting worse, their companions definitely noticed increasing psychological problems or mental distress.

One explanation for this finding is that HD affects the complex circuitry of the brain in a way that hinders insight. This could be due to gradual damage in many connected parts of the brain that sync up to control self-awareness. Or it could simply be that when behaviors and habits deteriorate slowly over long periods of time, the change is easier to see from an outside vantage point. A person at risk for HD almost always completes their own health assessments, which may be part of the reason why the psychiatric symptoms have been difficult to link with disease progression.

Considerations and conclusions

There are a couple of caveats to reflect on when we consider these results. The psychological questionnaire is very general, and it only asks about the previous week of the participant's life, so their answers might not always capture their feelings about the whole *year* since they last responded.

Another consideration is that all the participants and their companions were aware of their mutation status from the beginning to the end of the study. Getting tested is an extremely personal choice made by only a small fraction of those at risk for HD, and that knowledge could affect how a person and their friends and family perceive changes in behavior.

Nevertheless, PREDICT-HD is the largest and longest study that has ever been completed about the prodromal phase of HD, and there are many new results emerging from the data. The questionnaire responses show that there are a great variety of psychological and behavioral symptoms experienced by people with prodromal HD.

The results also imply that patients may not always be aware of how their symptoms change, confirming that the input and support of trusted companions can be a great advantage. Importantly, the combined data from thousands of helpful volunteers has turned individual anecdotes into solid data that will inform how we can better evaluate and treat the early symptoms of HD.

Acknowledgement: www.hdbuzz.net

Huntington's NSW
Holiday Break

Our office will close on
Wednesday 23rd December 2015
and re-open on
Monday 11th January 2016.

Huntington's New South Wales

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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: Therese Alting
Members: Felicity O'Neil & Katy Clymo

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