



HDNL 2017 - Issue No 68, Q1 17

## GENETICS

For information on Genetic testing contact the Genetics Centre - Tel: 01 409 6902 or [www.genetics.ie](http://www.genetics.ie)

## ENROLL HD

You can express your interest by emailing [enrollhd@beaumont.ie](mailto:enrollhd@beaumont.ie)



## MEMBERS MEETING & RESPITE WEEKEND - CUISLE

9th-11th June, 2017



**SPEAKERS: Dr Emma Yhnell,** Health and Care Research Wales Fellow, Neuroscience and Mental Health Research Institute, Cardiff University.

Dr. Yhnell has a strong research interest in Huntington's disease's and her work includes exploring computerised cognitive training for people with HD.

**Áine Ennis,** Senior Psychiatric Social Worker at Bloomfield Health Services is a member of a multidisciplinary team providing continuing care for people with Huntington's Disease.

**Information enclosed.**

# NEWSLETTER OF THE HUNTINGTON'S DISEASE ASSOCIATION OF IRELAND

# Hope

IRELAND

## HD Papal Audience

On May 18, for the first time ever, Pope Francis will recognise the devastating plight of those living with and affected by HD. He will host the world's largest gathering of the HD community.



POPE FRANCIS' SPECIAL AUDIENCE WITH THE HUNTINGTON'S DISEASE COMMUNITY IN SOLIDARITY WITH SOUTH AMERICA. May 18, 2017 - Vatican City



### YOU ARE INVITED

TO JOIN THE HUNTINGTON'S DISEASE (HD) COMMUNITY AT THE FIRST EVER PAPAL AUDIENCE WITH HD FAMILIES

Pope Francis' Special Audience with the Huntington's Disease Community in Solidarity with South America

**Hddenomore** (pronounced Hidden No More), an international coalition of HD patient advocates has come together to bring global attention to HD. The coalition includes Charles Sabine, former NBC war correspondent and television journalist; Dr. Ignacio Muñoz-Sanjuán, CHDI and Professor Elena Cattaneo, distinguished Huntington's researcher and Lifetime Senator in the Italian Parliament. The purpose of the event is to raise awareness of the condition and to lift the stigma around the disease. A warm welcome is extended to the entire HD community to attend. You can also help to raise awareness by sharing news on social media. See <http://hddenomore.com/>

## HDAI's Information Seminar



At HDAI's Information Seminar in Dublin

on March 4th, eighty five family members gathered to receive information on Huntington's Disease (HD) and get an update on research. The Seminar took place during National Brain Awareness Week, which was one of over thirty events organised around the country by neurological and brain related groups. Thomas Lillis, HDAI Chair, welcomed all, acknowledged funds received from Ireland Fund France to host the Seminar and introduced our three distinguished speakers.

**Dr. Niall Pender,** Principal Clinical Neuropsychologist, Beaumont Hospital commenced by noting the significant advances in research, the understanding of HD and of potential treatments, and recently the emergence of clinical trials to target the disease. He acknowledged the frustration in Ireland due to slow progress with clinical care, multi-disciplinary support and specialist services. Specialist clinics are necessary to improve treatment and care, there is work underway to improve HD services at Beaumont Hospital but this is dependent upon increased resources.

As current President of The Irish Brain Council, Dr Pender advocates for a national brain strategy. He noted that 38% of the EU population experience some form of brain disorder. The burden of brain disorder is twice that of cancer yet funding for brain disorder research and care is significantly less. Dr Pender observed that people facing a genetic neurological illness often tend to worry unduly over normal brain errors and assume that it is the start, or progression of neurological symptoms. A perfectly healthy brain is prone to errors including: forgetting a birthday, burning the dinner, texting the wrong person or not remembering where the car is parked. Undue worry over normal brain errors can increase stress and anxiety unnecessarily.

Dr Pender discussed the cycle of neurological illness and the need for effective interventions. Symptoms including involuntary movements, fatigue and tension impact on mood and can lead to depression and anxiety. In turn, this effects function and may prevent people from getting out, socialising and connecting with others. This takes away opportunities to feel good and reducing a person's sense of well-being. As a result people experience an increased sense of disability.

Attendees were reminded that symptoms of HD vary from person to person even within the same family. HD affects the basal ganglia and frontal lobe regions of the brain.

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These regions are responsible for coordination of movement, aspects of thinking, personality and behaviour.

People with HD may experience difficulties with social cognition - an ability to interact with other people, use humour, connect and empathise. Dr. Pender referred to Dr. Tom Burke's research on the emotional impact of caring for a person with personality and behavioural changes.

Caring for a person with a complex neurological condition can have a huge impact on the caregiver and how they react to the changes in their loved one. Studies being conducted with MND carers suggest that high rates of depression and anxiety evident among caregivers, make it harder for them to effectively care for a patient. It is important to have systems in place to protect and support care givers.

*Carers can learn skills and develop effective attitudes to manage these issues such as:*

- Do not overload a person with lots of demands - one task at a time;
- Little and often - small amounts of information regularly can be processed easily;
- Slow down;
- Reduce distractions where possible - focus on one task at a time;
- People with HD fatigue easily - take rests;
- Use memory aids: diaries, calendars and phones to assist memory (this doesn't make your brain lazy);
- There is no uniform pattern of cognitive changes in HD - symptoms can vary a lot between people - each person needs an individualised plan.

Dr Pender suggests that disease self-management programmes, available for a range of chronic conditions, could be modified for HD so that the impact of symptoms can be reduced. This is being studied for people with Motor Neurone Disease patients at present.

Techniques such as mindfulness can be useful in reducing stress and anxiety. The Mindfulness and Relaxation Centre at Beaumont Hospital is a good resource - <http://www.beaumont.ie/marc>

HD is a condition that develops slowly therefore it is important to put strategies in place to allow people to stay well and stay functioning in their jobs and relationships for many years. Factors to assist include:

- Lifestyle;
- Nutrition;
- Exercise for fitness and general well-being;
- Medication management - discuss worries with clinician;
- Relaxation to manage anxiety;
- Education on the illness;
- Skills - memory, emotion etc.;
- Communicating fears and worries - don't be afraid to talk about it.

Dr Pender mentioned the positive changes he sees in people being more open and willing to discuss their concerns and fears around HD.

#### **Enroll HD**

Dr Pender thanked those who have already joined the study and those who have emailed to express interest. He acknowledged delays due to limited resources but expressed his hope of increasing capacity at Beaumont in order to increase registration numbers over the coming year.

**Professor Orla Hardiman** gave a very informative talk on the Importance of Clinical Trials in Rare Neurological Diseases. A Clinical trial tests new drugs and treatments to see: whether they are safe; whether they have a positive effect and to determine side effects. Trials are necessary to provide factual information on the effectiveness of treatments.

Professor Hardiman outlined the many checks and balances involved in the clinical trial process. Regulatory authorities such as the Health Products Regulatory Authority (HPRA) in Ireland, the European Medicines Agency and the FDA in the USA are responsible for determining whether a treatment is safe before it can be administered in a clinical trial.

Ethics committees with representatives from different disciplines within a hospital must give their approval before a trial can commence. Trials are then monitored by a Data, Safety and Monitoring Committee.

#### **Why Participate in Trials?**

- New drugs will not be developed without clinical trials;
- People participating in clinical trials do better;
- People participating have a higher chance of getting the drug at the end of the trial;

Dr. Hardiman spoke of the lack of clinical trials for HD in Ireland currently but pointed to Enroll-HD as a significant step in attracting future trials to Ireland. She highlighted the benefit of patients and families working together with clinicians and scientists to improve research outcomes.



**Dr. Ed Wild**, Consultant Neurologist and Clinician Scientist at University College London's Huntington's Disease Centre, commenced his keynote address by reminding us that HD research is much further forward today than ever before. Success is reliant on the global HD community of family members, researchers, clinicians, health professionals and voluntary organisations working together. The discovery of the HD gene in 1993 is the best example of the community working together. Over 12,000 family members contributed data which led to the HD collaborative group's discovery. Organisations including the European Huntington's Disease Network and the CHDI Foundation continue to facilitate this networking.

Much of the recent progress in HD research is due to CHDI, a non-profit foundation dedicated to progressing HD research. It is the largest financial contributor to HD research made possible because of an anonymous US billionaire who has donated \$200 million dollars per year over the last ten years.

HD is a problem scientists believe they can solve. Laboratory science and animal models provides important information to researchers but observational research studies with HD patients and family members is also crucial. Dr Wild emphasised the importance of the Enroll-HD study and noted the benefit of having a study site in Ireland.

Dr Wild provided a summary of various trials to date and noted that while some past trials did not generate a successful drug they have contributed to greater understanding for researchers. There are currently 15 different trials of drugs designed for HD ongoing throughout the world.



This includes a Phase 1 trial for the gene silencing compound IONIS-HTRx which is delivered directly into spinal fluid. Dr Wild commended the 36 people currently involved in this trial in the UK, Germany and Canada, 25% of whom are receiving a placebo. So far this trial is meeting expectations for safety, the next step is to measure its effectiveness.

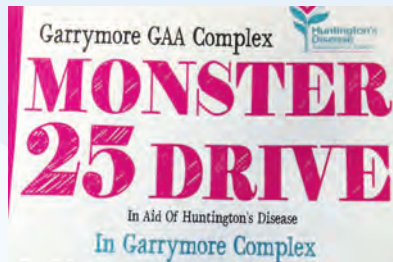
Dr Wild also gave an overview of other possible targets and new technologies being researched. In the past year virus technology has improved significantly and may facilitate delivery of drugs to the brain via the blood rather than through spinal fluid. Attendees were encouraged to contribute to the global research effort by participating in the Enroll-HD study. Enroll-HD is the database used to recruit participants for HD Clinical trials and the more people who register the better chance there is of bringing trials to Ireland.

**You can view all three talks on the Huntingtons Ireland You Tube channel or via our website link.**

## Fundraising - Thank You

### Garrymore GAA Club

Garrymore GAA Club in Co. Mayo held a Charity Card Drive on 31/1/2017 which raised an amazing €1,214 for HDAI.



Huge thanks to all who contributed prizes, sold tickets and helped organise the event, and in particular to James and Siobhan Heaney and to Ann Hannon for their hard work.

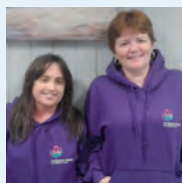
### Bank of Ireland Business Online

Thank you to David McGann and his colleagues at Bank of Ireland Business Online for their very generous charity contribution of €380. Bank of Ireland *Give Together* matched this contribution bringing the total donation to a very generous €760. Sincere thanks to all involved.

### Recently Received Donations

- Martina, Finnola and Zil - monthly Direct Debit donations
- Dulux Paint €300
- Porters Desk, Pearse College €45
- Online Donations €200
- Donations in memory:
  - Teresa Clarke €235
  - Mark Daly €130
  - Michael Towey €200

### Christmas Cards and HDAI Hoodies



Proceeds from HDAI Christmas cards continue to arrive. Thank you to Deirdre McInerney and colleagues at Mary Immaculate college for €257, Ann Hannon and her friends in Ballindine €264, Anne McErlean €70 and to Bernie McCoy and her colleagues at NCAD €90. We welcome themed photos or paintings for our 2017 card designs.

Thank you to Martin Conachey at Cabrini Clothing for providing fabulous HD Hoodies which have generated €390 for HDAI.

You can order hoodies online via [www.huntingtons.ie](http://www.huntingtons.ie)

## Membership

Thanks to all who have sent in 2017 membership subscriptions.

## Upcoming Events



Dublin Women's Mini Marathon entries now open. If you would

like to participate for HD we can provide t-shirts and sponsorship cards.

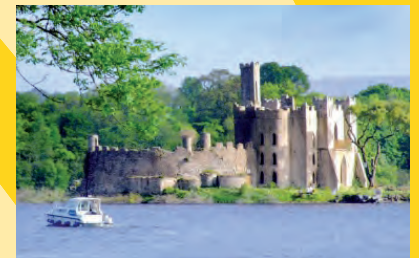
### Dunnes Stores Bag Packing

Dunnes Stores have kindly allocated **22nd - 24th June** to HDAI for charity bag packing in Donaghmede. We are looking for volunteers to bag pack for 2 hour slots over the 3 day period. If you are available to help please contact us on [info@huntingtons.ie](mailto:info@huntingtons.ie)

### Sunrise up the Reek

Family and friends of the late Eleanor Breen are planning another Reek climb in her memory. The Croagh Patrick Climb will take place on **Saturday July 1st 2017** and everyone is welcome to join in. If you want more info email: [John@hansonregan.com](mailto:John@hansonregan.com)

### HD Awareness Walk at Lough Key Forest Park - 15th July



Come walk with us to help raise awareness and funds (suggested donation €5.00 or sponsorship cards available). We will gather from 10.00am to start walking by 10.45am. Lough Key Forest Park, located just off the N4 between Carrick on Shannon and Boyle, is a beautiful spot for a stroll and a picnic!

**Minister Simon Harris** (above right), pictured with Thomas Lillis, HDAI Chair and Dr Higgins at Bloomfield Health Services in January for the launch of their HD Booklet.



## Light It Up 4HD

Huntington's Disease Awareness week takes place from **May 15th - 19th, 2017**. Iconic buildings including The mansion House, Christ Church Cathedral and some Dublin City Council buildings will light up to raise awareness.



Former Lord Mayor of Dublin, Criona Ní Dhálaigh with HDAI Chair Thomas Lillis

You can support this international #LightItUp4HD campaign by taking a photo or a selfie with a lit up building or by sharing images on social media. Contact [info@huntingtons.ie](mailto:info@huntingtons.ie) for more information.

## Follow HDAI on Social Media

**Facebook:** [https://www.facebook.com/pages/Huntingtons-Disease-Association-of-Ireland/121707387897701?ref=br\\_tf](https://www.facebook.com/pages/Huntingtons-Disease-Association-of-Ireland/121707387897701?ref=br_tf)

**Twitter:** @HDAI\_ie  
**Thank you to our volunteers for their social media work.**



**Huntington's Disease Association of Ireland**

Carmichael Centre  
North Brunswick Street  
Dublin 7

Tel: 01 872 1303

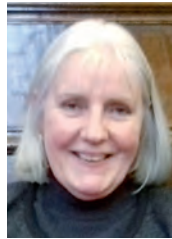
FreeFone: 1800 393939

Email: [info@huntingtons.ie](mailto:info@huntingtons.ie)

[www.huntingtons.ie](http://www.huntingtons.ie)

## Rare Disease Day 2017

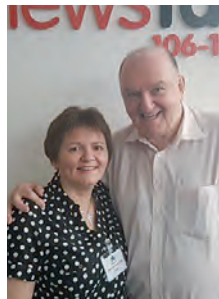
The 2017 Rare Disease Day Conference was a joint North / South event held at The Royal Hospital Kilmainham, Dublin on February 28th. The themes for this year's conference included: Updates on the Rare Disease Plans for Ireland and Northern Ireland, Access to new and innovative medicines and The Role of Genomics and Research in Rare Diseases. Speakers included patients and family members impacted by rare diseases, clinicians and researchers. Professor Eileen Treacy, National Clinical Programme for Rare Diseases gave an update on progress at a European level.



Rosemarie Kelly, Genetic Counsellor and Anne Lennon-Bird gave a joint presentation on The Patient Journey- Living with a Genetic Diagnosis. In her talk Rosemarie Kelly gave an overview of Genetic Testing and also highlighted the critical underfunding of genetic services in Ireland. Ireland has one of the lowest staff to population ratios in Europe.

Anne, a HD family member, powerfully described her family's experience of the genetic testing process and the implications of a HD diagnosis and predictive test results. She movingly described the unfairness of her young children being forced to become caregivers for their parent suffering from HD and the lack of psychological support available for her family.

Presentations from Rare Disease Day are available to view at: <http://www.mrcg.ie/go/news/news/rare-disease-day-speaker-presentations>



Also on the day, Anne joined Avril Daly, the Chair of the Genetic and Rare Disorders Organisation to speak with George Hook on Today FM's High Noon talk show. They discussed the impact of rare diseases on individuals and families and the need for greater awareness and services for people affected. Issues discussed included: access to

timely diagnosis; to care, treatment and therapies and to research studies and clinical trials. Avril Daly also outlined the grave lack of genetic services here in Ireland compared with other European countries. Anne discussed the symptoms of HD and it's hereditary implications in addition to discussing it's impact on her family. George thanked Anne and acknowledged that he was **"in awe of her courage"**.

## Brain Awareness Week

National Brain Awareness Week took place from March 6th to 12th with over thirty events organised around the country by neurological and brain related groups. The Neurological Alliance of Ireland and the Irish Brain Council held a keynote conference at Trinity College, Dublin on 7th March.

A position paper, Building a Supportive Framework For Brain Research in Ireland, was launched at the conference. The paper aims to outline the challenges and opportunities for brain research in Ireland and highlight the need for a supportive infrastructure to attract future investment, retain highly qualified personnel and ensure that research findings are effectively translated into better outcomes for people with brain conditions.

*"Brain disorders are a grand challenge of our time, a challenge that is growing with our changing demographics... Crossing the divide requires communities to come together; patients, clinicians, policy makers, researchers and more."*

Dr Graham Love, CEO, Health Research Board. You can download the paper from NAI's website and at: <http://bit.ly/2nfLd74>

## Acknowledge Kindness



HDAI's board nominated **Rosemarie Kelly** for flowers to acknowledge her work as

a Genetic Counsellor for the predictive testing process for over 19 years. Rose took early retirement in March.

## HDAI Support Meetings

All those impacted by HD are welcome to join us at our Support Meetings in Cork, Dublin, Longford and Limerick. Our Family Support Officer or a HDAI representative will host the meetings. Tea, coffee and scones are provided. Contact the HDAI office on 1800393939 for more information.

**DUBLIN**

May 6th  
Aug. 12th

**CORK**

July 1st  
Oct. 7th

**LONGFORD**

Sept. 9th

**LIMERICK**

Oct. 14th