

NTSAD Science Symposium & Workshop For Researchers & Healthcare Professionals

Hyatt Regency – Reston, VA
April 16, 2015

AGENDA

Co-Chairs

Fran Platt, PhD, Professor of Biochemistry and Pharmacology, University of Oxford, UK

Cynthia Tift, MD, PhD, Director of Pediatrics for the NIH Undiagnosed Diseases Program

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| 8:00-8:45 | Arrival, coffee and registration |
| 8:45-9:00 | Welcoming remarks
<i>Shari Ungerleider, NTSAD Board President</i> |
| 9:00-9:45 | Common Elements of the Pathogenesis of Neurodegenerative diseases
<i>Fran Platt, PhD, University of Oxford</i> |
| 9:45-10:00 | Questions & Discussion |
| 10:00-10:45 | Clinical trial design and outcome measures for diseases affecting the CNS: FDA perspective
<i>Yao-Yao Zhu, MD, PhD, Center for Biologic Evaluation and Research, FDA</i> |
| 10:45-11:00 | Questions |
| 11:00-11:30 | Coffee and Informal Networking Discussions |
| 11:30-11:50 | Specific issues relating to the gangliosidoses
<i>Alessandra d'Azzo, PhD, St. Jude Children's Research Hospital</i> |
| 11:50-12:00 | Questions |
| 12:00-12:20 | Specific issues in the leukodystrophies
<i>Adeline L. Vanderver, MD, Children's National Medical Center</i> |
| 12:20-12:30 | Questions |

12:30-1:30 Lunch

1:30 – 2:30 Workshops – first session (choose 1)

❖ Small molecule approaches that could be disease-modifying for our diseases. Are there opportunities in repurposing therapies? What is the path to translation?
Gustavo Maegawa, MD, PhD, Johns Hopkins University School of Medicine
Steven U. Walkley, DVM, Ph.D, Albert Einstein College of Medicine

❖ Gene therapy: current status and future – we will touch on different approaches e.g., AAV, lentiviral gene therapy, and use in different diseases,
Miguel Sena-Esteves, PhD, University of Massachusetts Medical Center
Douglas McCarty, PhD, Nationwide Children’s Hospital

2:30 – 2:45 Break

2:45 – 3:45 Workshops – second session (Choose 1)

❖ Newborn screening: What is the role of NBS in developing therapies for pediatric neurodegenerative diseases?
Joan Keutzer, PhD, Genzyme, a Sanofi Company
Michael S. Watson, PhD, FACMG American College of Medical Genetics

❖ NTSAD’s diseases: What are the outstanding issues in clinical trial readiness - natural history studies, registries, clinical trial design and proposed outcome measures, biomarkers
Florian Eichler, MD, Massachusetts General Hospital
Cynthia Tift, MD, PhD, National Institutes of Health

3:45 – 4:00 Break

4:00 – 5:00 Panel discussion for workshop leaders and attendees: Identify critical issues that people have raised during the day. What are the gaps that have been identified?

5:00 – 6:00 Reception with families attending conference