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Patrick van der Wel

I have a longstanding interest and expertise in RNA regulation, non-coding RNAs, and genome-wide approaches in genetic neurological diseases. Thanks to training at Massachusetts General Hospital and Harvard Medical School, I gained extensive experience in the human genetics of rare disorders, particularly Huntington's Disease (HD).

In the lab, we utilize molecular genetics in human and mouse systems that replicate causative human mutations, leveraging genome-wide methods, epigenetic/chromatin analysis, and RNA sequencing. Our research focuses on the understanding of alternative mRNA splicing and the back-splicing mechanisms generating circular RNAs (circRNAs). Specifically, we characterized how mutant huntingtin affects linear and back-splicing genome-wide and locally, at the HTT locus, where we described circHTT(2,3,4,5,6), the first characterized circRNA stemming from the HTT locus. We are also interested in evaluating if and how circRNAs could function as valuable biomarkers in the HD pathology. Our program is supported by a multi-round Telethon grant, alongside funding from the Marie Skłodowska-Curie actions, the EMBO fellowship, Huntington's Society of Canada, HDSA, and EHDN.

Beyond research, I serve as an active member of the EHDN Scientific and Bioethics Advisory Committee (SBAC) since September 2022, contributing to the review of biannual Seed Funds. I am also an experienced ad hoc reviewer for journals including Nature Biotechnology, EMBO reports, EMBO Molecular Medicine, Neurobiology of Disease. Additionally, I regularly evaluate international grants for AFM-Telethon (2024, 2025), Association Huntington France, FRM (MND-Maladie Neurodégénérative), the Medical Research Council (UK), and the Swiss National Science Foundation (SNSF) on topics spanning neurodegeneration, genetic nervous system disorders, and neurodevelopmental syndromes.

Given this expertise in RNA biology and neurogenetics, my interest and commitment to the EHDN, and my extensive international reviewing record, I'll be happy to continue contributing to the SBAC's evaluation tasks in Basic Biology, Neuroscience, RNA Biology, and Genetics.

Dr Giulia Coarelli

Paris Brain Institute, Pitié-Salpêtrière University Hospital

FRANCE

I am a Neurologist with a subspecialty in neurogenetics, working at a tertiary referral center for rare inherited neurological diseases, including the Reference Center for Huntington Disease at Pitié-Salpêtrière Hospital, and in the research laboratory led by Alexandra Durr and Sandrine Humber at the Paris Brain Institute. My clinical and research activities focus on repeat-expansion disorders, particularly Huntington disease and spinocerebellar ataxias. I am involved in diagnostic evaluation, longitudinal patient management, and translational research, with a strong focus on biomarker development and clinical trial readiness. I have extensive experience in clinical research as principal investigator and sub-investigator in multiple French and international clinical trials in Huntington disease and spinocerebellar ataxias, as well as in natural history cohorts. This work has provided me with a solid understanding of methodological issues related to clinical trial design, endpoint selection, and feasibility in rare neurodegenerative diseases. I am also involved in presymptomatic and prenatal testing, as well as early diagnosis in inherited neurodegenerative disorders, which has strengthened my awareness of the ethical challenges associated with predictive medicine and emerging disease-modifying therapies.

In addition, I have experience in academic peer review for international neurology and neurogenetics journals, as well as in the evaluation of research projects in the field of neurodegeneration.

Within the EHDN Scientific and Bioethics Advisory Committee, I would aim to contribute by combining clinical expertise, translational research experience, and ethical awareness. I am interested in supporting discussions on clinical trial methodology, biomarker qualification, and the implementation of emerging therapies in HD. I would also contribute to reflections on ethical issues related to presymptomatic and prenatal testing, disease-modifying trials, and the evolving definition of premanifest disease. Overall, my goal would be to support SBAC activities by bridging clinical practice, research innovation, and ethical reflection to advance patient-centered care and research in Huntington disease.

Dr Sarah Gunn

University of Leicester

UNITED KINGDOM

I am a practising clinical psychologist and researcher, with my main focus on understanding and supporting mental wellbeing difficulties in HD. I have published numerous peer-reviewed papers on HD psychology, held an EHDN Seed Fund award (recently completed), created successful mental health therapeutic interventions for people affected by HD, and I was recently awarded an eight-year Wellcome Career Development Award (£1.96 million) to research mental health and HD. I lead the active University of Leicester HD Psychology Research Group and co-lead the East Midlands HD Network. I have longstanding collaborations in the HD third sector through which I offer mental health support for HD families UK-wide, and I recently co-wrote national guidance with the HD Association on cognition and mental health support for HD families. I contribute across multiple EHDN WGs, and co-lead the newly-established End-of-Life WG.

I review for various high-quality journals focused on neurology and physical health conditions (e.g., Journal of Huntington's Disease, Journal of Rehabilitation Medicine, Disability and Rehabilitation). I have reviewed funding applications for the ESRC Peer Review College and MS Society, and I am an expert member for an NHS Research Ethics Committee (reviewing research ethics applications). I review academic book proposals for Routledge and Cambridge University Press, and I am an editor for the upcoming textbook "Clinical Neuropsychology Essentials" and the recently-published "Pathways into Clinical Neuropsychology".

I hope bringing my clinical psychology background to SBAC would provide psychological knowledge and skills which may be currently less well-represented on the Committee, but which are of increasingly recognised importance in HD research and clinical care. I am also from an HD family myself, and my book on psychological wellbeing in HD (from clinical and personal perspectives) will be published this summer. Accordingly, I hope I could contribute to SBAC from academic, clinical and personal perspectives.

Dr Davina Hensman Moss

University College London

UNITED KINGDOM

I am a Clinician Scientist, passionate about advancing understanding of Huntington's disease in order to improve the lives of people affected by the condition. I have worked on Huntington's disease (HD) for 14 years, initially doing a PhD under Professor Sarah Tabrizi at UCL (UK). Following this, I focused on clinical work before gaining a lectureship at St George's, and subsequently returned to UCL to establish my own research group. My research has focused on the identification of genetic modifiers of HD and other trinucleotide repeat disorders, resulting in several prizes and highly cited papers. Over recent years I have been studying genetic anticipation and germline CAG repeat instability in HD. I have been co-Lead Facilitator of the EHDN Genetic Modifiers Working Group since January 2022.

In addition to my academic work I am also a Neurology Consultant. I do both specialist Huntington's disease clinics and General Neurology clinics; I particularly value this clinical work because the issues and questions raised by patients help guide and motivate my research.

I have experience reviewing journal articles for journals including Movement Disorders, European Journal of Neurology, Journal of Huntington's Disease and EBioMedicine. I have served on the SBAC since 2022, during which time I have developed my skills in assessing Seed Fund Grant applications and clinical trial protocols, always aiming to provide balanced and constructive feedback.

The EHDN's role of supporting research in Huntington's disease, promoting collaboration, facilitating clinical trials and improving clinical care is invaluable to the HD community. I would welcome the opportunity to contribute both clinical and scientific perspectives to help support the EHDN's evolving priorities. It would be an honour to continue supporting the EHDN's goals through the SBAC for an additional term.

Dr Nicola Hobbs

University College London

UNITED KINGDOM

Area of expertise:

I am a Senior Research Fellow at the Huntington's Disease Centre, within the UCL Institute of Neurology. My research focuses on the development of neuroimaging biomarkers for HD and their implementation in clinical trials. I have worked on observational studies of HD whilst at UCL (TRACK-HD, PADDINGTON, HD-YAS), and interventional trials of HD whilst working for an imaging CRO, so I have experience of planning, acquiring and analysing neuroimaging data in both academic and industry environments.

Previous reviewing tasks:

I regularly perform peer review for a range of journals, including Biological Psychiatry, Human Brain Mapping, Brain Imaging and Behavior, Journal of Huntington's Disease, Biomarker Research and Scientific Reports. I have reviewed book chapters on neuroimaging in HD and grant proposals for the European Research Council and for the SBAC. I have also provided peer review of clinical trial protocols as a member of the SBAC.

How I hope I can make a contribution to the SBAC:

Grant proposals and clinical trial protocols for review by the SBAC often include a neuroimaging component. I hope I can use my knowledge of neuroimaging in HD to provide constructive and clear feedback to applicants and potentially improve the quality and robustness of findings when the study is executed. I am passionate about collaborative research, and I think the SBAC is a wonderful example of the HD community working together to facilitate advancements in the field.

Prof. Peter Holmans

Cardiff University

UNITED KINGDOM

I have over 30 years' experience in the statistical analysis of complex genetic traits, both in data analysis and developing novel genetic methodology. I have led the statistical analysis of large multicentre collaborations in genome-wide linkage of schizophrenia and association in Alzheimer's disease and schizophrenia. I have a particular interest in the use of pathway analysis to infer disease relevant biology from genomic data of multiple types, and have developed novel methodology to perform pathway analyses in genome-wide association data. I have led the pathway analysis of multiple large international collaborations, studying copy number variants in ADHD and genome-wide association studies of Parkinson's disease, Alzheimer's disease and psychiatric disorders.

Of particular relevance to HD and other repeat disorders, I led the pathway analysis of the GeM genetic modifiers GWAS, and also a GWAS of HD progression, implicating DNA repair pathways as modifiers both of age at onset and progression in HD. I also led the statistical analysis implicating SNPs from these pathways as age at onset modifiers in other repeat disorders. Currently, I am working in the GeM consortium to study genetic modifiers associated with psychiatric and behavioural phenotypes in HD.

I have been a member of the EHDN SBAC since 2011, and the Enroll Scientific Review Committee since 2018, reviewing seed fund and data access applications. More generally, I have reviewed grants as part of the MRC Methodology Research Panel, the Health Research Board of Ireland, and the Swedish Research Council, as well as several grants and journal papers on an ad hoc basis. I am also an Editor for the Annals of Human Genetics.

I can contribute my extensive knowledge of statistical genetics to the SBAC, and the HD field in general. I can also contribute general statistical and bioinformatic knowledge.

MD José Luis López-Sendón Moreno

Hospital Ramon y Cajal (Madrid)

SPAIN

I am a Spanish neurologist with 18 years of experience in Huntington's disease and movement disorders at Hospital Universitario Ramón y Cajal, a national reference centre (CSUR) for rare movement disorders in Madrid, Spain. My clinical and research expertise focuses on Huntington's disease, neurodegenerative disorders, clinical trials, biomarkers, and patient-centered care. I have been actively involved in longitudinal cohort studies including REGISTRY & ENROLL-HD, as well as multiple international clinical trials in Huntington's disease, consistently achieving high recruitment and retention rates.

I have experience in scientific reviewing and evaluation activities, including peer review for journals and grant evaluation for the Instituto de Salud Carlos III (Spanish Ministry of Science and Innovation). I have also contributed to regulatory and advisory activities, including the Therapeutic Positioning Reports for the Spanish Medicines Agency (AEMPS), participation in Data Safety Monitoring Boards, and advisory boards with pharmaceutical companies involved in neurodegenerative diseases.

A particularly important aspect of our work is the close collaboration with the Spanish Huntington's Disease patient association, including joint educational activities, patient engagement initiatives, dissemination of scientific knowledge, and support for families and caregivers.

I believe I could contribute to the SBAC through a combination of clinical expertise, experience in clinical trials and scientific evaluation, strong patient-centered commitment, and active participation in collaborative HD networks and translational research initiatives.

Dr Saül Martínez-Horta

Hospital de la Santa Creu i Sant Pau, Barcelona

SPAIN

I am a neuropsychologist in the Movement Disorders Unit of the Neurology Department at Hospital de la Santa Creu i Sant Pau in Barcelona, where I also coordinate the monographic Neuropsychology Clinic of the CSUR for rare diseases associated with movement disorders.

I am a researcher in the Parkinson's Disease and Other Movement Disorders Research Group at the Sant Pau Research Institute (IR Sant Pau), where I lead the Huntington's disease research line. I am also affiliated with CIBERNED, the Biomedical Research Networking Centre for Neurodegenerative Diseases of the Instituto de Salud Carlos III. Scientifically, I have authored 130 indexed publications in high-impact journals, with an h-index of 34 and 4,021 citations.

I am also a Clinical Associate Professor in the Department of Medicine at the Universitat Autònoma de Barcelona and I teach in several postgraduate programmes. In addition, I coordinate the Neuropsychology Section of the Spanish Society of Neurology and I am a member of the International Neuropsychological Society, the International Parkinson and Movement Disorder Society, and the European Huntington's Disease Network. Within the latter, I coordinate the Cognitive Phenotype Working Group and serve as a member of the Scientific and Bioethical Advisory Committee.

Dr Tom Massey

Cardiff University & University Hospital of Wales

UNITED KINGDOM

I am a clinical academic Neurologist specialising in Huntington's disease. I originally trained in Biochemistry, receiving my D.Phil from the University of Oxford. I subsequently qualified in Medicine and specialised in Neurology, completing my clinical and academic training in 2021. My practice spans basic wet lab science, clinical trials and clinical work in both HD and general Neurology. I am a group leader in the UK Dementia Research Institute at Cardiff and aim to use genetic modifiers in HD to identify and prosecute new drug targets.

I have been a member of the SBAC for the last 2 years and so understand the role in reviewing seed funds and clinical trial protocols. Given my scientific and clinical background, I am able to make useful contributions across a range of different funding areas. I also regularly review papers for top-level journals including Nature Neuroscience and EMBO journal, and have reviewed grants for various funding bodies both in the UK and internationally.

Dr Duncan McLauchlan

Cardiff University

UNITED KINGDOM

I currently work as Consultant Neurologist for and Cardiff and Vale University Health Board. I am an Honorary Senior Research Fellow for Cardiff University. My clinical work is focused on movement disorders and Huntington's disease in particular.

My research interest is in the biological basis underlying the neuropsychiatric symptoms of Huntington's disease. I have used clinical assessment, and complex modelling of behaviour on cognitive tasks to explore the cognitive processes leading to behavioural change in Huntington's disease. I have a developing interest in using large datasets of 'real world' data to explore the efficacy of antidepressant medication for depression in large populations. Most recently, I have been developing expertise in statistical analysis and health economics used in clinical trials. I have particular expertise in target trial emulation and have used this to demonstrate a possible effect of antidepressants on disease progression in HD.

I have experience in using R, and some experience in Stata. From my previous research I have used a number of different modelling techniques to explore outcomes in large datasets, and have knowledge about the pitfalls and limitations of these techniques as well as the necessary pre-requisites for robust data analysis.

I have published in high impact neurology journals (Brain, Movement Disorders, Journal of Neurology, Brain Communications) and reviewed for a number of journals, including Movement Disorders, Movement Disorders Clinical Practice, Psychological Medicine, Journal of Huntington's Disease, Practical Neurology, Clinical Medicine, Journal of Neurology, Neurodegenerative Disease Management, JNNP & the European Journal of Neurology. I have reviewed some seedcorn grants for the MRC, and am an active grant reviewer for the SBAC. I am keen to continue my work on the SBAC, bringing expertise in marrying clinical assessment, neuropsychological assessment and statistical analysis techniques.

Prof. Dr. Andreas Neueder

University Medical Center Hamburg-Eppendorf

GERMANY

As a trained biochemist with a strong focus on molecular and systems biology, my research sits at the intersection of basic science and translational neuroscience, with Huntington disease (HD) as a primary model. My core expertise spans RNA biology and homeostasis, the molecular mechanisms underlying toxic huntingtin (HTT) fragment generation, mitochondrial biology, and extracellular vesicle (EV) biology in health and disease. A key discovery in my scientific career demonstrated for the first time that the smallest and most toxic HTT fragment arises through incomplete splicing of the HTT pre-mRNA, a finding subsequently validated in human post-mortem HD tissue. Alongside this, my group has established standardised EV purification workflows from biological fluids and is building a reference dataset of EV biology in HD, supported by both academic and industry funding. A major pillar of my work is bioinformatics and systems biology. I have developed state-of-the-art analysis pipelines for RNAseq, epigenomics, proteomics, and multi-omics integration, including correlation network analyses, machine learning-based disease classifiers, and nanopore third-generation sequencing applications.

With respect to reviewing experience, I have served as a regular peer reviewer for journals including Human Molecular Genetics, Acta Neuropathologica, EMBO Reports, and Nature journals, and have reviewed EHDN Seed Fund applications.

I believe my combined background in HD basic science, pre-clinical models, multi-omics methodology, and bioinformatics infrastructure positions me well to make a productive and balanced contribution to the scientific evaluation duties of the EHDN Scientific and Bioethics Advisory Committee.

Christian Saß

Asklepios Klinikum Harburg

GERMANY

I am a Neurologist with special expertise in neurodegenerative disorders, mainly HD, neuropsychiatry, and stroke medicine. I work at a big non-university hospital in the south of Hamburg which is affiliated with the Asklepios Campus Hamburg, a faculty of the Semmelweis-University in Budapest.

My scientific work in HD took place at the Universities of Aachen and Münster and includes preclinical (cell culture, mouse models, stem cell transplantation) and clinical (development of the Q-Motor system, trial design, taking part in multiple clinical trials) projects. I have been a long standing member of the EHDN and took part in the REGISTRY study.

My main areas of expertise are mouse models, stem cell transplantation, gene silencing/ modification, and clinical studies including the development of feasible and robust outcome measures. I see myself mainly as a clinical nowadays, albeit with a firm background in preclinical scientific work in the HD field.

I've been *ad hoc* member of the SBAC since January 2025 and have greatly enjoyed working with the other SBAC members. I would be honoured to be able to continue contributing to the EHDN community through the SBAC.

Dr Chiara Scaramuzzino

IGBMC (Strasbourg)

FRANCE

I focused my career on understanding the molecular mechanisms underlying the pathogenesis of polyglutamine diseases. During my PhD training, I demonstrated that a specific postranslational modification, arginine methylation, regulates the toxicity of androgen receptor that is mutated in SBMA, a polyglutamine disorder. Being convinced that understanding the function and the post-translational modifications of these polyglutamine-containing proteins is the bona fide approach to develop appropriate therapies, I joined the lab of F. Saudou. I developed and used microfluidic devices to reconstitute HD neuronal circuitry in which we can address physiological and pathological cellular mechanisms. Through this approach, I validated a compound of therapeutic interest for HD, found that arginine methylation is not restricted to SBMA but also modulates HTT function. Finally, I identified a new molecular mechanism involving HTT in the transport of signaling endosomes and that contribute to the maintenance of neuronal circuits in the brain. These transport is disrupted in HD and impairs unknown mechanisms of local translational. I am starting my lab at the IGBMC where I will focus on deciphering these mechanisms and develop strategies to restore such disrupted transport in HD.

I co-reviewed with the PI of the team several papers (Neuron, EMBO J, Curr Biol, PLOS Biol). I am regular reviewer at Neurobiol of Disease, editorial board member at Journal of Huntington Disease, and I reviewed for French National Research Agency (ANR), FC3R, BioConvS equipment grants.

Beyond technical expertise, I work collaboratively and with integrity, and I will support clear, constructive discussions. I will combine my expertise with critical and strategic thinking to help the SBAC make decisions. Finally, being part of the SBAC will be an incredible opportunity to be part of the EHDN community and connect deeper with scientists and clinicians that all have in common to contribute finding a cure for HD.

Dr Yury Seliverstov

University Hospital of Ulm

GERMANY

I am a clinical neurologist with a focus on movement disorders and neurodegenerative diseases. HD has been at the center of my clinical and research work since 2012. Since then, I have contributed to numerous HD clinical trials as an investigator.

I also serve as a peer reviewer for journals such as Movement Disorders, BMJ, and the JHD, and have reviewed grant applications in clinical research. Since 2025, I am a coordinator of the ERN-RND Disease Group for Chorea and Huntington Disease.

I will be glad to contribute to the review, development, and facilitation of clinical research projects on HD.

I am a protein biochemist and structural biologist who has worked on the molecular biology of Huntington disease since ca 2010. My lab has revealed the structure of misfolded protein aggregates, mechanisms of misfolding, as well as small molecules and antibodies that can redirect the aggregation process. Via collaborative work we study also the underpinnings of differences in neurotoxic properties of different states of the mutant protein. We have published >15 primary research papers and multiple review articles on polyglutamine and/or huntingtin protein studies.

I am acting as (academic) editor for multiple journals, I have been an invited reviewer for dozens of different scientific journals, and I have acted as grant reviewer for funding agencies from the USA (NIH), EU (ERC), The Netherlands, UK, Germany (DFG, others), France (ANR), New Zealand, and various (international) Foundations. This includes participation in multiple grant review panels.

3. Historically a key focus of the EHDN was to ensure the support of, and attention for, fundamental mechanistic research on Huntington's disease. In this context, there is a critical role for basic research on key mechanisms, such as the mutant gene, the mutant protein and the native role of huntingtin. I would see myself as offering a deep expertise and knowledge of the fundamental mechanisms, thus contributing this expertise to the SBAC. Secondly, I can make sure to represent and consider the needs of fundamental mechanistic research in the SBAC. Regarding contribution to HD research in general: my lab has used its unique expertise to shed new light on multiple key processes in the life cycle of the mutant protein. This includes the structure of the mutant protein in its aggregated state, but also increasingly connections between aggregate strains and their cellular toxicity, and how we can modulate or diagnose/detect these effects.