# WORLD*Symposium* 2016
# Program

## Monday, February 29

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| **1:00 – 5:00** | **Pre-Conference Symposium** | Emerging Trends: State of the art for experts<br>*(Registration required)* |
| **6:00** | **Satellite Symposium**<br>*Supported by PTC Therapeutics* | MPS I: New horizons and opportunities for change<br>*(This session not available for CME credit)* |

## Tuesday, March 1, 2016

## Basic Science I — Co-Chairs: Walter Low, Danuta Krotoski, Gregory Grabowski

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| **6:30** | **Satellite Symposium**<br>*Supported by BioMarin Pharmaceutical, Inc.* | Optimizing Treatment in Morquio A: Capturing multi-domain impact through patient-directed outcomes<br>*(This session not available for CME credit)* |
| **7:50** | **Chester B. Whitley**<br>University of Minnesota<br>Minneapolis, MN, United States | Welcome and Opening Remarks |
| **8:00** | **Emil Kakkis**<br>Ultragenyx Pharmaceutical<br>Novato, CA, United States | WORLD*Symposium* 2016 Award for Innovation and Accomplishment |
| **8:30** | **Chrissa Dwyer**<br>University of California San Diego<br>La Jolla, CA, United States | Lysosomal degradation of heparan sulfate is required for normal development of the neural circuitry |
| **8:45** | **Camila de Aragao**<br>CHU Sainte-Justine Mother and Child University Hospital Center<br>Montreal, QC, Canada | Synaptic dysfunction in Sanfilippo syndrome type C |
| **9:00** | **Vincent Puy**<br>CHU Amiens, Centre de Biologie Humaine<br>Amiens, France | Alteration of cerebral iron metabolism in Sanfilippo syndrome |
| **9:15** | **S. Pablo Sardi**<br>Genzyme, a Sanofi company<br>Framingham, MA, United States | Glucosylceramide synthase inhibition reduces α-synuclein pathology and improves cognition in murine models of synucleinopathy<br>*(This session not available for CME credit)* |
| **9:30** | **Mia Horowitz**<br>Tel Aviv University<br>Ramat Aviv, Israel | Presence of mutant GBA allele leads to ER stress and development of Parkinson's disease |
| **9:45** | **Yvonne L. Latour**<br>National Institutes of Health<br>Bethesda, MD, United States | Development of isogenic human cerebral organoids with beta-galactosidase deficiency |
| **10:00** | **Break & Exhibits** | |
| **10:15** | **Manoj K. Pandey**<br>Cincinnati Children's Hospital Medical Center<br>Cincinnati, OH, United States | Immune cells attack and neurodegeneration in Gaucher disease |

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| **10:30** | **Debora Bertholdo**<br>DAPI - Diagnóstico Avançado por Imagem<br>Curitiba, Brazil | Structural changes in the brain of patients with Gaucher disease |
| **10:45** | **Volkan Seyrantepe**<br>Izmir Institute of Technology<br>Izmir, Turkey | Deletion of sialidase NEU3 causes progressive neurodegeneration in Tay-Sachs mice |
| **11:00** | **Andreas Schaaf**<br>Greenovation Biotech GmbH<br>Freiburg, Germany | Moss−aGal: preclinical evaluation of a plant made enzyme replacement for Fabry disease<br>*(This session not available for CME credit)* |
| **11:15** | **Jin-Song Shen**<br>Baylor Research Institute<br>Dallas, TX, United States | Sortilin expression and uptake of α-galactosidase A: a general mechanism of endocytosis in Fabry disease cell types |
| **11:30** | **Lunch** | Council of Patient Advocates (COPA) lunch meeting<br>or *Lunch Satellite Symposium supported by Ultragenyx*<br>or Lunch on-your-own<br>*(Lunch sessions not available for CME credit)* |

## Basic Science II — Co-Chairs: Scott McIvor, Rashmi Gopal-Srivastava

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| **1:00** | **Takahiro Tsukimura**<br>Meiji Pharmaceutical University<br>Kiyose, Japan | Anti-α-galactosidase A antibodies and serum-mediated inhibition in Fabry disease |
| **1:15** | **Derrick T. Deming**<br>University of Massachusetts Amherst<br>Amherst, MA, United States | The molecular basis of Pompe disease: crystal structure of acid alpha-glucosidase |
| **1:30** | **Nina Raben**<br>National Institutes of Health<br>Bethesda, MD, United States | Pompe disease: from pathophysiology to therapy and back again |
| **1:45** | **Richard Steet**<br>University of Georgia<br>Athens, GA, United States | Cathepsin-mediated alterations in TGF-β related signaling underlie the cartilage and bone defects associated with impaired lysosomal targeting |
| **2:00** | **Zhirui Jiang**<br>The University of Adelaide<br>Adelaide, Australia | Reduced chondrocyte proliferation and hypertrophy contribute to delayed endochondral bone formation in murine mucopolysaccharidosis VII |
| **2:15** | **Alessandra d'Azzo**<br>St.Jude Children's Research Hospital<br>Memphis, TN, United States | Pathogenic cascade downstream of NEU1 regulated lysosomal exocytosis |
| **2:30** | **Jonathan H. LeBowitz**<br>BioMarin Pharmaceutical, Inc.<br>Novato, CA, United States | Utilizing activity assays and population-wide allele frequencies to assess the contribution of novel mutations in NAGLU to MPS IIIB incidence<br>*(This session not available for CME credit)* |
| **2:45** | **Break & Exhibits** | |
| **3:00** | **Maria Fuller**<br>SA Pathology<br>North Adelaide, Australia | Manipulation of regional brain bis(monoacylglycero)phosphate in the MPS I mouse by dietary fatty acid supplementation |
| **3:15** | **Kanut Laoharawee**<br>University of Minnesota<br>Minneapolis, MN, United States | AAV9 mediated correction of iduronate-2-sulfatase deficiency in the central nervous system of mucopolysaccharidosis type II mice |

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| **3:30** | **Kanagaraj Subramanian**<br>The Scripps Research Institute<br>La Jolla, CA, United States | Quantitative analysis of the proteome response to histone deacetylase inhibitor in Niemann-Pick disease |
| **3:45** | **Li Ou**<br>University of Minnesota<br>Minneapolis, MN, United States | ZFN-mediated correction of murine MPS I model by expression of the human IDUA cDNA from the albumin “safe harbor” locus |
| **4:00** | **Richie Khanna**<br>Amicus Therapeutics<br>Cranbury, NJ, United States | Co-administration of the pharmacological chaperone AT2221 with a proprietary recombinant human acid alfa-glucosidase leads to greater plasma exposure and substrate reduction compared to alglucosidase alfa<br>*(This session not available for CME credit)* |
| **4:15** | **Mustafa A. Kamani**<br>University Health Network<br>Toronto, ON, Canada | Reduced glucocerebrosidase activity improves acid ceramidase deficient mice |
| **4:30** | **Poster Reception & Presentation** | *(Poster session not available for CME credit)* |
| **6:30** | **Dinner Satellite Symposium**<br>*Supported by Amicus Therapeutics, Inc.* | The Many Faces of Lysosomal Disease: A Global Perspective<br>*(This session not available for CME credit)* |

## Wednesday, March 2, 2016

### Translational Research I — Co-Chairs: Jill Morris, Raphael Schiffmann

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| **6:30** | **Breakfast Satellite Symposium**<br>*Supported by Shire* | Discovery and Research Platforms in Rare Disease<br>*(This session not available for CME credit)* |
| **7:50** | **Chester B. Whitley**<br>University of Minnesota<br>Minneapolis, MN, United States | Announcements |
| **8:00** | **Christopher P. Austin**<br>National Institutes of Health<br>Bethesda, MD, United States | Keynote Address:<br>Catalyzing translational innovation |
| **8:30** | **Lalitha Belur**<br>University of Minnesota<br>Minneapolis, MN, United States | Intranasal gene delivery of AAV9 iduronidase: a non-invasive and effective gene therapy approach for prevention of neurologic disease in a murine model of mucopolysaccharidosis type I |
| **8:45** | **Tammy Kielian**<br>University of Nebraska Medical Center<br>Omaha, NE, United States | Adeno-associated virus 9 gene therapy for juvenile neuronal ceroid lipofuscinosis |
| **9:00** | **Walter L. Acosta**<br>BioStrategies LLC<br>State University, AR, United States | Lectin-mediated delivery of α-L-iduronidase: a novel approach for MPS I enzyme replacement therapy<br>*(This session not available for CME credit)* |
| **9:15** | **Elma Aflaki**<br>NIH/NHGRI<br>Bethesda, MD, United States | iPSC-derived dopaminergic neurons from patients with Gaucher disease and Parkinsonism demonstrate the potential of a new glucocerebrosidase chaperone |
| **9:30** | **Allison Bradbury**<br>University of Pennsylvania<br>Philadelphia, PA, United States | Natural history study and preliminary assessment of therapies in canine globoid cell leukodystrophy |

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| **9:45** | **Haiyan Fu**<br>Research Institute at Nationwide Children's Hospital<br>Columbus, OH, United States | Functional benefits of systemic rAAV9-HIDS gene delivery in MPS II mouse model<br>*(This session not available for CME credit)* |
| **10:00** | **Break & Exhibits** | |
| **10:15** | **Behzad Najafian**<br>University of Washington<br>Seattle, WA, United States | Podocyte globotriaosylceramide (GL-3) content in male adult patients with Fabry disease reduces following 6-12 months of treatment with migalastat |
| **10:30** | **Baodong Sun**<br>Duke University School of Medicine<br>Durham, NC, United States | New perspectives for ERT in Pompe disease: extending the action of the enzyme to cytosolic targets |
| **10:45** | **Mark Tarnopolsky**<br>McMaster University<br>Hamilton, ON, Canada | Exosome-mRNA and exosome-protein therapy for Niemann-Pick disease type C |
| **11:00** | **Rasa Ghaffarian**<br>University of Maryland<br>College Park, MD, United States | ICAM-1 targeting by direct conjugation enhances gastrointestinal transcytosis and encapsulation enables gastric protection and controlled released for oral enzyme delivery |
| **11:15** | **Sang-oh Han**<br>Duke University Medical Center<br>Durham, NC, United States | Minimum effective dose for immune tolerance induction with an adeno-associated virus vector in Pompe disease |
| **11:30** | **Lunch Break** | Lunch on-your-own, or Lunch Satellite Symposium s*upported by Shire International*<br>*(Lunch session not available for CME credit)* |

## Translational Research II — Co-Chairs: Danilo Tagle, Dolan Sondhi

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| **1:00** | **Rachel L. Manthe**<br>University of Maryland<br>College Park, MD, United States | Enhanced lysosomal enzyme delivery across the blood-brain barrier by modulating the valency of ICAM-1-targeted nanocarriers |
| **1:15** | **Adeel Safdar**<br>McMaster University<br>Hamilton, ON, Canada | Exosome-mRNA (EXERNA) therapy for Pompe disease<br>*(This session not available for CME credit)* |
| **1:30** | **Heather L. Gray-Edwards**<br>Auburn University<br>Auburn University, AL, United States | Long term survival after gene therapy in a feline model of Sandhoff disease |
| **1:45** | **Alia Ahmed**<br>University of Minnesota<br>Minneapolis, MN, United States | Association of physical symptom score (PSS) with age and cognitive measures in attenuated mucopolysaccharidosis types I, II and VI |
| **2:00** | **Mika Aoyagi-Scharber**<br>BioMarin Pharmaceutical Inc.<br>Novato, CA, United States | Time- and dose-dependent normalization of pathological lysosomal storage and biochemistry in the mucopolysaccharidosis IIIB (MPS IIIB, Sanfilippo B) mouse model by intracerebroventricular enzyme replacement therapy with BMN 250, a NAGLU-IGF2 fusion protein<br>*(This session not available for CME credit)* |
| **2:15** | **Lauren C Boudewyn**<br>Albert Einstein College of Medicine<br>Bronx, NY, United States | Assessment of n-butyl-deoxynojirimycin as a therapeutic option for mucolipidosis type IV |

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| **2:30** | **Russell DeKelver**<br>Sangamo BioSciences<br>Richmond, CA, United States | ZFN-mediated in vivo genome editing results in supraphysiological levels of human iduronate 2-sulfatase and phenotypic correction in a murine MPS II model<br>*(This session not available for CME credit)* |
| **2:45** | **Break & Exhibits** | |
| **3:00** | **Anita Grover**<br>BioMarin Pharmaceutical, Inc.<br>Novato, CA, United States | Intracerebroventricular administration of BMN 250 to cynomolgus monkeys results in elevated tissue levels and superior biodistribution in the central nervous system in comparison to intravenous delivery<br>*(This session not available for CME credit)* |
| **3:15** | **Zoheb B. Kazi**<br>Duke University<br>Durham, NC, United States | Prophylactic immune modulation in infantile Pompe disease using low-dose methotrexate induction: a safe, inexpensive, widely accessible, and efficacious strategy |
| **3:30** | **Yedda Li**<br>Washington University in St. Louis<br>Saint Louis, MO, United States | Combination therapy increases lifespan and improves clinicobehavioral performance in the murine model of globoid cell leukodystrophy |
| **3:45** | **Aaron Meadows**<br>Research Institute at Nationwide Children's Hospital<br>Columbus, OH, United States | Functional correction of mucopolysaccharidosis I in adult mice by a systemic rAAV9-IDUA gene delivery |
| **4:00** | **Angela Schulz**<br>University Medical Center Hamburg-Eppendorf<br>Hamburg, Germany | Intracerebroventricular cerliponase alfa (BMN 190) in children with CLN2 disease: Interim results from a phase 1/2, open-label, dose-escalation study |
| **4:15** | **Gizely N. Andrade**<br>Albert Einstein College of Medicine<br>Bronx, NY, United States | Multisensory processing in lysosomal disorders: a behavioral and high-density electrophysiology investigation in Niemann-Pick disease type C and cystinosis |
| **4:30** | **Poster Reception & Presentation** | *(Poster session not available for CME credit)* |
| **6:30** | **Dinner Satellite Symposium**<br>*CME Satellite Sponsored by MedIQ. Supported by an educational grant from Sanofi Genzyme* | How Early is Early? When to Start ERT and Other Considerations for Optimizing Treatment of Fabry Disease<br>*(Satellite session available for CME credit through MedIQ)* |

## Thursday, March 3, 2016

### Clinical Trials I — Co-Chairs: Stephen Groft, Elsa Shapiro

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| **6:30** | **Breakfast Satellite Symposium**<br>*Supported by BioMarin Pharmaceutical, Inc.* | Recognizing the signs of CLN2 disease – emerging evidence for a paradigm shift in CLN2 diagnosis<br>*(This session not available for CME credit)* |
| **7:50** | **Chester B. Whitley**<br>University of Minnesota<br>Minneapolis, MN, United States | Announcements |
| **8:00** | **Barbara K. Burton**<br>Ann & Robert H. Lurie Children's Hospital<br>Chicago, IL, United States | Newborn screening for lysosomal diseases in Illinois |

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| **8:15** | **Renuka P. Limgala**<br>Lysosomal and Rare Disorders Research and Treatment Center<br>Fairfax, VA, United States | Selective large scale screening for lysosomal disorders in minority groups shows higher incidence rates |
| **8:30** | **Arunabha Ghosh**<br>St. Mary's Hospital<br>Manchester, United Kingdom | IDUA mutational profile and genotype-phenotype correlations in mucopolysaccharidosis type I |
| **8:45** | **Hernan Amartino**<br>Hospital Universitario Austral<br>Buenos Aires, Argentina | New measure to assess severity of MPS II: the disease severity score |
| **9:00** | **Nathan J. Rodgers**<br>University of Minnesota<br>Minneapolis, MN, United States | Thirty year follow-up in Hurler syndrome after hematopoietic cell transplantation: the University of Minnesota experience |
| **9:15** | **Christian J. Hendriksz**<br>Salford Royal Foundation NHS Trust<br>Manchester, United Kingdom | Impact of long-term elosulfase alfa treatment on pulmonary function in patients with Morquio syndrome type A |
| **9:30** | **Paul R. Harmatz**<br>UCSF Benioff Children's Hospital Oakland<br>Oakland, CA, United States | Impact of elosulfase alfa in patients with Morquio syndrome type A who have limited ambulation: an open-label, phase 2 study |
| **9:45** | **Deborah Elstein**<br>Shaare Zedek Medical Center, affiliated with the Hebrew University-Hadassah Medical School<br>Jerusalem, Israel | Therapeutic goals and normal clinical values achieved within 4 years of initiating velaglucerase alfa in treatment-naïve patients with Gaucher disease in phase 3 studies |
| **10:00** | **Break & Exhibits** | |
| **10:15** | **Timothy M. Cox**<br>University of Cambridge<br>Addenbrooke's Hospital<br>Cambridge, United Kingdom | Four-year follow-up from the ENCORE trial: a randomized, controlled, non-inferiority study comparing eliglustat to imiglucerase in patients with Gaucher disease type 1 stabilized on enzyme replacement therapy |
| **10:30** | **Patrick B. Deegan**<br>Addenbrooke's Hospital<br>Cambridge, United Kingdom | Risk factors for fracture in imiglucerase-treated Gaucher disease type 1 patients in the ICGG Gaucher Registry |
| **10:45** | **Magy Abdelwahab**<br>Cairo University Pediatric Hospital<br>Cairo, Egypt | Long-term follow up and sudden unexpected death in Gaucher disease type 3 in Egypt |
| **11:00** | **Ari Zimran**<br>Shaare Zedek Medical Centre<br>Jerusalem, Israel | Long-term efficacy and safety results of taliglucerase alfa through 5 years in adult treatment-naïve patients with Gaucher disease |
| **11:15** | **Gerald Cox**<br>Sanofi Genzyme<br>Cambridge, MA, United States | Functional performance in patients with late-onset Tay-Sachs and Sandhoff diseases |
| **11:30** | **Lunch Break** | *Lunch Satellite Symposium supported by Sanofi Genzyme*<br>or lunch on-your-own<br>*(Lunch session not available for CME credit)* |

## Clinical Trials II — Co-Chairs: James Cloyd, Ari Zimran

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| **1:00** | **Luciana Giugliani**<br>Hospital de Clínicas de Porto Alegre<br>Porto Algre, Brazil | Disease duration and survival in Brazilian Niemann-Pick disease type C patients: Preliminary data on potential impact of miglustat |

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| **1:15** | **Forbes D. Porter**<br>National Institutes of Health<br>Bethesda, MD, United States | Phase 1/2 evaluation of intrathecal 2-hydroxypropyl-β-cyclodextrin for the treatment of Niemann-Pick disease, type C1 |
| **1:30** | **Christine Dali**<br>Department of Clinical Genetics, Rigshospitalet<br>Copenhagen, Denmark | Intrathecal delivery of recombinant human arylsulfatase A in children with late-infantile metachromatic leukodystrophy |
| **1:45** | **Loren Pena**<br>Duke University<br>Durham, NC, United States | Phase 1 exploratory efficacy of the novel enzyme replacement therapy neoGAA in treatment-naïve and alglucosidase alfa-treated late-onset Pompe disease patients |
| **2:00** | **Mark Friedman**<br>Alexion Pharmaceuticals, Inc.<br>Lexington, MA, United States | Safety findings from 3 trials of treatment with sebelipase alfa in children and adults with lysosomal acid lipase deficiency<br>*(This session not available for CME credit)* |
| **2:15** | **Simon A. Jones**<br>Manchester Centre for Genomic Medicine, St Mary's Hospital, Central Manchester Foundation Trust, University of Manchester<br>Manchester, United Kingdom | Effect of sebelipase alfa on survival and liver function in infants with rapidly progressive lysosomal acid lipase deficiency: 2-year follow-up data |
| **2:30** | **Robert J. Desnick**<br>Icahn School of Medicine at Mount Sinai<br>New York, NY, United States | Evolution of cardiac pathology in type 1 classic Fabry disease: progressive cardiomyocyte enlargement leads to increased cell death and fibrosis, and correlates with severity of ventricular hypertrophy |
| **2:45** | **Break & Exhibits** | |
| **3:00** | **Franklin K. Johnson**<br>Amicus Therapeutics<br>Cranbury, NJ, United States | Comparison of integrated white blood cell alpha-galactosidase A activity exposure between every-other-day orally administered migalastat and biweekly infusions of agalsidase beta or agalsidase alfa<br>*(This session not available for CME credit)* |
| **3:15** | **Derralynn Hughes**<br>University College London<br>London, United Kingdom | Novel treatment for Fabry disease: IV administration of plant derived alpha-GAL-A enzyme safety and efficacy interim report |
| **3:30** | **Patricio Aguiar**<br>Centro Hospitalar Lisboa Norte<br>Lisbon, Portugal | Urinary type VI collagen: better than albuminuria to identify incipient Fabry nephropathy |
| **3:45** | **David G. Warnock**<br>UAB<br>Birmingham, AL, United States | Anti-proteinuric therapy and Fabry nephropathy; factors associated with preserved kidney function during agalsidase-beta therapy |
| **4:00** | **Dau-Ming Niu**<br>Taipei Veteran General Hospital<br>Taipei, Taiwan | Revisited later-onset cardiac type Fabry disease: cardiac damages progressed in silence, the experiences from an extremely high prevalent area, Taiwan |
| **4:15** | **Suma P. Shankar**<br>Emory University School of Medicine<br>Atlanta, GA, United States | Eye findings in Fabry disease and correlation with disease severity |
| **6:00** | **Banquet and Award Ceremony** | *(Not available for CME credit)* |