



**MARCH
12-14, 2025**

Bürgerhaus
Bahnstadt •
Gadamer Platz 1
• 69115
Heidelberg •
Germany

Hosted by:



CONSENSUS MEETING Demonstrating Meaningful Change in HSPs and Ataxias

ANNUAL MEETING TreatHSP and the Tom Wahlig Foundation

Wednesday, March 12, 2025

12:00 – 13:00	Registration & Light Lunch	
13:00 – 14:40	Demonstrating Meaningful Change – State of the Art	
13:00 – 13:10	Welcome and introduction to the programme	Rebecca Schüle & Matthis Synofzik
13:10 – 13:30	Clinical outcome assessments in HSP – current status	Rebecca Schüle
13:30 – 13:50	Clinical outcome assessments in ataxia – current status	Matthis Synofzik
13:50 – 14:30	Preparing the stage: key points from the FDA PFDD guidance on meaningful endpoints	Michele Potashman, Biohaven
14:30 – 15:00	Coffee Break	
15:00 – 17:00	Demonstrating Meaningful Change – Current Use Cases	
15:00 – 15:30	Clinical meaningfulness of stride velocity 95th percentile in Duchenne Muscular Dystrophy: pitfalls and learnings from the EMA qualification	Damien Eggenpieler, Vernon
15:30 – 15:55	Anchoring measures for ataxia COAs: experiences with the SARA, PGI, PROM-Ataxia and FARS-ADL	Andreas Träschütz, Tübingen
15:55 – 16:20	Application of the PFDD to the validation of the f-SARA and FARS-ADL	Michele Potashman
16:20 – 16:40	Anchoring the Upright Stability Score (FARS-E) in patient-meaningful milestones: methodology and FDA guidance compatibility	Christian Rummey, Clinical Data Science
16:40 – 17:00	Anchoring change in digital-motor outcomes in patient voice: results on successful anchoring approaches and variables from MOBILISE-D	Clemens Becker, Heidelberg
17:00 – 17:30	Break	



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17:30 – 18:30	Demonstrating Meaningful Change – Round Table Discussion	
17:30 – 18:00	Learnings from the use cases: anchor variables and anchoring methods – strengths and limitations for the ATX/HSP field	Panel
18:00 – 18:30	Prioritized candidate anchor variables and anchoring methods – action points towards consensus / implementation for the ATX/HSP field	Panel
19:30	Dinner: Da Baggio, Römerstr. 24-26, 69115 Heidelberg	Scan me 

Thursday, March 13, 2025

07:00 – 08:00	Registration	
08:00 – 09:25	Meaningful Change – Specifications and Consensus for HSPs	
08:00 – 08:10	Consensus from day 1	Rebecca Schüle, Matthis Synofzik
08:10 – 08:35	Outcome validation study – master protocol for adult patients	Rebecca Schüle, all
08:35 – 09:00	Outcome validation study – master protocol for pediatric patients	Darius Ebrahimi-Fakhari, Boston, all
09:00 – 09:25	HSP consensus paper series - strategies and responsibilities	Rebecca Schüle, all
09:25 – 10:40	Meaningful Change – Specifications and Consensus for Ataxias	
09:25 – 09:50	Specification and consensus on best ataxia anchors and anchoring methods	Matthis Synofzik, all
09:50 – 10:15	Application paths of best anchors & anchoring methods for outcome domains / AGI working groups: ClinRO, DMO, imaging outcomes, molecular outcomes	Matthis Synofzik, all
10:15 – 10:40	Writing strategy for AGI consensus paper: establishing meaningful change in ataxia outcomes	Matthis Synofzik, all
10:40 – 10:50	Group photo (in front of the building)	
10:50 – 11:20	Coffee Break	



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11:20 – 12:30	News from TreatHSP	
11:20 – 11:30	Personalized genomic therapies for Hereditary Spastic Paraplegias	Rebecca Schüle
11:30 – 11:40	Mobile gait outcomes in HSP – a window for digital biomarkers	Heiko Gassner, Erlangen
11:40 – 11:50	Investigating axon-specific phenotypes in spastin-deficient iPSC-derived neurons	Stefan Hauser, Tübingen
11:50 – 12:00	Pathophysiology of REEP1 associated axonopathies	Christian Hübner, Jena (<i>remote</i>)
12:00 – 12:10	Human stem cell based disease modeling in HSP and beyond	Beate Winner, Erlangen
12:10 – 12:20	Understanding the function of the ERLIN1/2-RNF170 complex and its role in HSP	Elena Rugarli, Cologne
12:20 – 12:30	Tracking and targeting maladaptive immune reactions in SPG11	Janos Groh, Munich
11:20 – 12:30	Internal Meeting: SCA27B Natural History Study Group	
11:20 – 12:30	Multi-center ataxia outcome validation study for trial-readiness of SCA27B	Matthis Synofzik, NH study group
12:30 – 13:30	Lunch Break	
13:30 – 15:40	Surrogate Endpoints in HSP and Related Diseases	
13:30 – 14:00	Surrogate endpoints in Charcot-Marie-Tooth Disease	Mary Reilly, London
14:00 – 14:20	Acetylated alpha-tubulin: a biomarker candidate for SPG4	Gautam Wali, Sydney
14:20 – 14:40	Molecular biomarkers in SPG11: from neurodegeneration to neuroinflammation	Martin Regensburger, Erlangen
14:40 – 15:00	Spinal cord imaging in HSPs: current state and perspectives	Marcondes Franca, Campinas (<i>remote</i>)
15:00 – 15:20	Brain imaging in HSPs: current state and perspectives	Tobias Lindig, Tübingen (<i>remote</i>)
15:20 – 15:40	Peripheral nerve imaging in HSPs: current state and perspectives	Heike Jacobi, Heidelberg Jennifer Hayes, Ann Arbor (<i>remote</i>)
15:40 – 16:10	Coffee Break	

End of ERDERA programme track



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Thursday, March 13, 2025 (ct'd)

16:10 – 17:10 Poster Flash Talks		
16:10 – 16:17	Defining the distinct clinical and molecular spectra of <i>de novo</i> and inherited <i>SPAST</i> -related HSP (SPG4)	Julian E. Alecu, Boston
16:17 – 16:24	Short-term fluctuations of ataxia severity in SCA3: a multicentre, cross-sectional study	Thomas Klockgether, Bonn
16:24 – 16:31	Genetic and phenotypic complexity in early-onset HSP	Natalia Espasandin-Hueter, Barcelona
16:31 – 16:38	Novel complex spastic paraplegia and neurodevelopmental disorder caused by variants in the ribosomal protein S6 kinase C1 (RPS6KC1)	Aurora Pujol, Barcelona
16:38 – 16:45	Diagnostic yield of short-read whole genome sequencing in a cohort of children with progressive movement disorders	Luca Schierbaum, Boston
16:45 – 16:52	Longitudinal study to correlate genetic variants, cellular imaging data and clinical outcome in HSP-SPG4	Francesca Sardina, Rome
16:52 – 16:59	High-throughput phenotyping of fibroblast-derived induced neurons in SPG4-HSP	Klara Metzner, Erlangen
16:59 – 17:06	Establishing a patient-derived <i>in vitro</i> model for SPG11 and SPG15 hereditary spastic paraplegia for drug testing	Tim Böstler, Erlangen
17:10 – 18:10 Poster Session		
18:20	Leave for the joint walk to the dinner location	Guided by Sabrina Stängle
19:00	Dinner: Kulturbrauerei Heidelberg, Leyergasse 6, 69117 Heidelberg	Scan me 



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INITIATIVE
worldwide platform for clinical research in ataxias

E:DERA
European Rare Diseases
Research Alliance

 Co-funded by
the European Union

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 **TWS**
TOM WAHLIG
STIFTUNG

Friday, March 14, 2025

09:00 – 10:45 Pathways and Therapeutic Targets

09:00 – 09:25	Modulation of PI4K2A activity improves autophagy in SPG11	Mukhran Khundadze, Berlin
09:25 – 09:50	Emerging molecular-mechanism based therapies for AP-4-associated hereditary spastic paraplegia	Darius Ebrahimi-Fakhari
09:50 – 10:15	Cul-4 inhibition rescues spastin levels and reduces defects in hereditary spastic paraplegia models	Cinzia Rinaldo, Rome
10:15 – 10:45	Genetics of HSP: genes, mechanisms and challenges	Stephan Züchner, Miami

10:45 – 11:15 Coffee Break

11:15 – 13:15 Gene and RNA Therapies in HSP

11:15 – 11:45	Modelling an AAV9 gene therapy for SPG56 (<i>CYP2U1</i>) in patient-specific brain organoids	Ernst Wolvetang, Brisbane
11:45 – 11:55	TBC	TBC
11:55 – 12:20	Gene replacement therapy for SPG47 (<i>AP4B1</i>) – current status of the preclinical development	Mimoun Azzouz, Sheffield
12:20 – 12:50	Antisense oligonucleotide therapy in an individual with <i>KIF1A</i> -associated neurological disorder	Wendy Chung, Boston (<i>remote</i>)
12:50 – 13:15	Small molecule FICD inhibitors suppress endogenous and pathologic FICD-mediated protein AMPylation – implications for HSP	Matthias C. Truttmann, Ann Arbor (<i>remote</i>)

13:15 – 14:00 Lunch Break



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14:00 – 15:30 Gene and RNA Therapies in HSP (ct'd)

14:00 – 14:30	AAV gene therapy for SPG50 (AP4M1): a phase 1 trial in a single patient	James J. Dowling, Toronto (<i>remote</i>)
14:30 – 14:50	Journey to cure Michael	Terry Pirovolakis, Elpida Therapeutics (<i>remote</i>)
14:50 – 15:00	Rewriting the future for childhood-onset SPG4: the Lilly and Blair Foundation's mission to turn hope into reality	Katie Gregg, Lilly and Blair Foundation (<i>remote</i>)
15:00 – 15:20	Ethical considerations and challenges pursuing individualized genomic interventions as rare as N-of-1	Lynn Bush, Boston (<i>remote</i>)

15:20 – 16:00 Award Ceremonies and Farewell

15:20 – 15:30	Awarding of the Adolf Struempell Prize	Gerald Fischer, Euro HSP
15:30 – 15:40	Adolf Struempell Prize Lecture: Mobile digital gait analysis captures effects of botulinum toxin in hereditary spastic paraplegia	Alzhraa Ibrahim, Erlangen
15:40 – 15:50	Awarding of the Tom Wahlig Poster Prizes	Henry and Susanne Wahlig, Bochum
15:50 – 16:00	Farewell and closing remarks	Henry and Susanne Wahlig, Rebecca Schüle

End of Meeting