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Discussants

Jonathan D. Cooper, PhD
Reader in Experimental Neuropathy
The James Black Centre
King's College London

Kathy Simms, MD
Director, Developmental Neurogenetics
Clinic
Director, Neurogenetics Diagnostic Lab
Massachusetts General Hospital
Professor of Neurology
Harvard Medical School

Pascale Tiger, PhD
BioMarin Pharmaceutical Inc.

Stephen Kuhn, MD
V.P., Head of the CNS Program
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November 11—12, 2010

National Institute of Neurological
Disorders and Stroke

Drew's Hope Research Foundation

Fight for Nicolas

Hope 4 Bridget

Jasper Against Batten

Mary Payton's Miracle Foundation

Noah's Hope

Our Promise to Nicholas Foundation

Day 1

Welcome

8:00 am.—8:10 a.m.

David Pearce, PhD

Sanford Children's Health Research Center

University of South Dakota

“Welcome”

Introduction to NCLs

(Session I Chair—David Pearce)

8:10 a.m.— 8:35 a.m.

Denia Ramirez-Montealegre, MD, MPH, PhD

University of Rochester Medical Center

“Update on Clinical and Genotypic Aspects of INCL and LINCL”

8:35 a.m.— 9:00 a.m.

Peter Lobel, PhD

Robert Wood Johnson Medical School

Center of Advanced Biotechnology & Medical School

“Basic & Translation Research on Classical Lae Infantile Neuronal Ceroid Lipofuscinosis”

Therapeutic Studies-delivery?

9:00 a.m.—9:25 a.m.

Robert Steiner, MD

Oregon Health & Science University

“CNC Transplantation of Purified human Neural Stem Cells in NCL: Phase I Trial”

9:25 a.m.—9:50 a.m.

Sandra Hoffmann, MD

UT Southwestern Medical Center at Dallas

“Enzyme Replacement Therapy for CLN1/PPT deficiency in mice”

Coffee Break 9:50 a.m.—10:10 a.m.

10:10 a.m.—10:35 a.m.

Mark Sands, PhD

Washington University Medical School

“Combination Therapy for Infantile Batten Disease”

10:35 a.m.—11:00 a.m.

Beverly L. Davidson, PhD

University of Iowa Healthcare

“Vector Mediated Delivery of Lysosomal Enzymes to the Brain”

11:00 a.m.—11:25 a.m.

Silvia Muro, PhD

Center for Biosystems Research

“ICAM-1 Targeted Nanocarriers For Delivery of Lysosomal Enzymes to the Brain”

11:25 a.m.—11:50 a.m.

Steven Gray, PhD

University of North Carolina at Chapel Hill

“Strategies and Practical consideration for AAV-mediated global CNS Delivery”

Discussion #1

11:50 a.m.—1:30 p.m.

Lunch

12:30 p.m.—1:30 p.m.

New Therapeutic Approaches to INCL and LINCL—Small Molecules?

(Session II Chair—Beverly Davidson)

1:30 p.m.—1:55 p.m.

Rozzy Finn

Sanford Research Center

“Glutamate Receptors as a Potential Therapeutic Target For the Treatment of INCL”

1:55 p.m. - 2:20 p.m.

Kalipada Pahan, PhD

Rush University Medical Center

“Protection of Dopaminergic Neurons by Simvastatin in a Mouse Model of Parkinson's Disease”

2:20 p.m.—2:45 p.m.

Ravi Singh, PhD

Iowa State University

“Antisense-mediated correction of Aberrant Splicing in a Model Genetic Disease of Children”

2:45 p.m.—3:10 p.m.

David Bedwell, PhD

University of Alabama at Birmingham Department of Microbiology

“Modulating Translation Termination to Treat Genetic Diseases”

Coffee Break 3:10 p.m. —3:30 p.m.

3:10 p.m.—3:35 p.m.

Michael Jackson, PhD

Sanford-Burnham Medical Research Institute

“Screening for Compounds (drugs) Using High Content Imaging and Patient Derived Cells. New Hope of Finding Treatment Options for Rare Diseases”

3:35 p.m.—4:00 p.m.

Christopher Austin, MD

National Institute of Health

“The NIH Therapeutics for Rare and Neglected Diseases Program”

Discussion #2
4:00 p.m.—5:30 p.m.

Day 2

Clinical Trials?

(Session III Chair—Mark Sands)

8:00 a.m.—8:25 a.m.

Edward M. Kaye, MD

Genzyme Corporation

“From Small Molecules to Proteins Lessons Learned in Developing Therapies for Gaucher, Fabry and Pompe Diseases”

8:25 a.m.—8:50 a.m.

Katherine A. High, MD

Children's Hospital of Philadelphia

“AAV-Mediated Gene Therapy for Leber's Congenital Amaurosis: Regulatory Considerations, Clinical Endpoints, and results of a Phase I/II Study”

8:50 a.m.—9:15 a.m.

Ronald G. Crystal, MD

Weill Cornell Medical College

“Gene Therapy for the CNS Manifestations of the Lysosomal Storage Disorders”

9:15 a.m.—9:40 a.m.

Niklas Mattsson, MD

Sahlgren's University Hospital

“Cerebrospinal Fluid Biomarkers of Neurodegeneration”

9:40 a.m.—10:05 a.m.

Anne Pariser, MD

White Oak Campus

“FDA Review and Regulation of Rare Clinical Trials”

Coffee Break 10:05 a.m.—10:20 a.m.

Discussion #3
And Meeting Summary