

CONTINUING THE FIGHT ON BEHALF OF OUR MEMBERS



HDNL 2016 - Issue No 64, Q1 16

INFORMATION ONLINE

www.huntingtons.ie

Please follow us on **Facebook**
and **Twitter**

SUPPORT MEETINGS

See dates on page 4

SHOW YOUR SUPPORT

Membership forms for
2016 enclosed with this
newsletter

GENETICS

For information on Genetic
testing contact the Genetics
Centre - Tel: 01 409 6902 or
www.genetics.ie

SAVE THE DATE

Members Meeting and
Respite Weekend - Cuisle,
10th-12th June, 2016



Headline speaker -
Jimmy Pollard, International
HD Speaker and author of
"Hurry Up and Wait"

ENROLL HD

You can express your interest by
emailing enrollhd@beaumont.ie



NEWSLETTER OF THE HUNTINGTON'S
DISEASE ASSOCIATION OF IRELAND

Hope

IRELAND

HD Café

Family members are invited to attend a HD Café event with tips on **"Minding your Mental Wellbeing"** at Bloomfield Health Services Rathfarnham, Co Dublin on Wednesday 27th April at 7pm.

Refreshments served. Please contact HDAl on 1800 393939 for booking and more information.

Neurological Alliance of Ireland Conference

The Neurological Alliance of Ireland, with the support of the National Clinical Programme in Neurology, organised a half day conference in Dublin on 29th January 2016 to explore a range of issues in relation to research and treatment of rare neurological conditions in Ireland.

Titled **"Rare neurological conditions in Ireland: Exploring challenges and solutions in research and treatment"**, clinicians, researchers, patient representative organisations and people with neurological conditions gathered to discuss both challenges and possible solutions to improving access to diagnostics and treatments, as well as enhancing Irish research into rare neurological diseases.

Panel discussion with (l to r): Professor Tracey, Professor Lynch, Ms Patricia Towey, HDAl, Professor Hardiman and Professor Andrew Green (Geneticist)



Professor Orla Hardiman, Professor of Neurology at Trinity College Dublin, outlined the current landscape with regard to research and discussed necessary improvements needed before service development can proceed to adequate standards.

They are:

- Population-based databases: to allow accurate mapping of a disease and to ensure that nobody is lost to follow-up. This would also facilitate Irish inclusion in international clinical trials of novel agents.
- Centralised services with a *"hub and spoke"* model putting the *"patient at the centre"* and providing specialist treatment, while maintaining the person's quality of life within the community. Professor Hardiman discussed research showing that attendance at multidisciplinary clinics results in lower numbers of hospital admissions and better patient outcomes.
- Research embedded into clinical care. Professor Hardiman stressed that this approach allows population-based incidence and prevalence studies, identifies prognostic indicators, and can also identify sub-populations that can help discover new genes or susceptibility factors. Research also helps to inform health services, while good quality research can facilitate international collaboration.

Professor Hardiman explained that as many rare diseases have a genetic component, the integration of genetics into clinical care is vital. Genome-wide association studies (GWAS) can help to paint a picture of rare genetic diseases within the Irish population, while helping to clarify the genetic basis of a disease.

Patricia Towey, HDAl, gave an overview of rare neurological conditions from the perspective of a patient representative organisation.

(continued on page 2)

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Patricia echoed Professor Hardiman's comments in relation to the difficulties involved in a person's diagnostic journey when they have a complex condition that is not widely understood. She outlined the issue of delays in accessing services such as neurological and genetic services in addition to the negative impact of genetic discrimination. Patricia stressed the need for condition-specific expertise and care pathways to determine timely referral to multidisciplinary treatments and care. Adequately resourced neuro-rehabilitation services - including neuro-psychiatric and neuro-psychological services - community services and accessible palliative care are crucial services for people with HD and other rare neurological conditions. Patricia also mentioned positive international HD developments such as the Enroll-HD study and the collaborative work of the European HD Network.

Adult Neuro-genetics Clinic: Mater Hospital The first adult neuro-genetics clinic in Ireland, was piloted in the Mater in 2015. This combined clinic, which is led by one neurologist and one geneticist, seeks to address gaps in care of those with a heritable neurological disease. The pilot has been deemed a success and requires resources to develop.

Professor Tim Lynch, Clinical Lead for National Clinical Programme for Neurology and Edina O Driscoll, Programme Manager, outlined perspectives on rare neurological conditions from the neurology model of care: the framework document for neurology services which will be published by the HSE later this year.

Prof Lynch stated that as neurological conditions cross all health divisions from acute hospitals, mental health, social care, primary care and health & wellbeing, it requires an integrated approach.

Ms O'Driscoll stressed that staffing and resourcing within neurology must be addressed.

Clear lines of communication between hospitals and community, as well as support for GPs in managing chronic neurological conditions are essential. Equitable access to diagnostics including neuroimaging, as well as high cost medications, was also highlighted.

Development of Joint Genetics Laboratory: Collaboration between the Mater Hospital and Ireland East Hospital Group. Professor Des Fitzgerald, Professor of Molecular Medicine and UCD Vice-President for Health Affairs presented on an important collaboration to develop genetics services within the Ireland East Hospital group.

Professor Eileen Treacy, National Clinical Lead for Rare Diseases, delivered an update on the Rare Disease Strategy and the work of the Clinical Programme for Rare Disease. She discussed **Orphanet** a database of rare disease resources and the development of European Reference Networks (ERNs).

ERNs will facilitate Europe's top specialists working together to tackle complex conditions ERNs will allow the exchange of expertise and clinical data, and the aim is to improve high quality specialised care for patients with rare or low prevalence complex diseases. **(EHDN and the European Huntington Association are participating in the setting up of an ERN for rare neurological conditions).**

The Rare Disease programme is also seeking to develop care pathways for more common rare diseases.

Ronan Clarke spoke eloquently about living with Freidreich's ataxia, a rare neurological condition. He outlined how little people know or understand about his condition and often mistake it for other conditions. For further detail see: http://www.nai.ie/go/resources/nai_documents/report-of-nai-conference-on-rare-neurological-conditions

Rare Disease Day 2016 - "The Patient Voice"



Attendees at Rare Disease Conference

A conference entitled '**The Patient Voice**' took place in Dublin Castle to mark Rare Disease Day on February 29th. The conference was organised by the Genetic and Rare Disorders Organisation (GRDO), the Medical Research Charities Group (MRCG) and the Irish Platform for Patient Organisations, Science and Industry (IPPOSI). Presentations included the following:

Philip Watt, Chair of the Rare Disease Task Force, an umbrella group of patient networks and advocacy organisations, highlighted the need for progress on the implementation of the National Rare Disease Plan (RDNP) launched in July 2014. The resourcing of clinical genetics services in Ireland is fundamental to the diagnosis, care and treatment of people with rare diseases yet current clinical genetics staffing levels fall far short of European recommendation. "We need to see information on waiting lists for genetic services being made publicly available so that we can put in place the appropriate resourcing of this crucial speciality". Philip called for publication of the Smyth Report which outlines key proposals for the future of genetic services in Ireland.

Rachel Martin gave a moving account of living with Loeys-Diets syndrome, a rare genetic connective tissue disorder. She raises awareness and advocates for Marfan syndrome and Loeys-Diets syndrome.

Dr John Devlin, Deputy Chief Medical Officer and Chair of the RDNP Oversight Committee and Prof. Eileen Treacy National Lead for the Rare Disease Clinical Programme discussed progress on implementing the RDNP.

The National Rare Disease Office was established in June 2015 by the HSE and is based at the Mater Hospital, Dublin 7. Rita Marron, Administrator gave an update on their work - www.rarediseases.ie

Avril Daly, GRDO Chairperson, spoke of the progress being made across a range of disease areas in the development of new treatments and therapies. "We are hearing about ground-breaking research, with the ongoing development of clinical trials and emerging gene therapies. This brings so much hope." However, Irish healthcare delivery needs to progress without delay in order to take advantage of these developments.

Dr Derick Mitchell, Chief Executive IPPOSI discussed access to orphan medicine for people with a rare disease. Currently, the Irish health system does not have a funding process in place for orphan medicines. Information is required regarding the basis of funding decisions, the number of orphan medicines currently available, reimbursement costs and how these medicines are being prescribed / monitored. The patient's perspective should be incorporated in the process; in addition to the cost of not treating someone. Greater communication and transparency is required.

Dr Kevin Mitchell, Senior Lecturer in Genetics, Trinity College Dublin, and Wiring the Brain blogger, pointed to genetic diagnostics changing the future of patient care: *"The advances we are now seeing will place genetic testing at the front line of diagnostics - the question is how long are we prepared to wait for that to happen here.... If the health service in Ireland is to keep pace with international developments and provide the best care for patients, the role of genetic services will have to be greatly expanded."*

Richard Corbridge, Chief Information Officer with the Health Service Executive, discussed the need for Electronic Health Record (EHR) across the Irish Health System. *"This will mean faster diagnosis and more timely access to cutting-edge treatment and care"* See further info at <http://www.ehealthireland.ie/>

Eilish Hardiman, Group CEO of the Children's Hospital Group, **Denis Costello**, EURORDIS and **Dr Liam Farrell**, curator of #IrishMed tweet chat also spoke at the conference. Presentations are available online at <http://ipposi.ie/index.php/news-a-events-mainmenu-28/439-rare-disease-day-speaker-presentations>

HDYO European Youth Camp

August 7th - 13th - Applications Open

Four places are available for young people aged between 18 - 25 years, who are living in Ireland and are impacted by HD. The camp offers young people the chance to meet others from across Europe - to attend information workshops, to share experiences and take part in many fun activities.



Further information and link to application form: <http://www.huntingtons.ie/content/youth-support>

Fundraising - Thank You

Afternoon Tea for HD

Bernie, Micheál, Kevin and Oisín would like to thank all who attended and so generously donated to our fundraiser last December. A special mention goes to Ruth Bodin, Sandra Canning, Brigid Mc Manus and Majella McEvoy for their delicious home baking, your time and generosity are greatly appreciated. Austin Mann printed and delivered invitations, a superb job as always. Cecile Greneche Glynn donated one of her precious paintings for auction on the day. John Kelleher provided lovely airs on his concert flute. Thank you to the many friends who donated spot prizes on the day. Some local businesses generously gave spot prizes - Lilac Wines, Philipsburgh Avenue, Costello's pharmacy, Marino, Kennedy's coffee shop, Fairview, Lloyd's Pharmacy, Omni Shopping Centre and Life Pharmacy, Santry.



Thanks to all who donated after the event. It is so uplifting to experience such goodwill and we are ever amazed at the extra mile our friends, neighbours and family go to supporting us and the HD cause.

November for HDAI



Kevin Moran raised an excellent €370 for HDAI by raising sponsorship to shave off this impressive beard! "It was a pleasure to raise the funds for such a worthwhile cause".

Fingal Gospel Choir

A huge cheer for Fingal Gospel Choir for their marvellous energy and the beautiful sounds created at their Carol Singing Fundraiser on December 17th at the OMNI Park Shopping Centre.



Thanks to Choir Master Dave, to the wonderful singers and to all who volunteered and contributed on the night.

Waterfront Marathon



Michael Connern braved very challenging weather in early February to complete the Clonakilty Waterfront Marathon in aid of HDAI. Thanks to Michael, Sharon and all their family and friends for their remarkable efforts in raising €1,544.

Mount Sackville PA

Mount Sackville Parents Association Secondary School very kindly held a fundraising lunch for Huntington's Disease. Sincere thanks to all those who donated so generously raising a magnificent €3,000 for HDAI

Christmas Jumpers



A big thanks to Selina McQuillan who organised a Christmas

Jumper Coffee morning and raised €400 for HDAI. Thanks also to Veolia Energy who donated €322 through their charity matched funds initiative.

Christmas Cards

Thanks to all who supported and/or sold HDAI's Charity cards including Pat Davis, Angelina O'Shea, Deirdre & friends at Mary Immaculate College, Bernie Spellman & friends at Pearse College, Gurteen Post Office, Ann Hannon, Ann Long Hennessy and John Hennigan at Centra in Ballindine, Co Mayo.

UPCOMING EVENTS

HD Car Boot Sale - Clara Market on 8th May, 2016

Dublin to Wexford 180km Cycle on June Bank Holiday Weekend

VHI Women's Mini Marathon on Monday, 6th June

Donations

HDAI was heartened by the very generous donations received in December and early 2016. These included:

- Martina and Finnola for their monthly Direct Debit donations.
- Dulux Paint - €500.
- Pauline & Joe €150.
- Pearse College €200 donation.
- Anonymous donation of €80 received by post.
- Donations received online - Karen H, Rohana.

(donations continued overleaf)

In Memory



"Our beloved and devoted mother **Eleanor Breen**. She coped well with the

condition. We all miss her more than we can say!"

HDAI received €1,203.05 of donations in her memory. Our condolences and gratitude to her family and friends.



Michael John Higgins died on 21st January 2016, a beloved husband, father and

grandfather, he fought HD with dignity and good humour. Sincere condolences to his wife Rosemary, his children, grandchildren, siblings, relatives and friends.



Leo Grant died on 29th January 2016 aged 46 years. He will be lovingly remembered

by his brothers, sisters, nieces, nephews extended family and friends.



Theresa Clarke died on February 29, 2016 having struggled with HD for 20

years. Sincere condolences to her husband Larry, her brothers, sisters, nieces, nephews extended family and friends.



Huntington's Disease Association of Ireland

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FreeFone: 1800 393939

Email: info@huntingtons.ie

www.huntingtons.ie

(donations continued from page 3)

- Jack O'Shea €250 in lieu of payment from RTE News & Current Affairs.
- St Colmcilles Girls National School donated €150 in appreciation of the voluntary work of a retiring Board member.
- Mishcon de Reya's generous donation of €631.05 in remembrance.
- Kevin McCarthy, Tim Flood, Andrew Wolfin, Niki Stephens, Richard, Michelle & Finlay Tyler for their kind donations of remembrance of €322.

Acknowledge Kindness



We would love to nominate our stoic and loyal neighbours **Brigid Mc Manus and Noel Doyle** for a bouquet of flowers in recognition of their

constant practical help and support to our family over a long number of years.

We have been lucky enough to avail of their hospitality on many occasions - be it the finest herbal tea, strong coffee, full bodied well chosen red wine or a dinner party to die for, we've had it all in their house. Brigid and Noel showed their true colours at our last fundraiser on 5th December 2015. Brigid baked dozens of mini quiches and enough lemon tart to feed 60 people.

When I asked her for help she cleared her diary and produced the goods and her husband Noel delivered her baking to our kitchen on the day. This is just one of many examples of Brigid and Noel's generosity to our family and we are truly thankful to them.

Bernie & Micheál

EHA Website Launch

The European Huntington Association have launched a website:

<http://eurohuntington.org/>



Hospital Saturday Fund Grant



Paul Jackson, Chief Executive, HSF; Patricia Towey HDAI and Ardmhéara, Críona Ní Dhálaigh

Twenty-four Irish charities received donations from the Hospital Saturday Fund (HSF) at a special reception hosted in Dublin recently. In total, €106,500 was donated to the medical charities and organisations at the event, which was hosted by Ardmhéara/Lord Mayor of Dublin, Críona Ní Dhálaigh. HDAI received a generous grant of €4,000. Also among the recipients were Trinity College Dublin which received a donation to help support vital training by funding student Medical Electives.

Speaking at the reception, The Lord Mayor celebrated the extraordinary work of all the charities receiving donations from the HSF. The Lord Mayor, Críona Ní Dhálaigh, who nominated Our Lady's Hospice & Care Services, Harold's Cross as her chosen charity, remarked that "this donation means that the Hospice now has the benefit of a special rise recline chair which will provide comfort and support and benefit both patients and staff".

Paul Jackson, Chief Executive, Hospital Saturday Fund said: "We are delighted to continue the tradition of supporting many charities in Ireland, many of which are less well-known. The Hospital Saturday Fund is honoured to support the efforts of such deserving charities and help in some way towards the exceptional, tireless work that they do."

In 2016, the Hospital Saturday Fund will give €1,200,000 in donations and grants to medical charities for care and research, hospices and hospitals across the UK and Ireland. Assistance will also be given to individuals whose illness or disability has caused financial difficulties.

HDAI Coffee Mornings/Support Meetings



You are very welcome to join us at our Coffee Mornings/Support Meetings from 11.00am to 1.00pm on a Saturday in Cork, Dublin and Limerick (dates below). Our Family Support Officer or a board member will host the meetings. Tea, coffee and scones are provided. Contact the HDAI office on 1800393939 for more information.

DUBLIN

May 7th

CORK

Mar 19th
July 2nd

THE WEST

Cuisle, Roscommon (AGM),
10th - 12th June, 2016

LIMERICK

April 9th