



Gateway

News from Huntington's NSW & ACT

Volume 21 No 2

Winter 2018

Walk 4 Hope

5 years on and still going strong

We would love you to join us for one of our walks in September to mark the 5th Anniversary of our very successful awareness and fundraising venture.



ORANGE SUNDAY 2ND SEPTEMBER

PARRAMATTA SUNDAY 9TH SEPTEMBER

SPEERS POINT SATURDAY 15TH SEPTEMBER

CANBERRA SUNDAY 23RD SEPTEMBER

REGISTER NOW AT

[HTTPS://WWW.EVERYDAYHERO.COM.AU/EVENT/WALKFORHOPE2018](https://www.everydayhero.com.au/event/walkforhope2018)

Support Group Meetings

West Ryde Carers Support Group

21 Chatham Road, West Ryde
Wednesdays at 10.30am—12.30pm

18th July	15th August
12th September	24th October
7th December, Friday, Christmas Lunch	

For further information contact Cecelia Lincoln on 8890 7528 or Robyn Kapp on 9874 9777.

Newcastle Carers Support Group

Jesmond Neighbourhood Centre
42 Mordue Parade, Jesmond
Thursday, 10.00am—12noon

12th July	9th August
13th September	11th October
8th November	13th December

For further information contact John Conaghan on 4922 3076 or 0434 605 999

Port Macquarie Support Group

Westport Club, 25 Buller Street.
Friday, 1.00am—1.00pm

6th July	3rd August
7th September	5th October
2nd November	7th December

For further information contact Kim Frumar on 0432 148055 or Robyn Kapp on 9874 9777

Wollongong Support Group

Wollongong Golf Club,
151-161 Corrimal Street.
Monday, 10.00am– 12 noon

2nd July	6th August
3rd September	October to be advised
5th November	3rd December

For further information contact Julie Lozano on 0429 720 478 or Robyn Kapp on 9874 9777

Canberra Support Group

Ainslie Football Club
52 Wakefield Ave, Ainslie
Saturday from 12.30pm

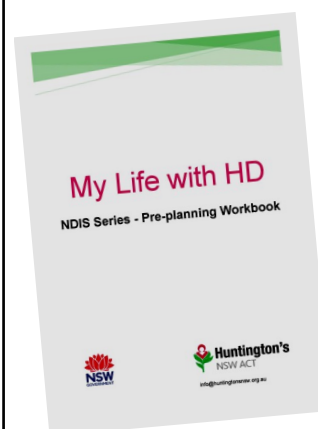
4th August	6th October	1st December
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For further information contact Felicity O'Neil at blis27@bigpond.com

New Resources

The following books are now available from the Association or on our website.

NDIS Pre-planning Workbook

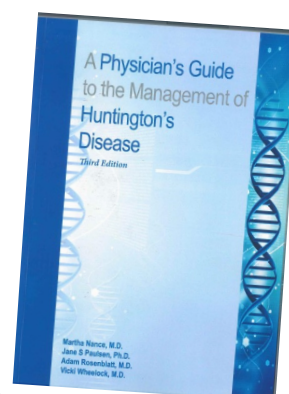


The Association received funding from the NSW Department of Family and Community Services to develop this booklet specifically for people with HD. Staff from the HD Service at Westmead

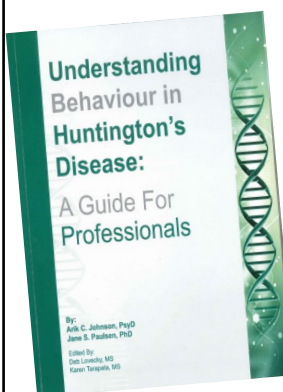
Hospital and John Hunter Hospital provided valuable information and advice. The workbook is designed for each individual to record information about their own particular circumstances. It's a must for everyone applying for NDIS funding!

Physician's Guide and Understanding Behaviour

The Huntington's Disease Society of America gave permission for the Association to publish these two publications.



The Physician's Guide covers topics such as the movement, cognitive and psychiatric aspects of HD as well as genetic counselling and the management of late stage HD.



Many topics are covered in this book including the stages of HD, communication, perception, apathy, impulsivity, denial and depression.

If you would like a copy of these books to give to your doctor or health professional, please ring us on 9874 9777.

From the Executive Officer

Dear Families, Members and Colleagues

Since 2004 the Association has received funding from the NSW Government to provide a Carer Support Program. Initially it was for carers throughout the state, however in 2010 we decided to concentrate on carers in rural and regional NSW due to their social and geographical isolation.

Last August, the NSW Department of Family & Community Services (FACS) advised that they were planning a significant redesign of the Carers Grants Program to take effect from 1st July 2018. We applied for funding from the new Carers Investment Program, but unfortunately we were not successful and therefore our funding has ceased. Consequently, the Rural Social Work position will not be continuing in its present form.

Nevertheless, we will be investigating other opportunities for funding and also making representation to all Members of the NSW Parliament regarding the loss of funding and the impact that this will have on our families.

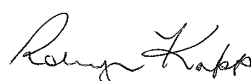
Please be assured that the Board is well aware of the situation and we want to stress that we will be doing our very best to continue supporting our families in rural and regional NSW.

Here is our plan of support for the next six months:

1. You can ring me on 9874 9777. I am usually in the office on Monday – Wednesday and Friday. I can also be contacted on 0456 013 612.
2. Our Youth Worker, Amy Hale, who is a social worker, will also be helping out and she is available by ringing 9874 9777.
3. The HD Service at Westmead Hospital provides a valuable service to all HD families throughout NSW and they can be contacted on 8890 9960.
4. John Conaghan, Social Worker at John Hunter Hospital has many years of experience working with our families and he is also willing to take calls – he can be contacted on 4922 3076 or 0434 605 999.
5. We intend to run more Talk-Link programs in conjunction with Carers NSW.
6. We will continue to support the existing Support Groups in rural and regional areas.
7. I am planning to undertake a number of rural visits from July to December.

I would like to emphasise that I am personally committed to supporting our rural and regional families and I encourage you to contact me if you would like to discuss this further.

In friendship



Membership Renewal

Yes it's that time of year again. Membership renewals for 2018/2019 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. Alternatively you can renew your membership on line at www.huntingtonsnsw.org.au

Donations to further the work of the Association are very welcome and we would be most grateful!

Research Update



7 June 2018

June update: Getting to know Roche & Genentech, RG6042 (formerly known as IONIS-HTTRx) **Huntington's disease development programme**

Dear global Huntington's community,

As many of you are still getting to know us (and vice versa), we would like to take this opportunity to tell you more about our company, philosophy on working with the HD patient community, commitment to collaborate to advance science, and the investigational molecule RG6042 development programme.

Roche & Genentech: one company, two names

Roche is a global biotech company focused on advancing science to improve people's lives. We were founded 122 years ago in Basel, Switzerland and now have a network of more than 94,000 employees working in 100+ countries. We believe in:

- *Investing in and following the science.* We invest more on research and development than any other healthcare company – last year alone over 10 billion Swiss Francs (~\$10.5 billion US dollars) – and we've translated that science into approved therapies that have fundamentally changed the way numerous conditions such as cancer, haemophilia, and multiple sclerosis are treated.
- *Innovation and focusing on areas of unmet need.* We aim to transform how diseases can be treated, and we've earned various Health Authorities designations – including 21 breakthrough therapy designations from the US Food and Drug Administration. We certainly hope to transform the way in which HD impacts your families.

Given all the communications about Roche in HD, we want to clarify our company name. Globally and in most parts of the world, you know us as Roche, but in the United States our pharmaceutical division is called Genentech. This is due to the 2009 company integration of Roche and Genentech, a US-based company and the world's first biotech company. What's important for you to know is that Genentech = Roche

pharmaceuticals in the US, and we are one company working seamlessly together.

Partnering with the Huntington's disease community

At Roche and Genentech, we are proud of our history of working with patient groups. Our goal is to be a trustworthy partner and for all partnerships to reflect common values of integrity, maintenance of independence, respect, equity, transparency and mutual benefit.

We have dedicated people and teams at both the global- and country-level focused on developing sustainable collaborations with patient communities. Open and constructive dialogue is crucial. This helps you know what can be expected from us, and it helps us better understand how to serve patients, carers and physicians, and to focus our activities on areas that are most beneficial to the communities we serve.

Collaborations in HD to advance scientific progress

Since our partnership with Ionis Pharmaceuticals started five years ago, we have had the privilege of working with leading experts and HD groups to advance the scientific understanding of HD and mutant huntingtin lowering. Collaborations have led to:

- Design of the first-in-human huntingtin lowering clinical study and follow-on open label extension study,
- Optimisation of a mutant huntingtin protein (mHTT) assay or measurement test, and
- Development of clinical and digital endpoints to better understand and measure the impact of HD and disease progression.

Since taking over development of RG6042 from Ionis at the end of 2017, we have and will continue to engage with the community (e.g.,

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South America & HD – One Year after the Vatican Visit

We have known for some time that Huntington's disease exists all over the world. Many people are aware of the contribution that Venezuelan families made in helping find the gene that causes the disease, but HD also exists in other countries in South America.

In May of last year, HD families from around the world, but especially from South America, met the Pope in the Vatican to let the world know more about living with the disease. Australian HD families know all too well the life-long challenges they face. In many other countries, people also have to cope with additional problems. Services are either non-existent or very limited - there is no Medicare or Disability or Aged Pensions, and poverty is an additional barrier to the help and support which can lessen the load a little.

In February of this year, I had the honour of meeting three amazing people working to help HD families in Colombia, South America. Social worker Mariela Campo Oviedo is a powerhouse of a woman. HD is in her family. In 1998 she was living in Medellin, and on a trip home she saw the toll the disease was taking in her home town of El Difícil, a community of about 20,000 people in the north of the country. She decided she had to do something to help. In her words, "I couldn't stand to see how much suffering was in the one home." There is a very large cluster of HD in the north of Colombia, and she formed a patient organisation called FUNCOVULC (Colombian Foundation for Vulnerable Communities) in 2011. She works directly with families, lobbies the government and health services and has highlighted their plight in the media. As a result of the Vatican trip, there are plans for a rehabilitation centre in El Difícil for HD families.



Therese Alting, Dr Gustavo Barrios & Mariela Campo Oviedo

In 2011 she met Dr Gustavo Barrios, a neurologist from Bogota who cares for HD families. For the past few years, every three months he travels to the north of the country, on the Caribbean coast, on his own time and at his own expense. There he sees the families Mariela has organised who have the greatest need. I had the privilege of

sitting in on one of his clinics. People had travelled for hours to come to see him and were so grateful for the help they received from Mariela and Gustavo.



Dr "Nacho" Ignacio Munoz-Sanjuan and Mariela Campo Oviedo with some of the children they help.

In Medellin, I managed to meet up with another Huntington's hero in Colombia - neuropsychologist Sonia Moreno, who has been helping HD families for over 30 years. She works at the University of Antioquia - their Neuroscience group currently provides services to 38 extended families. In addition to disease specific help, they also assist with housing, helping the children get an education and sometimes providing food. There are many other people working to help HD families, though there is no national organisation and help very much depends on where you live and what you can afford.

The difficulties faced by HD families throughout South America have been brought to a wider audience in the last few years. Many people have been involved, with one of the prime movers being Dr "Nacho" Ignacio Munoz-Sanjuan. Originally from Spain, he is involved in HD research at CHDI in Los Angeles. In 2012 he launched Factor H (<https://factor-h.org>), and in just a few short years the organisation has helped many HD families throughout South America. In Colombia, Factor-H assists Sonia, Mariela, Gustavo and many others in their work.

The most well-known achievement was the Vatican trip of May 2017. The teams from Factor H, HDdenmore and many other people around the world organised the meeting of Pope Francis with families, from Argentina, Colombia and Venezuela. The three HD advocates I met in Colombia all reported that the visit had achieved positive things. The attention that HD received, and acknowledgment of their struggle had given families hope - that crucial ingredient that we all

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patient groups, medical professionals, health authorities, payors, etc.). We commit to incorporating diverse perspectives in the design of the RG6042 development programme, as well as contributing to the advancement of the broader scientific understanding of HD. This is a commitment and journey we share with the HD community.

RG6042 development programme update

The Phase I/IIa study evaluating RG6042 in people with early HD has completed. This is an exciting time for HD, but there is still much work to be done before it can be determined if RG6042 can slow the relentless progression of HD. Big questions exist such as:

- What are the effects on lowering mHTT over a period of time longer than the 13-week Phase I/IIa study?
- Do any unexpected safety concerns emerge when we treat a larger group of people for a longer time?
- Does sustained treatment slow or stop the progression of HD?

We recognise the medical urgency that exists in HD and our team is committed to answering these big questions with other studies in early HD, collaborations with the HD community, and engaging global Health Authorities on the design of a global clinical development programme.

What's happening next?

We are planning studies that can provide Health Authorities with enough data to assess the benefits and risks of the investigational molecule RG6042, while also balancing speed and efficiency.

- A longer, larger global study. As previously announced, we are in the planning stages of a global study designed to detect clinical benefit and evaluate longer-term safety in early stage HD. Details about the study, including eligibility criteria, planned start date, and study sites around the world, will be announced as soon as these aspects are finalised.
- Additional studies. We are also committed to conducting smaller, targeted studies including:
 - ◇ The ongoing open-label extension study of RG6042 for those who participated in the Phase I/IIa study. This study looks at the safety and tolerability of longer-term dosing of RG6042, among other measures.
 - ◇ A "natural history" study to further understand the role of mHTT and

disease progression in the absence of any active treatment. This small study is also in the planning stages and not yet open or enrolling.

We understand that families may wish to seek access to investigational medicines as soon as possible. However, access to RG6042 can only be through clinical trial participation at this time. Because the benefits and risks of RG6042 are not fully understood, we are not able to grant pre-approval, compassionate use or "right-to-try" requests.

With the support of the HD community we are working with urgency and care to develop an appropriate clinical development programme that answers important questions around RG6042. We look forward to providing you with additional updates in September.

Sincerely,



Mai-Lise Nguyen, on behalf of the Roche HD team
Patient Partnership Director, Rare Diseases

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need in life - especially those living with the challenges of HD.

Further good news is that in July of this year, the first ever Huntington's Disease meeting covering the whole of South America will be held in Colombia. The Enroll-HD study has already begun in Argentina and Chile, and two sites in Colombia, Bogota and Medellin, are close to beginning.

One of the strengths of the Huntington's community is our desire to reach out to all people with the disease. In Australia, families from the 1970s up to the present day have joined forces to end the decades of isolation they had experienced. This activism resulted in the thriving HD organisations, better services and research collaborations which have led to these exciting times, especially with the recent advances in gene silencing. Let's hope there is also help for people in less well-developed countries. If you would like to know more about the disease in Latin America, the Factor-H website is a great starting point.

Therese Alting
theresealting@yahoo.com.au

Enroll-HD Congress in Quebec City

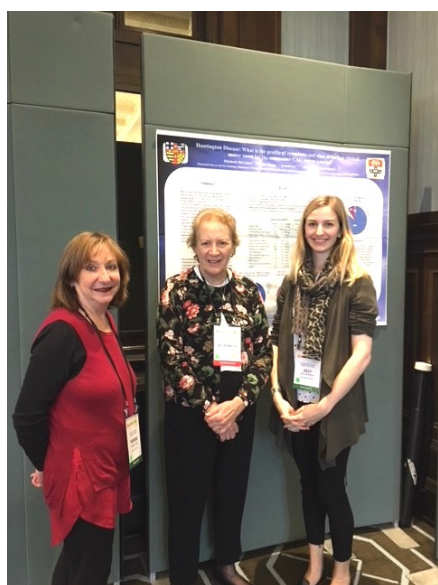
Many of you may have heard of the Enroll-HD study, which is now the biggest observational study of HD ever conducted. Beginning with 305 participants in 2012, over 17,500 people globally have now joined the study.

In May, 2018, the first Enroll-HD Congress was held in Quebec City. The meeting was organised to bring together those involved in the study, as well as other researchers, to discuss the exciting new developments in HD research, and the future directions for Enroll-HD.

Dr Bernhard Landwehrmeyer, the study's Principal Investigator, gave an inspiring talk on research already using Enroll-HD data. The blood samples each participant provides are held in a repository in Italy, and the study organisers have approved 150 separate requests to access this data for further research into HD. Dr Jim Gusella, one of the main people involved in finding the gene in 1993 gave a talk and there was a combination of both long-term and new researchers who are doing their best to find effective treatments for HD.

Three people from the Huntington Disease Service at Westmead Hospital had the privilege of attending this inspiring meeting, alongside other colleagues from Western Australia and Victoria.

Dr Elizabeth McCusker was invited to speak and gave a thought-provoking talk on the subject of patient-reported outcomes in HD studies,



Therese Alting, Dr Elizabeth McCusker & Jillian McMillan

focussing on the importance of factors such as a person's insight into their situation. Her wide-ranging talk was based on her long years of clinical and research experience, backed up by evidence from previous research especially the Predict-HD study. Jillian McMillan and Therese Alting attended in their role as Study Co-ordinators – much information was given about future directions for the study.



Westmead Hospital Study Co-ordinators, Jillian McMillan & Therese Alting

Two sessions were set aside for Study Co-ordinators to share experiences and challenges in conducting the study.

The Enroll-HD study is still open for those who are interested in being involved. The current focus of recruitment is for people who are gene positive but without symptoms, or in the very early stages of the disease.

If you would like to see the conference programme, click here: <https://www.enroll-hd.org/wp-content/uploads/2018/05/Enroll-HDCongressProgramBook.pdf>
The Enroll-HD website has a lot of information about the study: www.enroll-hd.org

If you want to know more about joining the study, feel free to contact us:
therese.alting@health.nsw.gov.au

The Enroll-HD study is funded by CHDI, who also funded the travel and accommodation costs of all of the Congress participants.

Westmead Hospital Research Team.

New collaboration seeks to speed Huntington's disease drug licensing

Critical Path Institute launches new initiative to get HD drugs licensed as quickly as possible

Recently the Critical Path Institute announced a new effort - the Huntington's Disease Regulatory Science Consortium, or HD-RSC. This collaboration with many international partners aims to speed the development of new HD treatments. What's all this about, and how could it help HD patients?

What is C-Path?

The Critical Path Institute is likely new to most Huntington's disease community members, but they're not new to the fight against human diseases.

In 2004, the US Food and Drug Administration (FDA) launched an effort to modernize and speed the development of new therapies. As a result of that effort, in 2005, the FDA and partners founded and funded the Critical Path Institute (or C-Path) with a goal of speeding the development and reducing the costs of bringing new drugs to patients who need them.

To do its work, C-Path brings together teams of organizations that are working on a specific problem. These C-Path consortia work to streamline drug trials in their area of interest, as well as developing new tools to improve the quality of trials.

As an example, C-Path has a consortium focused on Alzheimer's Disease which they call Critical Path for Alzheimer's Disease, or CPAD. This consortium of regulatory agencies, drug companies, researchers and advocates focuses on developing new tools to accelerate the development of new therapies for Alzheimer's Disease.

What does C-Path do?

How do groups like CPAD work to improve or

speed clinical trials? As an example, CPAD has developed a sophisticated computer model that tracks the progression of Alzheimer's Disease by compiling huge amounts of existing information about Alzheimer's Disease patients. Alzheimer's Disease clinical trials are done by a number of different individual organizations, and the data is not always shared between trials.

This is where C-Path comes in - they specialize at pulling together information about the disease stored with drug companies and academic researchers around the world. Based on real data from dozens of drug trials, C-Path's computer model allows organizations thinking of

developing a new Alzheimer's Disease model to essentially do a practice run of their trial in a computer simulation.

This allows any organization with a new Alzheimer's Disease drug to plug in a few numbers about how well they think their drug will work, and get feedback about how likely a given trial design is to be successful. That can be enormously powerful for

trying to figure out how many patients to enroll in a study, and how to divide the people in the study between the arms of the study.

C-Path's consortia have also worked on other critical roadblocks to rapidly completing trials. Several of their consortia have focused, for example, on helping researchers develop a new biomarker to simplify or shorten clinical trials.

There are a few kinds of biomarkers, but ultimately they are precise measurements that serve to tell us about how a disease is progressing in a human - say, a brain scan for HD. They can also report on how well a drug is doing based on changes that drug leads to in body chemistry or other measurements.

Because of their history and membership, C-

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Regulatory agencies like the FDA and EMA need very specific evidence before drugs will get licensed. The HD-RSC will work to speed up this process.

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Path understands how regulators, like the FDA and the European Medicines Agency (EMA), think about drug approval. This puts them in a great position to translate the science that researchers generate, into the measurements and outcomes that regulatory agencies need, to prove that drugs are safe and effective.

C-Path's Newest Consortium: HD-RSC

"This puts them in a great position to translate the science that researchers generate, into the measurements and outcomes that regulatory agencies need, to prove that drugs are safe and effective"

Recently, the official launch of C-Path's newest consortium was announced - the **Huntington's Disease Regulatory Science Consortium**, or HD-RSC. The consortium was launched in partnership with the CHDI foundation, a non-profit dedicated to rapidly developing meaningful treatments for HD.

The HD-RSC involves a huge range of players in the HD world beyond C-Path and CHDI, including drug companies (ten in fact, including Roche, Sanofi, Teva, Wave Life Sciences and others), regulatory agencies and patient advocacy organizations (including the HDSA, HSC and EHDN). These diverse organizations all have their own expertise and interests, but all of them are committed to developing new therapies for HD patients.

Last November, representatives from all these organizations came together in Silver Spring Maryland for a kickoff meeting. Your humble HDBuzz author attended to get a sense of what to expect from the HD-RSC. Over two days of meetings (program available at the link), dozens of participants talked about how best to speed clinical trials for HD.

Excitingly, the attendees included very high-ranking members of the FDA, including Eric Bastings (Deputy Director), and Billy Dunn (Division Director, Division of Neurology Products). These are the folks that are in charge of the review of new HD drugs being tested. Far from standing on the sidelines, the FDA attendees sat front and center in the first row, taking notes and asking a number of questions. It seems that these regulators get HD, and they're interested in trying to speed the safe development of new therapies for HD families. This consortium sums up the collaborative spirit of the HD community and our determination to

make progress as quickly and efficiently as possible.

By the end of the meeting, the organizations comprising the HD-RSC had organized themselves into 5 working groups. Each of these smaller groups is focused on helping solve a specific problem - developing new biomarkers, for example.

Another exciting goal embraced by the HD-RSC is the problem of how to design trials of drugs for presymptomatic HD mutation carriers - meaning people who carry the HD mutation, but who don't yet have HD symptoms.

Stopping HD symptoms before they start is the goal for everyone working on HD, but it's complicated to understand how we would design a trial and monitor people who don't yet have any symptoms to measure. Excitingly, this working group suggests that some really savvy folks think this is an important problem and have agreed to spend time working on creative solutions.

Take home message for families

We're entering a new phase in the fight against HD. Incredible drugs, designed specifically for HD, have moved from labs around the world into the clinic. The feeling many in the community had that "no one cares about HD" is being replaced by the realization that HD is a very hard, but solvable, problem.

Groups like the HD-RSC are great news for us because they mean that smart people are working hard, that they're collaborating and most importantly - that all these organizations feel that HD drug development is an exciting place to work. Stay tuned for exciting new ideas from the HD-RSC.

Acknowledgements:

*By Dr Jeff Carroll; Edited by Dr Ed Wild
HD Buzz; <https://en.hdbuzz.net/259>*



Darkness and Light

By SIMON ROACH

The confirmation that a person will develop Huntington's disease can bring them more uncertainty — but also relief.

Mark Newnham has seen the future, and it's etched on his father's face. Despite being in good health, the 31-year-old knows that Huntington's disease is coming — he just doesn't know when.

Like many people living under the shadow of the condition, Newnham, who lives in London, first heard about Huntington's disease when it struck older members of his family. A great-uncle had been diagnosed with it at the end of his life. So when Newnham's father started to develop the involuntary movements associated with the condition, he got tested for the gene mutation responsible. His father's diagnosis meant that Newnham — who was 20 years old at the time — had a 50% chance of carrying the gene. "I didn't know what Huntington's disease was when my Dad told me that he had it," Newnham says.



Mark Newnham chose to discover whether he carries the gene mutation for Huntington's disease.

In the ten years since, his father's symptoms have progressed to include more severe involuntary movements, memory difficulties and mood swings. Later, his driving became worryingly erratic. Thinking about those years, in which his father's mental health began to decline, is painful, Newnham says — he feels as though he has been witnessing someone "at war with himself, every day".

Throughout his early twenties, and despite his father's illness showing him what might await, Newnham did not want to take the genetic test that would reveal whether he had inherited the mutation for Huntington's disease. "I was more of a free spirit," he says. "I thought, 'I don't need to know.' I can get on with it and just see if it happens later on in life."

That attitude reflected his general approach to life. As an actor and musician, he launched from one project to the next with little thought about what would come later. "I wanted to headline the Glastonbury Festival, and I wanted to become the next Johnny Depp," he laughs. "Those were my goals."

He continued on that path, he says, until he met his partner. Finding happiness and stability changed his perspective on Huntington's disease — especially when the couple thought about having children. Could he face rolling the dice when he might pass the condition to his offspring?

Newnham sought genetic counselling through the UK National Health Service, during which he explored the impact that testing could have on his life. This involved considering his motivation for being tested, as well as changes that he might need to make in the event that he did have the mutation. Aside from the emotional strain that such testing can bring, it also raises questions about physical care and finances; the certainty of knowing you have the mutation can make it more difficult to get long-term health or life insurance. Genetic counsellors can help those who might be affected to pick through the entangled pros and cons.

After three sessions, and given his and his partner's desire to have children, Newnham concluded that he needed to know his status with respect to Huntington's disease. "We didn't want to have a

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child without that certainty," he says. The result was not what he had hoped for. Like his father, he carries the mutated gene.

Newnham is yet to experience symptoms, and it could be decades before he shows signs of the disease. He still works as an actor and musician, but says that his priorities have changed. "The test results made me realize that what really drove me as a person before, and what ambitions I had, they're not as important now," he says. The dream of performing at Glastonbury will never be gone, but spending time with family and friends seems more important. This shift in perspective has given him a quiet contentment, he adds.

More willing to look ahead, Newnham and his partner immediately began to explore how they could have a child who would not carry the Huntington's disease mutation.

"I wanted to make sure that I don't pass this on to the next generation," he says. That meant going through a process called preimplantation genetic diagnosis (PGD).

In PGD, embryos created through in vitro fertilization (IVF) are screened for specific genetic disorders; only those without the related mutations are implanted. In England, up to three rounds of PGD are available at no cost to people who meet certain criteria. (In the United States, many health-insurance plans won't cover the process, so people typically pay US\$15,000–25,000 for IVF with PGD.)

Not everyone with a family history of Huntington's disease goes to these lengths. Some leave it to chance. And various religious groups have reservations about prenatal genetic-screening methods such as PGD because any embryos found to have genetic abnormalities will be destroyed.

For those who do elect for PGD, the chances of success are low — as Newnham and his partner found out when they received the news that their journey towards parenthood had, for now, come to an end. The IVF part of the process, itself a complicated procedure, had failed and there were no embryos to test.

For now, the couple are weighing up the options. Adoption is a possibility, but people who will go on to develop conditions such as Huntington's disease tend to be at the bottom of the list because of their own care needs later in life, Newnham says.

Despite the prospect of a life without children, Newnham does not regret his decision to get tested. At least, he explains, he is moving forward with his eyes open. And advances in research fill him with "immense hope" that some form of treatment will be available in his lifetime — too late for his father, perhaps, but soon enough that the risk of having a child with Huntington's disease might no longer be one of life or death.

*Acknowledgement: Simon Roach is a freelance writer in Glasgow, UK.
Nature, Vol 557, 31 May 2018*

Do you have a story to share?

If you have a story that you would like to share in "Gateway" please contact Robyn at robyn.kapp@huntingtonsnsw.org.au



Huntington's NSW & ACT Inc.

PO Box 178, West Ryde, NSW 1685
21 Chatham Road, West Ryde, NSW 2114
Telephone: (02) 9874 9777
Free Call: 1800 244 735 (Country NSW only)
Email: info@huntingtonsnsw.org.au
Web Site: www.huntingtonsnsw.org.au

Huntington's NSW & ACT Inc.

The Association is a not-for-profit organisation established in 1975.

Our Mission

The energies and resources of Huntington's NSW & ACT 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: Stephen Guthrie
Secretary: Therese Alting
Members: Katy Clymo
Alison Hill

Association and Other Useful Contacts

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Hunter HD Service

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Predictive Testing

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