

# HD News

WINTER  
2023

RESEARCH 3  
formation of Huntington  
protein found

FUNDRAISING 4  
Walk 4 Hope dates

HA UPDATE 6  
the countdown is on

## A-GLIMMER PROJECT

ARTICLE FROM MONASH UNIVERSITY

### BACKGROUND

In Australia, life insurers are legally allowed to use applicants' genetic test results to discriminate – meaning that they can deny applicants life insurance cover or increase the cost of their cover based on genetic test results. This is called genetic discrimination.

Health insurance is protected from genetic discrimination by a specific Australian law and therefore applicants cannot experience genetic discrimination when applying for health insurance.

Research shows that because of fears of genetic discrimination, many people who are at risk of genetic conditions choose not to have clinical genetic testing (which could lead to preventative action or early treatment) or choose not to be involved in genetics research.

### PROJECT RATIONALE

Given there is no current government oversight of life insurers' use of genetic test results, it is critical that the impact, effectiveness and appropriateness of the moratorium is monitored, taking into account different stakeholder perspectives (consumers, healthcare providers, researchers and the financial services industry). That is the goal of the Australian Genetics and Life Insurance Moratorium: Monitoring the Effectiveness and Response (A-GLIMMER) project

The aims of the recommended policy change in this area are:

- To reduce consumer fears related to insurance, which deter the uptake of clinical genetic testing and/or research participation
- To eliminate genetic discrimination in the Australian life insurance industry

- To remove a barrier currently compromising the success of genetic medicine in Australia
- To ensure government oversight and monitoring to combat concerns with industry self-regulation

### STUDY DESIGN

A-GLIMMER will apply a mixed-method, multi-site approach to evaluate the impact of the insurance moratorium across four different stakeholder groups: Patients and consumers; Health Professionals; Financial Industry and Genetic Research Community.

Now that the recommendations have been made, the real work begins in seeking implementation from government! We encourage you to reach out to your local MP and ask them to raise this issue with the Ministers for Health, Financial Services, and the Attorney-General.

Download the Final Stakeholder Report here <https://tinyurl.com/z49sefya>



## FROM THE DESK *Lewis Kaplan*

### LEWIS KAPLAN, CEO Huntington's NSW ACT

As I write this, we are examining every aspect of the Association's current and past operations to ensure that, as we move ahead to merge with Huntington's Australia, the organisation's rich history is preserved in the right way. Our most important records, such as management committee minutes, are to be kept in perpetuity and have been scanned. Some of the oldest paper records were looking pretty faded!

We continue to expand in the lead up to the merger, having just advertised for a 0.4 FTE (2 days a week) HD Specialist who will provide professional facilitation for a number of our support groups as well as on-going support for our growing counselling service.

We have also been in the fortunate position of making an internal promotion: Justine Amos has been appointed to a development position

to become our next NDIS support coordinator over coming months.

A brief reflection on the merger process demonstrates that people of goodwill and with similar objectives can come together (almost completely virtually due to COVID-19) and forge something which will be significantly greater than the sum of its parts. The first draft document about a possible merger was written in July 2020, only a few months after five of the six state associations had agreed to collaborate. We signed a collaboration MoU in July 2020, and the first formal monthly meeting of what became the "Consortium of Australian Huntington's Associations" was held (online) in August 2020. Following a detailed feasibility study undertaken in 2021-22, we were ready to ask members if they agreed to merge! Our Annual General Meeting in November 2022, along with members from the other four collaborating State HD Associations all agreed.

On a sadder note, we have learned that St Joseph's hospital is closing down next month, but the Sister Maria Cunningham Centre which houses the state's only specialist HD aged care beds will remain open under the continuing management of St Vincent's Care Service. The 4-bed short-stay

behavioral management beds will be transferred to a different provider. We are liaising closely with the Westmead HD Service in relation to this change. We will provide updates via social media and our website as soon as they are available.

On another sad note, Amanda Dickey and I will not be transferring with the other Association staff to Huntington's Australia when the merger is completed as there are no suitable roles for either of us at this early stage in its development. The board and the rest of the staff join me in thanking Amanda for her many years of service to the Association and to the HD community – initially as a community advocate/leader, then as a board member and for the past nearly eight years as a valued member of staff.

The board has asked me to stay around after the merger to look after the formalities of winding up the Association while Huntington's Australia carries our mantle forward, so this is not goodbye quite yet.

## MEMBERSHIP

It's that time of year again - membership renewals for 2023-2024 are now due.

The more active members we have, the better we can understand the needs of the HD community and speak on your behalf. We are asking members to renew their membership at the state level and during September you will be invited to become a member of Huntington's Australia at no cost for the first year.

You can renew your membership online by visiting the link below. If you are wanting to renew your membership but are suffering from financial hardship, please know that you can seek an exemption from the fee. We encourage you to reach out to us at the office.

<https://huntingtonsnswact.org.au/get-involved/become-a-member/>

# INITIAL STEPS IN HUNTINGTIN PROTEIN'S TOXIC FOLDING IN HUNTINGTON'S FOUND

**WORDS** MARISA WEXLER

Scientists have figured out the first molecular events in the formation of toxic huntingtin protein clumps in Huntington's disease.

The findings shed light about why only mutations that result in at least 36 repeats of glutamine (one of protein's building blocks) in the huntingtin protein cause the disease — and open new avenues toward possible treatment.

"For three decades, we've known that Huntington's and related fatal diseases occur when proteins contain more than around 36 Qs [glutamines] in a row, causing them to form chains of proteins in the brain, but we didn't know why," Randal Halfmann, PhD, the study's senior author at the Stowers Institute for Medical Research, said in an institute news release. "We've now figured out what the first link in the chain looks like, and, in doing so, have discovered a new way to stop it."

The study, "Pathologic polyglutamine aggregation begins with a self-poisoning polymer crystal," was published in eLife.

Huntington's is caused by mutations in the gene that provides instructions for making the huntingtin protein. As a result, cells produce an abnormal huntingtin protein that has a long tail, called the polyQ region, where glutamine is repeated excessively.

Mutated huntingtin protein containing a long polyQ region is known to form clumps, called amyloid aggregates, inside cells. Amyloid clumps made from other proteins are found in other neurodegenerative diseases like Alzheimer's and amyotrophic lateral sclerosis.

It's well established that Huntington's will only develop if there are at least 36 glutamine

repeats in the huntingtin protein, but the reason for this numeric cutoff hasn't been clear, prompting scientists to examine the amyloid nucleus of mutated huntingtin protein, looking at the earliest physical changes in the protein that makes it form clumps.

## EARLIEST STEPS IN FORMING AMYLOIDS

"This is the first time anyone has experimentally determined the structure of an amyloid nucleus even though most major neurodegenerative diseases are associated with amyloids," Halfmann said.

In simple terms, the setup involved engineering huntingtin proteins with specific numbers of polyQ repeats that were tagged with molecules that would light up when the protein folded in on itself.

They then made small adjustments to the polyQ region, changing its length or altering the types of amino acids at different positions. By examining how folding altered when these changes occurred, they came up with a model for the earliest steps in amyloid formation.

"A key innovation was to minimize the volume of the reaction to such an extent that we can witness its stochasticity, or randomness, and then we tweak the sequence to figure out what is governing that," Halfmann said.

Through these experiments, the researchers showed that polyQ folds in on itself like a zipper, with the individual glutamines acting as its teeth.

The scientists showed the initial step of this zipper-like folding occurs within a single huntingtin protein that folds in on itself. This suggests the amyloid nucleus is monomeric (requiring only one molecule) rather



than polymeric (requiring multiple molecules), a question that's long been debated.

This self-folding was only possible when there were at least 36 glutamine repeats. With shorter repeats, the polyQ region isn't long enough to fold into the zipper shape.

"Prior work in test tubes supports a monomeric nucleus, but this model has been controversial," Halfmann said. "We now have strong evidence that 36 Qs is the critical number for nucleation to happen in single protein molecules, and moreover, that this is how it happens inside living cells."

"Once formed, this minimal Q zipper germinates in all three dimensions to ultimately produce amyloid fibers with ... zippers longer than that of the nucleus proposed here," the researchers wrote.

These structures exhibited "self-poisoning" properties, where aggregate growth would slow at very high huntingtin concentrations because small protein aggregates start interfering with each other's conversion into the growing structure, results indicated.

This finding may explain why the severity of Huntington's doesn't seem to differ between people with one or two mutated copies of the disease-associated gene, the researchers noted. It also may open avenues toward new, "admittedly radical" possibilities for treatments, they said.

"We suggest that therapies designed to (further) [aggregate] huntingtin preemptively will delay [amyloid nucleus formation] and thereby decelerate the disease," they said.

## HIGH TEA 4 HD

During May we held three High Tea 4 HD events to raise awareness of Huntington's disease and fundraise. With over 250 people in attendance, across the three events, we have received \$18,956 in income.

We were fortunate to get some help along the way from Mulgoa Valley Receptions, The Hotel Canobolas, HD Awareness Orange, Collins Booksellers Orange, and Novotel



Newcastle Beach. A big shout out to these venues and organisations who partnered with us to run the High Tea 4 HD, we truly appreciate your support. I'd also like to acknowledge all of the people who helped secure prizes for our raffle and auction, many hands make light work. Lastly to our amazing guest speakers Ian, Kelly, Rachael and Jaime – you all have so much courage and told your story so well. Thank you.



## WALK 4 HOPE

Dates have been set for the Walk 4 Hope 2023 and we are really hoping the community will get behind us. When you sign up for Walk 4 Hope, you can choose the location and date that suits you best, or you can create your own virtual walk in your area. Remember to ask your family, friends and colleagues to sponsor your walk!

- **Newcastle** – Speers Point Park – Sunday 10 September
- **Canberra** – Weston Park – Saturday 16 September
- **Orange** – Robertson Park – Sunday 24 September
- **Parramatta** – Parramatta Park – Sunday 24 September

To find out more information visit our website <https://huntingtonsnswact.org.au/get-involved/walk-4-hope/>



**WALK4  
HOPE**  
HUNTINGTON'S NSW ACT

Newcastle - Sunday 10 September  
Canberra - Saturday 16 September  
Orange - Sunday 24 September  
Parramatta - Sunday 24 September

# ALL THINGS NDIS

## WORDS MALISHA FERNANDO

The last few months have been busy as we prepare for the merger as an organisation and as a Support Coordination team whilst going through the process of the annual external audit to continue NDIS registration and provide NDIS services.

We are thrilled to announce that Huntington's NSW ACT passed the audit with flying colours! The external auditors made mention of the positive feedback and specialised support Participants and Guardians experience working with a service that understand the spectrum of experiences and needs of those with Huntington's disease. The purpose of the audit

is to ensure that as an NDIS registered organisation, Huntington's NSW ACT follows and abides by the NDIS guidelines and practice standards that underpin the work we do every day. Undergoing regular audits ensures we focus on continuous improvement and provide support to our clients to the highest standard possible.

As business grows, so does our team and we will be training up our Support Services Administrator, Justine Amos, to become a Support Coordinator. Justine is eager to learn and start assisting our NDIS Support Coordination Clients with their NDIS Plans and ongoing support needs. Welcome Justine!

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## NEW HD YOUTH SPECIALIST

### WORDS GRACE JOO

My name is Grace, and I am excited to come on board the HD Youth Connection program. With a background in Social Work, empowering young people has always been a passion of mine.

Before joining Huntington's NSW ACT, I have worked with youth from all walks of life from the foster care system to refugee and asylum seeker background. I am always inspired by the resilience and strength that young people demonstrate

despite their circumstances. My role here will be to provide emotional and practical support to young people and their families navigating the complexities of Huntington's disease, whether it is in the form of weekly catch ups, advocacy or connecting with other organisations.

Perhaps one of the highlights of Youth Connection is our scholarship program, Ailsa's Legacy. Since its launch last year, we have had 7 successful recipients.

We understand how difficult it can be to continue education when dealing with extra responsibilities, limited finances and time with a family member affected by HD. The funds from the scholarship can be put towards courses, school excursions, camps, uniforms, computers, IT software, extracurricular activities and more.

To be eligible to apply you must meet the following criteria:

- Are you under 25?
- Are you enrolled in school, university, TAFE or other educational institution? OR are you planning to in 2023?
- Are you living with an HD affected parent or carer (or previously done so)?
- Do you live in NSW or ACT?
- Would receiving this grant help you start, continue or return to education in 2023?

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**Visit the website for more information**



WELCOME

*Grace*

Grace manages our Youth Connection program. As a social worker, Grace is experienced in community organising, education delivery and case management. However, her special interest lies in project development for young people and has worked in various youth sectors since 2015.

# UPDATE ON HUNTINGTON'S AUSTRALIA

## AN UPDATE FROM HA CEO LENNI DUFFIELD



It doesn't seem that long ago that we were all attending AGMs to vote to enable the proposed merger of the 5 state Huntington's associations. Fast forward 8 months and here we are, less than 4 months away from the merger and well and truly in the thick of it, with a fury and hive of activity! What have we learnt along the way ... that mergers are never easy, and they are rarely simple.

But why are they so challenging? Well, for a start you have the combination of three main things: people, operational structures/processes and technology/ services. The path to merger success isn't a steady, consistent, incremental process where each effort you

make toward integration is matched by parallel gains. In the early stages of consolidation, you put forth a lot of effort but seem to get little accomplished. Then you hit the yield point, and that last little bit of effort required gives great rewards. It is this last bit of effort that makes an imperishable difference and will be the catalyst for lasting change.

Right now, we are at the "yield point" – it is like an invisible barrier, it's turbulent just before you reach it, but smooth as silk once you break through to the other side. We would not have gotten to this stage without each of the states all working together to make a difference. Along the way we have shared resources, and team members, supported each and may have sworn and shed a tear or 2! But we are nearly there, and we can see the new beginning, and I know that our families, communities and staff across Australia will benefit from our collective and collaborative efforts to make Huntington's Australia a reality.

## AN UPDATE FROM HA CHAIR CHRIS GLASSON

Dear Members, Clients and Volunteers

Progress towards the merger is in full swing, so I thought I would give you a bit of an update on some important things that are happening.

Due to NDIS registration approval timeframes we have made the decisions to delay the merger date to 1 October. Although we still may obtain that approval earlier, time is running out for us to be able to manage all the things we need to, such as the transfer of all our existing grants, funding arrangements, service contracts etc. Whilst on the one hand disappointing, on the other it allows us to have things to a greater level of completion than originally planned, especially around our systems. There is always a silver lining. When the merger happens, it is our intention to make the changeover process for our current NDIS clients as smooth as possible.

The spirit of cooperation and collaboration that has been a hallmark of how we have gone about this merger from the very start is still being evidenced. Even though we have not actually merged yet, we have seen great examples of a willingness to work together and share resources to enable better outcomes for the HD community.

Some examples are:

- NSW is supporting Queensland to start early NDIS service operations in that State (under NSW's existing NDIS registration) by employing an NDIS Support Coordinator who will be located in Brisbane.
- WA has one of its experienced NDIS Coordinators heading off to Queensland for 2 weeks to support the roll out of that State's new NDIS operations.
- Queensland has agreed to second one of its HD Practitioners for 6 months to Tasmania to help provide much needed community support and service provision in that State. Other State HD Practitioners have been taking client cases for Tasmania to help them out in the interim.
- WA is lending one of its HD Practitioners to NSW to provide specialist support remotely to NSW youth clients while they recruit for a replacement.

These are wonderful examples of how working together provides tangible outcomes for our community nationally, but also provides great opportunities for staff to gain new experiences. The power of working together!

Regards  
**Chris Glasson**  
**Chair HA**

## HEAR FROM MEMBERS OF OUR COMMUNITY



Community Member

*Anne McMahon*

**Q: How did you get involved in the HD world?**

My mum had Huntington's and it has been in the family for a very long time. I recently lost my brother,

Peter, who had Huntington's for about 20 years before he passed. One of my sons (Christian) has the gene, my second child (Simon) doesn't want to be tested, and my other two children don't have the gene (Karen and Shane). My goal is to end Huntington's with my grandchildren – what we can't control we must endure.

**Q: What excites you about Huntington's Australia?**

Quite a few things! With a national identity Huntington's will get more attention, more awareness, more funding, and more resources. So many people I talk to haven't heard of Huntington's and it would be lovely to be recognised like Parkinson's and other diseases. Families will definitely benefit from more services and support.

**Q: What is your ideal Sunday?**

We have two granddaughters that sleep over every weekend and I love waking up with them on Sunday mornings. We usually have pancakes and then go the beach to play in the water and sand. If we aren't at the beach, we like to feed the Magpies in our garden.



Stakeholder

*Therese Alting*

**Q: How did you get involved in the HD world?**

My first introduction to HD was way back in 1993. I saw a person from a HD family as part of the

predictive testing programme. The enormity of the decision to get tested and his reaction really moved me.

**Q: What excites you about Huntington's Australia?**

For decades, the Huntington's community has lacked a national voice. It is so exciting that we'll be able to pool the resources of the State Associations to improve the lives of HD family members, regardless of where they live in Australia. It's a scary time, but full of possibilities.

**Q: What is your ideal Sunday?**

Warning - nerd-alert! Early morning walk bird watching, either in the bush or by the sea. Home for late brekkie of eggs from my chooks, plus wordle. Pottering around the garden with our kelpie and greyhound in tow, reading non-fiction, family dinner with lots of laughing. New York Times mini crossword to end the day.



Staff Member

*Cathy Holland*

**Q: How did you get involved in the HD world?**

Applied for a job which looked interesting (it was and still is) with Huntingtons Queensland. I have a big interest in genetic health after my daughter was born with a rare genetic condition and I remember how hard it was sourcing information when she was little. Being able to help people connect to find answers and support is what makes it so rewarding.

**Q: What excites you about Huntington's Australia?**

So many things! The collaboration of people from all over the country, bringing together all that knowledge and expertise. Becoming a louder collaborative voice to get federal government support. Provide more education to doctors and allied health professionals to enable individuals to get consistent and improved care. And most importantly our clients will benefit with services and information that will meet their needs and be more accessible across the country. Such an exciting time.

**Q: What is your ideal Sunday?**

Either a really lazy day with brunch and Netflix or an early start with a good long walk (preferably near water or bush). But my favourite Sunday is always spending time with my children (who are now grown up).



*The NSW ACT team enjoying lunch with Basil from Queensland.*



# Huntington's NSW ACT

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## Our team

**LEWIS KAPLAN**  
Chief Executive Officer

**MALISHA FERNANDO**  
NDIS Senior Support  
Coordinator

**MIA SMALL**  
NDIS Specialist Support  
Coordinator

**KATE MCNAMARA**  
NDIS Support Coordinator

**JUSTINE AMOS**  
NDIS Support Coordinator

**AMANDA DICKEY**  
Community Programs Officer

**GRACE JOO**  
HD Youth Specialist

**JULIE LETO**  
HD Counsellor

**STEWART SWALES**  
Administration Co-ordinator

**ALISON WEIR**  
Fundraising & Marketing

## HD SERVICE

**Westmead Hospital**

**DR CLEMENT LOY  
(DIRECTOR), DR SAM KIM,  
DR FLORENCE CHANG**  
Neurologists  
(02) 8890 6793

**FELICITY STEHOUWER**  
Social Worker  
(02) 8890 6699

**SANDRA BARISIC**  
Clinical Nurse Consultant  
(02) 8890 9960

**PETER KWONG**  
Clinical Nurse Specialist  
(02) 8890 9960

HD Clinic Appointments  
Outpatients Department  
(02) 8890 6544

**HUNTER HD SERVICE**  
**John Hunter Hospital**

**JOHN CONAGHAN**  
Social Worker  
(02) 4922 3076

**HUNTINGTON'S UNIT**  
**St Joseph's Hospital**  
(02) 9749 0215

**PREDICTIVE TESTING**  
Find your nearest Genetics  
Clinic at <https://www.genetics.edu.au/SitePages/Genetic-Services.aspx>

**EDUCATION RESOURCES**  
**Huntington Study Group**  
Global HD cooperative  
therapeutic research org.  
<https://huntingtonstudygroup.org>

**HDYO**  
Support and education to  
young people (aged up  
to 35) impacted by HD  
around the world.  
<https://en.hdyo.org/>

**RESEARCH QUERIES**  
**Dr Therese Alting**  
Enroll-HD Study  
(Westmead Hospital)  
(02) 8890 6310 or  
0438 604 719

**HDBuzz**  
Research news. In plain  
language. Written by scientists.  
For the global HD community.  
<https://en.hdbuzz.net/>

**Huntington's Disease Network  
of Australia (HDNA)**  
The map-HD registry.  
(03) 9902 0081  
<https://hdna.com.au>

# WHAT'S HAPPENING? COMMUNITY SUPPORT GROUPS



Take a look  
at how your  
region is  
choosing to  
connect.

## PEOPLE LIVING WITH HD SUPPORT

**WOLLONGONG (Social)**  
Wollongong, 11:30am – 1:30pm  
1st Monday monthly.

## CARER SUPPORT

**CENTRAL COAST**  
Postponed until a facilitator is recruited.

**COFFS HARBOUR**  
Korora, 11:00am - 1:00pm  
3rd Friday monthly.

**NEWCASTLE**  
Jesmond, 10:00am - 12:00pm  
2nd Thursday monthly.

**SYDNEY**  
Postponed until a facilitator is recruited.

## COMMUNITY SUPPORT

**ACT (Social)**  
Ainslie, 12:30am - 2:30pm  
1st Saturday bi-monthly.

**ORANGE & CENTRAL WEST (Social)**  
Quarterly  
Details to be confirmed, refer to our website.

**PORT MACQUARIE**  
Northhaven, 11:30am – 1:30pm  
1st Thursday bi-monthly

**Latest details and to register your  
interest can be found on our website at:  
<https://huntingtonsnswact.org.au/support-groups/>  
or contact us on 02 9874 9777.**