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EHDN *News*

EUROPEAN **HUNTINGTON'S DISEASE** NETWORK

Credit: Steven Pesket

The documentary film 'Dancing at the Vatican', which recounts the journey of a number of families from Latin America affected by HD, was shown at the British Academy of Film and Television Arts (BAFTA) in London on 5 February. It was presented by HD campaigner Charles Sabine, pictured here.

A special issue dedicated to ethics

Laura Spinney

The rise of predictive genetic testing and new technological advances such as whole genome sequencing have confronted HD family members, carers, clinicians and scientists – not to mention policymakers – with a range of new ethical challenges that are not easy to solve. This edition of the newsletter is therefore dedicated to the ethical aspects of HD, and in particular to the question of how to balance a person's right-to-know with their right-not-to-know, when it comes to genetic information.

Sharing their experiences with us on this important topic, in three separate but linked articles, are a gene carrier and his partner, a clinical geneticist and a clinical psychologist who work together, and a neurologist. Next, **Mette Gilling Nielsen** gives us her thoughts on navigating an ethical minefield in HD research, and **Gráinne Bird** discusses the potential of social media to do good, both within and beyond the HD community. The remainder of the issue is filled with our regular updates on clinical trials, Enroll-HD, the fellowship exchange programme and recently awarded seed funds, along with a renewed invitation to send us your photos. Last but not least, lawyer **Heidi Beate Bentzen** – the newest recruit to EHDN's Scientific and Bioethics Advisory Committee – brings us back to the issue's main theme, addressing it from a lawyer's perspective. Enjoy!

CALLING ALL YOUNG CLINICIANS: APPLY NOW FOR THE 2020 FELLOWSHIP EXCHANGE PROGRAMME. DEADLINE 20 MARCH!

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The view of a gene carrier and his partner

Here, a couple share their views with us on how test results should – or should not – be shared. They start by talking in one voice, then they give their personal views. Because they wish to remain anonymous, we have distinguished the gene carrier's sole voice by putting it in italics.

We have been together since 2011, and from the first months of our relationship we discussed the presence of HD in our lives. We were both aware that at some point we would have to go through the genetic testing procedure. Before that happened, we would occasionally talk about it and discuss the possible effects of the result on us and on our relationship. A positive result meant significant decisions would have to be taken, and adjustments made, in terms of our family, careers and personal plans.

As a partner, I was worried about how I would deal with a positive result personally, and how I could support my partner in the best way possible.

As a gene carrier, when I felt I was ready to face my fear, I announced to my partner and my family that I wanted to be tested for the HD gene. At that point, I felt a strong need to know what would come next in my life. In 2014, I made my decision definitively and arranged an appointment at the neurology clinic for the testing procedure. About a month later, I received the result, which was positive. It was a difficult time for me and for the people around me. At first my feelings were mixed – relief that I finally knew, fear of what lay ahead, anger and disappointment for finding myself in this situation. Soon, though, there was hope. Hope is what keeps us going. I decided to share my status with people I knew would not use the information against me or in any way that would affect me negatively. People I felt could give me strength and be positive. I asked those closest to me – my mother, partner, brothers and close friends – to use the information in the same spirit. They could share it but only with trustworthy people.

As a partner, I have to say that the period following the diagnosis was one of the most difficult of our life together. However, the positive attitude of my partner was what helped us both cope and manage our emotions. We continued to discuss how we felt about the situation and to share our thoughts and worries openly. We managed to replace many of the negative feelings that HD produces with hope. In my opinion, the person who undergoes the genetic test is the only one who is entitled to decide with whom he or she wishes to share both that experience and the test result. In our case, the fact that both our families were informed from the beginning helped significantly. The role of the physician you partner up with in this journey is equally significant. Physicians need to provide proper guidance to the person tested, so that they receive the necessary psychological support to prepare for the result, and are able to build a support network that will give them strength and positivity in the face of whatever the future holds. Nobody should fight HD alone.

Marcela Gargiulo

Alexandra Durr

On families and their secrets

Marcela Gargiulo and Alexandra Durr

Secrets can interfere with the duty to inform. We would like to illustrate this with a case study from the clinic.

A 64-year-old man at risk for HD had never told his children about his risk or the fact that his mother had died of the disease. Since his wife had passed away, this made him the only person who possessed this information. Having been diagnosed with metastatic cancer, his thoughts turned to his own end and he decided to take the presymptomatic test for HD. In light of his age, he felt sure that he would find out he was not a carrier, but to his surprise the test revealed that he was – his *HTT* gene harbouring a small, but still pathological,

stretch of CAG repeats. After several interviews with our genetic counselling team, he remained determined only to mention his test result in his will, which would be read after his death by his son – a doctor. "Dear Son," he wrote, "I was a carrier of the genetic mutation that causes Huntington's disease. This concerns all of you, including your children. Please forgive me, Papa."

What can we learn from this case? This gentleman's response to his own risk was strongly influenced by his family history, because in his family HD carried a powerful stigma and had always been kept secret. It was also shaped by his personality, because he was a person for whom projecting an unblemished image of himself was very important. Finally, his concept of parental responsibility required him, first and foremost, to protect his family from information that could potentially destroy his children's future.

Two photos from Adamos Papantoniou's 2018 exhibition "Settled and Sacred Landscapes of Cyprus: Images for Huntington's Disease"

Sun of Hope

Sometimes Hope is only in the unseen

His case illustrates how a refusal to tell the truth is not always the result of indifference or neglect of the other; it can also stem from a fear of harming the other, making it "a lie for love", in this particular father's words. One might legitimately ask if protecting his family was merely a pretext for preserving his self-image. A study that our group carried out last year sheds some light on this (RISQUinfo, Durr and Gargiulo 2019). We found that only 48% of people from HD families had been informed of their risk by their parents. The case we have described is therefore not exceptional, and informing one's children of their risk of HD remains a taboo in many families.

Is not informing one's children a lie? A lie is a deliberate assertion of false information, made with the intent to mislead. Some types of lie are generally considered morally permissible – pious lies that keep the peace, for example, and white lies that have no consequence. But there are other types of lie that, while altruistically motivated, affect the lives of others and do have consequences. Such lies spring from a disconnect between the ideal image one holds of oneself, and reality. A person desperate to maintain an ideal image of himself is vulnerable to lying in this way, and to becoming trapped in his own lie – organising his life around it. This was the case of the gentleman whose story we told. Unable to tell his family the truth in his lifetime, he condemned them to a terrible solitude after his death.

A secret often lies buried deep in family histories. Among the subjects that are often treated as taboo are sexual orientation, adoption, illegitimate children, suicide, psychiatric illness, and heritable diseases of all kinds. When the secret concerns the nuclear family, it can disrupt the harmony of the home, which should be a place of trust, loyalty and mutual support. Some secrets are harmless, but others can pose a physical and moral threat. This is the case with genetic diseases, when the

withholding of information places others at risk or prevents them from making informed choices.

Secrets are kept alive by the inability to accept that our existence, and that of those we love, rarely lives up to our ideals. And since this inability is part of human nature, and family secrets have been at the core of family life since the beginning of time, we believe that it is unrealistic to think that people could ever be convinced to inform their descendants of a genetic risk, if their deep conviction was that it was better not to.

Marcela Gargiulo is a clinical psychologist and Alexandra Durr is a clinical geneticist. They both work at the Pitié-Salpêtrière Hospital in Paris.

The neurologist's view

Patrick Weydt, Co-Chair,
EHDN Executive Committee

For a neurologist, caring for a patient with HD often begins with delivering bad news. Sometimes the diagnosis has been expected or dreaded for many years, sometimes it comes out of the blue. Never is it welcome.

When informing a person about a genetic or clinical diagnosis of HD, the neurologist has to be mindful of that person's right-to-know and her equally important right-not-to-know. Ethically balancing these two mutually incompatible propositions in an individual case is of course challenging. A good patient-physician relationship is essential, so that the patient can take the time to discover her own personal need to know – or not.

Because the HD mutation is an autosomal dominant trait that follows fundamental and well known laws of genetics, a positive test result has immediate implications for the closest relatives of the individual tested. Since each of these may position themselves differently with respect to their right-to-know their risk status, or not to know it, this can create dilemmas. A parent may not wish to know her genetic or disease status, for example, while her adult child may have compelling reasons for learning it. Often different family members are cared for by different specialists: a symptomatic patient is cared for by a neurologist or a psychiatrist, while asymptomatic, at-risk individuals may be counselled by a geneticist. While it is not always possible to resolve such conflicts of interest to the satisfaction of all involved, a close working relationship between specialists can help defuse problematic situations without compromising patient confidentiality.

In addition there are cultural or national differences in how the balance between the right-to-know and the right-not-to-know is handled. In Germany, for example, the right-not-to-know genetic information is explicitly protected in law, while in other European countries it is not.

One practical way to improve the testing experience for those who undergo it is to implement tandem teams consisting of a geneticist and a neurologist or psychiatrist, who deliver the diagnostic information together. This requires good teamwork between the specialties, but it means that the person receiving the test result is not lost between two systems at a time when she needs all the support she can get.

With the uptake of predictive genetic testing on the rise in the wake of trials of potentially effective gene-silencing therapies, these conundrums will likely become more common rather than less. Open communication respectful of patient confidentiality is the best way forward.

IN BRIEF

Should doctors have a legal duty to warn relatives of their genetic risk?

Last November, Anna Middleton of the Society and Ethics Research Group at the Wellcome Genome Campus in Cambridge, UK, and colleagues, published a Comment with this question for its title, in the medical journal *The Lancet*. They described cultural differences in the relative importance that people attach to protecting patient confidentiality. Americans and Israelis tend to consider confidentiality paramount, for example, and disapprove of doctors sharing a person's genetic information without that person's consent. In contrast, the researchers found that British people support a legal duty to share such information, even in the absence of consent. Their cross-cultural survey continues. You have to have an account with the journal to read the article, but it's free to register.

Ethics in research

Mette Gilling Nielsen, Enroll-HD Scientific Project Manager

After the HD gene was discovered in 1993, it quickly became apparent that even though there was only one genetic cause of HD – the pathological CAG expansion in the *HTT* gene – the size of that expansion was not the sole determinant of when motor symptoms started. Since then, researchers have been searching for other genetic variants that influence disease onset, with the ultimate goal of refining the prognosis for individual HD expansion carriers, and developing new treatments that delay disease onset.

Today, technologies exist that enable rapid screening of large portions of the human genome affordably and in days, making identification of such variants easier. There is much excitement around these new technologies, and the knowledge they will generate, but they also present novel ethical challenges.

A key concern is how to deal with incidental findings – that is, previously unknown genetic predispositions to disease that are discovered unintentionally. Before the days of whole genome sequencing, researchers and clinicians only looked at specific, predefined genes in their analyses, so there was no risk of stumbling across a disease-causing variant in another gene. That is no longer the case. So what should be done if in the course of HD research, for example, a study participant is found to be a carrier of another genetic disease?

Ethical guidelines that speak to this and similar situations are emerging in some European countries, but they tend to deal with prospective genetic studies rather than with data or samples that have already been collected, and they are not always consistent between countries. The informed consent form used to obtain a sample also provides guidance on how the sample can be used, but it contains some ambiguity too. How do we respect both the individual's right-to-know about incidental findings, and her right-not-to-know? In a global study like Enroll-HD, how do we make sure we comply with different countries' regulations while still ensuring consistency in our methods? These are the questions we are currently grappling with.

Another challenge is our obligation to protect the identity of study participants. This is important because inadvertently revealing a participant's identity could have real-life consequences for her, such as higher health insurance premiums or difficulties in getting a job. Certain types of personal data – or combinations of such data – are considered highly identifying (including name, address, date of birth, phone number and postcode) and are never released to researchers. Moreover, each time a dataset is released, the potential risk of identification is assessed for each participant, given the specific combination of variables requested. If the risk is too high, the dataset is adapted to reduce that risk to a minimum. The advent of whole genome sequencing data has made this more difficult, because an individual's complete DNA sequence is unique, making them identifiable if that sequence is matched to genetic data obtained elsewhere – via online genealogy databases, for example.

From genetic data it is possible to infer biological kinship, and online genealogy services such as MyHeritage exploit this potential to enlighten their customers about their ancestry and potential health risks. Such commercial datasets have proliferated in recent years, and since they may combine more or different types of identifying information with genetic data, than our dataset does, their existence potentially increases the risk of identification of study participants from the dataset that we deliver.

It is normal that legislation and guidance should lag behind the cutting edge of science, leaving a temporary ethical void, and it is the responsibility of the researchers conducting that science to fill the void until states or professional bodies catch up. We in the HD research community are defining new ways to protect study participants without impeding valuable research that promises better diagnosis and care for those affected by the disease.

HD and social media

Gráinne Bird, Ireland

I grew up in a family affected by HD. My Dad, who has the illness, became symptomatic at an early age and his health has steadily declined ever since. He currently requires 24/7 care.

As a child, I was a carer to my parent. When you're affected by such a rare and isolating illness it can alienate you from your peers. The only ones who understood what I was going through were living under the same roof as me. They were my siblings and my mother.

Circumstances like these can make or break a family. Thankfully ours became strong. My Mam became heavily involved in the Huntington's Disease Association of Ireland (HDAI), and in the HD community internationally. Because of this, I grew up in an informed and supportive environment. Unfortunately, not everyone is so lucky. My Dad was shunned by his own family, and left to deal with this shattering illness in a cloud of familial denial, secrecy and shame. This cycle of stigma needs to end.

The benefit of using social media to promote a rare illness such as HD is first and foremost that it helps build a community. It is unlikely you will meet another family with the same rare illness by chance, in the real world, but you can find them online.

I participate in many HD-related groups on social media platforms such as Facebook and Twitter. These groups are filled with people who want to share their stories, ask for advice, or just feel that they're not alone. This is vital for those affected by HD, because feeling that you are part of a community is very often the difference between existing with the disease and living with it. It helps you feel less isolated or alienated. For those who cannot talk publicly about the presence of HD in their family, social media allows them to connect with others, and ask for information and support, while maintaining their anonymity.

Generally speaking, people outside the community are not aware that HD exists or are not well informed about it. Raising awareness is crucial if we inside the community are to obtain the help we need – and to help ourselves – in dealing with the problems that HD presents to us. Promoting awareness online often encourages friends to do their own research into the illness. That is so satisfying and means so much. It's also a great way to stay on top of new developments in research, and to inform others about them. I have also found social media to be a very useful tool for raising funds – in my case, for the HDAI. It has helped me reach a wider audience when promoting events, not just locally but globally too. And advances in technology have made it easy – and safe – for people to donate online.

Social media platforms do have flaws, of course – the most notable one being that they can spread misinformation – and they should be used with caution as a result. But as long as we are aware of these potential dangers I think social media holds huge promise for us in the HD community. Long may we embrace it and benefit from it.

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 Kristina Bečanović: kristina.becanovic@euro-hd.net

Update: Clinical trials

Jenny Townhill and Tim McLean, Central Coordination

The following trials have been endorsed by EHDN. EHDN endorsement of a study protocol follows review by the EHDN Scientific and Bioethics Advisory Committee, which makes its recommendations to the Executive Committee. A formal letter of endorsement may then be issued to the study sponsor, signalling to the HD community, regulatory authorities and/or ethics committees that the study protocol has been reviewed and endorsed by a group of expert HD scientists and clinicians.

PRECISION HD1 AND HD2: On 30 December 2019, sponsor WAVE Life Sciences announced topline data from the ongoing phase 1b/2a PRECISION HD2 trial of the allele-specific stereopure oligonucleotide WVE-120102. For patients treated with multiple doses of WVE-120102, there was a statistically significant reduction of 12.4% in mutant huntingtin in cerebrospinal fluid (CSF), compared to patients receiving placebo. This reduction was found at the highest doses tested, but no difference in total huntingtin was reported in the treatment group compared to placebo. The analysis also examined the presence of neurofilament light chain in CSF, finding no difference between the treatment and placebo groups. WVE-120102 was generally safe and well tolerated across all dose cohorts (72% of those who received WVE-120102 experienced an adverse event, versus 83% of those who received a placebo). This finding supports the inclusion of higher dose cohorts in the trial, and WAVE is planning to launch a 32mg cohort in early 2020. Recruitment to both trials continues at sites in Australia, Canada, Denmark, France, Poland and the UK, and further sites are due to be activated soon (see ClinicalTrials.gov for the current list). Based on the PRECISION HD2 results to date, a 32mg dose cohort is also due to be added to the PRECISION HD1 study. Topline results from PRECISION HD1 are expected this spring.

GENERATION HD1: Recruitment to this Roche global phase 3 trial of the huntingtin-lowering antisense oligonucleotide RG6042 for participants with early manifest HD is ongoing, subject to regional approvals for the latest version of the protocol. The protocol has undergone two amendments since the start of the trial, with a change to the dosing frequency announced in March 2019, and an increase in the number of trial participants (from 660 to 801) announced the following October. The additional participants will provide more data to facilitate evaluation of the two dosing groups, and to increase the statistical power of the study. A list of participating sites and their current status can be found at ClinicalTrials.gov.

HD-DBS: This multicentre trial of pallidal deep brain stimulation for HD, sponsored by the University of Düsseldorf, continues at sites in Austria, France, Germany and Switzerland. The target number of participants has nearly been met, and the trial is expected to complete recruitment in the first half of this year. For further information, please contact: dbs@euro-hd.net

PACE-HD: Recruitment to this activity intervention study, sponsored by Cardiff University, was completed in May 2019. Follow-up visits continue at seven sites in Germany, Spain and the US, with results expected in the first half of this year.

Update: Enroll-HD

Olivia Handley

Enroll-HD collects information about individuals' health and wellbeing, including information on their HD genetic status. Such information is sensitive and potentially identifying, and Enroll-HD has the responsibility of ensuring that it is collected, stored and shared in accordance with local regulations relating to data protection and confidentiality. In the European Union, for example, this would be the General Data Protection Regulation, which came into effect in 2018.

At the start of Enroll-HD, the Data Safety Monitoring Committee (DSMC) was set up to address concerns regarding participant safety and the integrity of safety-related data collected in the study and in selected studies that use the Enroll-HD platform. The following is an explanation of what the DSMC is and what it does.

Who is/are the DSMC?

The five members of the DSMC – Ken Cheung of Columbia University, Susan Fox of the University of Toronto, Arvid Heiberg of Oslo University Hospital, Aad Tibben of Leiden University and Daniel Weintraub of the University of Pennsylvania – have been selected based on their independence from the Enroll-HD study and lack of vested interests in it. The committee is chaired by Arvid Heiberg who has decades of clinical and research experience in the field of HD.

Why is the DSMC needed for a study like Enroll-HD?

There are several important considerations regarding the safety and integrity of the data collected in Enroll-HD.

First, every participant in the study is required to donate a blood sample, which undergoes testing for the HD gene expansion. This includes individuals who have already been tested locally and are either carriers (premanifest or manifest for HD) or non-carriers, and individuals who have not been tested locally, meaning

Photo: G. Stautner, Artifox

their HD genetic status is unknown. The DSMC reviews aggregate and individual data, including CAG repeat length discrepancies between the Enroll-HD test result and, if available, the local predictive/diagnostic test result. If the review identifies any significant discrepancies that could have an impact on the care of the participant, the DSMC reports these to the Enroll-HD site investigator.

Second, Enroll-HD captures data on reportable events which include but are not limited to the aforementioned research/diagnostic genotyping discrepancies, suicide attempts, completed suicides, mental health events requiring hospitalisation, and deaths from any cause. The DSMC is responsible for regularly reviewing all reported events and communicating its findings and recommendations to the Enroll-HD operations team. The operations team then disseminates those findings and recommendations to the study investigators, institutional review boards or ethic committees, and regulatory agencies, as necessary.

All data provided to the DSMC, all DSMC discussions and all outcomes of its review process are treated as confidential.

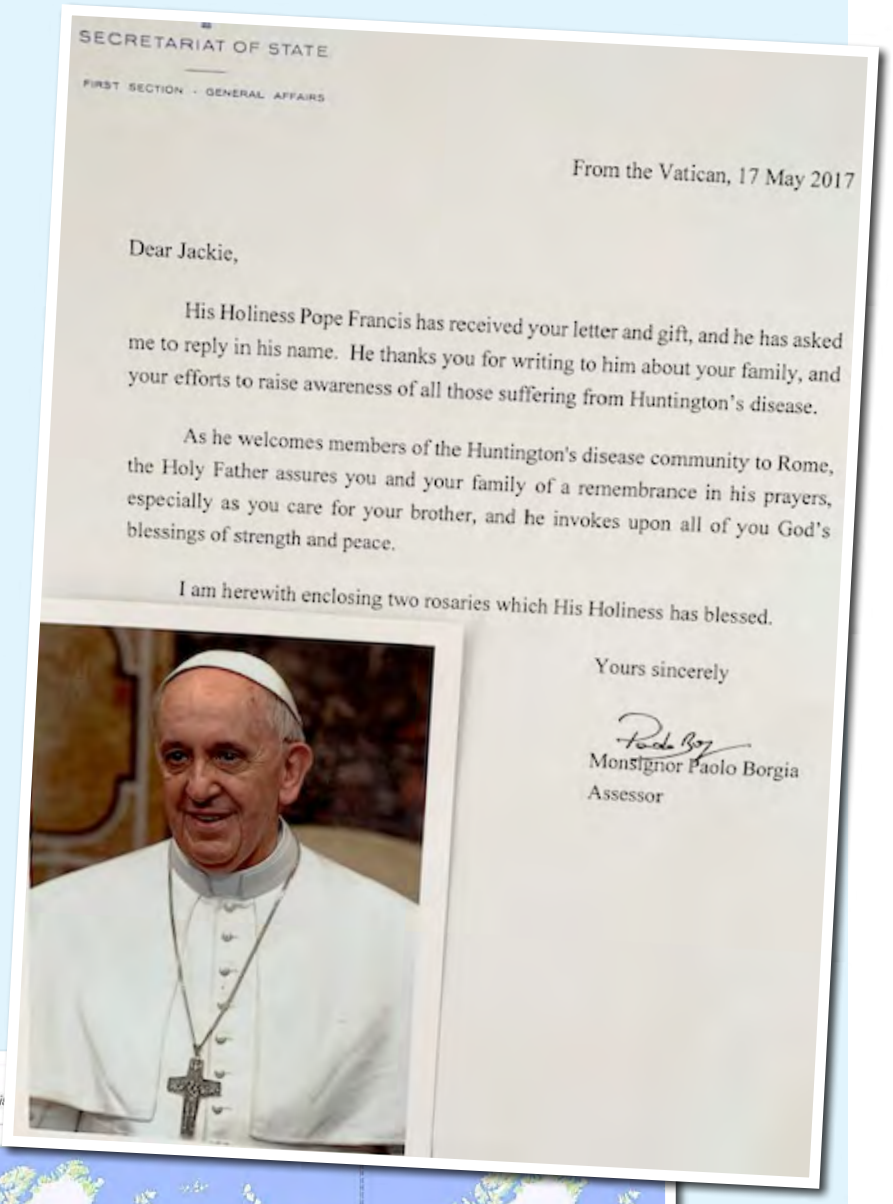
Fellowship exchange programme

The 2020 round of the fellowship exchange programme, which is sponsored by EHDN and the European section of the International Parkinson and Movement Disorder Society (MDS), is now accepting applications, but the deadline is 20 March 2020 so hurry and apply now! Six places are available this year. Any queries should be addressed to: fep@euro-hd.net

Send us your photos!

This quarter our photo is generously provided by **Jackie Harrison of Brighouse in Yorkshire, UK**, who cared for her brother Mark – who had HD – before he passed away last year. Jackie is a tireless campaigner, with her "Hounds4Huntingtons" project, and her little felt dogs – all called Sybil after her real dog – have travelled the world with friends and acquaintances. They have also been gratefully received as gifts by celebrities, royalty and the pontiff, who wrote a nice thank you letter, pictured here.

A sample of Sybil's wanderings is shown on the map [*Editor's note:* she came to Los Angeles with me]. She has even made it to the top of Mount Everest, and she keeps on going. Jackie writes, "The project has raised thousands for the [UK Huntington's Disease Association] and [Huntington's Disease Youth Organization] and hopefully a little awareness." Go Sybil!



SECRETARIAT OF STATE

FIRST SECTION - GENERAL AFFAIRS

From the Vatican, 17 May 2017

Dear Jackie,

His Holiness Pope Francis has received your letter and gift, and he has asked me to reply in his name. He thanks you for writing to him about your family, and your efforts to raise awareness of all those suffering from Huntington's disease.

As he welcomes members of the Huntington's disease community to Rome, the Holy Father assures you and your family of a remembrance in his prayers, especially as you care for your brother, and he invokes upon all of you God's blessings of strength and peace.

I am herewith enclosing two rosaries which His Holiness has blessed.

Yours sincerely

Monsignor Paolo Borgia
Assessor

Our photo experiment continues!

Whether you're affected by HD personally, or you're a carer, clinician or scientist working in the field, we'd like to publish your images in the newsletter. If you have a photo that provides an insight into your daily life, that you think might interest or inspire other EHDN members – or make them think differently about the disease – please send it to us along with a few words explaining who you are and what the image shows: newsletter@euro-hd.net

Darren Monckton and Sarah Cumming

Nayana Lahiri

Sebastian Iben

New seed funds awarded

EHDN has approved seed funding for a project proposed by **Darren Monckton** and **Sarah Cumming** of the University of Glasgow, and **Nayana Lahiri** of St George's University Hospital NHS Foundation Trust in London, to investigate whether the precise CAG repeat sequence of intermediate and reduced penetrance alleles of the *HTT* gene influence likelihood of expansion into the pathogenic range.

De novo mutations in the *HTT* gene, which occur at a low rate, are thought to arise from intergenerational expansion of the CAG repeats from the intermediate range (27-35) into the disease range (36 or more). A recent study showed that approximately 6% of the general population carries an intermediate allele, so it would appear that only a small proportion of these expand into the disease range when they are passed on to the next generation.

The researchers will study individuals identified through the Registry observational study as having no family history of HD, and families with intermediate/reduced penetrance alleles, using single nucleotide polymorphism haplotype analysis and next-generation sequencing. The goal is to gain insights into the sequence and architecture of the CAG expansion, to determine their influence on CAG repeat stability. Understanding these mechanisms better will improve the accuracy of genetic counselling for patients with intermediate/reduced penetrance alleles, as well as advance knowledge of HD pathogenesis.

A second project, proposed by **Sebastian Iben** of the University of Ulm, has also been approved for seed funding. The balance of protein synthesis, maintenance and degradation – protein homeostasis – is disrupted in HD, as in all proteinopathies. Iben hypothesises that, as in an unrelated childhood progeria with neurodegeneration that his group recently described, disturbances in protein synthesis at the ribosome might contribute to the overall disruption of protein homeostasis in HD. He will investigate the synthesis, processing, assembly and performance of ribosomes of HD patients, with the aim of discovering if the HTT protein acts as a transcription factor that is recruited to ribosomal biogenesis by RNA polymerase I.

Seed funds are intended to support pilot studies that will eventually kickstart larger projects. The next deadline for applications is 1 November 2020. More information about the programme and how to apply can be found here.

Mapping a moral grey zone:

Interview with Heidi Beate Bentzen

A lawyer specialising in data protection and health law, Heidi Beate Bentzen of the University of Oslo in Norway has just joined the EHDN's Scientific and Bioethics Advisory Committee. Her role is to consider the ethical and legal aspects of HD-related research projects that the network has been asked to fund or provide with data. Her academic interests lie in the legal regulation of precision medicine, including the use of biological samples and data.

What is the significance of HD for you?

Over the last few decades, the number of genetic tests available has expanded enormously, but many of the laws relating to the sharing and protection of health-related data are modelled on HD, since it was one of the first diseases for which a reliable genetic test became available. So it's really a poster child for our field.

How does legislation on the sharing of genetic test results differ across Europe?

A couple of years ago I was part of an EU initiative to survey European countries about their legislation in this area. In most of them, a clinician can't share the patient's test results with relatives without the patient's consent, but there are exceptions. In Norway, for example, there are circumstances in which the clinician is permitted to bypass the patient and share the results with relatives – if there is a conflict in the family, say. The situation becomes more complex when the clinician's duty to rescue becomes relevant.

Could you explain what you mean by the clinician's duty to rescue?

If not sharing the results of a test poses an immediate danger to someone's life, in most European countries a health professional is obliged to share it. A professional could make that judgement in the case of HD (though a positive test result for HD does not, on its own, fulfil the disclosure criteria for a clinician's duty to rescue), but it's rare in connection with most diseases for which tests exist. Usually we're talking about a high risk of developing the disease, at most.

Photos kindly provided by Heidi Beate Bentzen

As a lawyer, do you think Europe-wide?

Yes, but not only. On a global level, I'm a member of the United Nations' Special Rapporteur on the Right to Privacy's Task Force on privacy and the protection of health data. Last year we published a recommendation that can be used, either as a model for legislation by those countries that want to legislate in this area, or as a model of best practice.

Is the law keeping up with the technology?

I think the law is always a step behind the technology, actually, and that can be a double-edged sword. On the one hand, people may look to the law for guidance and find it offers little. On the other, you could argue that we benefit from being able to accumulate experience in using the technology and reflect on how best to regulate it.

Given that practically every case is different, when it comes to disputes over the sharing of sensitive health-related information, is there an argument for not legislating at all in this area – for leaving decisions to the discretion of the clinicians involved?

A lot of European legislation, at least in civil law countries – those of continental Europe as opposed to the UK, which practises common law – already has that flexibility built into it. The laws are not that detailed, and you have leeway in how you apply them.

Do good legal tools exist for protecting people's privacy, or do we need better ones?

We have strong legal protection for privacy at the EU level. However, I think we need better tools for incorporating privacy into digital systems, especially given the vast amounts of data that are now being generated. At the European level, there's currently a push for privacy by design – where the privacy solution is built into and prioritised by the technology. Dynamic consent is also emerging as a useful tool. This allows people to log into an electronic portal, for example, and adjust their preferences regarding what they want to know and what they don't want to know. Another promising approach involves encryption tools that strip away all unnecessary information, so that researchers receive only what they need to know about patients and no more.

"Direct-to-consumer testing is a loophole that needs closing. Europeans aren't sufficiently aware of what they are signing up to."

Are there any loopholes in the current legislation, in your view?

Yes, direct-to-consumer (DTC) testing is a loophole that needs closing. Europeans aren't sufficiently aware of what they are signing up to, or that by sending off their information – typically to the United States – they lose a lot of the protection that they enjoy here. Some DTC companies collaborate with law enforcement, for example, so that by sharing your data you could potentially incriminate yourself or a relative. People should bear in mind that if the companies target European consumers, they are obliged to abide by European privacy laws, but many of them don't. It is the European citizens who reach out to them, thereby stripping away their own protection.

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Dates for your diary

Save the dates for:

- Huntington's Disease Youth Organization's 1st International Young Adults Conference, Glasgow, UK, 9-11 May 2020
- The 10th European Conference on Rare Diseases & Orphan Products, Stockholm, Sweden, 15-16 May 2020
- EHDN2020 plenary meeting, Bologna, Italy, 10-12 September 2020 (details to come)

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