## 2nd Annual Robert J. Gorlin Symposium

**Title:** Precision Medicine: A Multidisciplinary Approach  
*(Registration required)*

**Speaker:** Jeanine R. Jarnes  
**Affiliation:** University of Minnesota  
**Location:** Minneapolis, MN, United States

**Time:** 1:45 PM

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## Emerging Trends: State-of-the-art for Experts

**Speaker:** Filippo Pinto e Vairo  
**Affiliation:** Mayo Clinic  
**Location:** Rochester, MN, United States

**Title:** Case Studies of Multi-Omic Approach for the Diagnosis of Lysosomal Diseases

**Time:** 2:15 PM

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**Speaker:** Jennifer Goldstein  
**Affiliation:** UNC-Chapel Hill  
**Location:** Chapel Hill, NC, United States

**Title:** NIH-Funded Resources: ClinGen and ClinVar

**Time:** 2:35 PM

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**Speaker:** Jeanine R. Jarnes  
**Affiliation:** University of Minnesota  
**Location:** Minneapolis, MN, United States

**Title:** Implementation of Pharmacogenomics Programs within Clinical Settings

**Time:** 2:55 PM

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**Speaker:** Chester B. Whitley  
**Affiliation:** University of Minnesota  
**Location:** Minneapolis, MN, United States

**Title:** Introduction and Course Overview

**Time:** 4:00 PM

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**Speaker:** Gregory A. Grabowski  
**Affiliation:** Cincinnati Children's Hospital Research Foundation  
**Location:** Cincinnati, OH, United States

**Title:** Lysosomal Function and Pathogenesis

**Time:** 4:01 PM

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**Speaker:** Marc C. Patterson  
**Affiliation:** Mayo Clinic Children's Center  
**Location:** Rochester, MN, United States

**Title:** Clinical Features

**Time:** 4:15 PM

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**Speaker:** Amy Gaviglio  
**Affiliation:** Centers for Disease Control and Prevention  
**Location:** Minneapolis, MN, United States

**Title:** Newborn Screening

**Time:** 4:30 PM

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**Speaker:** Jeanine R. Jarnes  
**Affiliation:** University of Minnesota  
**Location:** Minneapolis, MN, United States

**Title:** Lysosomal Disease Therapies

**Time:** 4:45 PM

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**Speaker:** Christine Yuen-Yi Hon, PharmD  
**Affiliation:** Office of New Drugs | CDER | FDA  
**Location:** Silver Spring, MD, United States

**Title:** Regulatory Review

**Time:** 5:00 PM

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**Speaker:** Jennifer Klein  
**Affiliation:** National MPS Society  
**Location:** Durham, NC, United States

**Title:** Patient Perspective

**Time:** 5:15 PM

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**Speaker:** N. Matthew Ellinwood  
**Affiliation:** National MPS Society  
**Location:** Durham, NC, United States

**Title:** Rare Disease Research

**Time:** 5:30 PM

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**Speaker:** Chester B. Whitley  
**Affiliation:** University of Minnesota  
**Location:** Minneapolis, MN, United States

**Title:** Open Q&A

**Time:** 5:45 PM

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**Speaker:** Be the Catalyst Event

**Time:** 6:00 PM
# WORLD Symposium™ 2023 Preliminary Program*

**Platform Presenters**

**Wednesday, February 22, 2023: Basic Science**

**Moderators:** Brian Bigger, Lalitha Belur, and Michael Przybilla

## Satellite Symposia

<table>
<thead>
<tr>
<th>Time</th>
<th>Presenter(s)</th>
<th>Institution(s)</th>
<th>Location(s)</th>
<th>Topic</th>
</tr>
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<tbody>
<tr>
<td>6:15 AM</td>
<td>Chester B. Whitley</td>
<td>University of Minnesota</td>
<td>Minneapolis, MN, United States</td>
<td>Welcome &amp; Announcements  Presentation of 2023 Roscoe O. Brady Award to William A. Gahl</td>
</tr>
<tr>
<td></td>
<td>William A. Gahl</td>
<td>National Human Genome Research Institute</td>
<td>Bethesda, MD United States</td>
<td>Roscoe O. Brady Award Presentation: Pursuing Advances in Rare and Undiagnosed Diseases</td>
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<tr>
<td>7:30 AM</td>
<td>Chester B. Whitley</td>
<td>University of Minnesota</td>
<td>Minneapolis, MN, United States</td>
<td>A brain penetrant progranulin-derived biologic protects against neuronopathic Gaucher disease *2023 Young Investigator Award Recipient</td>
</tr>
<tr>
<td>8:00 AM</td>
<td>Xiangli Zhao</td>
<td>New York University Grossman School of Medicine</td>
<td>New York, NY, United States</td>
<td>Earlier-onset, more severe neurodegeneration in PGRN KO mice with a decreased dose of D409V Gba1</td>
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<td></td>
<td>Yi Lin</td>
<td>Cincinnati Children’s Hospital Medical Center</td>
<td>Cincinnati, OH, United States</td>
<td>A multifaceted evaluation of microgliosis and differential cellular dysregulations of mTOR signaling with fluctuating lysosome function in neuronopathic Gaucher disease *2023 Young Investigator Award Recipient</td>
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<td></td>
<td>Zhenting Zhang</td>
<td>Cincinnati Children’s Hospital Medical Center</td>
<td>Cincinnati, OH, United States</td>
<td>Study of miRNA expression profiles depending on the severity of bone involvement in patients with Gaucher disease</td>
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<td>Irene Serrano Gonzalo</td>
<td>Fundación Española para el Estudio y Terapéutica de la Enfermedad de Gaucher y otras lisosomales</td>
<td>Zaragoza, Spain</td>
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<tr>
<td></td>
<td><strong>Moderated Q&amp;A</strong></td>
<td>Xiangli Zhao, Yi Lin, Zhenting Zhang, and Irene Serrano Gonzalo</td>
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<tr>
<td>9:00 AM</td>
<td>Maria Fuller</td>
<td>SA Pathology</td>
<td>North Adelaide, Australia</td>
<td>Signature biomarkers for diagnosis, screening, and biochemical monitoring of the mucopolysaccharidoses</td>
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<td></td>
<td>Rebecca C. Ahrens-Nicklas</td>
<td>The Children’s Hospital of Philadelphia</td>
<td>Philadelphia, PA, United States</td>
<td>Biomarkers of disease severity in multiple sulfatase deficiency</td>
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<td></td>
<td>Hannah Best</td>
<td>Cardiff University</td>
<td>Cardiff, United Kingdom</td>
<td>The Batten disease associated protein CLN3 is required for the efflux of lysosomal K+ *2023 Young Investigator Award Recipient</td>
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<td></td>
<td>Tyler M. Pierson</td>
<td>Cedars-Sinai Medical Center</td>
<td>Los Angeles, CA, United States</td>
<td>Modeling CLN6 with IPSC-derived neurons and glia</td>
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<tr>
<td></td>
<td><strong>Moderated Q&amp;A</strong></td>
<td>Maria Fuller, Rebecca C. Ahrens-Nicklas, Hannah Best, and Tyler M. Pierson</td>
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<tr>
<td>10:00 AM</td>
<td><strong>Break</strong></td>
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<tr>
<td>10:30 AM</td>
<td>Francyne Kubaski</td>
<td>Greenwood Genetic Center</td>
<td>Greenwood, SC, United States</td>
<td>Sensitivity and specificity of four lysosomal disorder biomarkers in dried blood spots</td>
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</table>
### Platform Presenters

<table>
<thead>
<tr>
<th>Name</th>
<th>Affiliation</th>
<th>Topic</th>
<th>*2023 Young Investigator Award Recipient</th>
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</thead>
<tbody>
<tr>
<td>Neil Kasaci</td>
<td>Lysosomal and Rare Disorders Research and Treatment Center, Fairfax, VA, United States</td>
<td>Caspase inhibitors can counteract inflammasome activation and caspase-1 mediated fibrosis in Fabry disease</td>
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<tr>
<td>Saida Ortolano</td>
<td>Galicia Sur Health Research Institute, Vigo, Spain</td>
<td>PBXs: New pharmacological chaperones to increase α-galactosidase A activity in Fabry disease cellular models</td>
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<tr>
<td>Efecan Aral</td>
<td>University of Massachusetts - Amherst, Amherst, MA, United States</td>
<td>Establishing personalized medicine in Fabry disease through functional analysis of disease mutants</td>
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<td>*2023 Young Investigator Award Recipient</td>
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### Moderated Q&A

- Francyne Kubaski, Neil Kasaci, Saida Ortolano, and Efecan Aral

### 11:30 AM Break and Satellite Symposia

<table>
<thead>
<tr>
<th>Time</th>
<th>Name</th>
<th>Affiliation</th>
<th>Topic</th>
<th>*2023 Young Investigator Award Recipient</th>
</tr>
</thead>
<tbody>
<tr>
<td>1:00 PM</td>
<td>Behzad Najafian</td>
<td>University of Washington, Seattle, WA, United States</td>
<td>The spectrum of podocyte injury in later onset (LO) variants of Fabry disease (FD)</td>
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<td></td>
<td>David Smerkous</td>
<td>Oregon State University, Corvallis, OR, United States</td>
<td>Development of an online cloud-based tool for automatic measurement of foot process width (FPW) using deep learning (DL): Applications in assessment of podocyte injury in Fabry disease (FD)</td>
<td>*2023 Young Investigator Award Recipient</td>
</tr>
<tr>
<td></td>
<td>Alex J. Shamoun</td>
<td>University of Florida, Gainesville, FL, United States</td>
<td>Differences in organ abundance of iduronate 2-sulfatase and intravenous recombinant enzyme delivery: Potential implications for clinical response to ERT in MPS II</td>
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<td></td>
<td>Marta Artola</td>
<td>Leiden University, Leiden, Netherlands</td>
<td>1,6-epi-cyclophellitol cyclosulfamidate is a new superior lysosomal α-glucosidase stabilizer for the treatment of Pompe disease</td>
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<td>*2023 Young Investigator Award Recipient</td>
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</tbody>
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### 2:00 PM Moderated Q&A

- Behzad Najafian, David Smerkous, Alex J. Shamoun, and Marta Artola

### 2:00 PM Break and Satellite Symposia

<table>
<thead>
<tr>
<th>Time</th>
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<th>Topic</th>
<th>*2023 Young Investigator Award Recipient</th>
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</thead>
<tbody>
<tr>
<td>2:00 PM</td>
<td>Mahsa Taherzadeh</td>
<td>McGill University, Montreal, QC, Canada</td>
<td>Severe neuronal demyelination in Sanfilippo disease</td>
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<td>Frederick Ashby</td>
<td>University of Florida, Gainesville, FL, United States</td>
<td>Bone pathology within Sanfilippo syndrome type B mice as a novel biometric for peripheral disease correction</td>
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<td></td>
<td>Chloé Dias</td>
<td>Université Toulouse III Paul Sabatier, Toulouse, France</td>
<td>Microglia-derived extracellular vesicles promote neuropathology in Sanfilippo syndrome</td>
<td>*2023 Young Investigator Award Recipient</td>
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<td></td>
<td>Angela J. Espejo</td>
<td>Pontificia Universidad Javeriana, Bogotá D.C., Colombia</td>
<td>Magnetite nanoparticles as a vehicle to transport recombinant hexosaminidase A and B through an in vitro model of the blood-brain barrier</td>
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### 3:00 PM Speed Mentoring Session

### 5:15 PM Speed Mentoring Session
WORLDSymposium™ 2023 Preliminary Program*
Platform Presenters
Thursday, February 23, 2023: Translational Research
Moderators: PJ Brooks, Amy Gaviglio, and Francyne Kubaski

6:15 AM Satellite Symposia

7:30 AM Chester B. Whitley
University of Minnesota
Minneapolis, MN, United States
Welcome & Announcements
Presentation of 2023 Patient Advocate Leader (PAL) Award to Christine Waggoner and 2023 Young Investigator Awards Presentation

8:00 AM Anna-Maria Wiesinger
Paracelsus Medical University Salzburg
Salzburg, Austria
A precision medicine tool for high utilization and quality of individual treatment trials with immunomodulatory drugs in mucopolysaccharidosis
*2023 Young Investigator Award Recipient

Barbara K. Burton
Northwestern University Feinberg School of Medicine
Chicago, IL, United States
Newborn screening for mucopolysaccharidosis type II

Stuart M. Ellison
University of Manchester
Manchester, United Kingdom
Validation of a GMP stem cell gene therapy manufacturing process for mucopolysaccharidosis type II (MPS II) in preparation for an approved phase I/II clinical trial

Anna Luzzi
The Lundquist Institute for Biomedical Innovation at Harbor-UCLA Medical Center
Torrance, CA, United States
Decreased regulatory T-cells in patients with Sanfilippo syndrome may allow the development of autoimmune disease

Moderated Q&A
Anna-Maria Wiesinger, Barbara K. Burton, Stuart M. Ellison, and Anna Luzzi

9:00 AM Kim M. Hemsley
Flinders University
Bedford Park, Australia
A prohibitin-targeting drug modifies aspects of disease in a mouse model of Sanfilippo syndrome

Simon Jones
St. Mary's Hospital
Manchester, United Kingdom
Sustained biochemical engraftment and early clinical outcomes following ex-vivo autologous stem cell gene therapy for mucopolysaccharidosis type IIIA

Oriana Mandolfo
University of Manchester
Manchester, United Kingdom
Developing an iPSC-based neural gene therapy approach for MPS IIIA

Nissrine Ballout
Université Toulouse III Paul Sabatier
Toulouse, France
Development and validation of a novel adeno-associated viral gene therapy for mucopolysaccharidosis type IIIB (MPS IIIB)
*2023 Young Investigator Award Recipient

Moderated Q&A
Kim M. Hemsley, Simon Jones, Oriana Mandolfo, and Nissrine Ballout

10:00 AM Break & Exhibits

10:30 AM Troy Lund
University of Minnesota
Minneapolis, MN, United States
Decreases in CSF neuro-inflammatory markers are associated with gain in neurocognitive function after ERT + HCT in Hurler syndrome

Roselena S. Schuh
Universidade Federal do Rio Grande do Sul
Porto Alegre, Brazil
Nasal administration of laronidase-loaded liposomes aiming at mucopolysaccharidosis type I treatment
*2023 Young Investigator Award Recipient

Michael J. Przybilla
University of Minnesota
Minneapolis, MN, United States
Treating murine Hurler syndrome utilizing small-activating RNA following bone marrow transplant

01/24/2023
**WORLD Symposium™ 2023 Preliminary Program*  
Platform Presenters**

**Thursday, February 23, 2023: Translational Research (cont.)**

**Moderators: PJ Brooks, Amy Gaviglio, and Francyne Kubaski**

**Betul Celik**  
University of Delaware  
Newark, DE, United States  
Lentiviral gene therapy for mucopolysaccharidosis type IVA

**Moderated Q&A**  
*Troy Lund, Roselena S. Schuh, Michael J. Przybilla, and Betul Celik*

**11:30 AM**  
**Break, Exhibits and Satellite Symposia**  

**1:00 PM**  
**Leigh Fremuth**  
St. Jude Children's Research Hospital  
Memphis, TN, United States  
AAV-mediated gene therapy for galactosialidosis: A long-term safety and efficacy study

**Sandra Vranic**  
University of Manchester  
Manchester, United Kingdom  
Defect-free graphene enhances enzyme delivery to fibroblasts derived from the patients with lysosomal disorders

**Paul J. Orchard**  
University of Minnesota  
Minneapolis, MN, United States  
Compassionate use of OTL-200 for patients with metachromatic leukodystrophy

**Laura A. Adang**  
Children's Hospital of Philadelphia  
Philadelphia, PA, United States  
Developmental delay can precede neurologic regression in metachromatic leukodystrophy

**Moderated Q&A**  
*Leigh Fremuth, Sandra Vranic, Paul J. Orchard, and Laura A. Adang*

**2:00 PM**  
**Lars Schlotawa**  
University Medical Center Goettingen  
Goettingen, Germany  
Screening of approved drugs identifies 3rd generation retinoids as in vitro therapeutic agents in multiple sulfatase deficiency

**Aimee Donald**  
University of Manchester  
Manchester, United Kingdom  
Sustained improvement of clinical CNS and somatic features of Gaucher disease type 3 after haematopoietic stem cell (HSC) gene therapy: A first-in-world report

**Andreas Hahn**  
University Hospital Giessen  
Giessen, Germany  
Treatment of CLN1 disease with a blood-brain barrier penetrating lysosomal enzyme AGT-194

**Jason A. Weesner**  
St. Jude Children's Research Hospital  
Memphis, TN, United States  
Preclinical enzyme replacement therapy with a recombinant β-galactosidase-lectin fusion for CNS delivery and treatment of GM1-gangliosidosis  
*2023 Young Investigator Award Recipient*

**Moderated Q&A**  
*Lars Schlotawa, Aimee Donald, Andreas Hahn, and Jason A. Weesner*

**3:00 PM**  
**Poster Session in the Exhibit Hall**

**5:15 PM**  
**Satellite Symposia**
### Satellite Symposia

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<td>6:15 AM</td>
<td>Welcome and Keynote Speaker Introduction</td>
<td>Chester B. Whitley</td>
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<td>University of Minnesota</td>
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<td>Minneapolis, MN, United States</td>
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<tr>
<td>8:00 AM</td>
<td>Francesca Fumagalli</td>
<td>Long-term clinical outcomes of atidarsagene autotemcel</td>
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<td>(autologous hematopoietic stem cell gene therapy [HSC-GT] for metachromatic leukodystrophy) with up to 11 years follow-up</td>
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<td>San Raffaele Telethon Institute for Gene Therapy, IRCCS San Raffaele Scientific Institute Milan, Italy</td>
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<tr>
<td>8:30 AM</td>
<td>Maria Jose De Castro Lopez</td>
<td>Twice weekly dosing with sebelipase alfa rescues severely ill infants with Wolman disease</td>
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<td>Hospital Clínico Santiago</td>
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<td>Santiago, Spain</td>
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<tr>
<td>8:30 AM</td>
<td>Robert J. Hopkin</td>
<td>STAAR, a phase I/II study of isaralagene civaparvovec (ST-920) gene therapy in adults with Fabry disease: Dose escalation phase results</td>
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<td>Cincinnati Children's Hospital Medical Center</td>
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<td>Cincinnati, OH, United States</td>
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<tr>
<td>8:30 AM</td>
<td>Valeria Calbi</td>
<td>Lentiviral haematopoietic stem cell gene therapy for metachromatic leukodystrophy: Results in 5 patients treated under nominal compassionate use</td>
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<td>San Raffaele Telethon Institute for Gene Therapy, IRCCS San Raffaele Scientific Institute Milan, Italy</td>
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<td>9:00 AM</td>
<td>Moderated Q&amp;A</td>
<td>Francesca Fumagalli, Maria Jose De Castro Lopez, Robert J. Hopkin, and Valeria Calbi</td>
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<tr>
<td>9:00 AM</td>
<td>Joseph Muenzer</td>
<td>Interim analysis of key clinical outcomes from a phase 1/2 study of weekly intravenous DNL310 (brain-penetrant enzyme replacement therapy) in MPS II</td>
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<td>University of North Carolina Chapel Hill</td>
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<td>Chapel Hill, NC, United States</td>
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<tr>
<td>9:00 AM</td>
<td>Paul Harmatz</td>
<td>Interim results of a phase 1/2 study of JR-171 (lepunafusp alfa), a novel brain-penetrant enzyme replacement therapy for MPS I</td>
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<td>UCSF Benioff Children's Hospital Oakland</td>
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<td>Oakland, CA, United States</td>
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<tr>
<td>9:00 AM</td>
<td>Raymond Y. Wang</td>
<td>RGX-111 gene therapy for the treatment of severe mucopolysaccharidosis type I (MPS I): Interim analysis of data from the first in human study</td>
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<td>Orange, CA, United States</td>
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<td>9:30 AM</td>
<td>Cara O'Neill</td>
<td>Development of consensus guidelines for the clinical care of individuals with Sanfilippo syndrome</td>
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<td>Cure Sanfilippo Foundation</td>
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<td>Columbia, SC, United States</td>
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<td>Joseph Muenzer, Paul Harmatz, Raymond Y. Wang, and Cara O'Neill</td>
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<td>10:00 AM</td>
<td>Break &amp; Exhibits</td>
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<tr>
<td>10:30 AM</td>
<td>Barry J. Byrne</td>
<td>Long-term follow-up of cipaglucosidase alfa/miglustat in ambulatory patients with Pompe disease: An open-label phase I/II study (ATB200-02)</td>
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<td>Gainesville, FL, United States</td>
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<td>Platform Presenters</td>
<td>Description</td>
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<tr>
<td><strong>Erin Huggins</strong>&lt;br&gt;Duke University&lt;br&gt;Durham, NC, United States</td>
<td>Longitudinal follow up uncovers an early emerging phenotype in children with late-onset Pompe disease diagnosed via newborn screening</td>
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<td><strong>Priya S. Kishnani</strong>&lt;br&gt;Duke University Medical Center Durham&lt;br&gt;Durham, NC, United States</td>
<td>Efficacy and safety of avalglucosidase alfa in participants with late-onset Pompe disease after 145 weeks of treatment during the COMET trial</td>
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<tr>
<td><strong>Jordi Diaz Manera</strong>&lt;br&gt;Newcastle University&lt;br&gt;Newcastle Upon Tyne, United Kingdom</td>
<td>AT845 gene replacement therapy for late onset Pompe disease: An update on safety and preliminary efficacy data from FORTIS, a phase I/II open-label clinical study</td>
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</table>

**Moderated Q&A**<br>Barry J. Byrne, Erin Huggins, Priya S. Kishnani, and Jordi Diaz Manera

**11:30 AM**<br>Break, Exhibits and Satellite Symposia

**1:00 PM**<br>**Eric Wallace**<br>University of Alabama<br>Birmingham, AL, United States<br>First results of a head-to-head trial of pegunigalsidase alfa vs. agalsidase beta in Fabry disease: 2 year results of the phase 3 randomized, double-blind, BALANCE study

**John Bernat**<br>University of Iowa Hospitals and Clinics<br>Iowa City, IA, United States<br>Long-term safety and efficacy of pegunigalsidase alfa administered every 4 weeks in patients with Fabry disease: Two-year interim results from the ongoing phase 3 BRIGHTS1 open-label extension study

**Melissa P. Wasserstein**<br>Albert Einstein College of Medicine/Children's Hospital at Montefiore<br>Bronx, NY, United States<br>Plasma lyso-sphingomyelin as a biomarker for acid sphingomyelinase deficiency: Correlations with baseline disease and response to olipudase alfa treatment in clinical trials

**Roberto Giugliani**<br>Federal University of Rio Grande do Sul<br>Porto Alegre, RS, Brazil<br>Long-term catch-up growth in children with acid sphingomyelinase deficiency treated with olipudase alfa enzyme replacement therapy in the ASCEND-Peds trial

**Moderated Q&A**<br>Eric Wallace, John Bernat, Melissa P. Wasserstein, and Roberto Giugliani

**2:00 PM**<br>**Pramod K. Mistry**<br>Yale University School of Medicine<br>New Haven, CT, United States<br>Changes in hematologic and visceral manifestations over time following imiglucerase initiation in Gaucher disease type 1 and type 3 pediatric patients in the ICGG Gaucher Registry

**Jeanine R. Jarnes**<br>University of Minnesota<br>Minneapolis, MN, United States<br>Updated interim safety, biomarker, and efficacy data from Imagine-1: A phase 1/2 open-label, multicenter study to assess the safety, tolerability, and efficacy of a single dose, intra-cisterna magna (ICM) administration of PBGM01 in subjects with type I (early onset) and type II (late onset) infantile GM1 gangliosidosis (GM1)

**Yoshikatsu Eto**<br>Institute of Neurological Disease<br>Kawasaki City, Japan<br>Real-world data of enzyme replacement therapy with pabinafusp alfa for neuronopathic MPS-II: Updated clinical data from Japan

**David L. Rogers**<br>Nationwide Children's Hospital<br>Columbus, OH, United States<br>Intravitreal enzyme replacement therapy to prevent retinal disease progression in children with neuronal ceroid lipofuscinosi type 2 (CLN2): Interim safety report

**Moderated Q&A**<br>Pramod K. Mistry, Jeanine R. Jarnes, Yoshikatsu Eto, and David L. Rogers

**3:00 PM**<br>Poster Session in the Exhibit Hall

**5:30 PM**<br>Satellite Symposia
6:15 AM  **Satellite Symposia**

7:45 AM  **Chester B. Whitley**  
University of Minnesota  
Minneapolis, MN, United States  
**Welcome and New Treatment Award**

8:00 AM  **Shababa T. Masoud**  
Denali Therapeutics  
South San Francisco, CA, United States  
ETV:SGSH, a brain-penetrant enzyme transport vehicle for SGSH, corrects heparan sulfate accumulation, lysosomal lipid storage and inflammation in MPS IIIA mouse brain

**Asuka Inoue**  
JCR Pharmaceuticals Co., Ltd.  
Kobe, Japan  
Nonclinical pharmacodynamics, pharmacokinetics and safety profiles of anti-human transferrin receptor antibody-fused N-sulfoglucosamine sulfohydrolase for mucopolysaccharidosis type IIIA

**Andrew Hedman**  
M6P Therapeutics  
St. Louis, MO, United States  
Novel dual promoter AAV gene therapy platform ensures production of therapeutic soluble lysosomal enzymes with high M6P content to enable broad cellular uptake and cross correction in vivo

**Charu Reddy**  
Codexis  
San Carlos, CA, United States  
An engineered β-galactosidase with improved stability and cross-correction for the potential treatment of GM1 gangliosidosis via AAV gene therapy

**Moderated Q&A**  
**Shababa T. Masoud, Asuka Inoue, Andrew Hedman, and Charu Reddy**

9:00 AM  **Stephanie Cherqui**  
University of California San Diego  
La Jolla, CA, United States  
Phase 1/2 clinical trial of autologous hematopoietic stem and progenitor cell (HSPC) gene therapy for cystinosis

**Shyam Ramachandran**  
Sanofi  
Waltham, MA, United States  
AAV-ARSA-mediated gene replacement for the treatment of metachromatic leukodystrophy

**Mathews Adera**  
AVROBIO, Inc.  
Cambridge, MA, United States  
The Guard1 clinical trial - A first in-human, phase 1/2 study evaluating AVR-RD-02, a hematopoietic stem cell (HSC) gene therapy for Gaucher disease: Preliminary safety, pharmacodynamic and clinical efficacy results from the subjects observed for up to 24 months post-infusion

**Maria L. Escolar**  
Forge Biologics  
Grove City, OH, United States  
First-in-human phase 1/2 trial of intravenous FBX-101 following hematopoietic stem cell transplantation increases GALC activity, supports brain development, and improves motor function in patients with infantile Krabbe disease: RESKUE clinical trial

**Moderated Q&A**  
**Stephanie Cherqui, Shyam Ramachandran, Mathews Adera, and Maria L. Escolar**

10:00 AM  **Break & Exhibits**

10:30 AM  **Raphael Schiffmann**  
4D Molecular Therapeutics  
Emeryville, CA, United States  
Cardiac effects of 4D-310 in adults with Fabry disease in a phase 1/2 clinical trial: Functional, quality of life, and imaging endpoints in patients with 12 months of follow up

**Russell Gotschall**  
M6P Therapeutics  
St. Louis, MO, United States  
M021: rhGAA with optimal glycosylation profile containing very high levels of bis-phosphorylated N-glycans clears accumulated glycogen and rapidly normalizes muscle strength in treated Pompe disease mice
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<td>Developing treatments for rare diseases on a shoestring: The Batten disease (CLN1) enzyme replacement therapy experience</td>
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<td>Michael H. Gelb</td>
<td>A glimpse into the feasibility of next generation sequencing for newborn screening of lysosomal and other diseases with second-tier biochemical assays as part of the screening process</td>
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<td>Raphael Schiffmann, Russell Gotschall, Ana C. Puhl, and Michael H. Gelb</td>
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<td>AZ-3102 significantly increases survival and decreases neuroinflammation in a mouse model of Sandhoff disease</td>
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<td>Small molecule inhibition of glycogen synthase 1 restores autophagolysosomal and metabolic pathway dysfunction in a mouse model of Pompe disease</td>
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<td>RTB-lectin facilitates the distribution of enzymes across the blood-brain-barrier and correction in the MPS IIIA mouse model</td>
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<td>Centyrin-targeted glycogen synthase-1 siRNA conjugates: A novel therapeutic modality for the treatment of Pompe disease</td>
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<td>Nagy Habib</td>
<td>Drugging transcription factors with small activating RNAs: A novel approach for enhancing bone marrow therapy for monogenic rare diseases</td>
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**11:30 AM Break, Exhibits and Satellite Symposia**

**1:00 PM Kyle Landskroner**
Azafaros AG
Basel, Switzerland
AZ-3102 significantly increases survival and decreases neuroinflammation in a mouse model of Sandhoff disease

**2:00 PM Nagy Habib**
MiNA Therapeutics Ltd
London, United Kingdom
Drugging transcription factors with small activating RNAs: A novel approach for enhancing bone marrow therapy for monogenic rare diseases
Sunday, February 26, 2023: Late-Breaking Science
Moderators: Elizabeth Braunlin, Roberto Giugliani, and Rebecca Ahrens-Nicklas

8:00 AM

**Li Ou**
Genemagic Bio
Agoura Hills, CA, United States

A meta-analysis of 39 AAV clinical trials for lysosomal diseases: Immunogenicity, toxicity, and durability

**Lucas Tricoli**
Children's Hospital of Philadelphia
Philadelphia, PA, United States

Improved gene therapy for metachromatic leukodystrophy

**Andrés Felipe Leal**
Pontificia Universidad Javeriana
Bogotá D.C., Colombia

Assessment of an iron oxide-coupled CRISPR/nCas9 gene editing in mucopolysaccharidoses type IVA mouse model

**Chester B. Whitley**
University of Minnesota
Minneapolis, MN, United States

The PS Gene-editing (PSG) System for treatment of lysosomal diseases

**Moderated Q&A**

**Li Ou, Lucas Tricoli, Andres Leal, and Chester B. Whitley**

9:00 AM

**Can Ficicioglu**
The Children's Hospital of Philadelphia
Philadelphia, PA, United States

RGX-121 gene therapy for the treatment of neuronopathic mucopolysaccharidosis type II (MPS II): interim analysis of data from the first in human study

**Kelly George**
Sanofi
Cambridge, MA, United States

Anti-mouse-TfR-GAA fusion proteins for the treatment of Pompe disease targeting the central nervous system and peripheral tissues

**Julie C. Ullman**
Maze Therapeutics
South San Francisco, CA, United States

Results from a first in human study of MZE001, an orally bioavailable inhibitor of glycogen synthase 1 and potential substrate reduction therapy for Pompe disease

**Benedikt Schoser**
Ludwig-Maximilians-Universität München
Munich, Germany

Long-term efficacy and safety of cipaglucosidase alfa/miglustat in ambulatory patients with Pompe disease: A phase III open-label extension study (ATB200-07)

**Moderated Q&A**

**Can Ficicioglu, Kelly George, Julie C. Ullman, and Benedikt Schoser**

10:00 AM

Break

10:15 AM

**Xiomara Rosales**
Neurogene Inc.
New York, NY, United States

Evidence from a study of CLN5 -/- sheep supporting dose escalation in an ongoing clinical trial of NGN-101 in pediatric patients with CLN5 Batten disease

**Patricia I. Dickson**
Washington University in St. Louis
Saint Louis, MO, United States

Intraventricular recombinant human N-acetylgalcosamine-6-sulfatase corrects lysosomal storage in mucopolysaccharidosis type IIID mice

**Eric H. Zanelli**
Allievex Corporation
Boston, MA, United States

Tralesinidase alfa modifies the course of Sanfilippo syndrome type B

**Akos Herzeg**
Center for Maternal-Fetal Precision Medicine, UCSF
San Francisco, CA, United States

A phase 1 clinical trial of in utero enzyme replacement therapy for lysosomal disorders: Interim results

**Moderated Q&A**

**Xiomara Rosales, Patricia I. Dickson, Eric H. Zanelli, and Akos Herzeg**

11:15 AM

**WORLDSymposium 2023 Adjourns**

*The Preliminary Program is subject to change without notice. Any updates to the program will be posted on the website: worldsymposia.org*