7th Meeting

Translational Research Conference for the Management of NCLS

Chicago, Illinois
November 3-4, 2022
The Westin Chicago Lombard

We would like to thank you all for joining us for the 7th gathering of the Translational Research Conference for the Management of NCLs. It's hard to believe that a meeting that started 12 years ago in a small hotel conference room in Bethesda with ~30 attendees has evolved into what we are hosting today. Although the agenda and attendance have grown, the initial goal has not changed: to create a forum to bring together scientists, clinicians, and patient advocates to explore new and innovative approaches for treating Batten Disease. That initial meeting in 2010 focused entirely on treatment approaches for CLN1 and CLN2 disease, but recognizing we are all stronger together, our program and focus has evolved to include strategies for all forms of Batten disease.

Over this last decade plus, this meeting and others have been the spark for collaborative research partnerships between academia, industry and patient advocacy groups - really accelerating the path towards treatments for many forms of Batten disease. From the development and approval of an ERT for CLN2 Batten disease to the more recent gene therapies and biologics being developed for other forms of Batten disease, our research community has persevered in thinking innovatively and strategically. As the march toward novel therapies continues, leading to increased clinical trials, this meeting creates a forum to ensure we are leveraging experience and know-how from both within our Batten disease community as well as from experts outside our field.

This conference is structured to promote dialog and drive conversations. Speakers have been grouped by topics with a moderated panel discussion at the end of each session. Although we have aim to invite a wide variety of speakers for each session, time restrictions prevent us from having everyone working in that space present. Therefore, we kick off each panel discussion by opening the floor to the audience for anyone working in that session key focus area but who is not a speaker to provide a 2-3 min snapshot of their team's progress. After that, we will take questions and comments from the audience and look forward to a productive meeting.

We also want to say thank you to all the wonderful sponsors that make this meeting possible and to say thank you to each of you for being a part of this exciting meeting.

The Conference Organizing Committee
Heather and Chris Dainiak - Our Promise to Nicholas Foundation
Jill Weimer - Sanford Research
David Pearce – Sanford Research
Tracy VanHoutan – Noah's Hope- Hope4Bridget
Karen Kahn – Forebatten Foundation
Sandi and Stephen Lehrman

Agenda

Wednesday, November 2nd	Day 0
Cocktail Reception	
6:30-8:00pm	**Cypress Room**
Thursday, November 3rd	**Day 1**
Breakfast	
7:00-8:00am	**Magnolia Foyer**
Welcome and Opening Remarks **Magnolia Room**	
8:00-8:10am	**Jill Weimer, PhD & Chris Dainiak, MD**
Clinical Trial Updates & Lessons Learned: Session Chair: Dr. Steven Gray and Sharon King (Taylor's Tale)	
8:10-8:50am	**PJ Brooks, PhD; National Institute of Health** **Keynote Speaker** **"**Getting beyond "One disease at a time": Platform therapeutic approaches for monogenic disease"
8:50-9:15am	**Steven Gray, PhD; UT Southwestern Medical Center** *"Three different translational journeys for three different NCL gene therapies: CLN1, CLN5, CLN7"*
9:15-9:40am	**Angela Schulz, MD, PhD; University Medical Center Hamburg-Eppendorf Children's Hospital** ***"****Real world evidence on Intraventricular cerliponase alfa as standard of care in CLN2 disease: 4-year update from an independent ongoing observational study compared to clinical trials results"*
9:40-10:05am	**Xiomara Rosales, MD, MPH; Neurogene** "*Gene Therapy for CLN5 Neuronal Ceroid Lipofuscinosis: Seeking to Change the Natural History of CLN5 Batten Disease"*
10:05-10:30am	**Emyr Lloyd-Evans, PhD; Cardiff University (on behalf of Theranexus)** *"How glycosphingolipid accumulation impacts the pathophysiology of NCLs; development of substrate reduction therapy with Batten-1 (miglustat) for treating CLN3 disease and other NCLs"*
10:30-10:55am	**James Wawrzynski, MD; Great Ormand Street Hospital London** *"Intravitreal Cerliponase alfa for the treatment of CLN2 type Batten Disease related retinal dystrophy"*

10:55-11:20am	**Elizabeth Berry-Kravis, MD, PhD; Rush University Medical Center** *"Challenges to Translation of Disease-Targeted Therapies in Rare Neurogenetic Diseases"*
11:20-11:50am	**Moderated Session 1**
Lunch	
12:00-1:00pm	**Magnolia Foyer**
Natural History Studies, Biomarker Discovery & EMR Mining/ AI in Drug Discovery: Session Chair: Dr. Jon Mink and Dr. Chris Dainiak (Our Promise to Nicholas)	
1:00-1:20pm	**Jon Mink, MD, PhD; University of Rochester** *"NCL Natural History Studies in Preparation for Clinical Trials"*
1:20-1:40pm	**An Dang Do, MD, PhD; NICHD/NIH** *"CLN3 Natural History and Biomarker Discovery"*
1:40-2:00pm	**Forbes D. Porter, MD, PhD; NICHD/NIH** *"Development of Tools to Facilitate Clinical Care and Drug Development: Biomarker Discovery and Characterization for Niemann-Pick Disease, type C1"*
2:00-2:20pm	**Erika Augustine, MD, MS; Kennedy Krieger** *"Advancing Natural History Knowledge in the NCLs"*
2:20-2:40pm	**Melissa Pratt; Sanford Research** *"Translational Biomarker Discovery for CLN3 Batten Disease: Happenings and Hurdles"*
2:40-3:00pm	**Robert Ellis; Koneksa Health** *"Do multi-component digital biomarkers in diseases like Parkinson's have potential in Batten disease?"*
3:00-3:25pm	**Moderated Session 2**
3:25-3:30pm	***Break***
Nontraditional Clinical and Pathological Changes: Session Chair: Dr. Jon Cooper and Dr. David Pearce	
3:30-3:55pm	**Michael Boyne, PhD; COUR** *"Addressing Immunogenicity in the Treatment of Rare Disease using Antigen-Specific Tolerance"*
3:55-4:20pm	**Heather Adams, PhD; University of Rochester** *"Characterizing sleep dysfunction in individuals with NCL Disorders"*

4:20-4:45pm	**Robert O. Heuckeroth, MD, PhD; Children's Hospital of Philadelphia and Perelman School of Medicine at the University of Pennsylvania** *"Human bowel motility disorders and the "second brain" in the bowel"*
4:45-5:10pm	**Jonathan D. Cooper, PhD; Washington University in St. Louis** *"Defining and treating disease outside the brain"*
5:10-5:35pm	**Stephen Waggoner, PhD; Cincinnati Children's Hospital Medical Center** *"Loss of CLN3 delivers a gut punch to neuro-intestinal homeostasis"*
5:35-6:00pm	**Moderated Session 3**
Dinner	
6:15-9:00pm	**Junior Ballroom**

Friday, November 4th	**Day 2**
Breakfast	
7:00-8:00am	**Magnolia Foyer**
Small Molecules, ERTs, Chaperones, and Biologicals: **Magnolia Room** Session Chair: Dr. May Khanna and Dr. Stéphane Lefrancois	
8:00-8:30am	**Marco Sardiello, PhD; Washington University** *"Combined administration of trehalose and miglustat in a mouse model of CLN3 disease"*
8:30-9:00am	**Paul Trippier, PhD; University of Nebraska** *"Compounds and Screening Modalities to Drive Small Molecule Drug Discovery in CLN3 Disease"*
9:00-9:30am	**Diego Medina, PhD; TIGEM** *"Repurposing tamoxifen to treat multiple NCLs"*
9:30-10:00am	**Elena Batrakova, PhD; UNC at Chapel Hill** *"Extracellular Vesicles as Next Generation Drug Delivery Vehicles for Treatment of NCL"*
10:00-10:30am	**May Khanna, PhD; New York University** *"Developing small molecule chemical chaperone"*
10:30-10:55pm	**Moderated Session 4**

10:55-11:00am	***Break***
Viral Mediated Gene Correction: Session Chair: Dr. Jill Weimer and Tracy Van Houtan (Noah's Hope/Hope 4 Bridget)	
11:00-11:30am	**Xin Chen, MD, PHD; UT Southwestern** *"Crucial considerations during the development of preclinical gene replacement therapy for CLN7 Batten disease"*
11:30-12:00pm	**Russell Gotschall; M6P Therapeutics** *"Defining and overcoming challenges for developing more effective treatments for LSDs."*
Lunch	
12:00-1:00pm	**Magnolia Foyer**
Continuation of Viral Mediated Gene Correction: Session Chair: Dr. Jill Weimer and Tracy Van Houtan (Noah's Hope/Hope 4 Bridget)	
1:00-1:30pm	**Miguel Esteves, PhD; University of Massachusetts** *"Designing AAV vectors for CNS gene therapy applications"*
1:30-2:00pm	**Arun Srivastava, PhD; University of Florida** *"Development of NexGen, GenX, and Opt AAV vectors for human gene therapy"*
2:00-2:25pm	**Moderated Session 5**
2:25-2:30pm	***Break***
Emerging nucleic acid therapeutic approaches: Session Chair: Dr. Michelle Hastings and Gavin Ferrandino (Drew's Hope)	
2:30-3:00pm	**Michelle Hastings, PhD; Rosalind Franklin** *"Antisense Oligonucleotides for the Treatment of CLN3 Disease"*
3:00-3:30pm	**Gopi Shanker, PhD; Tevard Biosciences** *"Transforming Therapeutic Development for Rare Diseases with Transfer RNA"*
3:30-4:00pm	**Kiran Musunuru, MD, PhD, MPH, ML; University of Pennsylvania** *"Genome editing to prevent human disease"*
4:00-4:30pm	**Matt Stanton, PhD; Generation Bio** *"Solving key scientific challenges to unlock non-viral gene therapy"*

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Dr. Jonathan Cooper
on behalf of NCL 2021

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