Center for Rare Disease Therapy

World-Renowned Experts | Novel Therapies | Hope for Families

RARE DISEASE DAY 2024 POSTER SESSION

Our Posters and Presenters

February 29, 2024 12 pm - 2pm
UPMC Children's Hospital of Pittsburgh Atrium

Nadene Henderson, MS, CGC
More frequent dosing with agalsidase beta – an update to our Fabry cohort

Ashley Lahr, MS, CGC and Emily Stuebing, MS, CGC
One Center's Experience of Infants Identified with Variants of Uncertain Significance During Newborn Screening For Lysosomal Diseases

Jirair K. Bedoyan, MD, PhD, FACMG
Are asymptomatic carriers of OTC always asymptomatic? A multicentric retrospective study of risk using the UCDC longitudinal study database;
Progress towards novel target-based small molecule therapeutics for pyruvate dehydrogenase complex deficiency due to specific recurrent E1α amino acid replacements

Robert D. Nicholls, D.Phil.
Rationale synthetic design of a broadly expressed NRF1-array mini-promoter for application to disease modeling and gene therapy

Anokhi Kashiparekh, BS, BA
Multi-omics approach to characterize the TANGO2 function: dance partners and potential therapies;
Investigating a novel PNPT1 variant in Amish population

Kelly Schooping Tripi, MS, CGC
Congenital Corneal Opacification in 22q Deletion Syndrome

Hannah Scanga, MS, CGC
Calcified Sclero-Choroidal Choristomas in Mosaic RASopathies

Eduardo Vieira Neto, MD, PhD
Functional and in silico analyses of variants found in mitochondrial trifunctional protein deficiency patients

Keaton Solo, BS, PhD Candidate
Medium-chain acyl-carnitines target muscular phenotypes of long-chain fatty acid oxidation disorders

Shakuntala Basu, MSc, PhD
A Novel Muscle VLCAD/LCAD Double Knock Out Mouse Model Explores VLCAD Deficiency Induced Skeletal Muscle Myopathy and Its Therapies

Xuejun Zhao, MD
Long-term correction of very long-chain acyl-CoA dehydrogenase deficiency in Acadvl-/- mice using AAV gene therapy

Anuradha Karunanidhi, MS
Lysine hyposuccinylation in human MCAD deficient fibroblast cells alleviated with heptanoic and medium branched-chain fatty acids and in Acadm-/- mice with triheptanoin

Our Posters and Presenters (continued)

Bianca Seminotti, MSc, PhD
Mavodelpar, A Selective Peroxisome Proliferator-activated Receptor Delta (PPARδ) Agonist, Improves Cellular Bioenergetics in Fibroblasts from Patients with Primary Mitochondrial Myopathy and Complex I Deficiency

Deepa Rajan, MD
A conditional mouse model of GEMIN5 neurodevelopmental disorder with motor dysfunction and cerebellar atrophy;
Nerve conduction studies in Krabbe disease: effect of hematopoietic stem cell transplantation (HSCT)

Alyssa Powers, BS
Structure and functional studies of TANGO2 protein

Oluwaseun Akinyele, PhD and Dwi U. Kemaladewi, PhD
Characterizing the phenotypic abnormalities of a mouse model of Snyder-Robinson Syndrome: for therapeutic development

Yonne Menezes, PhD and Dwi U. Kemaladewi, PhD
Where immunology meets genetics: A snapshot of immunophenotypes in LAMA2-CMD pathophysiology

Xuejun Zhao, MD and Dwi U. Kemaladewi, PhD
Upregulation of LAMA1 using CRISPR activation as a therapeutic approach for LAMA2-deficient congenital muscular dystrophy

Ruvi Ranatunga BS, BA and Dwi U. Kemaladewi, PhD
Overview of phenotyping assays to study rare neuromuscular and neurological diseases

Marie Johnson, BS and Dwi U. Kemaladewi, PhD
Development of a miniaturized CRISPR activation for a single AAV delivery method for LAMA2-CMD

Al-Walid Mohsen, PhD
New class of anaplerotic compounds ameliorates substantial loss of lysine succinylation in propionyl-CoA carboxylase deficient cells

Joshua Owens, MD
The Phenotypic Spectrum of the Cornelia de Lange-like "Alazami-Yuan Syndrome": A Case Report of the 7th Diagnosed Patient and Review of the Literature;

Early initiation of celecoxib slows but does not prevent symptom progression in a female with primary hypertrophic osteoarthropathy;

A Dual Diagnosis of Okur-Chung Neurodevelopmental Syndrome and Becker Muscular Dystrophy Delineates the Lower Limits of Neurodevelopmental Functioning Attributable to Muscular Dystrophy;

Arginine, Glycine, and Creatine Supplementation Improves Symptoms in a Female with SLC6A8 Creatine Transporter Deficiency;

Expanding the phenotypic spectrum of RAB11B-related disorder with a case report of the oldest reported patient;

Mosaic Turner syndrome presenting with early-onset inflammatory bowel disease;

Lateral Meningocele Syndrome without Lateral Meningoceles: A Case Report to Expand the Phenotype;

Long-Term Renal Transplant Success is Possible in Hypoparathyroidism, Sensorineural Deafness, and Renal Dysplasia Syndrome: A Familial Case Series

Ashley Lahr, MS and Evgenia Sklirou, MD, FACMG
Atypical presentation of central precocious puberty in a patient with RHOA-related disorder