Day 1

Registration/Light breakfast 8:30 – 9:00

I. Introduction

9:00 – 9:10 Welcome
  Dr. Wendy Introne
  Dr. William Gahl

9:10 – 9:30 Living with Chediak-Higashi disease
  Selena Tenhoeve, CHS Association
  Heather Alvey, CHS Association

9:30 – 10:00 Overview of Chediak-Higashi Disease
  Dr. Wendy Introne
  Dr. Marjan Huizing

10:00 – 10:30 Cell Biology and Genotype-Phenotype Correlation
  Dr. Andrew Cullinane

10:30 – 10:45 Break

II. LYST function – What is known?

Chair: Dr. Andrew Cullinane

10:45 – 11:00 Introduction to mouse models
  Dr. Andrew Cullinane

11:00 – 11:45 Molecular aspects and pathophysiology of LYST
  Dr. Genevieve de Saint Basile

11:45 – 12:30 Lunch

12:30 – 1:15 LYST in vesicle development
  Dr. Diane McVey Ward

1:15 – 2:00 Function of p14 in a mouse model
  Dr. Christoph Klein

2:00 – 2:15 Break
III. Neurologic Manifestation of CHS

Chair: Dr. Camilo Toro

2:15 – 3:00 Neurologic Spectrum of Atypical CHS
   Dr. Camilo Toro

3:00 – 3:30 Cell Biology Parallels with other Neurologic Disorders, such as SPG11 and SPG15
   Dr. Craig Blackstone

3:30 – 4:00 Clinical Similarities with other Lysosomal Disorders
   Dr. Ellen Sidransky

IV. Roundtable Discussion

Chairs: Dr. Camilo Toro and Dr. William Gahl

4:00 – 4:45
   All participants to discuss clinical, cell biological, and animal model plans for advancing understanding of the neurologic manifestations.

Day 2

Light Breakfast 8:30 – 9:00

V. Hemophagocytic Lymphohistiocytosis and Bone Marrow Transplantation

Chair: Dr. Wendy Introne

9:00 – 9:45 NK cell function and role in CHS
   Dr. Konrad Krzewski

9:45 – 10:30 Familial Forms of HLH—Functional assays and genotype/phenotype correlation
   Dr. Yenan Bryceson via Skype

10:30 – 10:45 Break

10:45 – 11:30 Advances in Management of HLH and Success and Timing of BMT in CHS
   Dr. Lisa Filipovich
11:30 – 12:00 Challenges of BMT in CHS

Dr. Kirk Schultz

VI. Roundtable Discussion and Summary

Chairs: Dr. Wendy Introne and Dr. William Gahl

12:00 – 1:00

All participants to discuss monitoring and treatment of HLH, eligibility and timing of BMT.

Lunch and Adjourn