



EHDN & Enroll-HD
Strasbourg 12–14 Sep 2024

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Photos: Vladimir Marciano/Factor-H

Looking Back with Gratitude, Moving Forward with Purpose

Catherine Deeprise

What an impressive start to 2024! As we drive forward progress in HD research, we will take time to remember what we have learned on HD Gratitude Day on 23 March. Read on to find out more about this global demonstration of HD solidarity, a personal account of HD care in Venezuela, and Charles Sabine's latest achievements in championing awareness about HD.



We gratefully recognise our partnership and support the work of Factor-H serving the Venezuelan HD families. Team up with your colleagues and friends, share photos on social media, celebrate the gene discovery, and donate to Factor-H.

On 23 March 2024, we are planning to have a **live stream from Venezuela** (more details to come), which will include participation from some clinicians who were part of the original team when the gene was discovered. We want to bring together the global HD community and the Venezuelan families in a day of awareness and gratitude.

Join us: [#hdgratitudeday](#)
[Donate](#) to Factor-H



23 March is HD Gratitude Day!

Asun Martinez, Cristina Ferreira, Katrin Barth,
 Ignacio Muñoz-Sanjuan, James Pollard

23 March 2023 marked the 30th anniversary of the discovery of the HD gene. On that day, the HD community showed their gratitude to the many families from around the world, with a big proportion coming from Venezuela and other places in Latin America, for their outstanding contributions to the discovery of the HD gene. In doing so, these families provided a vital vision of hope for the future.

We were overwhelmed by the incredibly positive response and outreach when HD Gratitude Day was initiated last year. Many people from all around the world joined us with their unique, creative, touching, and all incredibly important, contributions. The sense of unity among all people brought together by HD, family members, scientists, clinicians, and other professionals was tremendous.

We are proud to say that 'H-Hands' has become a symbol for HD solidarity. The generous donations provided to Factor-H in 2023 are being used to advance Factor-H's medical, educational, and social programmes.

This year, we want to continue our quest to honour all families who made the cloning of the HD gene possible in bringing the worldwide HD community together once again in a Day of Gratitude.





Alex Fisher



Current HD Care in Venezuela

Alex Fisher, Senior Occupational Therapist, Birmingham and Solihull Mental Health NHS Foundation Trust

Armed with school-level Spanish, Google translate, and the goodwill of her new colleagues and friends, in September 2023, Alex Fisher became the first foreign healthcare professional to join Factor-H in visiting the Venezuelan communities. She shares with us her thoughtful reflections on the trip.

Four hours from crumbling Maracaibo (the State capital), riding in a second-hand bus from China packed to the gills with food parcels, medical professionals, and humanitarian personnel, I'm here with Factor-H and Habitat Luz. These two organisations work together to provide advocacy and humanitarian aid for impoverished HD communities throughout Latin America. Habitat Luz provides an administrative and operative arm focusing on Venezuela and the townships of San Francisco, St Luis, and Barranquitas. Ten percent of the remaining population in these areas have HD. I say 'remaining' as migration from Venezuela has been upwards of 7 million since 2015.

Whilst the HD families of St Francisco and St Luis live in perilous circumstances, being on the city's edge at least gives them access to piped (if not clean) water. Barranquitas does not have this luxury. So, alongside the

abandoned infrastructure courtesy of a divided and detached government, I join in the collective cries of 'La Luz' (meaning 'the light') when the electricity returns from its daily outages.

Sore from the roller coaster ride of the immense potholes on our journey, we stretch and step out into a space where trestle tables are being laid out under the cover of a basketball court and see an increasing throng of families waiting. I dodge a motorbike that pulls up loaded with four people, two of whom with profound chorea are sandwiched between the driver and rear pillion passenger. This is an outpatient clinic, but not as we might know it. It's (usually) a three-monthly visit by the teams, which combines the delivery of foodstuffs to HD families, educational and fun materials for the under 12s, and symptom check-ups and advice for everyone, provided by Factor-H and Habitat Luz.





The outpatient clinic is overseen by two (volunteer) neurologists and a paid GP (via Factor-H). Whilst volunteer medical students work alongside Habitat Luz local volunteers in the clinic, Dr Aura and Dr Soto head out on home visits to those who can't attend the clinic, having been advised of the need by Yoliz (Factor-H's Resident Social Worker).

We attend a home visit, driven by Samuel (Factor-H's local 'fixer'). The family structure and societal norms mean that as we make our way into people's homes, neighbours and interested parties appear, too. I make mental notes on the environment, how care is delivered, and ask permission to return.

Usually, the three-monthly visit to Barranquitas takes place over two days, with the teams either returning to their homes or repairing to a hotel in the prefecture's capital. However, the rainy season has better ideas, and Samuel receives news that the roads are flooded. With our precious cargo and the bus with its frayed tyres, it is too risky to attempt. Disappointed but not bowed, Marina (President of Habitat Luz) takes the opportunity to engage with the Prefecture's Mayor. Land purchased by Factor-H for a future project called Casa Dignidad (a dedicated day centre) in Barranquitas has been stolen and built on. The Mayor is sympathetic and agrees to explore what had happened (I later hear that he was good to his word, and in November, an alternative site was earmarked).

I return to Barranquitas for four days and stay in the bosom of Atalina and her family, whose home is next to



the Factor-H/Habitat Luz office. I'm bunking in the same room as Yoliz while Samuel and Gindel (a Caracas-based documentary maker/journalist and my translator) sleep in the office.

Marina has asked me to support a project to improve caregiver well-being. Meeting with the local team, it soon became clear that my prepped list of what we thought the community needed and what they actually wanted was poles apart.

Focusing on the community and the conditions in which they live, we set out to interview and film the stories of individuals. This created a natural, iterative foundation for 'The Cuidadores (Carers) of Barranquitas' project. First, we meet Selina, to whom Gindel brings joy through dancing, and we try to decipher what has happened to her belongings through the buzz of mosquitoes and the many voices of her neighbours. Crime is close at hand here, and her two sons, both of whom appear to have HD, may have sold the furniture for drugs but also for survival, leaving her only with a string chair, a hammock, a dirty mattress, and her cat. Despite

her sons masquerading as her carers, it is the community and her sister-in-law who tend to her. While clearly a very serious issue, reaction requires processing, and intervention has ramifications for Selina's safety. Both organisations have a humanitarian drive, and that's about equity of delivery which does not cause tensions elsewhere.

Onwards, we meet Olga and Maria and, later, Jorge, a homeless man with HD with three affected brothers in the same position. Jorge, sleeping in an old school building, is being fed by a number of ladies who ask us for more information on his condition and how to help. Despite their own desperate circumstances, the compassion is genuine.

Over the next few days, we continue to collate the stories of people's daily lives. As an HD professional in the UK, I meet suffering and inspiration in equal measure, but here it is magnified. On one of our last visits, Gindel draws me into a darkened room. As my eyes adjust, I see that I am surrounded by four moving bodies in varying stages of their illness. Although initially stunned, I am struck by the tenderness with which these people are cared for by a young man. Jesus cares not only for his father but also his two aunts and uncle. He knows he, too, is at risk, but his faith consoles him, and in the brief periods of time he has to himself, he reads and tends to the garden at the rear of the house he shares with the family. We watch as he carries his aunts into the open-air kitchen area, and his grandmother and a friend undertake precision feeding of their loved ones. His uncle, fed earlier, can now not eat fluidly as his rigidity is affecting his ability to open his jaw (and therefore hold and swallow adequate nutrition). I advise Jesus on back care, which may seem trivial but is vital to his well-being. I also advise on feeding his uncle and how to increase his hydration and make a mental note to discuss supporting safer manual handling with Marina.

Silently, we walk back to the Habitat Luz office. There is much to be done, and as we eat together, we share thoughts and plans. There's one last event: the formation of the new Cuidadores and Volunteers group. We all grab a plastic chair, walk to a nearby lakeside wharf and form a circle next to a group of piglets irritating the more established chickens. We talk about faith and suicide, but mostly about the hope of what we can achieve and love and pile in for many group hugs. On returning to Maracaibo and, for me, to the UK, the



teams have been building on the insights and ideas from the trip. In essence, the hope is to create a greater community of practice which can cater for the current and future delivery of care to these communities to whom we owe so much. Factor-H and Habitat Luz, as well as the patient organisations of Latin America, have formed the vital foundations and framework. But if we are to envisage a time when there will be the equitable delivery of the hard scientific hope of treatment, many barriers remain to be overcome.



Charles Sabine on stage

The Unlimited Capability of Every Human: TEDx Talk by Charles Sabine, OBE

Catherine Deeprose

TEDx talks are created as part of [TED's mission](#) to research and discover 'ideas worth spreading'. So who better than Charles Sabine – Emmy Award-winning television journalist, OBE recipient, and founder of [The Hidden No More Foundation](#) – to speak loud and clear in championing awareness about HD. You can view Charles's personal, powerful and inspiring presentation here: youtube.com/watch?v=RsUfCKfyQYY.

[X@HD HiddenNoMore](#)

instagram.com/hiddennomorefoundation

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Charles with his brother John

'This is an incredibly important talk – so informative, while being moving and powerful.'

Leslie Thompson
Neurology Professor
University of California Irvine

'Charles is an amazing example of "be the difference you want to see in the world". I can't begin to express how much I admire and respect his efforts.'

Caroline Benn
Chief Scientific Officer
LoQus23 Therapeutics

'An amazing talk. Charles has a true gift for captivating an audience; they will not forget their newfound awareness of HD.'

Gillian Bates
Professor of Molecular Neuroscience
University College London, UK



Niels H Skotte



Lauren Byrne

The Biomarker Working Group: Key Developments and a Call to New Members!

Niels H Skotte and Lauren Byrne

Dr Niels H Skotte is a research group leader at the University of Copenhagen in Denmark. He has worked extensively on Huntington's disease for nearly 15 years. His research group is focused on molecular and cellular neuropathology and clinical proteomics for biomarker discovery in saliva, plasma, and cerebrospinal fluid from humans as well as animal models.

Dr Lauren Byrne has been working in HD biomarkers since 2015. Her research lab at University College London combines a focus on biofluid, imaging, data harmonisation and modelling for both adult and Juvenile HD. She has spearheaded several high-profile publications on key biofluid biomarkers, including mutant huntingtin and neurofilament light chain.

A Brief History

The EHDN Biomarker Working Group (WG) is close to 20 years old. It has a long history of driving pivotal biomarker research studies for HD, including TRACK-HD, TrackOn-HD, and HDClarity. The WG was established in 2005 by Sarah Tabrizi as lead facilitator, followed by Maria Björkvist in 2010. In 2015, Ed Wild took on the lead role, with Niels H Skotte joining him as co-lead facilitator in 2017. Ed Wild stepped down in early 2023 to join the EHDN Executive Committee, and at this point, Lauren Byrne joined Niels H Skotte as co-lead facilitator.

We are deeply grateful to Sarah Tabrizi, Maria Björkvist, and Ed Wild for their many contributions to the field of HD biomarker discovery. Sarah Tabrizi is still pioneering biomarker research in HD with her HD Young Adult Study (HD-YAS), which aims to discover biomarkers of the earliest signs of pathogenesis in HD mutation carriers. Maria Björkvist's research seeks to understand the peripheral manifestations of HD and how these could translate to biomarkers. Ed Wild continues to act as the Global Chief Investigator of HDClarity – one of our flagship projects. All three remain HD biomarker experts contributing to the goals of the WG.

Moving Forward

Moving forward, the WG will be renamed the Fluid Biomarker WG to illustrate our focus and complementarity to other WGs. As the Fluid Biomarker working group, we want to be active and drive new initiatives for biomarker development whilst creating opportunities to update our members on the latest advances in HD biomarkers. For this reason, we are moving forward with meeting formats which each have their specific purposes: Core WG Meetings (quarterly virtual updates on all active projects), Topic-focused Workshops (small yearly in-person events), and Biomarker Showcase Seminars (biannual events, open to all, online data presentation).

We will have a strong focus on several topics, including obtaining external funding and establishing European collaborative projects; working with regulatory bodies to streamline validation for therapeutic development; exploring the newest technology advancements for biomarker discovery; employing data integration and AI-driven analyses to improve the predictive power of currently available biomarker data; and synthesising current best practices and presenting guidelines for biomarker development.

We want your input and commitment to increase synergy and collaborations across research labs, industry, and European borders to reach our goals. Hence, we have launched a [survey](#) to assess the current gaps in HD biomarker discovery, where we should focus our efforts, and who we have in our membership to actively contribute to future projects.

Furthermore, we will have two types of memberships to increase the number of active members and allow each to contribute according to their capacity. Active members will be encouraged to actively contribute to ongoing projects and attend Core WG meetings throughout the year. Passive members will receive general WG updates, may be invited for expert opinion on specific projects, and will be invited to the bi-annual biomarker showcase seminar. We will reach out to our current members, requesting that they actively opt-in to the membership level that best suits them.

A Call to New Members

Please complete our [sign-up survey](#) (alternatively scan the QR-code) if you are interested in HD biomarkers and believe you have relevant skills or expertise to contribute to the WG. We look forward to speaking with you soon!



Scan the code to complete our EHDN Biomarker WG Survey



EHDN & Enroll-HD 2024: Key Updates

Åsa Petersén (Lund University) and Sandrine Humbert (Paris Brain Institute ICM France)



Åsa Petersén



Sandrine Humbert

As programme chairs, we are working closely with the organisational team from EHDN and a dedicated programme committee to finalise the meeting programme. It will include many important aspects of HD, from genetic modifiers and molecular mechanisms to cognitive rehabilitation and psychological support. Experiences from people in families affected by HD will be shared, and the latest developments in clinical trials will be showcased. A

special evening session with L'Eurobuzz will summarise the highlights of the meeting! You are invited to submit abstracts and maximise the dissemination of novel and exciting HD research.

- **Public registration: 1 May to 30 June**
- **Abstract submission: 1 May to 31 May**

Nominations for the EHDN Executive Committee (EC) are now open at www.euro-hd.net/html/network/project/voting. The EC is the body responsible for governing the EHDN, directing and overseeing its activities and establishing its [strategy](#). There are four places available in this upcoming election, and one current EC member is standing for re-election. The nomination period will be open until 1 April 2024. The voting period will open in mid-July and remain open until shortly before the meeting in Strasbourg in September.



Update: Clinical Trial Developments

Jenny Townhill and Tim McLean, Central Coordination

Key updates since the last newsletter are provided below for EHDN-endorsed trials and studies; please refer to [Table 1](#) for a summary of all ongoing research. An expanded description of all EHDN-endorsed trials and studies (completed and in progress), as well as details of the EHDN endorsement process, is published on the [EHDN website](#).

On 9 November 2023, **Wave** shared an update on the SELECT-HD phase 1b/2a trial of WVE-003, an allele-selective ASO. Previous data reported in September 2022 showed that a single dose of WVE-003 reduced mutant huntingtin (mHTT) protein in cerebral spinal fluid (CSF) (mean reduction of 35%) compared to placebo, with wild-type huntingtin (wtHTT) preserved. Enrolment has now been completed into the first multi-dose cohort of the trial, which will evaluate the effects of 30 mg doses given every 8 weeks. Results from all single-dose cohorts and the first multi-dose cohort, including the longer-term follow-up data, will be shared in the second quarter of 2024.

In addition to the update on the SELECT-HD trial, Wave also reported positive results from a study of WVE-003 in non-human primates, which demonstrated significant levels of WVE-003 in the striatum and other deep brain regions, suggesting that it has the ability to reach the areas of the brain that are important in HD (see [press release](#) for further details).

On 19 December 2023, **UniQure** provided an update on the US and European surgical trials of AMT-130 gene therapy (HD-Gene TRX-1 and HD-Gene TRX-2, respectively) in participants with early-manifest HD, including results of the latest interim analysis on the combined data from both trials.

In the US trial, HD-Gene TRX-1, where participants are either randomised to treatment with AMT-130 or sham control surgery, 26 participants have been enrolled. The first 10 participants were enrolled in a low-dose cohort (6 treated, 4 control) and then 16 participants were enrolled in a high-dose cohort (10 treated, 6 controls). Thirty months of follow-up data were analysed for the high-dose cohort, and 18 months of follow-up data were analysed for the low-dose cohort. In the European open-label trial, HD-Gene TRX-2, 13 patients have been



Photos: Gabriele Stauner · arifox.com

enrolled (6 in the initial low-dose cohort and 7 in the subsequent high-dose cohort).

The latest analysis shows that AMT-130-1 continues to be generally well-tolerated across both dose cohorts with a manageable safety profile and provides additional evidence of favourable trends in clinical and functional measures, which are potentially dose-dependent, in comparison to a separate matched natural history cohort with similar characteristics to the trial participants.

Mean CSF NfL levels suggest a reduction in neurodegeneration compared to natural history data following an initial post-surgery increase. CSF mHTT levels continue to be variable and are not considered to be representative of mHTT at the targeted brain region. Post-surgical changes in total brain volume, which trended below natural history in treated patients, were considered not to be clinically meaningful and were not associated with prolonged increases in NfL (see [press release](#)). UniQure expects to start discussions with drug regulators regarding the next steps in development for AMT-130 early in 2024, with the next update on the trials expected in mid-2024.

The **Roche** GENERATION-HD2 phase 2 trial of tominersen has reached an important milestone in recruitment, with more than 50% of participants now enrolled in 15 countries across North America, Europe, South America and Oceania, with approximately 75 sites taking part.

The study is evaluating the safety, biomarkers and efficacy trends of two different dose levels of tominersen and aims to enrol 300 people with prodromal or early manifest HD.

Table 1: Current EHDN Endorsed Trials and Studies

Registration ID (CT.gov)	Sponsor	Trial name	Phase	Investigational Product	Mode of Action	Delivery	Treatment Goal	Target Enrolment	Location(s)	Status
NCT05541627	BrainVectis, a subsidiary of AskBio	ASK-HD-01-CS-101	1/2	AB-1001	Restoration of cholesterol metabolism dysfunction (AAV-delivered gene therapy)	Surgical, intrastriatal	Disease modification	18	France	Active, not recruiting
NCT05686551	Roche	GENERATION HD2	2	Tominersen	htt lowering ASO	Intrathecal	Disease modification	300	Argentina, Australia, Austria, Canada, Denmark, France, Germany, Italy, New Zealand, Poland, Portugal, Spain, Switzerland, UK, USA	Recruiting
NCT04556656	Prilena Therapeutics	PROOF-HD	3/OLE*	Pridopidine	Sigma-1 receptor agonist	Oral	Disease modification	480	Austria, Canada, Czech Republic, France, Germany, Italy, Netherlands, Poland, Spain, Switzerland, UK, USA	Active, not recruiting (Phase 3 complete, OLE* ongoing)
NCT05358717	PTC Therapeutics	PIVOT-HD	2	PTC518	mRNA splicing modifier	Oral	Disease modification	162	Australia, Austria, Canada, France, Germany, Italy, Netherlands, New Zealand, Spain, UK, USA	Recruiting
NCT05107128	Sage Therapeutics	DIMENSION	2	SAGE-718	NMDA receptor modulator	Oral	Symptomatic	178	Australia, Canada, UK, USA	Recruiting
NCT05358821	Sage Therapeutics	SURVEYOR	2	SAGE-718	NMDA receptor modulator	Oral	Symptomatic	80	Canada, USA	Recruiting
NCT05655520	Sage Therapeutics	PURVIEW	3/OLE*	SAGE-718	NMDA receptor modulator	Oral	Symptomatic	300	Australia, Canada, UK, USA	Recruiting
NCT05475483	SOM Biotech	SOMCT03	2b	SOM3355 (bevantolol hydrochloride)	VMAT2 inhibitor	Oral	Symptomatic	129	France, Germany, Italy, Poland, Spain, Switzerland, UK	Recruiting
NCT04120493	UniQure	HD GeneTRX2	1b/2	rAAV5-miHTT	miRNA AAV delivered gene therapy	Surgical, intrastriatal	Disease modification	15	Germany, Poland, UK	Recruiting
NCT05032196	Wave Life Sciences	SELECT-HD	1b/2a	WVE-003	Allele-selective htt lowering ASO	Intrathecal	Disease modification	36	Australia, Canada, France, Germany, Italy, Netherlands, Poland, Spain, Switzerland, UK	Recruiting

Note. AAV = Adeno-associated virus; ASO = antisense oligonucleotide; htt = huntingtin; mRNA = messenger ribonucleic acid; NMDA = N-methyl-D-aspartate; OLE = open-label extension; VMAT2 = vesicular monoamine transporter 2

GENERATION-HD2 aims to test the hypothesis that tominersen may provide clinical benefit in those with HD who are younger with a lower disease burden, based on the post-hoc analyses from GENERATION-HD1.

Dr Peter McColgan, Clinical Science Lead, explains 'Tominersen has conclusively shown dose-dependent lowering of mutant huntingtin protein in the cerebrospinal fluid in people with Huntington's disease; therefore this is an important study in addressing the role of huntingtin lowering as a potential treatment for Huntington's disease'.

The EHDN, in collaboration with Roche, hosted a webinar on 12 February to discuss the recently published data on GENERATION-HD1 and explain why the GENERATION-HD2 trial is needed and answer questions from the HD community. The recording can be accessed here: <https://ehdn.org/roche-ehdn-virtual-meeting-12feb2024/>



Update: Enroll-HD Biosamples Strategy to include RNA collection

Olivia Handley, Enroll-HD Global Platform Manager (EHDN),
Hilary Wilkinson (CHDI)

Enroll-HD has successfully established a large, high quality biosamples collection that can be [accessed](#) by the research community. More than 1 million aliquots of DNA (deoxyribonucleic acid), lymphoblastoid cell lines, and peripheral blood mononuclear cells (PBMCs) have been collected from approximately 30,000 participants. Within the last four years, Enroll-HD set up a smaller longitudinal collection of plasma, aiming for a cohort of 2,500 participants. Close to 1,700 participants have donated plasma so far.

Obtaining high-quality biosamples remains key. However, it is equally important to ensure that the platform continues to meet the demands of the scientific community. A recent refocus of the biosamples strategy for Enroll-HD has led to a new initiative being rolled out to collect RNA in 2024.

What is RNA?

RNA is a ribonucleic acid whose primary function is to convert genetic information from DNA into proteins. There are several kinds of RNAs, and they can also affect the behaviour of cells.

Why do we need RNA for HD research?

The structure and function of each cell in our body are determined by activating different combinations of genes. By studying the messenger RNAs that are transcribed from these genes, we can find out which genes are active in a particular cell type. Other RNA types are also found in blood, and their function is less well understood. Gathering information on all types of RNA helps us understand more about how a given cell carries out its function. By taking a much deeper look at the role of RNA in HD, we can discover potential biomarkers as well as identify possible new treatment strategies.

There is a high demand for RNA samples in HD research. Presently, within Enroll-HD, researchers need to



Photo: Gabriele Staliner · ariflox.com

use PBMCs to extract RNA, which is neither efficient nor economical. Also, several types of RNAs are found in blood but not in PBMCs. By switching to whole blood collection in PAXgene tubes, it will be possible to yield high-quality and diverse types of RNA for HD research.

How many Enroll-HD sites will be participating, and how were they selected?

Enroll-HD sites were sent a feasibility survey in 2023. They were asked about the availability of trained staff and other resources, such as monitoring systems in both -20 and -80 freezers and access to freezer space. Thirty sites, mainly from North America and Europe, met all feasibility requirements.

What will Enroll-HD sites and participants be asked to do for the RNA collection?

Enroll-HD sites will need to pass an RNA Collection Training Module in the Enroll-HD Clinical Training Portal. A mini remote RNA Collection site initiation visit (SIV) will also be done. Since there will be an extra payment for RNA collection, the Enroll-HD site agreement will be amended. There is no requirement to submit an amendment to the independent ethical review board since RNA collection is permitted under the current protocol. For Enroll-HD participants donating samples for RNA, they will be required to donate 10 ml of whole blood that will be collected in multiple small PAXgene tubes. There is no change to the maximum amount of blood (up to 30 ml) collected as part of the optional component for biosamples.

When will the RNA collection begin?

It is expected to begin in March 2024. The aim is to follow 3,000 HD gene expansion carriers (premanifest/early manifest 70:30 ratio) and approximately 300 control participants for five years. At the same time, the collection of whole blood for PBMCs will be suspended at all sites – this may resume at a few sites in the future.

Acknowledgements

Thank you to Elena Ngangom for her contribution to this article.

HDClarity

Update: HDClarity

Gail Owen, Principal Research Associate, University College London, UK

This week will be the seventh anniversary of the first HDClarity sample collection in Canada in 2017¹. Since then, hundreds of participants have given their time and their CSF. Thanks to the incredible input from participants and the hard work of the study site teams, the HDClarity repository continues to grow, and over 200 CSF samples were added in 2023.

We're grateful to participants for every sample provided, especially those who have returned several times. Approximately 300 participants have provided more than one CSF sample, and nearly 100 have provided three or more samples. This is a great indication of the tolerability and safety of the lumbar puncture procedure, and these longitudinal samples are extremely valuable for looking at changes in HD over time.

In addition to the growing number of samples, the HDClarity data set has also increased significantly. The first periodic dataset consisted of data from around 120 visits, and the current dataset includes 832 study visits – see <https://enroll-hd.org/new-hdclarity-periodic-dataset-pds3-now-available/> for more information. Researchers interested in accessing data and/or samples should visit <https://www.enroll-hd.org/for-researchers/biosamples>.

‘PDS3 represents a huge amount of work by the HDClarity teams at UCL and CHDI and the site staff across the world. Above all, PDS3 is a gift beyond value donated by HD family members and their friends, for the benefit of the global community. We are enormously grateful to them for their altruism and trust.’

Professor Ed Wild

Looking forward to 2024, we are pleased to announce that there will be an HDClarity investigator meeting taking place in September at the joint EHDN & Enroll-HD meeting in Strasbourg. The date of the HDClarity

¹ the University College London (UCL) HD-CSF study began in 2016 and CSF samples from this study were combined in part with the HDClarity sample collection with support from CHDI

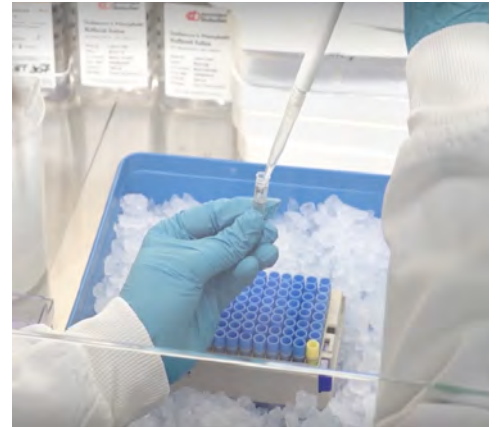


Photo provided by Gail Owen

meeting is scheduled for Sunday, 15 September (still to be confirmed), following the Enroll-HD investigator meeting. This will be the first time that HDClarity teams from around the world will have the opportunity to share their experiences of the study. Although there have been previous smaller meetings, this will be a great opportunity to share best practices and lessons learned, and it should be a very interactive and informative meeting.

Further information about HDClarity, including the current protocol, is available at www.hdclarity.net, and the Central Coordination team are always happy to answer any questions (hdclarity-cc@enroll-hd.org). Information about the study can also be found on a variety of platforms, as shown here.

Interested in Participating?

Enroll-HD  enroll-hd.org

 U.S. National Library of Medicine
ClinicalTrials.gov clinicaltrials.gov

HDClarity  hdclarity.net

 [@HDClarityStudy](https://twitter.com/HDClarityStudy)



Scan the code for
more site information:

Update: Funding Opportunities

Fionnuala Margreiter, Grants & Collaborations Manager

Current Funding Opportunities

- **EU Joint Programme – Neurodegenerative Disease Research** (JPND) Call for Proposals currently open: '[Mechanisms and measurement of disease progression in the early phase of neurodegenerative diseases](#)'. Applications involve a two-stage proposal and pre-proposals should be submitted by 5 March 2024. Country-specific information is available on the website.
- **Innovative Health Initiative** (IHI) funding call is now open: '[Clinical validation of biomarkers for diagnosis, monitoring disease progression and treatment response](#)' (Call 7, Topic 3). The full proposal submission deadline is 22 May 2024.

EHDN/MDS-ES Fellowship Update

2023 Fellowships

All fellows had the opportunity to gain experience in a multi-disciplinary clinic in the UK or Spain, and feedback has been very positive.

List of fellows (fellow country; host clinic)

- **Mehri Salari** (Iran; London, UK)
- **Victoria Zubiri** (Colombia; Barcelona, Spain)
- **Ninel Arakelyan** (Armenia; London, UK)
- **Mariana Andriievskia** (Ukraine; Cardiff, UK)
- **Elison Sarapura Castro** (Peru; Madrid, Spain)
- **David González Ruffino** (Venezuela; Madrid, Spain)

Fellowship Impact Project

A formal evaluation of the fellowship programme was undertaken to understand its overall success in meeting the original objectives, identify potential gaps and issues, and provide data-driven recommendations for adaptations and improvement.



Photo: Gabriele Stauner - artfox.com

Many thanks to all the past fellows and hosts whom we were able to interview and gain useful feedback. The results are currently being analysed, and a report is being prepared.

2024 Fellowship

Following the preliminary results from the impact project, four main adaptations were made to the 2024 programme. These include

- (1) ensuring that the programme provides equal opportunities globally for training to improve HD clinical care in underserved regions or specific aspects of HD care, including Europe,
- (2) making the application available as an online form for ease of application and administration,
- (3) centralised matching of fellow with host clinics after the selection process
- (4) increase in funding amount.

The 2024 application round was open until 21 February 2024. At the time of writing, 16 applications have been received. The applications will be evaluated together with MDS, and the successful fellows will then be matched with experienced host clinics. All placements are expected to take place during 2024.

For regular updates about funding opportunities, please see the [EHDN website](#) and the [Twitter\(X\) account @EHDN GRANTM](#).

Update: New Lesley Jones Seed Fund Awarded

The EHDN has recently awarded seed funding for an exciting new project.



Photo provided by Clare Eddy

Clare Eddy

Clare Eddy, at the National Centre for Mental Health and the University of Birmingham (UK), has been awarded funding for her project 'Social Cognition and Quality of Life in Huntington's disease'. Research has shown that HD can affect the ability to recognise emotions, and to reason about other people's thoughts. By using cognitive tasks and questionnaires, Clare intends to identify the social skills that are most important for everyday functioning and the quality of life of both people with HD and their close others. This information will help develop a new assessment about the social side of HD, which can be used in research and the development of new psychological interventions.

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Photo provided by F. Giorgini

Flaviano Giorgini

The Lesley Jones Seed fund programme is intended to support pilot studies that will eventually kickstart larger projects. The next deadline for applications is

1 November 2024.

More information about the programme and how to apply can be found [here](#) or you can contact Flaviano Giorgini (flaviano.giorgini@euro-hd.net) or Kinga Kolodziej (kinga.kolodziej@euro-hd.net) for further information.



Photo provided by K. Kolodziej

Kinga Kolodziej



Funding Awarded to Develop 'HD Academy'

Claudia Azañedo Vázquez, Communication Manager,
European Huntington Association

The [European Huntington Association](#), in close collaboration with the Ligue Huntington Francophone Belge, the Bulgarian Huntington Association, and the ACHE Corea Huntington Española, has just received an Erasmus+ Grant to develop an adult education project called HD Academy. The HD Academy will create an innovative, multilingual, and inclusive e-learning platform for the education and training of formal and informal caregivers of people impacted by HD. The project will run for 18 months and provide a new online resource to the global HD community. Our project scored 96 out of 100 in this highly competitive EU call, so we are really proud of this achievement. 2024 couldn't have started any better!



DOMINO-HD: A Long-term Study of Lifestyle in People with HD

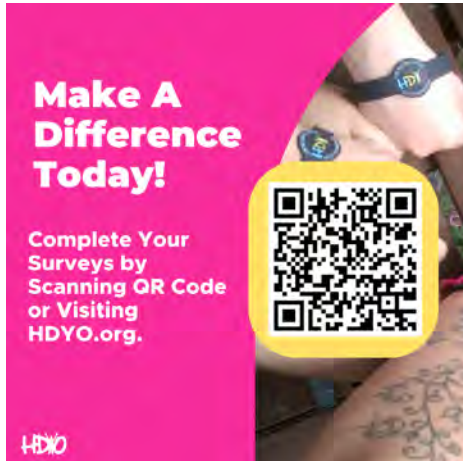
Catherine Deeprose

This project, which included a 12-month observational study is now complete. The researchers developed methods for combining digital data on lifestyle from wearable trackers and from questionnaires completed by HD participants. This allowed them to record activity measurements over a long period of time. All the 97 participants were part of Enroll-HD, meaning that data were available on each participant's disease status, which was then linked to their lifestyle data.

Outcomes from the study confirmed that the long-term study of lifestyle using digital tools in people with HD is possible. Findings also highlighted the need for flexibility and personalisation in the development of non-pharmacological interventions for people with HD.

‘The insights that DOMINO-HD have provided for the measurement and study of lifestyle in HD can be used to develop and test lifestyle interventions for improving the progression and general quality of life for people with HD. Additionally, the methods we have validated in this work, particularly the use of digital data, have broad applications across clinical research in HD.’

Monica Busse
Chief Investigator
Cardiff University, UK



Huntington's Disease Youth Organization Releases Second Survey

Jenna Heilman, Executive Director,
[Huntington's Disease Youth Organization](https://www.hdyo.org)

As a part of our global research surveys, we are seeking input from the HD community about different emotional and educational barriers to research. We already know some of the barriers to participating in research, like burdens with work, family, and so on. This isn't

replicating those findings but diving deeper into feelings and knowledge around different research initiatives. We want to use this information to enhance educational initiatives for HDYO and other organisations to better equip young people and community members about research. These surveys are currently translated into French, Italian, Polish, Portuguese and Spanish. Data from the first survey about how people are seeking resources and education will be analysed and available later this year. This survey also remains open. We are also thrilled to announce our second international congress will take place in Prague in 2025!

Save The Date!





10th Congress of the European Academy of Neurology

Diana Prioteasa, Promotion Exchange Coordinator,
[European Academy of Neurology](#)

The 10th Congress of the European Academy of Neurology (EAN) will take place in Helsinki, Finland (and online) from 29 June to 2 July this year.

The overarching theme for EAN 2024 is 'Neuromodulation: advances and opportunities in neurological diseases', which will be the focus of several sessions, with something for every category of participant, from trainees to specialists, for both clinicians and scientists. The congress programme will be bursting with symposia, workshops, teaching Sessions, and much more, including exciting [lectures](#) from a top-class line-up of speakers.

For more information about EAN 2024, please visit the [congress website](#).



Juliana Bronzova



Flaviano (Flav) Giorgini

Welcoming Our New Science Directors: Juliana Bronzova and Flaviano Giorgini

Juliana Bronzova, MD, DSc, and Flaviano (Flav) Giorgini, PhD, have a long history of involvement with the EHDN. Recently, they took up roles as Science Directors, and in addition to their contributions to the [EHDN Think Tank](#), they bring a wealth of knowledge and experience to the wider work of the EHDN. We find out more [here](#).

To start, can you tell us how you first got involved with the EHDN?

Juliana: As far back as 2011! At that time, I was deciding to leave the pharmaceutical industry, and when I heard about the HD community and the work being conducted by the EHDN, I knew I wanted to become involved in doing something very meaningful in the field of rare diseases. Right from the beginning, it was absolutely amazing – and it still is! That's why I'm still here, part of this community.

Flav: I moved from the USA to Leicester (in the UK) to take up a lectureship in 2006. I remember coming to the 2008 plenary meeting in Lisbon and thinking, 'This is great!'. I very quickly found out that the EHDN community was just really amazing, and as a basic scientist, my interactions with clinicians, clinical researchers, and families affected by HD were all incredibly positive. After becoming a member, I took on various roles over the years, including being Chair of the Scientific and Bioethics Advisory

Committee. In 2023, both Juliana and I took up positions as Science Directors.

In your current roles as Science Directors, how do you interact/complement one another?

Juliana: I think we are very complementary because the work of the EHDN is so far-ranging. Together, we can contribute to the entire spectrum of research – from very early science research to later clinical trials. There is a lot of work that we are involved in, from supporting research to promoting education through fellowships, courses, and collaborations.

Flav: That’s exactly right. Between the two of us, we span the academic science side and also the clinical trial and industry elements of HD research. We kind of fall into the aspects where we have the most to offer, and it feels like a very natural collaboration.

Will we see you both at EHDN & Enroll-HD 2024?

Flav: Of course! I’m going to be involved in the poster judging and lots of different things. I’m really excited about it. For me, the opportunity to hear from early career researchers is really valuable, and we’re going to do just that, alongside hearing from lots of the really big names in HD research.

Juliana: Definitely – I will be chairing a session as part of the Enroll-HD presentations, and I’m really looking forward to this exciting collaborative event!



Get in touch with the Think Tank!

The EHDN’s HD Science Think Tank brings together EHDN members and staff who are closely involved in supporting scientific research – including members of the Executive Committee, Central Coordination and the working groups – and it engages with the HD research community in three ways:

- Researchers may contact the Think Tank for help in identifying potential collaborators or funding opportunities, or to discuss scientific ideas
- The Think Tank welcomes suggestions of research topics, and has provided a [contact form](#) on its website via which these can be submitted
- The Think Tank may occasionally propose specific research topics that could be addressed by a dedicated task force working for a defined period of time

For more information about the [Think Tank](#), please contact Yury Seliverstov: yury.seliverstov@euro-hd.net

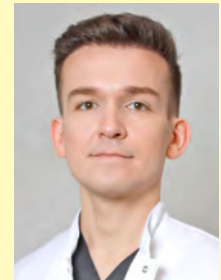


Photo provided by Yury Seliverstov



Dates for your diary

- **23 March 2024** is [HD Gratitude Day](#). Get involved!
- **16–19 May 2024:** The [MENA Congress for Rare Diseases](#) takes place in Abu Dhabi, UAE.
- **30 May to 1 June 2024:** The [39th Annual HDSA Convention](#) takes place in Spokane, Washington, USA. Early bird registration closes 31 May 2024.
- **29 June to 2 July 2024:** The [10th Congress](#) of the [European Academy of Neurology](#) (EAN) takes place in Helsinki, Finland (and online).
- **12–14 September 2024:** [EHDN & Enroll-HD 2024](#) takes place in Strasbourg, France. Registration and Abstract Submission both open on 1 May 2024. For details about past meetings, click here: <https://ehdn.org/plenary-meeting/>
- **6–9 November 2024:** The [Huntington Study Group](#) meeting takes place in Cincinnati, Ohio, USA. Find out more [here](#).
- **14–16 March 2025:** The [International Congress](#) of the [Huntington’s Disease Youth Organization](#) takes place in Prague, Czech Republic.

Would you like to share an upcoming event with our readers? Please email the details to newsletter@euro-hd.net

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