

Center for Rare Disease Therapy

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2ND ANNUAL

ANNUAL RARE DISEASE DAY POSTER SESSION

February 28, 2025

John G. Rangos Sr. Research Center, UPMC Children's Hospital of Pittsburgh

1. ***Elamipretide restores mitochondrial function in trifunctional protein deficiency mice and human fibroblasts.*** Eduardo Vieira Neto*, Meicheng Wang, Austin J. Szuminsky, Shakuntala Basu, Xuejun Zhao, Anuradha Karunanidhi, Yudong Wang, and Jerry Vockley.
2. ***Development of Anticipatory Guidance Materials for Families Attending an Interdisciplinary Prader-Willi syndrome Clinic: A Quality Improvement Project.*** Ann E. E. Johansson PhD, DNP, CRNP, FNP-BC*, Cynthia A. Danford PhD, CRNP, PPCNP-BC, CPNP-PC, FAAN
3. ***Lessons learned – Transition from research studies to clinical care.*** Nadene Henderson*, Emily Stuebing, Ashley Lahr, Raelynn Forsyth, Damara Ortiz.
4. ***Comparison of the efficacy of an AAV9.hVL vector, synthetic ACADVL-/- mRNA, and triheptanoin in a very long chain acyl-CoA (VLCAD) deficient mouse model.*** Shakuntala Basu*, Xue-Jun Zhao, Bianca Seminotti¹, Clinton Van't Land, Al-Walid Mohsen, and Jerry Vockley
5. ***Cardiac MRI findings among Fabry patients undergoing higher intensity therapy.*** Emily Stuebing, MS, CGC*, Steven Hopkins, MD, Timothy Wong, MD MS FAHA, Nadene Henderson, MS, CGC, Ashley Lahr, MS, CGC, Raelynn Forsyth, MD, FAAP, FACMG, Gavin Hickey, MD, Emilie Kurtz, DO, Damara Ortiz, MD, FAAP, FACMG
6. ***Exposure to infection increases inflammation and glucose metabolism in ganglioside-deficient Microglia.*** Sivakama Bharathi*, Venkatesh Subramanyam, Radha Gopal and Eric Goetzman
7. ***Evaluation of experimental mitochondrial ROS scavengers on their content in propionic and methylmalonic acidemias cell models.*** Anuradha Karunanidhi*, Al-Walid Mohsen, Peter Wipf, Jerry Vockley
8. ***TANGO2 expression in mouse and human tissues.*** Phillip Garretson*, Alyssa Powers, Lina Ghaloul-Gonzalez.
9. ***Sphrintzen Goldberg Syndrome in Siblings of Consanguineous Parents: A Case Series.*** Leen Alkalbani MD*, Hoda Abdel-Hami MD
10. ***Trimetazidine improves protein succinylation and bioenergetics, and decreases reactive oxygen species content in cells from patients with fatty acid oxidation disorders.*** Al-Walid Mohsen PhD*, Anuradha Karunanidhi MS, and Jerry Vockley MD, PhD
11. ***Medium-chain acyl-CoA dehydrogenase substrate binding site: a pharmacophore target for MCAD mutant chaperone therapy.*** Al-Walid Mohsen PhD*, Anuradha Karunanidhi MS, and Jerry Vockley MD, PhD
12. ***Unveiling novel insights into the pathophysiology of TANGO2 deficiency disorder using integrated functional and multiomic studies.*** Anokhi Kashiparekh*, Alyssa Powers, Al-Walid Mohsen, Jerry Vockley, Lina Ghaloul-Gonzalez
13. ***Expanding the Phenotype of EBF3-related Disorders – A Case of Pierre Robin Sequence with Cleft Palate.*** Kate Cioffi MS*, John Wang MD PhD, Linford Williams MS, Leslie KW Cyprych MS MPH, Prerna Venner MS, Evgenia Sklirou MD

* Presenting author