

TreatHSP/TWS Joint Symposium Wed November 30 – Fri December 2, 2022

Location:

Innovation Center ‚Westspitze‘, Eisenbahnstraße 1, 72072 Tübingen, Germany (<https://www.westspitze.com>)

Wednesday, November 30, 2022

Workshops of the TreatHSP consortium (12:00 – 18:00)

Thursday, December 1st, 2022

09:15 – 10:30	Session 1 – HSP around the world (chair: Schüle)
09:15	Landscapes of HSP in Japan and Japan Spastic Paraplegia Research Consortium (JASPAC) (Horoyuki Ishiura, Okayama, Japan) (25+5 min)
09:45	Landscapes of HSP in Africa (Jeannine M. Heckmann, Cape Town, South Africa) (25+5 min)
10:15	Landscapes of HSP in South America (Jonas Alexander Morales Saute, Porto Alegre, Brazil) (20+5 min)
10:45 – 11:00	Coffee Break (15 min)
11:00 – 12:30	Session 2 – Diagnosing and studying the natural history of HSP (chair: Schüle)
11:00	Development of Centers of Excellence to support a North American Registry and Natural History Study for HSP (Caig Blackstone, Darius Ebrahimi-Fakhari, Rebecca Schüle, Stephan Züchner) (25+5 min)
11:30	The clinical and molecular spectrum of ZFYVE26-associated HSP (SPG15) (Darius Ebrahimi-Fakhari, Boston, USA) (25+5 min)
12:00	Improving the diagnostic odyssey in Hereditary Spastic Paraplegia (Giovanni Stevanin, Bordeaux, France) (25+5 min)
12:30 – 14:15	Lunch Break and Posters
14:15 – 15:45	Session 3 – New HSP genes (chair: Depienne)
14:15	Dominant KPNA3 Mutations Cause Infantile-Onset Hereditary Spastic Paraplegia (Stefan Kindler, Hamburg, Germany) (25+5 min)
14:45	SPTAN1 mutations in spastic ataxia: new phenotypes expanding the neuro-spectrinopathies (Jonathan Baets, Antwerp, Belgium) (25+5 min)
15:15	Biallelic Variants in the Ectonucleotidase ENTPD1 Cause a Complex Neurodevelopmental Disorder with Intellectual Disability, Distinct White Matter Abnormalities, and Spastic Paraplegia (Daniel Clame, Houston, USA) (25+5 min)
15:45 – 16:00	Coffee Break (15 min)
16:00 – 17:30	Session 4 – New HSP genes, clinical overlap with other syndromes, genetic modifiers (chair: Depienne)
16:00	Closing the diagnostic gap: new genes in HSP and related disorders (Stephan Züchner, Miami, USA) (25+5 min)

- 16:30 Genetic modifiers of SPG4-HSP (Livia Parodi, Paris, France and Harvard, USA) (25+5 min)
- 17:00 Recognizing and exploiting the overlap between hereditary spastic paraplegia and ataxias (Bart van de Warrenburg, Nijmegen, Netherlands) (25+5 min)

19:30 Get Together and Dinner
 Restaurant Liquid Kelter, Schmiedtorstraße 17, 72070 Tübingen |
<https://www.liquid-kelter.de>
 Google Maps Location: <https://www.liquid-kelter.de>

Friday, December 2nd, 2022

09:00 – 10:30 **Session 5 – Deciphering mechanisms causing HSPs using cellular and animal models (chair: Klebe)**

- 09:00 (iPSC-based) advanced cell culture models to decipher disease mechanisms in HSP (Stefan Hauser, Tübingen, Germany) (25+5 min)
- 09:30 New mechanisms for motor neuron disease derived from human stem cell based models (Beate Winner, Erlangen, Germany) (25+5 min)
- 10:00 Mouse models for hereditary spastic paraplegia uncover a role of PI4K2A in autophagic lysosome reformation (Christian Hübner, Jena, Germany) (25+5 min)

10:30 – 11:00 **Coffee Break (15 min)**

11:00 – 12:30 **Session 6 – Monitoring HSP progression in clinical studies (chair: Klebe)**

- 11:00 5D gait analysis – perspectives for innovative digital precision medicine in HSP (Heiko Gassner, Erlangen, Germany) (25+5 min)
- 11:30 Iron-sensitive MR imaging of the primary motor cortex in Hereditary Spastic Paraplegia (Graziella Donatelli, Pisa, Italy) (25+5 min)
- 12:00 Gait Changes in Prodromal Hereditary Spastic Paraplegia Type 4 (Ludger Schöls, Tübingen, Germany) (25+5 min)

12:00 – 13:30 **Lunch Break and Posters**

13:30 – 14:30 **Session 7 – Awards Ceremony**

- 13:30 Investigating the role of spastin's binding partner protrudin in endosomal sorting (Julia Kleniuk, Cambridge, UK) (25+5 min)
- 13:45 Using Label-free Proteomics for Target Identification and Validation of Novel Treatments for AP-4-associated Hereditary Spastic Paraplegia (Afshin Saffari, Boston, USA) (25+5 min)
- 14:00 Measures of mobility and functioning in hereditary spastic paraplegia (Martin Regensburger, Erlangen, Germany) (25+5 min)
- 14:15 Nomination of 3 poster prizes

14:30 – 16:00 **Session 8 – Precision medicine approaches to treat HSP (chair: Schöls)**

- 14:30 Understanding the biology and pathology of upper motor neurons: one neuron at a time (Hande Ozdinler, Chicago, USA) (25+5 min)
- 15:00 Testing Miglustat in SPG11: a single center experience (Filippo Santorelli, Pisa, Italy) (25+5 min)
- 15:30 Development of an Intrathecal AAV9/AP4M1 Gene Therapy for Hereditary Spastic Paraplegia 50 (Xin Chen, Dallas, USA) (25+5 min)
- 16:00 End of the event