Gateway

News from Huntington's New South Wales
Volume 15 No 1 **Summer 2012**

From the Manager ...

It is both a privilege and a pleasure to be back working for Huntington's NSW after a break of six years. I am currently in the office two days per week (Tuesdays and Fridays) so please feel free to ring me or you can email me at robyn.kapp@ahdansw.asn.au.

So what am I trying to achieve in two days?

First and foremost I want to make sure that we are providing the best service we can to people with HD and their families throughout NSW and the ACT, given our financial resources. I have particular concern for those who live in rural NSW. To this end I am delighted to announce that Mark Bevan will be extending his rural visits to the NSW South Coast, South West and the ACT (details are on page 6). Please contact Mark (mark@ahdansw.asn.au) so that you can catch up with him when he is in your area.

We are currently developing a new website to bring us into the 21st century and we will let you know as soon as it goes 'live'. Computers, internet, websites, Skype, blogs, etc are now very much a way of life and I would like us to look at new and innovative ways of how we might assist families using such technology. Put your thinking caps and if you have any ideas please let me know.

We are now registered with Everyday Hero (www.everydayhero.com.au), a web site designed specifically for donating to and fundraising for charities. People can participate in major fundraising events such as the City to Surf and select **Huntington's NSW** as their charity for sponsorship and donations.

As there are more people wanting to raise funds for the Association we are also developing a Fundraising Kit. This will ensure that all fundraising events conducted on behalf of Huntington's NSW fulfil the legal requirements for fundraising in both NSW and the ACT.

I am delighted that our stalwart services, including the Social Club, "Gateway" newsletter, Information Resources and Holiday Program continue to be the backbone of our organisation and sincere thanks go to Lily, Ramona, Mark and Toni for ensuring that these programs continue.

I look forward to keeping you up to date with our progress in the near future.

In friendship

Robyn Kapp

Board Changes

In our last edition of Gateway we introduced you to Members of the Board. Since then, due to unforseen circumstances, there have been changes to the roles of some Board Members. Brian Rumbold is now Vice President and Judy Rough has taken over as Secretary. Anne Low has been granted leave of absence.

Meet the Staff Team ...

Manager: Robyn Kapp joined the Association shortly after its foundation in 1975, serving as President from 1978 to 1983 when she was appointed Executive Officer until her retirement in 2006. Throughout this period she worked to develop appropriate state, national and international approaches and programs for the care of people with Huntington's Disease and their families. She also served as National President (1985-1997) and President of the IHA (1993-1997). Robyn, who has a BA in psychology and mathematics, is delighted to be back at the Association, currently two days per week, to ensure the Association remains on track in fulfilling its aims and objectives to provide and enhance quality services for people with HD and their families. Robyn has a daughter and enjoys reading, gardening and travelling. She also works part-time in her local church.

Administration Officer: Toni Ling Zhang started with the Association in September 2009 initially as a volunteer to assist with accounting and administration. In March 2010 she was employed in her current role which involves accounting and administration duties. Toni has a Master of Professional Accounting from the University of Sydney and a background of accounting in private business. As she was more interested in using her skills in the not-for-profit sector, she is glad to be on staff of the Association. Toni enjoys working with the team and meeting our clients. In her spare time she likes to read, swim and travel.

Administrative & Activities Assistant: Lily Ma

Since joining the Association in September 2007, Lily has been working part-time, assisting in the running of our Lunch Club and Holiday Camps. Lily also undertakes a wide variety of everyday administration tasks. She considers her role at Huntington's NSW to be the most rewarding job of her career and is keen to contribute in any in way she can to the future of the organisation. Lily has two daughters and enjoys bushwalking, swimming and craft.

Family Support Worker: Ramona Watts has been with the Association for 2½ years and has loved every minute of it. Together with Lily, she manages our fortnightly social group get-togethers and has made some wonderful connections with our members and their families. Together with staff from the NSW Huntington Disease Service (Westmead) she assists with the running of the Family and Friends Support Group (Carers' Group) at Elsie Court Cottage. Ramona also provides regular in-service education and support to care facilities that have residents with HD. She has a background in Psychology and Communications, and has been working with people with disabilities in the not-for-profit sector for the past six years. Ramona has three children and likes to keep fit by going to the gym and swimming.

Family Support Worker: Mark Bevan has been on staff since June 2010, initially working in northern NSW. However this year he will also be visiting southern NSW and the ACT. His connection with the Association began in the late 1970s when his father-in-law was diagnosed with HD. Since that time he has been involved in various ways, including serving on the board and volunteering for our holiday camps. Mark is married to Karen and they have two children and ten grand-children. His working history includes the NSW Police Force, Pastor of a church and many years in corporate life. Mark also works part-time as Chaplain in a school. He enjoys spending time with family and friends, reading, riding his motorbike, and is involved in a local church.

Camp Breakaway

Our annual camp on the Central Coast is fast approaching. Interest is high! But we are still inviting members with HD who might be interested in joining us for five days at Camp Breakaway, San Remo on the Central Coast, Monday 12th March to Friday 16th March inclusive to apply to come to Camp.

The Camp is an excellent opportunity for our members with HD to connect with people who are in a similar situation to themselves, to talk, share experiences, participate in organised activities, indulge in great food, and be with friends.

All participants need to be independently mobile and able to dress, toilet, and shower with NO assistance. Planned activities include walks by the lake, a modified version of lawn bowls, time to relax, and craft. Preference will be given to people who have not previously attended one of our camps.

Please call us at the Association on 9874 9777 or email Lily on lily@ahdansw.asn.au for a camp application form.

Camp Volunteers Needed!!

If you are interested in coming with us to the beautiful Central Coast and helping us with the Camp we would love to hear from you. You can come for the whole 5 days or for just part of the week. If you want more information about volunteering at the camp, please call Ramona at the office on 9874 9777 or email her on ramona@ahdansw.asn.au

Social Club News

Social Club is back in full swing after a short break over the Christmas period following our successful Christmas party.

Our members at Social Club had a wonderful day to remember as we celebrated Christmas together at the end of last year. We spent the week before decorating our lunch room with tinsel,

putting presents under the tree and making colourful Christmas baubles together, and of course shopping for and preparing our wonderful feast.

We served up traditional mouth-watering delights like succulent turkey with rich gravy, creamy mashed potatoes, roast vegetables, Christmas pudding smothered in custard and ice cream, and lots and lots of chocolate!

After morning tea we had the highlight of our day — when we were presented with the beautiful voices of Caleb, Abigail, Noah, Beth, Bella and Jeziah who charmed their way into our hearts with their joyful renditions of Aussie Bush Christmas, We wish you a Merry Christmas, Jingle Bell Rock, and many other favourites.

Our first outing for this year saw us at Bobbin Head National Park despite the weather threatening to rain on our picnic. We were lucky enough for the rain to hold off, and the sun actually shone on us as we enjoyed a scrumptious barbeque under the shade of the trees!

Family & Friends Support Group

It was wonderful to see so many carers at our annual Christmas party at West Ryde in December. Our carers enjoyed a much needed break as they came together to celebrate a time of friendship and sharing amongst people who have seen them through good and bad times. It was a lovely opportunity to catch up, chat, laugh and share as we enjoyed the delicious local pub fare and good drink!

Our family and friends support group meets on Wednesdays each month, and we would welcome new members.

It's a great time to 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.

2012 Sessions

22 February, Wednesday, 10.30am
21 March, Wednesday, 10.30am
18 April, Wednesday, 10.30am
16 May, Wednesday, 10.30am
13 June, Wednesday, 10.30am
11 July, Wednesday, 10.30am
22 August, Wednesday, 10.30am
26 September, Wednesday, 10.30am
24 October, Wednesday, 10.30am
21 November, Wednesday, 10.30am
7 December, End-of-Year Get-Together - TBC

To RSVP and for further information,
please contact:
Jet Aserios: (02) 9845 7528
Social Work Department
Westmead Hospital

Have suitcase will travel – to the South Coast, South West and the ACT

It has been our desire to provide a consistent approach to support Huntington's families in regional areas across New South Wales. However, for various reasons and due to different circumstances, we have at times, not been able to achieve this.

So, with a view to improving the situation and providing that consistent and equitable support to all families in regional areas, we have made some changes in responsibilities for our support staff. The result is that Mark Bevan will take up responsibility for the support of the NSW South Coast, South West NSW and the ACT.

So if you live in or somewhere near Bega, Bermagui, Bateman's Bay or Bomaderry; Canberra, Cooma, Cootamundra, or Currarong; Gundagai, Griffith or Gerringong; Wagga Wagga, Willandra, or Wandandian, he would love to hear from you.

Mark has been providing support to Huntington's families in the northern half of the state for the past 18 months, visiting and communicating with people in those areas on a regular basis. He will arrange at least two visits to each region per year, and work with families in each area with a view to delivering support appropriate to each situation.

Mark would love to hear from you if you are connected with a Huntington's family in those areas. If you want to talk about arranging a visit when he is in your area, if you want to talk about any Huntington's related issues, just contact Mark emailing him at mark@ahdansw.asn.au or calling him on 0410 629 850. He would love to hear from you.

Launch of the Huntington's Disease Youth Organization's Website Celebrated Around the World

First organisation to focus on providing much needed support to young people affected by HD

The Huntington's Disease Youth Organization (HDYO) is pleased to announce the launch of **www.HDYO.org** on 6th February 2012. The organization developed specifically to support young people affected by Huntington's disease (HD).

The website began as a dream of one and a need by many. Matt Ellison, a 23-year-old from England, was the visionary to bring the HDYO website to life. As a young person witnessing his Dad's progression of HD, he knows firsthand of the difficulty that surrounds a young person impacted by HD.

"The impact of HD on your life can be huge, and yet the support available to young people is miniscule," said Ellison, Founder of HDYO. "I felt this lack of support and resources was unacceptable and that something needed to be done. Young people deserve to be recognized and provided appropriate support." said Ellison. With help from other young people affected by HD around the world, Ellison and the team have made great strides towards improving the much needed support with the launch of the HDYO website. The site contains educational information created specifically for young people, by young people. The material includes articles, illustrations, videos, audio and personal stories.

The goal of HDYO is to empower youth with knowledge about HD and provide a supportive community to help each other cope with realities of the disease.

"The website is truly a remarkable effort of Matt and many other young people who are dedicated to a cause," said BJ Viau, HDYO's U.S. Board Member. "Now the challenge is spreading the word and helping others realize we are here to support them."

In order to support young people worldwide, HDYO's material is currently being translated into a dozen different languages by a team of more than 20 translators – many of whom are young people from around the globe who want to help other young people in their region gain more access to information about HD.

HDYO Board of Directors

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BJ Viau (USA)
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Brynne Stainsby (Canada)
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Michelle O'Brien (Australia)
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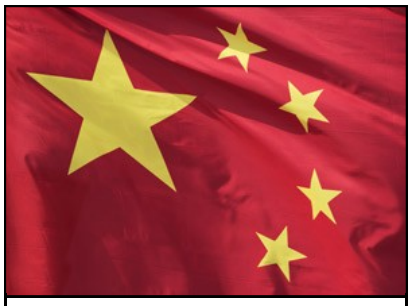
Chinese Huntington's Disease Network Launched



The Chinese HD Network will benefit HD families in China, and boost the global research effort to find effective

Chinese Huntington's Disease Network launched. Good news for China, good news for the global HD community

Huntington's disease is relatively rare. In western countries, estimates of the number of people with symptoms of HD are about 1 in 20,000 people. It's been suggested that HD is even rarer in China, but of course China is a huge place. HDBuzz attended the launch of the new Chinese HD Network to learn whether this is true, and what a Chinese HD Network could mean for HD patients around the world.

What do we know about HD in China?
Not much is known about how much Huntington's disease there is in China. In general, researchers believe it's less common than in Europe and America. But given the large size of the Chinese population, even if incidence is relatively low, there may be a large number of HD-affected people living in China.

Even in places with advanced medical systems and decades of knowledge about HD, accurate estimates of incidence are hard to come by. Recent work in the United Kingdom suggests that we may have underestimated the incidence of HD in that country by as much as half.

Many factors make it hard to produce accurate estimates of the incidence of HD. In some places, cultural or social factors mean that families care for HD patients, with little medical support. And stigma about neurodegenerative disease can be strong, which can worsen the isolation of patients and families.

The importance of clinical networks
Given the rarity of HD, it's important that patients have access to physicians with some expertise in the disease. The average neurologist or family doctor will have very little direct experience with HD, making it difficult for them to give good advice and care to patients and families.

One way of developing expertise for HD families is setting up networks of physicians who have expertise in HD. Centers in these networks can serve as sources of care and advice for otherwise isolated patients.

Many countries recognize that HD clinical networks are good for HD families. In the US, the HDSA supports 'centers of excellence' across the country. The European Huntington's Disease Network serves a similar role in Europe. And 2011 saw the launch of a new Latin American Network — the Red Latinoamericana de Huntington (RLAH). These networks support care, as well as fueling and organizing important research efforts.

The Chinese HD Network
There have been reported cases of HD in China, but so far the physicians seeing these patients have not been linked together in a formal way.

Jean-Marc Burgunder, a physician and scientist with positions in both Switzerland and China, says that approximately 300 Chinese HD patients have been recently seen in clinics there. Furthermore, he suggests, "these figures will probably change as time goes along", thanks to increasing awareness of HD among both physicians and families.

"All HD families deserve hope, regardless of language, culture or nationality"

To help speed this along, the Chinese HD Network was launched December 10th, 2011 at a meeting of scientists and physicians in Shanghai. Professor Burgunder, Zhong Pei, Shang Huifang and Chinese researchers hope to increase awareness about HD in China, and improve the quality of care available to families there.

The crowd at the launch of the Chinese HD Network

Chinese HD families

Patient advocate Ken To, from Hong Kong, addressed the launch meeting on behalf of patients and families. Ken's mother suffers from HD, and he himself is at risk for carrying the mutation that causes the disease.

Ken spoke eloquently about what he called the two big challenges of being an HD family member. First, the burdens of caregiving for an affected relative, and the feelings of "fear, guilt and powerlessness" that strikes the patient, as well as family members. Underlying this is another fear — the risk of inheriting the mutation from an affected parent.

The challenges highlighted by Ken To are recognizable to HD families regardless of their geographical location. It is clear that these challenges are compounded by isolation in countries with less developed awareness of HD. Ken will work with the Chinese HD Network to link families and share the burden of fighting against this disease.

Global benefits

Linking together specialist clinics serving HD patients in China would clearly be a huge benefit for them and their families. But could it have a broader impact for HD families elsewhere in the world?

One global benefit of building a Chinese HD network is to simply find more patients to participate in clinical research. Testing treatments for HD will require large numbers of subjects, and it's likely that in the near future, more drugs will need to be tested at the same time. Having access to more patients eager to participate in therapeutic trials helps the entire community run trials more quickly.

Less obviously, it's important that we learn about the diversity of HD to understand it more completely. We suspect that a large amount of the variation in HD symptoms is caused by environmental factors — like diet and lifestyle — and genes other than the HD gene, that might influence the disease. If HD behaves differently in China, that could give us important clues to both types of those factors.

But which of these factors are important, and can any of them be modified to alter the course of the disease? So far, we don't know. Having a diverse population of patients enables scientists to look at a broader range of factors when considering these types of questions.

All HD families deserve hope, regardless of language, culture or nationality. It's wonderful to see the launch of the Chinese HD network.

Acknowledgements: By Dr Jeff Carroll, Edited by Dr Ed Wild, HD Buzz

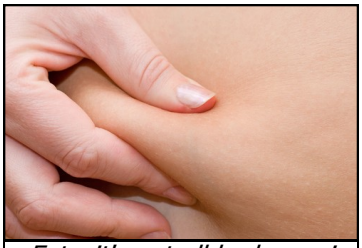
Do you have a Story to Share?

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

Special 'brain fat' injection helps HD mice

Directly injecting the brains of HD mice with a good type of fat called ganglioside GM1 provides dramatic benefit

The human brain is enriched in many different types of fat. Some of these greasy molecules are reduced in HD, and a new study demonstrates that replacing one specific type, called ganglioside GM1, leads to dramatic improvements in the behavior of HD mice.



Fat - it's not all bad news! Ganglioside fat molecules are one type of fat that's good for the brain

Fat molecules and the brain

'Fat' is a broad word with a bad reputation. But the word basically describes any greasy substance in cells and the body. The biology of fat is actually quite complicated - there are a number of different types of fat that have special roles, particularly in the brain.

The brain relies heavily on fat for its normal function. Like electrical wires, the projections of the brain are coated with an 'insulator' that helps signals travel long distances without being lost. The insulators that surround these projections in the brain are made of fat - so, far from being a bad thing, fat is critical to normal brain function.

Gangliosides in HD

In 2010, one interesting type of fat called ganglioside GM1 was shown to be reduced in HD patient cells and brains by a group lead by Simonetta Sipione at the University of Alberta.

'Gangliosides' are special types of fat that act, not just to insulate our brains' wiring, but to signal important messages themselves. We know that gangliosides are important, because terrible childhood diseases result from genetic mutations that affect the body's normal handling of them.

Brain problems are a common feature of the diseases caused by mutations affecting gangliosides, suggesting that whatever roles gangliosides play, they're critically important to brain function. When Sipione and her colleagues observed reductions in ganglioside GM1 they were left with a question - could replacing these important fats help with HD?

GM1 replacement

To address this question, Sipione and her team turned to mice carrying a mutant copy of the human HD gene. These mice develop symptoms, particularly movement problems, that might be similar to symptoms in HD patients. Like HD patients, these mice also have reduced levels of ganglioside GM1.

The simplest possible way to replace something that's missing is to just inject it, and that's just what this team of scientists did. To focus on the role for gangliosides in the brain, Sipione's team used tiny pumps to deliver GM1 directly into the brains of mice for 4 weeks. They were able to measure the levels of GM1 in the brain and determine that this approach was successful - levels of GM1 increased to normal after infusion.

The mice got better

So, did this replacement therapy work? Yes - and surprisingly well, judging by the observed improvements in the behavior of the mice. The mice were injected with GM1 at 5 months of age - while young for a human, this is about middle age for a mouse. By this stage, the mice already had problems with coordination, and infusion with GM1 led to a total reversal of these problems. The team used several different tests to understand how the behavior of the mice has improved, and the treated mice aced all the tests.

Mice treated with GM1 showed much better coordination when tested on a 'rotarod' apparatus

This is exciting stuff - many treatments in HD mouse models are begun at an early age, before any symptoms start. This would be much harder to do for humans because ethical and safety concerns make it very difficult to imagine treating HD mutation carriers from birth. Treating animals after they already have

symptoms of disease is much more relevant for understanding what might happen if we tried this in humans, but surprisingly most animal trials are not run this way.

How did it work?

Because we don't fully understand all the jobs performed by gangliosides in the brain, it's hard to say precisely why GM1 injections were so beneficial. But Sipione and her team were curious to know if there were any changes to things that we do understand that might explain the observed benefits.

The huntingtin protein, mutated in HD patients, is modified in cells after it is made. One of the ways it's modified is tagging the protein with small chemical labels that can change where the protein goes within the cell, as well as other, less well understood, effects.

To help understand the GM1 results, Sipione turned to Ray Truant, an expert on these types of modification of the huntingtin protein at McMaster university. The two teams looked at two specific 'addresses' within the huntingtin protein, called S13 and S16. We know that adding 'phosphate' tags at these spots makes the mutant huntingtin protein much less toxic.

In agreement with the idea that more phosphoryation at these two sites is good news, Sipione's team found more phosphorylation in mice treated with GM1. This gives some pointers to scientists as they try to understand the mechanism that GM1 uses to protect cells.

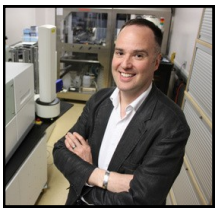
Human potential

The result from Sipione and her team is very exciting, but could it lead to trials in humans? Generally, it's very difficult to project how well these kinds of studies will 'translate' from animals to humans, but in this case there are some good signs.

First, GM1 has been used in humans in clinical trials with a good safety record. Simple safety is a huge hurdle for many drugs. Furthermore, in a small trial of 5 patients GM1 has actually been infused into the brains of Alzheimer's Disease patients for 1 year with no major adverse events. This type of brain delivery is tricky, so it's good news that GM1 seems well tolerated after this kind of direct infusion.
These are very early days for GM1 as a therapy for HD - many steps remain if it is to be tested in human patients. But the level of improvement of the mice is striking, and we should look forward to future studies on the potential role of GM1 as a therapy in HD.

Acknowledgements: By Dr Jeff Carroll, Edited by Dr Ed Wild, HD Buzz

Comments from Dr Ray Truant



Dr Truant said that while they expected to halt the disease's progression or minimize its neurodegenerative effects, they were surprised to see the mice back to "completely normal". "Not only did we halt the disease, but somehow the brain had managed to repair itself."

With their paper out, they are now at the phase where they are looking for drug compounds that can mimic the effects of GM1, Truant said. "I'm optimistic in that I think there's going to be, in the three-to-five-year timeframe, a major advancement in clinical trials as a result of this," he said. "It may not be GM1, but it may be a drug that acts like GM1."

And the only way to test whether this research applies to human therapy is to conduct small-level clinical trials, he said.
Pharmaceutical companies have encountered safety and efficacy issues with administering the lipid in humans because it is difficult to synthesize and the alternative option was to purify it from sheep brain, Truant said.
But interest in a possible drug has been recently resurrected because there are now apparently ways to make part of it synthetically and part of it by purifying from "super clean" sheep, he said.

Truant is cautiously optimistic. His fear is that people will start using that dreaded "C-word". "There are almost no cures in medicine, really. There are a lot of successful treatments. There are a lot of things that will improve the quality of life in people. And I think there's a real hope in this in that respect."

Acknowledgement: www.thespec.com/news/local/article/670711

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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: Don Ayres
Vice President: Brian Rumbold
Treasurer: Richard Bobbitt
Secretary: Judy Rough
Karen Bevan
John Conaghan
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