

CONTINUING THE FIGHT ON BEHALF OF OUR MEMBERS



HDNL 2017 - Issue No 71, Q4 17

CHECK OUT OUR WEB SITE

www.huntingtons.ie

SUPPORT MEETINGS

See dates on page 4

GENETICS

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

ENROLL HD

The ENROLL-HD clinic at Beaumont Hospital is now growing and they hope to increase registration numbers at a fortnightly Enroll-HD clinic from January 2018. You can express your interest in participating by emailing enrollhd@beaumont.ie

 A worldwide observational study for Huntington's disease families

HOPE

NEWSLETTER OF THE HUNTINGTON'S DISEASE ASSOCIATION OF IRELAND

IRELAND

ASO Drug Reduces Levels of Mutant Protein in HD Patients

Ionis and Roche announce (11/12/17) that the first human trial of a huntingtin-lowering drug 'Ionis-HTTRx' successfully lowers harmful huntingtin protein in the nervous system and is safe and tolerated.

This is the news the HD community have been hoping for. For more details see Hdbuzz who do marvelous work in bringing us reliable research news: <https://en.hdbuzz.net/249>



Professor Sarah Tabrizi, Director of the UCL HD Centre said:

"the key now is to move quickly to a larger trial to test whether IONIS-HTTRx slows disease progression..."

...This is a great day in the HD community, and it sets us on the path to even more exciting work in 2018. For the first time in history, HD patients are being treated with drugs known to reduce the amount of huntingtin protein in their brain. Until we conduct the next trial, we won't know if this reduces the impact of HD. And while we know the drug is safe in the short term, we will also have to watch carefully for any long-term adverse effects. But we're facing this problem with renewed excitement and hope. It's the best early Christmas present we could have hoped for".

(Source: HDBuzz)

Congratulations to Prof. Tabrizi who received the 2017 Leslie Gehry Brenner Prize for Innovation in Science. The award was presented by Dr. Nancy S. Wexler, President of the Hereditary Disease Foundation (HDF) at the HDF 50th Anniversary Symposium and Gala at the Metropolitan Club in New York on November 6.

HDAI Thanks

HDAI would like to thank all of the brave volunteers who signed up and committed to this trial. This is a very promising time for the HD community and a great Christmas present!



Drs Wild and Carroll had reason to dance again following the research announcement - Source: Huntington Study Group

Morning Ireland

Anne, a HD family member and Dr Ed Wild who works with Professor Tabrizi at University College London's Huntington's Disease Centre were interviewed on Morning Ireland on 12th December regarding the exciting IONIS research announcement. You can listen to the podcast from HDAI's website: www.huntingtons.ie or check out HDAI's Facebook and Twitter page to see Irish and international press coverage.

Senator John Dolan, Independent Senator and CEO of the Disability Federation of Ireland (DFI) discussed the need for a dedicated HD clinic and a HD Clinical Nurse Specialist in the Seanad on December 12th. HDAI are very grateful for Senator Dolan's commitment to disability advocacy.

HD Research Updates at NECTAR Meeting

The Network of European CNS Transplantation and Restoration (NECTAR) held its Annual meeting in Dublin from 6th - 8th December. NECTAR's aim is to bring together European groups who share the common goal of protecting, repairing and restoring the central nervous system damaged through degenerative disease or injury.

Scientists explore cell transplantation and gene therapy as therapeutic strategies to improve the lives of patients with various neurological illnesses including Huntington's disease, Alzheimer's disease and Parkinson's disease.

(continued on page 2)

Christmas Cards

Thanks to all who have supported and / or sold HDAI's Charity cards. We welcome Christmas themed photos or paintings for our Christmas Cards for 2018.



(continued from page 1)

Dr. Eilís Dowd, NUI Galway is current President of NECTAR and kindly offered HDAl representatives the opportunity to attend the HD research presentations at NECTAR 2017. *The very impressive line-up of HD research presentations included:*

Prof. Lesley Jones, Cardiff University, Wales.

A genetic perspective on Huntington's disease pathogenesis.

Prof. Jones is the Lead Facilitator of the Genetic Modifiers Working Group (GMWG) of the European Huntington's Disease Network (EHDN). She was part of the collaborative effort to identify the first genetic modifiers in HD in 2015 and continues to work with other HD researchers to progress this work. Discovering which molecular pathways might contribute to the age at onset and progression of human HD is vital in identifying the most appropriate therapeutic targets. Data collected from observational studies such as Registry and Enroll have facilitated this work.

Prof. Robert Lahue, National University of Ireland, Galway.

Slowing a runaway train: targeting histone deacetylase 3 in Huntington's disease.

Since joining NUIG in 2007, Prof Lahue's research work has been supported by Science Foundation Ireland, the Irish Health Research Board and the EHDN. In 2014, Prof Lahue began collaborating with Dr Silvia Ginés of the University of Barcelona to investigate the potential therapeutic effects of the HDAC3-selective inhibitor in HD mice.

Cognitive decline is a critical quality of life concern for HD patients and families. The enzyme histone deacetylase 3 (HDAC3) appears to be important in HD pathology by negatively regulating genes involved in cognitive functions. Research suggests HDAC3 also helps fuel CAG repeat expansions in human cells, suggesting it has a role in disease progression. This multifaceted role suggests that early HDAC3 inhibition offers an attractive mechanism to prevent HD cognitive decline and to suppress striatal expansions. This hypothesis was investigated by treating a HD mouse model with the

HDAC3-selective inhibitor RGFP966. Chronic early treatment in HD mouse models highlight RGFP966 as an appealing multiple-benefit therapy in HD.

Prof. Leslie M. Thompson, University of California, Irvine (UCI).

Neural stem cell transplantation for Huntington's disease.

Prof. Thompson has studied HD for most of her scientific career and was a member of the international consortium that identified the HD gene in 1993. She is a member of the Hereditary Disease Foundation HD Cure Committee, Huntington Study Group Scientific Affairs Committee, and is founding Co-Editor in Chief of the Journal of Huntington's Disease. Prof Thomson is involved in preclinical studies in a HD mouse model to investigate stem cell-based transplantation approaches for HD.

Prof. Anne-Catherine Bachoud-Lévi, Mondor Biomedical Research Institute, Créteil, France.

Works on basic research in cognition and clinical research in brain therapies.

Prof. Bachoud-Lévi and her team combine large scale studies in cell therapy and basic research in cognition. They use intracerebral grafting as a model of plasticity in human beings and integrate therapeutics in basic research in cognition. In addition, they develop cognitive programs in language and social cognition and transfer them to brain pathology.

Prof. Nicole Déglon, University of Lausanne, Switzerland.

The self-inactivating Kamicas9 system for HTT gene editing.

Prof. Déglon's research interests include the development of viral-based genetic models of neurodegenerative diseases and the treatment of neurodegenerative diseases from cell transplantation to gene therapy. Prof. Déglon and her team have developed the kamiCas9 self-inactivating CRISPR/Cas9 system. In the first application of this technology to neurodegenerative disorders, the gene responsible for Huntington's disease (HD) was targeted in adult mouse neuronal and glial cells. Mutant huntingtin (HTT) was efficiently inactivated in mouse models of HD,

leading to an improvement in key markers of the disease.

Genome editing has the potential to cure many diseases. Research in humans is at an early stage. CRISPR/Cas9 has the potential to treat many diseases but once the DNA for CRISPR and Cas have been inserted into a genome, they stay there forever. This may cause future problems. Prof. Déglon's research discovers a way to switch off Cas once it's finished editing the HTT gene, thereby reducing the chances of it triggering an immune reaction or cutting somewhere it shouldn't. (See HDBuzz recent article).

Prof. Sarah J Tabrizi, University College Hospital, London.

Meeting the therapeutic challenge in Huntington's disease.

Prof. Tabrizi is Director of the UCL HD Centre, which she co-founded with Professor Gill Bates in 2016. She is clinical global Principal Investigator for the world's first HTT lowering ('gene-silencing') trial in HD, IONIS-HTTRx, sponsored by Ionis pharmaceuticals, which started in September 2015. Prof. Tabrizi was elected as a Fellow of the UK Academy of Medical Sciences in 2014 for outstanding contribution to medical research.

Understanding of HD pathogenesis is evolving, and there are a number of candidate therapeutics with potential disease-modifying effects that are currently being tested. Prof. Tabrizi gave an update on new insights into HD pathogenesis and genetic modifiers of disease progression. She discussed her work to understand the neurobiology of neurodegeneration and neural compensation and plasticity in HD, and give an overview on exciting advances working towards HD gene silencing in humans.

Prof. Tabrizi mentioned the importance of family members and those with HD registering with the Enroll-HD study. Family members in attendance were delighted to have the opportunity to speak with her personally and thank her for her vital research work.

Organisations providing sponsorship for the NECTAR conference included:

Science Foundation Ireland, Campaign for Alzheimer's Research (Europe), Cure Parkinson's Trust, Neuroscience Ireland, Fáilte Ireland and Neuronal Signalling.



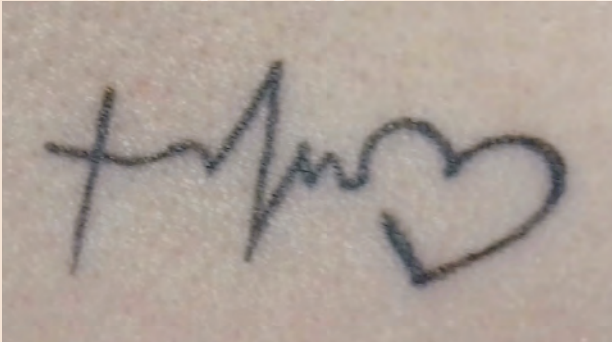
Joe Doran's Sports Person Award

Congratulations to Joe Doran for his marvellous achievement in winning the Longford Sports Partnership Sports Ability award. Joe and his family had an enjoyable night celebrating his achievement. Joe has been an amazing advocate and fundraiser for HDAl in recent years and was instrumental in establishing the HD Awareness walk held in July in Lough Key Forest Park.



Fundraising - Thank You

Faith, Hope and Love



Anna Porter held a unique charity fundraiser by offering a prize for anyone who correctly guessed the symbol she chose for her Tattoo to mark her 70th birthday. Anna received marvellous support from family and friends and raised a magnificent €1,250 for HDAl. Adam Collins at Wildcat Tattoos kindly donated €30, Theresa McBride at Craft Haven, Letterkenny donated €27 and Mick Scanlon donated €80.

Marathon Support



Jersey Marathon

Keith Posner very generously participated in the Jersey marathon to raise awareness and funds for HDAl in support of Violet O'Reilly who has been impacted by HD. Keith got great support from friends and colleagues and raised approximately €800 for HDAl. Sincere thanks to Keith, Paul and all who contributed to his excellent endeavours.

Josie's Half Marathon



Josie Leavy (right) and Kathleen Keogh participated in a half marathon recently and raised €700 for HDAl. Thanks to Josie for her much appreciated commitment to HDAl and to her family and friends for their support.

Coffee Morning



Huge thanks to Ann Maher who raised €408 from the sale of HDAl's Christmas cards. Ann also organised a Coffee Morning at St Andrew's Resource Centre, Dublin 2 on 16th of November 2017 and raised an additional €440.57.



Thanks also to Ann's friends and colleagues who helped and supported the event, in particular Joe McElliott (left), Patty Rogers, Rhonda Hill, Noel Clarke and Billy Ryan for their support with the event. Thanks to all who contributed and bought HDAl Charity cards.

Irish Life Health



Anne Doran, Damian Fadden and Geraldine O'Donovan Hennessy.

The Irish Life Health Sales team generously supported HDAl by organising a Cake Sale and raising sponsorship through a sponsored run in September. A very BIG thank you to Anne Doran, Damian Fadden, Natasha Grubb and all at the Irish Life Health Sales team for your very kind fundraising efforts raising €720 to support HDAl's work.

Postal Celtic Social Club

Special thanks to Pat Davis and her friends at the Postal Celtic Social Club for raising €100 for HDAl from their weekend outing. Pat also raised €25 from her Crochet Shawls.

Wedding Favours

Sincere thanks to Safa and Steven Feeley for their very generous donation of €500 to HDAl in lieu of Wedding Favours.

Recently received donations include:

- Rothco Advertising Company kindly nominated HDAl as their chosen charity for a very generous €1,000 donation.
- Martina, Finnola and Zil monthly Direct Debit donations.
- Patrick and Teresa O'Sullivan €1,000
- Edward and Mary €200
- Siobhan €150
- Christine Dempsey €200
- Pearse College Porters Desk Collection €35
- Sincere thanks to the family and friends of the late James Whelan who donated €850 to HDAl in his memory



HD Making Headlines -

A selection of newspaper features breaking the news.



Cork and Dublin Support Meeting and Christmas Lunch

We had a great gathering for the Dublin and Cork Christmas meetings. Unfortunately due to snow in the West, the Support meeting and lunch will be rescheduled for early 2018.



DUBLIN



CORK

Save The Date

Members Meeting and Respite Weekend Cuisle 8th -10th June, 2018

DO YOU HAVE ANY ARTICLES TO CONTRIBUTE OR TOPICS YOU WOULD LIKE DISCUSSED? THIS IS YOUR NEWSLETTER.



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When HD Came To Tea

The radio documentary 'When HD Came To Tea' was broadcast by Dublin City 103.2FM on Wednesday 29th November 2017 at 3pm. Sincere thanks to producer and presenter Peter Grogan and to Ken Tuohy editor and recordist.

Special thanks to all who contributed to the documentary which featured interviews with people living with HD, carers, family members and professionals. The programme features interviews with those affected by HD and up to date research in the search for effective treatments.

HDAI receive media requests from time to time - If you are interested in speaking to the media please let us know.

HD ID Card

A free Huntington's ID card is available. Please send your photograph, address, phone number and an emergency contact person's phone number to the HDAI office and we will take it from there.

Follow HDAI on Social Media

Facebook: https://www.facebook.com/pages/Huntingtons-Disease-Association-of-Ireland/121707387897701?ref=br_tf

Please Like our page.

Twitter: @HDAI_ie Thank you to our volunteers for their social media work.



HDAI Support Meetings

All those impacted by HD are welcome to join us at our Support Meetings throughout 2018 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 1800 393939 for more information.

DUBLIN

Feb 2nd

CORK

Mar 24th

ROSCOMMON

Feb 17th

LIMERICK

Jan 13th