POSTER PRESENTATIONS

Posters board dimensions should be <=3'10"x3'10" (46"x46"116x116cm)
Posters may be mounted between 1:00 and 6:00PM on Sunday, April 3. and removed by 10:30AM on Wednesday, April 6.
See listing of all posters, below.

Oral presentations — Selected Abstracts of Broad Interest
These abstracts were selected for oral presentation by the Program Committee from all submitted for poster presentation:
Curtis Coughlin  The genotypic spectrum of classic nonketotic hyperglycinemia due to mutations in GLDC and AMT

Matthew Olson  Quantitation of acyl-coenzyme A and acyl-carnitine in primary human hepatocytes from propionic acidemia patients

Parthasarathy Sonaimuthu  Functional characterization of NAGS protein using gene therapy

Ilona Tkachyova  Novel approach for the treatment of six mucopolysaccharidoses by targeting NDST1

Yudong Wang  Characterizing the molecular architecture of mitochondrial energy metabolism apparatus and its importance to clinical pathophysiology

Travel Award Poster Presentations
(5-minute presentation at each poster from travel award winners attended by SIMD Founders' Award Committee)

Poster #7  Joseph T. Alaimo, Ph.D.  Baylor College of Medicine, Houston, TX
Poster #38  Carmen Diez Fernandez, Ph.D.  University Children's Hospital Zurich, Zurich, Switzerland
Poster #44  Marta Friegeni, M.D.  University of Utah, Salt Lake City, UT
Poster #60  Hyu In Kim, Ph.D. Candidate  University of Pennsylvania, Philadelphia, PA
Poster #70 *withdrawn*  Ashwini Maratha, Ph.D.  National Centre for Inherited Metabolic Disorders, Temple Street Children's University Hospital, Dublin, Ireland
Poster #116  Nathanial Tolman, B.A.  Undiagnosed Disease Program NHGRI/NIH, Bethesda, MD

Travel Award Recipients and SIMD Hyperion Fellow
(Short oral presentations)

Oscar Aubi  Phenylalanine hydroxylase screening. Pharmacological chaperones for phenylketonuria treatment
Jirair K. Bedoyan  Diagnosis of genetically unresolved subjects with functional pyruvate dehydrogenase complex deficiencies using advanced genomic technologies with functional confirmation
Sean Froese  Structural insights into the MMACHC-MMADHC protein complex involved in vitamin B12 trafficking
Young Joon Kwon  Flunarizine rescues short lifespan of CLN3 knockout Caenorhabditis elegans model of Batten disease
Lisa Pan  Neurometabolic disorders: potentially treatable abnormalities in patients with treatment refractory depression and suicidal behavior
Zarazuela Zolkipli Cunningham  The diagnostic utility of aerobic exercise testing in mitochondrial myopathies
Didem Demirbas (SIMD Hyperion Fellow)  iPS cell-based modeling of galactosemia
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