TWS Symposium 2026

Filling the gaps in HSP and related disorders

March 12+13

Recklinghausen
Germany

Hospital
Knappschaft

Dorstener
Straße 151

Welcome to the Ruhr region!

Meeting booklet

KNAPPSCHAFT KLINIKEN

TWS TOM WAHLIG STIFTUNG

March 12+13

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This year's symposium of the Tom Wahlig Foundation focuses on improving understanding and care for people living with hereditary spastic paraplegia (HSP).

Guided by the vision of "**Filling the Gaps**," the symposium brings together leading international researchers, clinicians, and patient advocates to close the distance between scientific discovery and clinical reality. Remarkable progress in genetic diagnostics, next-generation and long-read sequencing, and molecular technologies has dramatically improved diagnostic precision and deepened our understanding of the complex pathophysiological mechanisms underlying HSP.

With a strong focus on careful progress and translation, the program highlights emerging therapeutic strategies, including precision medicine, small-molecule approaches, and gene- and RNA-based therapies. The long-term goal is to turn scientific advances into meaningful improvements in the lives of individuals and families affected by HSP worldwide.

The meeting takes place at the Knappschaftkliniken Recklinghausen, an institution deeply rooted in the history of the Ruhr region. Emerging from the miners' healthcare traditions of the "Knappschaft," the centre reflects a long-standing commitment to solidarity, care, and medical excellence — values that continue to shape its role as a modern hub for specialist medicine and research.

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General information for your pleasant time at our meeting

Public transport Recklinghausen main station <-> Hospital Knappschaft Bus stops in the city centre: Lohtor	SB25 214 219
Accomodation Both hotels are located in the city center	Holiday Inn Express Herrenstraße 8, Recklinghausen +49 2361 943020 info@express-recklinghausen.de Aspire Hotel, Lörhof 8, Recklinghausen +49 2361 4086600 palais@aspire-hotels.com
Restaurants Both restaurants are located in the city center	**March 11, 19h: Drübbelken, Münsterstraße 5, Recklinghausen** Who arrives already on March 11 can join us for a get-together in the pub "Drübbelken" in the city centre of Recklinghausen. There you can eat and drink in a cozy atmosphere at your own expense. **March 12, 19:15h: Wilma Wunder, Schaumburgstraße 1, Recklinghausen** Here we invite all who have registered for the attendance at our dinner.

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Site plan Knappschaft Hospital

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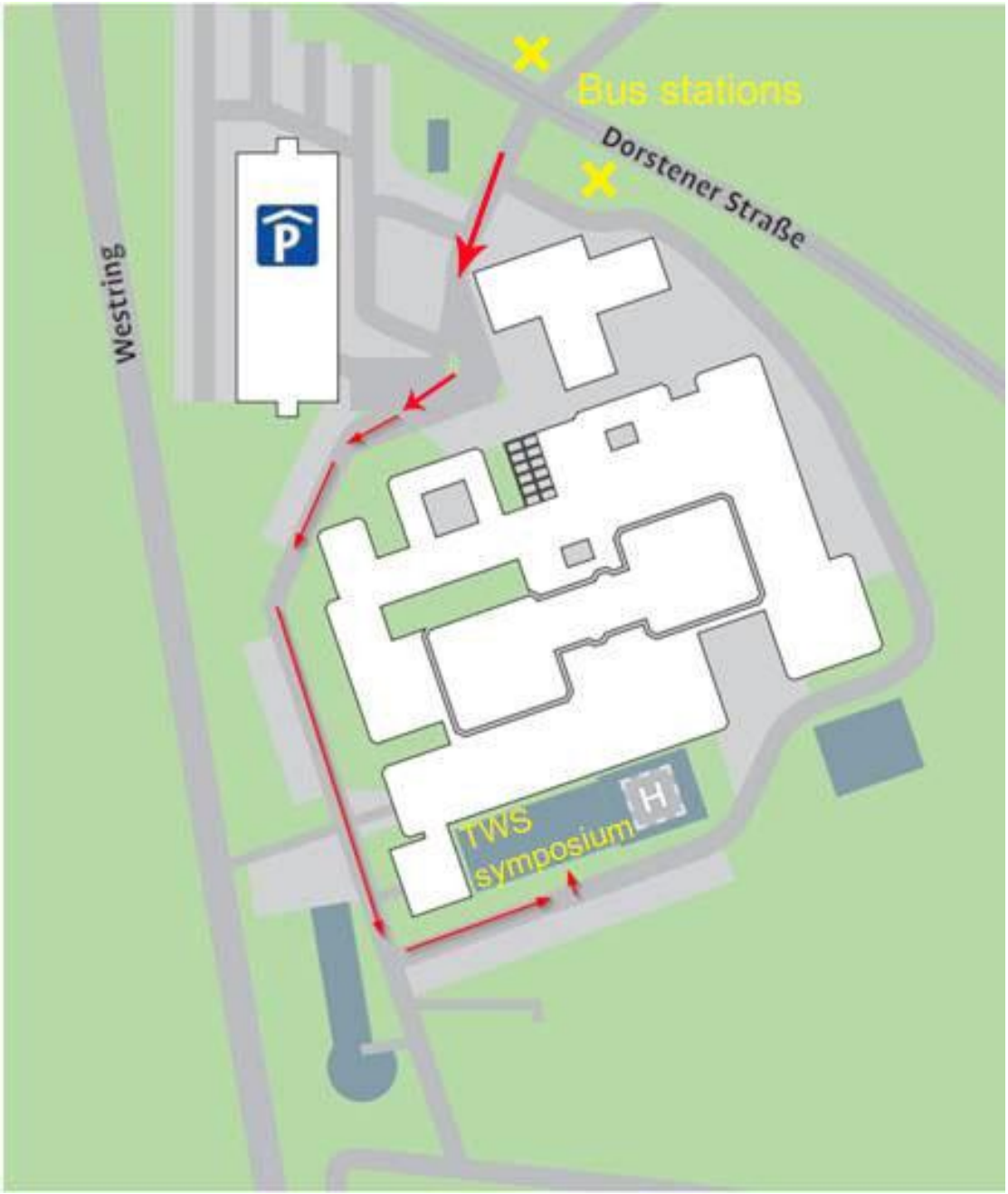

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Susanne & Henry Wahlig
Tom Wahlig Foundation (TWS)

Susanne and Dr. Henry Wahlig jointly run the Tom Wahlig Foundation (TWS), which is dedicated to research into the rare disease HSP.
The fact that hereditary spastic paraplegia is largely unknown and the difficult diagnosis of his son and wife prompted Henry's father, Dr. Tom Wahlig, to set up the foundation in 1998.
Since 2001, TWS has organized an annual symposium at which HSP researchers from all over the world come together at various locations in Germany to present and discuss the latest research findings.
Maintaining this network, the so-called "TWS family", is at the heart of the foundation's work, alongside research funding.

Dear participants — dear colleagues — dear friends!

We are very glad to welcome you all to our symposium 2026 – in Recklinghausen, which is located in the immediate neighbourhood of our home town of Bochum.

We are also delighted to be guests of Prof. Stephan Klebe. He has been actively involved with the foundation from the very beginning and, as a long-standing colleague and friend at our side, he is opening the doors of his clinic to us all in a spirit of warm connection and loyalty for this year's symposium, providing us with the facilities we need to experience scientific exchange and encounters with colleagues from the international HSP research community.

We feel at home here and hope to pass this feeling on to you during these two days of the meeting.

The annual symposium is at the heart of our foundation's work and, despite some routines, organising it is an exciting, varied and enjoyable task for us every year.
In addition to Stephan Klebe, we would also like to thank Prof. Christel Depienne, Prof. Filippo Santorelli and ao. Prof. Jonas Morales Saute for their tremendous support in putting together the programme. They are all presented in more detail in this booklet.

The cooperation with the clinic staff is also essential for the smooth running of the meeting – this year, our sincere thanks goes to Nicole Jeschonneck for taking on important organisational tasks, as well as to Frank Fabek and Sebastian Kähnert's team from SMD for their technical support.

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It is always a great motivation for us to see who accompanies us, supports us and travels to the symposium – from all parts of the world. We are delighted to see familiar and new faces and would like to extend our hand to all of you for a good and fruitful collaboration.

Together, we have the well-being of people with HSP in mind.

The people who live with the various forms of the rare disease HSP and cope with it in their everyday lives are the reason why we are here. They also enrich our work with great ideas and campaigns that help us to raise awareness of HSP.
And it is only with this awareness that we can collect donations, which in turn form the basis for our research work.

Scientific work is often lengthy, sometimes hard and requires a few detours, but – and the comparison is allowed here at the heart of mining – at some point you see the light at the end of the tunnel and can recover the gold. And that is our firm belief: that together with you, we will achieve many medical advances for people with HSP!

Yours, Susanne & Henry

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Stephan Klebe
Recklinghausen, Germany

Stephan Klebe is a board-certified neurologist and adjunct professor with longstanding expertise in movement disorders, neurogenetics, and neurological intensive care medicine. Hereditary spastic paraplegias (HSP) have been a central focus of his clinical and scientific work throughout his entire career, encompassing patient care, translational research, and international collaborations. He has maintained a close and long-standing collaboration with the Tom Wahlig Foundation. After completing his medical training in Göttingen and Kiel, he pursued an extensive academic and clinical career at leading university hospitals in Germany and France, including the Hôpital de la Pitié-Salpêtrière in Paris. Since 2024, he has been Head of the Department of Neurology at Knappschaft Hospital Recklinghausen, while continuing his strong commitment to medical education and specialist training.

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Christel Depienne
Essen, Germany

Christel Depienne is a molecular geneticist and Professor (W2) of Human Genetics at the University Hospital Essen, Germany, where she leads the Neurogenetics research group.
Trained as an Ingénieur des Grandes Écoles at AgroParisTech and holding a PhD in molecular and cellular biology, she has over 20 years of experience in human genetics.
Her research aims to identify genetic causes and molecular mechanisms underlying neurodevelopmental and neurodegenerative disorders.
Her work integrates short- and long-read genome sequencing, transcriptomics, bioinformatics and machine-learning approaches, and functional assays in patient-derived cellular models. More recently, her research has focused on the non-coding genome, including intronic repeat expansions and pathogenic variants affecting spliceosomal small nuclear RNAs.
She has contributed to the identification of more than 50 rare neurogenetic conditions and has authored over 230 peer-reviewed publications.
She maintains a long-standing collaboration with the Tom Wahlig Stiftung, having received its prize in 2005 for her work on spastin and a grant in 2018 to study repeat expansions using long-read sequencing, and has co-organised several TWS symposia since 2018.

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Filippo Santorelli
Pisa, Italy

Filippo M. Santorelli graduated with honors in MD from the University Federico II in Italy, where he developed a strong foundation in biomedical sciences and clinical practice, working with patients with inherited ataxias and neuromuscular disorders. His education continued with a residency program at the same University where he honed his skills in patient diagnosis and treatment, focusing on hereditary ataxias, spastic paraplegias, and neurogenetic disorders affecting mitochondrial and lysosome metabolisms. Driven by a thirst for knowledge, he pursued further training at the College of Physicians and Surgeons, the Presbyterian Medical Center, Columbia University, New York (1991-1996), first through a fellowship and then with the position of Associate Researcher in the genetics of neuromuscular disorders and neuropediatric mitochondrial encephalopathies, cementing his expertise and dedication to advancing medical practice. He got a PhD in Neuroscience from Sapienza University in Rome later. Since then, Dr. Santorelli has focused his independent research on the etiology and pathogenesis of neurodegenerative disorders of children and young adults. He also earned an extensive professional portfolio, holding positions in various esteemed institutions in Portugal, Brazil, and Italy, where he teaches courses in Neurology, Pediatrics, and Genetics.

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Programme

Thursday, March 12, 2026

9:00-9:10	Welcome & Introduction	Henry & Susanne Wahlig *TWS*

Session 1: HSP genetics – what´s known?
Chair: Christel Depienne

9:10-9:45	HSPs: Common pathways and pathogenetic mechanisms	Antonio Orlacchio *Perugia, Italy*
9:45-10:05	Gene curation: what about HSP?	Stephan Züchner *Miami, USA*

Session 2: HSP genetics – what´s new?
Chair: Christel Depienne

10:05-10:25	Exploring the genetics of childhood-onset HSP: partial loss of FITM2 function as a novel cause?	Ainara Salazar-Villacorta *London, UK*

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10:25-10:45	Phenotype associated with biallelic SPAST variants	Manon Degoutin *Bordeaux, France*

COFFEE BREAK
10:45-11:10

Session 3: HSP genetics – what´s next?

Chair: Stephan Züchner

11:10-11:50	How new technologies are bridging the gaps in genetics	Alex Hoischen *Nijmegen, Netherlands*
11:50-12:10	Long-read nanopore sequencing: applications in neurogenetics	Ingo Kurth *Aachen, Germany*
12:10-12:30	Long-read sequencing applications in HSP and related disorders	Liedewei Van de Vondel *Miami, USA*

LUNCH & POSTERSESSION
12:30-14:00

Session 4: Functions and disfunctions of HSP genes

Chair: Jürgen Winkler

14:00-14:30	Stress responses in Spg7-linked hereditary spastic paraplegia	Elena Rugarli *Cologne, Germany*
14:30-14:45	Investigating Wshc-5 (Strumpellin) Function in C. elegans: Implications for SPG8	Linda Nemetschke *Halle-Wittenberg, Germany*

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14:45-15:00	Developing an in vitro model of Hereditary Spastic Paraplegia Type 7 in human iPSC-derived i^{3}Neurons	Eleni Theiaspra and Isabelle Hall *Cambridge, UK*

Session 5: Natural history and longitudinal studies

Chair: Jonas Morales Saute

15:00-15:20	Natural history and genotype-phenotype correlations in SPG4	Julian Alecu *Tübingen, Germany*
15:20-15:40	Natural history and genotype-phenotype correlations in SPG7 (PROSPAX)	Emilien Petit *Paris, France*
15:40-16:00	Longitudinalbrain MRI study in HSP	Marcondes Franca Jr. *Campinas, Brazil*

COFFEE Break 16:00-16:30

Session 6: Refining outcome parameters in HSP

Chair: Martin Regensburger

16:30-16:45	Clinical and molecular characterization of SPG7 patients carrying the p.Ala510Val (A510V) variant	Giulia Coarelli *Paris, France*
16:45-17:00	Preliminary videoculographic (VOG) data in patients with genetically confirmed pure versus complicated HSP	Ludmila Novotná *Prague, Czech Republic*

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17:00-17:20	Blood-based biomarkers in HSP: Pitfalls and opportunities	*Darius Ebrahimi-Fakhari* *Boston, USA*
17:20-17:40	Patient priorities: What matters and why?	Mirlinda Ademi *Heidelberg, Germany*
17:40-18:00	Modified Spastic Paraplegia Rating Scale	Jonas Alex Morales Saute *Porto Alegre, Brazil*

19:15
TWS DINNER
At restaurant Wilma Wunder, located in the city centre

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Friday, March 13, 2026

Session 7: Therapies for HSP and related disorders

Chairs: Filippo Santorelli & Stephan Klebe

9:00-9:40	Huntington's disease: challenges and opportunities on the path to better therapies	Ralf Reilmann *Münster, Germany*
9:40-10:00	Treatment strategies for Adrenomyelo-neuropathy	Fanny Mochel *Paris, France*
10:00-10:20	Friedrich ataxia skyclarys	Valeria Gioiosa *Bordeaux, France*
10:20-10:40	Targeted small molecule treatment approaches for the most common genetic ataxia (SCA27B)	Matthis Synofzik *Tübingen, Germany*

COFFEE Break 10:40-11:10

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11:10-11:30	Fingolimod therapy in NCL3 children	Guido Goj *Datteln, Germany*
11:30-11:50	Physical therapy	Tim Rattay *Kiel, Germany*
11:50-12:05	High-throughput phenotyping of fibroblast-derived induced neurons (FiNs) in SPG4-HSP	Klara Metzner *Erlangen, Germany*
12:05-12:20	Personalized ASO therapy for ALS associated with KIF5A mutations around exon 27: a multi-strategy approach	Ingrid Kolen *Heidelberg, Germany*

LUNCH & POSTER SESSION
12:20-13:50

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Session 8: Treatable HSP forms

Chair: Stephan Klebe

13:50-14:10	Potentially treatable hereditary spastic paraplegia linked to COQ9 deficiency	Gaetan Lesca *Lyon, France*
14:10-14:30	4-Hydroxybenzoic Acid Therapy for Mitochondrial COQ2 Deficiency	Felix Distelmaier *Düsseldorf, Germany*

Session 9: Ceremony Award

Chair: Ludger Schöls

14:30-14:45	Naringenin and SMER28 target lysosomal reformation and rescue SPG11and SPG15 hereditary spastic paraplegia phenotypes	Giulia Guarato *Bosisio Parini, Italy* Awardee of Adolf-Struempell-Prize & Gerald Fischer *Hall in Tirol* EURO HSP Federation
14:45-15:15	TWS Poster Prize	Susanne & Henry Wahlig & 3 winners evaluated by participants
15:15-15:30	Conclusions and perspectives	

End of the meeting

Thank you and see you next year!

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Invited speakers in order of appearance

Antonio Orlacchio
Perugia, Italy

Professor Antonio Orlacchio, MD, PhD, is Director of the Medical Genetics Section at the Department of Medicine and Surgery, University of Perugia, Italy, where he teaches Human Genetics. He is an internationally recognized expert in neurogenetics and molecular medicine, with a career dedicated to translating genetic discoveries into clinical advances for patients with neurodegenerative diseases.

Previously, he directed the Laboratory of Neurogenetics at the European Center for Brain Research (IRCCS Santa Lucia Foundation, Rome) and served as Adjunct Professor of Neurology at the University of Rome “Tor Vergata”. He trained in Medicine and Neurology at the University of Perugia, earned his PhD in Neuroscience at the University of Rome “Tor Vergata”, and completed postdoctoral training at the University of Toronto, Canada. He is also a specialist in Medical Genetics.

His research focuses on uncovering the molecular mechanisms of disorders such as hereditary spastic paraplegia, Alzheimer’s disease, amyotrophic lateral sclerosis, and Charcot-Marie-Tooth disease. Professor Orlacchio has received multiple national and international awards and has authored more than 300 scientific publications.

His work has contributed to the identification of disease-causing genes in several neurodegenerative disorders, advancing diagnosis and paving the way for precision medicine approaches.

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Stephan Züchner
Miami, USA

Stephan Züchner, M.D., Ph.D., is a Professor of Human Genetics and Neurology in the role of Chief Genomics Officer at the University of Miami Miller School of Medicine. He received his degrees from the University RWTH Aachen, Germany and an honorary doctoral degree from the Semmelweis Medical School in Budapest. His research interests are focused on identifying strong genetic variation associated with disease. His lab been involved in identifying nearly 100 neuromuscular disease genes, including for spastic paraplegia and related disorders. He also leads the GENESIS genome database for rare neurological diseases, which is widely used in rare disease genomics studies. To further enhance the ability to identify pathogenic variation, his team has recently developed machine learning tools that have successfully supported gene identification. All this is directed towards the genomics-to-therapy concept, whereby progress in genomics will directly, and at times rapidly lead to therapeutic options to be tested in clinical trials. Other relevant roles include his leadership in the ClinGen efforts, the Board of Undiagnosed Disease foundation, and advisory roles to the Muscular Dystrophy Association, the Charcot-Marie-Tooth Association, and the HSP Research Foundation.

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Ainara Salazar-Villacorta
London, UK

Developmental Neurosciences Dept, UCL GOS Institute of Child Health, London, UK
Dept of Paediatrics, Donostia University Hospital, Biogipuzkoa Health Research Institute, Donostia-San Sebastián, Spain

Ainara Salazar-Villacorta is a paediatric neurologist and researcher currently working as a Clinical Research Fellow at the UCL Great Ormond Street Institute of Child Health, London. She is also a PhD candidate at the University of the Basque Country.
She completed her specialisation in Paediatric Neurology at Vall d'Hebron University Hospital, Barcelona. Since then, her clinical and academic career has focused on complex movement disorders and neurogenetics. She completed fellowships in Neurogenetics at UCL Queen Square Institute of Neurology and in Paediatric Movement Disorders at both Vall d'Hebron and Great Ormond Street Hospital.
Her PhD aims to improve molecular diagnosis and expand access to genetic testing for children with neurogenetic disorders, particularly in low- and middle-income countries, through active collaboration with clinical teams in Senegal.
Her work centres on advancing the diagnosis and understanding of complex motor disorders in children. She has contributed to multiple peer-reviewed publications and actively participates in international collaborative research, while continuing clinical practice as a locum paediatric neurologist at Great Ormond Street Hospital, London (UK), and Donostia University Hospital, San Sebastián (Spain).

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Manon Degoutin
Bordeaux, France

Service de Génétique Médicale, CHU Bordeaux (Pellegrin University Hospital); Centre de Référence Maladies Rares “Neurogénétique”, CHU Bordeaux; NRGEN Team, University of Bordeaux, CNRS, INCIA, UMR 5287, Bordeaux, France

Manon Degoutin is resident in Medical Genetics with a specialized focus on neurogenetics. She is currently conducting her Master 2 research at the INCIA laboratory (University of Bordeaux, CNRS) within the NRGEN Team. Her master's project focuses on the application of long-read sequencing technologies for the diagnosis of neurogenetic diseases caused by nucleotides repeat expansions.
In parallel with her research, she is completing her medical thesis dedicated to the diagnosis of Neurodegeneration with Brain Iron Accumulation (NBIA). Her clinical activity at the Bordeaux University Hospital mainly involves the management of patients with rare neurogenetic disorders, contributing to the "Neurogenetics" Rare Diseases Reference Centre.
She described the first published cohort of patients carrying biallelic SPAST variants, detailing a wide clinical continuum that extends from spastic paraplegia to infantile neurodegenerative disorders.

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Alexander Hoischen
Nijmegen, Netherlands

Medicine, Radboud university medical center, Nijmegen, The Netherlands

The research group 'Genomic Technologies for Immune-mediated and Infectious Diseases' (https://www.immuno-genomics.com/) at the Radboud University Medical Center have made significant contribution by pioneering genomic technologies across many rare diseases, with a particular focus ion rare immune diseases. They focused on developing novel genomic approaches for studying rare genetic diseases (RD) and translating these technological innovations into clinical applications, including the early adoption of exome sequencing and subsequent pioneering of long-read genome sequencing and optical genome mapping (OGM). Consequently, their institution is one of the first globally, to use long-read genome sequencing as a first tier generic assay across all rare diseases (launched Oct 2025, aim in 1st year 5,000 clinical long-read genomes). Their current research is focused on immunology and rare immune-mediated and infectious diseases, with a major aim to discover new Inborn Errors of Immunity (IEI). Their work led to the discovery of the X-linked TLR7 deficiency that predisposes to severe COVID-19. Alex is involved in international rare disease programs, most prominently he led a work-package for Solve-RD (H2020), followed-up by an ERDERA work-package lead (2024-2031). He is also supported by a ZonMW VICI grant (NWO, 1.5M€, 5 years, starting Sept-2024), which will strengthen innovative science for immune diseases. He serves as the Chief-Innovation officer of the Department of Human Genetics and Scientific Director of the Radboudumc Genomics Technology Center.

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Ingo Kurth
Aachen, Germany

Center for Human Genetics and Genomic Medicine, Uniklinik RWTH Aachen, Germany

Ingo Kurth is a human geneticist with many years of expertise in neurogenetics. His group is particularly interested in the function of nerve cells with long axons. These are affected in both peripheral neuropathies and HSPs, and there are also many functional connections between these fiber tracts. They are trying to understand these “axonopathies” using modern genomics and increasingly utilizing long-read sequencing technologies.

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Liedewei Van de Vondel
Miami, USA

Dr. Liedewei Van de Vondel specializes in the genetics and molecular mechanisms underlying neuromuscular and neurological disorders, including hereditary spastic paraplegias and spastic ataxias. She completed her PhD at the University of Antwerp under the guidance of Prof. Dr. Jonathan Baets. There, her work focused on genes with notable phenotypical heterogeneity, such as dominant mutations in SPTAN1 as a cause of HSP. Her research projects are carried out in the context of large-scale international consortia, such as the Solve-RD initiative. She is currently a postdoc at the University of Miami under the guidance of Prof. Dr. Stephan Zuchner, where she integrates bio-informatics, long-read sequencing technologies and disease-specific functional studies to uncover genetic origins of rare diseases.

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Elena Rugarli,
Cologne, Germany

Elena Rugarli studied Medicine at the University of Milan, Italy. After a postdoctoral experience at the Baylor College of Medicine in Houston, she worked as researcher at the Telethon Institute for Genetics and Medicine Italy. In 2000 she established her independent group at the Telethon Institute for Genetics and Medicine in Naples, Italy. In 2005 she moved to Milan as Associate Professor of Human Genetics, at the University of Milano-Bicocca. Since 2009, she is Professor of Molecular Biomedicine, at the Institute for Genetics and CECAD, University of Cologne.

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Linda Nemetschke
Halle an der Saale, Germany

Linda Nemetschke studied biology in Tübingen and Bristol, before earning her PhD at the University of Tübingen. During her doctoral and postdoctoral training, she developed a strong interest in the fundamental principles of developmental biology while gaining experience in investigating the molecular mechanisms of disease.
Since 2023, Linda has led the Developmental Biology Group at the Martin-Luther-University Halle-Wittenberg. There she is establishing the nematode Caenorhabditis elegansas a model system to study rare diseases, with a particular focus on Hereditary Spastic Paraplegia. This work brings together her interests in developmental biology and disease mechanisms and has been supported since 2024 by a Research Award from the Tom Wahlig Foundation.

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Eleni Theiaspra
Cambridge, UK

Eleni Theiaspra is a final-year, fully funded PhD researcher at the University of Cambridge, in the departments of Mitochondrial Biology and Clinical Neuroscience. She obtained her BSc in Cell and Molecular Biology from Glasgow Caledonian University, followed by an MSc in Medical Genetics and Genomics from the University of Glasgow, which solidified her interest in the genetic and molecular basis of human disease.
Her PhD research focuses on uncovering the mitochondrial mechanisms underlying Hereditary Spastic Paraplegia Type 7 (SPG7), a genetic, neurodegenerative disease. The main aim of her research is to eventually identify novel therapeutic strategies for this currently incurable disorder.
Using advanced cellular and molecular approaches, her work seeks to bridge fundamental mitochondrial biology with clinically relevant insights.

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Isabelle Hall
Cambridge, UK

Isabelle Hall completed her undergraduate studies in Medical Biosciences at Imperial College London, before going on to do a Master‘s degree in Neuroscience at University College London. In 2023 she began a PhD at the Cambridge Institute for Medical Research, under the supervision of Professor Evan Reid. This project aims to identify a novel therapeutically-targetable patho-mechanism for Hereditary Spastic Paraplegia 7, in collaboration with Eleni Theiaspra at the MBU. She will present on the human iPSC-derived neuronal SPG7 model established for this project.

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Julian Alecu
Tübingen, Germany

Dr. med. Julian E. Alecu, Boston Children's Hospital and Harvard Medical School & University Hospital Tuebingen, German Center for Neurodegenerative Diseases and Hertie-Institute for Clinical Brain Research

Dr. Julian Alecu is a physician-scientist and resident at the Department of Neurology, University Hospital Tübingen, and a postdoctoral research fellow at the German Center for Neurodegenerative Diseases (DZNE) in the laboratories of Dr. Stefan Hauser and Prof. Ludger Schöls. He received his medical degree from Friedrich-Alexander-University Erlangen–Nuremberg and completed his doctoral research in stem cell biology under the supervision of Prof. Beate Winner, with a focus on disease modeling in Parkinson's disease. He subsequently pursued postdoctoral training at Boston Children's Hospital and Harvard Medical School in the laboratory of Prof. Darius Ebrahimi-Fakhari. His research lies at the interface of clinical neurology and experimental neuroscience, leveraging computational approaches and stem cell–based disease models to investigate mechanisms of neuronal vulnerability in hereditary spastic paraplegias and related movement disorders, with the goal of translating these insights into meaningful advances for patients.

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Emilien Petit
Paris, France

Emilien Petit is a biostatistician at the Paris Brain Institute, working within the research group led by Alexandra Durr. His research focuses on the statistical analysis of longitudinal natural history data, with particular expertise in polyglutamine spinocerebellar ataxias (PolyQ SCAs). He has conducted retrospective analyses of pooled datasets from major natural history studies, including EUROSCA, RISCA, CRC, and SPATAX. A central aspect of this work has been the evaluation of the psychometric properties and longitudinal sensitivity to change of the SARA (Scale for the Assessment and Rating of Ataxia), with the aim of optimizing outcome measures for interventional studies. In parallel, he has analyzed prospective longitudinal data from the READISCA study, integrating clinical, neuropsychological, biomarker, and neuroimaging measures, this multimodal approach supporting a more refined characterization of disease progression and variability. The overarching objective of his work is to enhance the design and statistical power of clinical trials by identifying the most informative endpoints and selecting appropriate participant populations. His research has progressively extended beyond PolyQ SCAs to include Huntington disease and, more recently, hereditary spastic paraplegias through the PROSPAX study.

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Marcondes França
Campinas, Brazil

Marcondes França, MD, PhD is full professor and chair of the department of neurology at the university of Campinas (UNICAMP) in Brazil. He trained as a clinical neurologist (2001-2003), did a fellowship in Neuromuscular Disorders (2004-2005) and then PhD/Post-doc in neurogenetics (2006-2010) all at UNICAMP. He leads a research group interested in inherited ataxias and spastic paraplegias. His team has been involved in the genotypic as well phenotypic characterization of Brazilian patients with these conditions, in the investigation of potential biomarkers (especially neuroimaging-based) and in the development of therapies. Such research initiatives are done in collaboration with national and international groups, such as iAXON-HSP, ENIGMA and Treat-HSP.

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Giulia Coarelli
Paris, France

Sorbonne Université, Paris Brain Institute (ICM), AP-HP, Pitié-Salpêtrière University Hospital, Paris, France

Dr. Giulia Coarelli is a neurologist working in the Department of Genetics at Pitié-Salpêtrière Hospital, Paris, and a member of the NeuroGen team “Brain Development and Dysfunction in Neurogenetic Diseases” led by Prof. Alexandra Durr and Dr. Sandrine Humbert at the Paris Brain Institute (ICM).
She specializes in neurogenetics, with a focus on hereditary neurological disorders, particularly cerebellar ataxias, spastic paraplegias, and Huntington disease. She obtained her PhD in Neuroscience from Sorbonne University (Doctoral School ED3C) at the Paris Brain Institute, where her research focused on the identification of biomarkers in spinocerebellar ataxias. Her work combines clinical and translational research aimed at identifying the molecular bases and novel genes involved in inherited cerebellar ataxias and spastic paraplegias, studying genotype–phenotype correlations, characterizing disease natural history, including premanifest stages of polyglutamine spinocerebellar ataxias, and developing biomarkers of disease progression. She is also involved in innovative therapeutic approaches, including antisense therapies, and participates in Phase I–III clinical trials in hereditary neurodegenerative disorders.

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Ludmila Novotná
Prague, Czech Republic

Dr. Ludmila Novotná is a paediatric neurology resident at Motol University Hospital and a postgraduate student in Neurosciences at the Centre for Hereditary Ataxias, Motol University Hospital. Her research focuses on non-motor symptoms in both adult and paediatric patients with hereditary spastic paraplegia (HSP). Her recent work investigates oculomotor impairments in patients with pure and complex forms of hereditary spastic paraplegia.

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Darius Ebrahimi-Fakhari
Boston, USA

Movement Disorders Program, Department of Neurology Boston Children's Hospital, Harvard Medical School, Boston, MA, USA

Darius Ebrahimi-Fakhari is a child neurologist and neuroscientist at Boston Children's Hospital and Harvard Medical School, where he directs the Movement Disorders Program and the Pediatric Movement Disorders and Neurogenetics Fellowship. He is a physician-scientist dedicated to patient-centered translational research, with a focus on advancing clinical trial readiness and developing molecular therapies for rare genetic movement disorders. His work (https://www.def-lab.org/) integrates deep clinical phenotyping with experimental disease modeling and has led to first-in-human therapeutic programs for childhood-onset neurogenetic diseases. Clinically, he cares for children with severe and progressive movement disorders and is a strong advocate for individuals and families affected by rare diseases.

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Mirlinda Ademi
Heidelberg, Germany

Mirlinda Ademi studied medicine in Vienna before moving to Cambridge for an MRC-funded PhD in the laboratory of Prof. Michael Coleman, where she investigated mechanisms of Wallerian degeneration in human disease. Following a short postdoctoral period in the same lab, she began clinical training at the Department of Neurology in Heidelberg in 2023. She is currently a research associate in the group of Prof. Rebecca Schüle at the Division of Neurodegenerative Diseases in Heidelberg. Her research focuses on the development and validation of clinical outcome measures for HSP supporting future clinical trial readiness.

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Jonas Alex Morales Saute
Porto Alegre, Brazil

Jonas Alex Morales Saute, MD, PhD, is a neurologist and medical geneticist, Associate Professor in the Department of Internal Medicine at the Universidade Federal do Rio Grande do Sul (UFRGS), Brazil. He is affiliated with the Neurology and Medical Genetics services at Hospital de Clínicas de Porto Alegre (HCPA), where he coordinates the Neuromuscular Genetics Clinic and leads the Neuromuscular Genetics Research Group (NeMuG).
Dr. Saute has over 15 years of experience in neurogenetic and neuromuscular disorders and has authored or co-authored more than 137 peer-reviewed publications (H-index 29; 2,392 citations, Web of Science). Over the past decade, he has directed the Neuromuscular Genetics Clinic at HCPA, gaining extensive experience with clinical trials, clinical outcome assessments (COAs), digital health technologies (DHTs), and comprehensive genetic and genomic testing in hereditary spastic paraplegias (HSP) and related disorders. He currently coordinates the Brazilian HSP network, iAXON-HSP Brazil, a multicenter initiative across seven sites in four regions of the country, and actively collaborates with the European TreatHSP network with research periods at Heidelberg University, Germany.

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Ralf Reilmann
Münster, Germany

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Dr. Ralf Reilmann is the Founding Director and CEO of the George-Huntington-Institute (www.ghi-muenster.de) in Muenster, Germany, where he and his team of neurologists, researchers and coordinators follow one of the largest groups of Huntington's disease (HD) families globally.
Dr. Reilmann served on several HD related bodies, e.g. the Executive Committees of both the Huntington Study Group and the European Huntington Disease Network.
His main interests is the conduct of clinical trials in HD, several of which he leads as global coordinating principle investigator, including the first gene therapy trials for HD.
He is specifically committed to the advancement of objective endpoints for clinical trials and developed the "Q-Motor" assessment, which is meanwhile widely applied in multiple global clinical trials in HD and beyond.
He holds appointments at the Section for Neurodegenerative diseases at the Department of Neurology & the Hertie Institute for Clinical Brain Research at the University of Tuebingen and at the Institute of Clinical Radiology at the University of Muenster.
He trained at the University of Muenster (Germany) and at Columbia University (New York, USA) and studied as scholar of the "Studienstiftung des Deutschen Volkes" (German Academic Scholarship Foundation) in Muenster, Paris, London, and New York.

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Fanny Mochel
Paris, France

Fanny Mochel is a professor of genetics at Sorbonne University. She received her MD in Genetics in 2005 at the University Paris Descartes, her PhD in Neuroscience in 2010 at Sorbonne University and is board certified in inherited metabolic disorders. Prof. Mochel leads a national reference center for neurometabolic diseases in adults as well as a national reference center for leukodystrophies in adults. She is also the co-team leader of the MIND team ("Metabolism, Immunity and NeuroDegeneration") at the Paris Brain Institute of La Pitié-Salpêtrière University Hospital in Paris. From 2018 to 2024, she served as chair of the Adult Metabolic Physicians group of the Society for the Study of Inborn Errors of Metabolism (SSIEM) and she is co-chair of the French society for inborn of errors of metabolism in adults since 2014. Her research has focused on the characterization and treatment of brain energy deficiencies in neurometabolic and neurodegenerative diseases. Her major areas of expertise are the identification of neurometabolic biomarkers in vitro (metabolomics) and in vivo (metabolic imaging) as well as therapeutic approaches targeting the Krebs cycle. Recently, she has developed a new area of research, together with Angela Garcia-Cazorla, on the connections between physics and metabolism in brain functions and diseases.

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Valeria Gioiosa
Bordeaux, France

Affiliations: Department of Medico-Surgical Sciences and Biotechnologies, Sapienza University of Rome, Italy; and Service de Génétique Médicale and Rare Diseases Reference Centre "Neurogenetics", CHU Bordeaux; NRGEN Team, University of Bordeaux, CNRS, INCIA, UMR 5287, Bordeaux, France.

Valeria Gioiosa is a neurologist with experience in diagnosis and management of rare neurological diseases. After training at the Neurogenetics outpatient clinic of Sapienza University of Rome in Latina, she is now working as a contract hospital practitioner at the Neurogenetics Department at Pellegrin University Hospital (CHU Bordeaux). Since February 2024, her clinical activity has been focused on patients with rare neurogenetic disorders, in particular on treatment and follow-up of patients with Friedreich's ataxia with omaveloxolone.

Currently she is a PhD student in Neurosciences in joint supervision between the University of Rome La Sapienza and the University of Bordeaux. Her clinical work mainly involves patients with hereditary ataxias, hereditary spastic paraplegias, Huntington's disease, and other neurogenetic conditions. Her translational research is conducted at the INCIA laboratory (University of Bordeaux, CNRS), where she is currently carrying out her PhD research, with a focus on neurodegenerative diseases, particularly hereditary ataxias and neurodegeneration with brain iron accumulation (NBIA), integrating clinical, genetic, and functional data. She is a member of the European Academy of Neurology and the Italian Society of Neurology.

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Matthis Synofzik
Tübingen, Germany

Professor Matthis Synofzik is a clinician scientist focussing on unraveling the molecular basis of neurodegenerative diseases - particularly hereditary ataxia-spasticity-spectrum diseases , motor neuron diseases, and dementias - while identifying translational biomarkers and developing individualized mechanistic treatments. He has extensive expertise in next-generation genomics, deep phenotyping, fluid and digital biomarkers, and translational trial-readiness research. He has authored more than >550 PubMed-indexed peer-reviewed publications, including in leading journals such as New England Journal of Medicine, Brain, Nature Genetics, Neuron, Annals of Neurology, and Neurology.
As PI or co-PI of multiple international consortia, including Ataxia Global Initiative, he has advanced genomic stratification and biomarker validation across hereditary ataxia-spasticity-spectrum diseases, ALS and FTD, His work has contributed to the identification of more than 30 novel ataxia-spasticity-spectrum disease genes and mutational mechanisms, including GAA-FGF14 (SCA27B). He pioneers the development and clinical and regulatory implementation of ultra-individualized n-of-1 antisense oligonucleotide (ASO) therapies in Europe, following a platform approach of ASO development (now already covering >18 ASO programs) that might be applicable and scalable to many rare neurological disease mutations alike, here also serving as part of the steering committee of "1 Mutation 1 Medicine" (1M1M) Consortium.

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Guido Goj
Datteln, Germany

Dr. Guido Goj is a consultant in the department of pediatric neurology at the University of Witten-Herdecke, Datteln, Germany. He has high expertise in the diagnosis and treatment of children with developmental disorders. The main focus of his work is the treatment of multiple sclerosis and other autoimmune diseases of the central nervous system in the department of Prof. Rostasy. He has done research work on the neuroimaging features in children with acute flaccid myelitis. He leads the CLN3 study group in which the influence of the immunmodulatory drug Fingolimod on brain volumes and disease outcome in CLN3 patients is investigated.

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Tim Rattay
Kiel, Germany

Tim W. Rattay is the head of the Center for Rare Neurologic Diseases at the University Medical Center Schleswig-Holstein campus Kiel. He completed his HSP-specific clinical and scientific training in Tübingen (Ludger Schöls and Rebecca Schüle, now based in Heidelberg). His research focuses on deep phenotyping ("precise symptomatic characterization of different genetic HSP etiologies"), MRI-based imaging in HSP (in close collaboration with Tobias Lindig), non-motor symptoms in HSP, and, more recently, the prodromal phase of the disease, using SPG4 as a model.
He completed his habilitation on biomarkers in HSP in November 2022 in Tübingen and has been based in Kiel since January 2024. As a senior attending physician in the Department of Neurology, he has been providing specialized care for HSP patients in his HSP outpatient clinic since 2025. In addition, he has expanded his longstanding involvement in the treatHSP network to his current institution in Kiel.

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Klara Metzner
Erlangen, Germany

Klara Metzner is a doctoral researcher at University Hospital Erlangen specializing in neuroscience with a focus on neurodegeneration. She obtained her Bachelor's degree in biochemistry / molecular biology followed by a Master's degree in Molecular Medicine, both at Friedrich Schiller University Jena. She then moved to Canada for the competitive Master's program in Molecular and Cellular Medicine at Université de Montréal in cooperation with the Montréal Clinical Research Institute (IRCM), where she developed a strong foundation in cell biology and neuroscience. She is currently pursuing a PhD in neuroscience at the Department of Stem Cell Biology at University Hospital Erlangen. Her research focuses on the molecular mechanisms underlying neurodegenerative disorders, particularly amyotrophic lateral sclerosis (ALS) and hereditary spastic paraplegia (HSP).

Ingrid Kolen
Heidelberg, Germany

Ingrid Kolen is a PhD student in the group of Prof. Dr. Rebecca Schüle at the University Hospital in Heidelberg. She studied Biomedical Engineering at the Technical University of Eindhoven in the Netherlands, focusing on protein engineering. During her studies, she worked on developing a multiplex RNA detection assay using CRISPR Cas9-based luciferase complementation. Currently, her research is focused on developing antisense oligonucleotides for ALS-associated KIF5A mutations. She is part of the Medicine Made to Measure Marie-Curie doctoral Network, which aims to develop single-patient-tailored antisense oligonucleotide treatments for patients with nano-rare disease mutations.

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Gaëtan Lesca
Lyon, France

Department of Medical Genetics, University Hospitals of Lyon, Lyon, France

Gaëtan Lesca is a medical geneticist involved in molecular diagnosis and genetic research specializing in neurogenetic disorders, including epilepsies hereditary spastic paraplegias. In his diagnostic practice, Lesca applies comprehensive genetic testing and interpretation to identify causative mutations in patients with spastic paraplegia, helping to distinguish between the many genetic subtypes and guide clinical management. His recent expertise is based on the integration of genome sequencing approaches to improve diagnostic yields and refine genotype–phenotype correlations. In research, Lesca has contributed to expanding the understanding of genes implicated in spastic paraplegia. He was a co-author on a landmark publication reporting that pathogenic mutations in ALS2 are responsible for early-onset hereditary spastic paralysis, linking dysfunction of the ALS2 gene to degeneration of upper motor neurons. More recently, Gaëtan Lesca contributed to a study identifying mutations in COQ9 as a novel cause of hereditary spastic paraplegia, which not only expands the known genetic causes of the disease but also suggests potential therapeutic strategies involving Coenzyme Q10 supplementation

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Felix Distelmaier
Düsseldorf, Germany

Felix Distelmaier is a pediatric neurologist with more than 20 years of clinical experience and specializes in the diagnosis and treatment of neurometabolic diseases (especially mitochondrial disorders). He is division head of the pediatric-neurology department at the University Children's Hospital in Düsseldorf (Germany). He has a strong research focus on the identification and characterization of novel genetic diseases. During the last years, he was part of numerous international collaboration studies and was involved the first description of 26 novel neurometabolic diseases (for example Long-Olsen-Distelmaier syndrome, OMIM # 620609). In some of these disorders, he successfully facilitated novel treatment concepts (e.g. CAD-deficiency, TXN2-deficiency, NAXE deficiency). For the last 8 years, he conducted a DFG-funded research project that was dedicated to inherited disorders caused by impaired coenzyme Q10 biosynthesis. A main goal of his research work was the identification of therapeutic substances that can reactivate the endogenous CoQ10 production. His cell-based work identified 4-hydroxybenzoic acid (4-HBA) as a promising candidate for human COQ2 deficiency. He conducted the first compassionate-use 4-HBA treatments in children with COQ2 deficiency.

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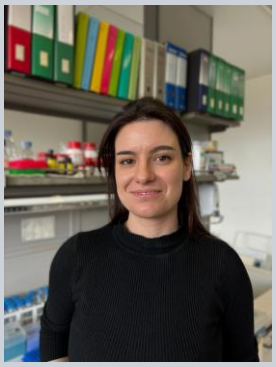
Giulia Guarato
Bosisio Parini, Italy



IRCCS Eugenio Medea

Giulia Guarato holds a degree in Pharmaceutical Chemistry and Technology from the University of Padova and obtained her PhD in Pharmacological Sciences in 2025. She is currently a postdoctoral researcher at the IRCCS Eugenio Medea Institute and specializes in using Drosophila melanogaster models to investigate disease mechanisms and identify candidate compounds for therapeutic intervention in HSP. Her work centres on autophagy and lysosomal dynamics and function, particularly the pharmacological restoration of impaired lysosomal pathways.
By integrating genetic, cellular, and pharmacological approaches, her research advances understanding of disease biology and supports the development of targeted therapeutic strategies, while establishing scalable platforms for rapid drug screening and mechanistic discovery, accelerating therapeutic development for rare diseases.

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We are delighted that poster presentations have become an integral part of our symposium in recent years.

Please feel free to use your lunch breaks to listen to the presentations and then vote for your three favorite posters using the QR codes which you find in the room.

Based on this evaluation, we will award three poster prizes at the end of the meeting.

Two of the posters presented are not participating in the evaluation.

Further information on these topics can be found at the following link, where you can download the abstracts:

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