

## 2020 International SSADHD Conference – Boston, MA July 9-10, 2020

### Conference Agenda

#### Day 1 – Newborn Screening, Clinical Studies, Epilepsy

- 7:00 AM – 8:00 AM: Registration/Breakfast:
- 8:00 AM – 8:15 AM: Introduction/Welcome by Conference Organizing Committee members (Dr. Pearl, Dr. DiBacco, Dr. Roulet)
- 8:15-8:45 AM: Keynote Speaker: The Global Impact of SSADHD on Intermediary Metabolism – Mike Gibson, WSU College of Pharmacy, Spokane WA USA
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#### Morning – Newborn Screening, Clinical Studies-I

Session 1: Newborn screening (*Moderator*: Dr. J.-B. Roulet, Washington State Univ, USA) 8:45 AM – 9:45 AM

1. Metabolomic Paradigms for Newborn Screening of SSADHD – Dr. Madalyn Brown, WSU College of Pharmacy, Spokane, WA
2. Refining Prevalence/Incidence Estimates of SSADHD – Dr. Sarah Elsea, Baylor College of Medicine, Houston, TX, USA

*15-min Break*

Session 2: SSADHD Clinical Studies – I (*Moderator*: Dr. Philip Pearl) 10:00 AM – 12:00 PM

3. Outcomes of the SGS-742 Clinical Trial and Next Steps - Dr. William Theodore, NINDS, NIH
4. SSADHD Natural History Study – *Half-Way Point* - Dr. Phillip Pearl, BCH Neurology, USA
5. Clinical symptom prevalence/Longitudinal REDCap survey - Dr. Melissa DiBacco, BCH, USA
6. Research Opportunities in Rare Neurological Diseases – Dr. Jill Morris, NINDS OR Dr. XX, Program Director/Imaging/Phil's contact

*12:00 PM – 1:15 PM Lunch*

#### Afternoon – Clinical Studies-II/Imaging, Epilepsy

Session 3: SSADHD Clinical Studies – II (*Moderator*: Dr. Thomas Opladen, Germany) 1:15 PM – 2:45 PM

7. Imaging Component – Simon Warfield PhD, Onur Afacan PhD, BCH Radiology
8. TMS Evidence of Abnormal Innervation in SSADHD – Alex Rotenberg MD, PhD, BCH Neurology
9. International Neurotransmitter Disease (iNTD) Registry- Dr. Kathrin Jeltsch, University Children's Hospital, Heidelberg, Germany

*15-min Break*

Session 4: Targeted Therapeutics-I (*Moderator*: Dr. Mike Gibson, Washington State Univ) 3:00 PM – 4:30 PM

10. Enzyme Replacement Therapy for SSADHD – Dr. Henry Lee, Boston Children's Hospital.
11. Dysregulation of Phototransduction Pathway by Vigabatrin – Dana Walters, WSU, Spokane, USA
12. Development of an InVitro Model of SSADHD Using Patient iPSC-Derived Neurons to Support Unbiased Screening of Novel Therapeutic Approaches to Treatment – Dr. Mustafa Sahin, Boston Children's Hospital

Q/A Session for families: SSADHD Panel of Experts (Pearl, DiBacco, Gibson, Jeltsch, Opladen, Hoffman, Fliesler, Roulet) 4:30 PM – 5:30 PM

*[Poster Session: 5:00 PM – 6:00 PM]*

## Day 2

- 7:00 AM – 8:00 AM: Breakfast
- 8:15 AM - 8:45 AM Keynote speaker: GABAergic Status in Diverse Neurometabolic Disorders. Dr. Angeles Garcia and/or Dr. Alfonso Oyarzábal, Barcelona, Spain

### Morning – Targeted therapeutics-II

#### Session 1: GHB/GABA receptors (*Moderator*: Dr. Senthil Natesan) 8:45 AM - 10:15 AM

1. High affinity  $\gamma$ -hydroxybutyrate (GHB) receptor ligands for SSADHD – Justin Chen, WSU, USA
2. Patient-customized therapy for rare genetic diseases – Tim Yu, MD, PhD, BCH Genomics
3. Targeting the GABA<sub>A</sub> receptor with farnesol as an antiepileptic agent”- Christopher Szlenk, WSU, USA

15-min Break (coffee)

#### Session 2: ALDH5A1 (*Moderator*: Dr. Gajja Salomons) 10:30 AM – 12:00 PM

4. Chaperone Therapy for SSADHD- Dr. Ritva Tikkanen, Justus-Liebig Universität Gießen, Germany
5. Mutations in the ALDH5A1/Gene Characterized with Overexpression Analyses”- Dr. Ana Pop/Dr. Gajja Salomons, Amsterdam, Netherlands
6. In silico ALDH5A1 mutation analysis – Senthil Natesan, PhD, WSU, USA

12:00 PM – 1:15 PM Lunch

### Afternoon – Patients, Caregivers

#### Session 3: Patient Contributions to Research (*Moderator*: Claudio Cinquemani) 1:15 PM – 2:45 PM

7. Integration of Clinical and Basic Research for SSADHD: Traversing International Borders. Dr. Claudio Cinquemani, Cologne, Germany
8. Engaging Patients and Families in Research: The Power of Social Media, Registries & Fund Raising – Alice McConnell/Carolyn Hoffman
9. Patient Reported Outcome Measures: Why We Need It for SSADHD – Mousumi Bose, PhD Montclair University, USA

15-min Break

#### Session 4: Living with SSADHD (*Moderator*: Alice McConnell/ Francisco José Peñarrubia) 3:00 PM – 4:30 PM

10. Sleep disorders in SSADHD – Dr. Kiran Maski, Boston Children’s Hospital, Boston, USA
11. Obsessive Compulsive Disorders: What it looks like and how to manage it – Dr. Joseph Gonzalez-Heydrich, Boston Children’s Hospital, Boston, USA
12. Understanding SUDEP for SSADHD patients – Dr. Phillip Pearl, BCH Neurology, USA

Q/A Session for families: SSADHD Panel of Experts (Pearl, DiBacco, García Cazorla, Jeltsch, Opladen, Hoffman) 4:30 PM – 5:30 PM

**Meeting adjourned/ SSADH Association Medical Advisory Board meeting**