

Interactive clinical cases

Transforming patient outcomes for neurodegenerative LSDs through early diagnosis

Thursday, February 6, 2025

3:45-4:45 PM PST

Palm Foyer outside the Exhibit Hall (second level)

Manchester Grand Hyatt San Diego, CA, USA



Prof. de los Reyes and Prof. Gissen will present real-world clinical cases in an interactive discussion with the audience. They will focus on the diagnostic journeys and differential diagnoses of patients with pediatric neurodegenerative lysosomal storage disorders (LSDs), including CLN2 (neuronal ceroid lipofuscinosis type 2) disease. The key topics of this session are the importance of genetic testing to identify patients early, as well as the impact of early diagnosis on patient outcomes.

Grasp the opportunity to share your opinion and ask questions during this expert theater!

Biographies

Emily C. de los Reyes, MD

Nationwide Children’s Hospital, The Ohio State University, Columbus, OH, USA

Emily C. de los Reyes is a clinical professor of Pediatrics and Neurology at The Ohio State University College of Medicine. She is a pediatric neurodevelopmental specialist at Nationwide Children’s Hospital, serves as director for the Batten’s disease center of excellence, and is the lead neurodevelopmental specialist of the Neurodevelopmental clinic, Prader-Willi and Rett syndrome center of excellence. She is also the neurology subspecialty chair of the Autism Treatment Network at Nationwide Children’s.

Prof. de los Reyes is the primary investigator for the Batten’s disease research programs, and is the lead investigator for the natural history studies for Batten’s disease. Her research focuses on the development of therapeutic approaches for neurodegenerative diseases that affect the central nervous system (CNS), particularly Batten’s disease. Other research interests include autism, La Crosse encephalitis and other rare mitochondrial and genetic disorders.

Paul Gissen, MBChB, PhD, FRCPCH

Great Ormond Street Hospital for Children, NHS Foundation Trust, London, UK

Paul Gissen is a clinical professor of Pediatric Metabolic Medicine at UCL Great Ormond Street Institute of Child Health, and honorary consultant Pediatric Metabolic Diseases at Great Ormond Street Hospital (GOSH) for Children NHS Foundation Trust. He is a National Institute for Health and Care Research (NIHR) senior investigator and leads the Gene, Stem and Cellular Therapies Theme at the NIHR GOSH Biomedical Research Centre. He is also a UK chief investigator for a number of industry and academic sponsored clinical trials of novel therapies.

Prof. Gissen obtained his medical degree from the University of Glasgow and trained in pediatrics at Manchester, Sheffield and Birmingham Children’s Hospitals specializing in inherited metabolic disorders. He undertook his PhD at Birmingham University where he identified genetic causes of several rare pediatric diseases. His research interests are in developing novel gene-based therapies for rare pediatric metabolic disorders.

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