Amanda Helip-Wooley, Ph.D.

STEM Learning Transformation Coordinator
University of South Florida
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EDUCATION AND TRAINING

Diplomate in Clinical Biochemical Genetics 2005 - 2015
American Board of Medical Genetics and Genomics

Postdoctoral Research Fellow 2003 - 2008
National Human Genome Research Institute, NIH

Ph.D. in Human Genetics 1997 - 2002
Tulane University
Dissertation: Aspects of Lysosome Biogenesis and Gene Expression with Emphasis on the Lysosomal Cystine Transporter Gene, CTNS

B.S. in Zoology with Honors 1993 - 1997
University of Florida

RESEARCH EXPERIENCE

Postdoctoral Research **National Human Genome Research Institute, NIH**
Principal Investigator: William Gahl, M.D., Ph.D.

- Led the Hermansky-Pudlak syndrome cell biology research group in investigating the function of five novel proteins and their role in lysosome-related organelle biogenesis and human disease.
- Uncovered the mistargeting of mutant peroxisomal EHHADH to mitochondria that results in inherited renal Fanconi's syndrome
- Characterized protein trafficking defects in BLOC-2 and BLOC-3 deficient primary human melanocytes.
- Studied the role of Rab small GTPases and their effectors in melanosome biogenesis.
- Utilized confocal fluorescence microscopy and protein biochemistry to uncover the association of HPS3 with clathrin.

Doctoral Research **Human Genetics Program, Tulane University**
Principal Investigator: Jess Thoene, M.D.

- Utilized microarrays to examine gene expression following experimentally induced lysosomal vacuolation.
- Investigated the use of aminoglycosides for premature termination codon read-through of nonsense mutations in cystinosis.

TEACHING EXPERIENCE

Adjunct Biological Sciences Instructor **Hillsborough Community College, FL**
2015 –

- Developed lesson plans and instructed Anatomy and Physiology II.
- Interacted with a diverse student population in lecture, during office hours and via an online learning management system.

Adolescent Program Teacher **Butler Montessori, Darnestown, MD**
2012 – 2014

- Created and implemented a two-year multidisciplinary science curriculum promoting active and collaborative learning in theme based units.
- Completed AMI Montessori Certificate of Adolescent Studies (2012).

VOLUNTEER EXPERIENCE

Special Volunteer 2008 - 2014
National Human Genome Research Institute

Institutional Biosafety Committee Extramural Member 2011 - 2014
American University, Washington, D.C.

Assistant Leader 2011 - 2014
Parent Encouragement Program, Kensington, MD

PROFESSIONAL SOCIETY MEMBERSHIPS

Society for Inherited Metabolic Disorders 2005 - 2009
American Society for Cell Biology 2003 - 2009
American Society of Human Genetics 2001 - 2012

HONORS AND AWARDS

Fellows Award for Research Excellence, National Institutes of Health	2005
Neil Buist Award, Society for Inborn Errors of Metabolism	2005
LEQSF/BORSF Scholarship, Tulane University	1998 - 2002
Florida Scholars Scholarship, University of Florida	1993 - 1997

PUBLICATIONS

Peer-Reviewed Articles

1. Klootwijk E, Reichold M, **Helip-Wooley A**, Tolaymat A, Broeker C, Robinette SL, Reinders J, Peindl D, Renner K, Eberhart K, Assmann N, Oefner PJ, Dettmer K, Sterner C, Schroeder J, Zorger N, Witzgall R, Reinhold SW, Stanescu HC, Bockenhauer D, Jaureguiberry G, Courtneidge H, Hall AM, Wijeyesekera AD, Holmes E, Nicholson JK, O'Brien K, Bernardini I, Krasnewich DM, Arcos-Burgos M, Izumi Y, Nonoguchi H, Jia Y, Reddy JK, Ilyas M, Unwin RJ, Gahl WA, Warth R, Kleta R. Mistargeting of Peroxisomal EHHADH and Inherited Renal Fanconi Syndrome. *N Engl J Med* 370(2):129-38, 2014.
2. Cullinane AR, Yeager C, Dorward H, Carmona-Rivera C, Wu HP, Moss J, O'Brien KJ, Nathan SD, Meyer KC, Rosas IO, **Helip-Wooley A**, Huizing M, Gahl WA, Gochuico BR. Dysregulation of Galectin-3: Implications for Hermansky-Pudlak Syndrome Pulmonary Fibrosis. *Am J Respir Cell Mol Biol* 50(3):605-13, 2014.
3. Huizing M, Pederson B, Hess RA, Griffin A, **Helip-Wooley A**, Westbroek W, Dorward H, O'Brien KJ, Golas G, Tsilou E, White JG, Gahl WA. Clinical and cellular characterization of Hermansky-Pudlak Syndrome Type-6. *J Med Genet* 46:803-810, 2009.
4. Rouhani FN, Brantly ML, Markello TC, **Helip-Wooley A**, O'Brien K, Hess R, Huizing M, Gahl WA, Gochuico BR. Alveolar Macrophage Dysregulation in Hermansky-Pudlak Syndrome Type-1. *Am J Respir Crit Care Med* 180:1114-1121, 2009.
5. Stanescu H, Wolfsberg TG, Moreland RT, Ayub MH, Erickson E, Westbroek W, Huizing M, Gahl WA, **Helip-Wooley A**. Identifying putative promoter regions of Hermansky-Pudlak syndrome genes by means of phylogenetic footprinting. *Ann Hum Genet* 73: 422-428, 2009.
6. Westbroek W, Tuchman M, Tinloy B, De Wever O, Vilboux T, Hertz JM, Hasle H, Heilmann C, **Helip-Wooley A**, Kleta R, Gahl WA. A novel missense mutation (G43S) in the switch I region of Rab27A causing Griscelli syndrome *Mol Genet Metab*. 94: 248-254, 2008.
7. Nazarian R, Huizing M, **Helip-Wooley A**, Starcevic M, Gahl WA, Dell'Angelica EC. An immunoblotting assay to facilitate the molecular diagnosis of Hermansky-Pudlak syndrome. *Mol Genet Metab* 93: 134-44, 2008.
8. Westbroek W, Adams D, Huizing M, Koshoffer A, Dorward H, Parkes J, **Helip-Wooley A**, Kleta R, Tsilou E, Duvernay P, Digre K, Creel D, White JG, Boissy RE,

Gahl WA. Chediak-Higashi Syndrome: Severity of cellular defect correlates with genotype and clinical phenotype. *J Invest Derm* 127: 2674-2677, 2007.
9. **Helip-Wooley A**, Westbroek W, Dorward H, Koshoffer A, Huizing M, Boissy RE, Gahl WA. Improper trafficking of melanocyte-specific proteins in Hermansky-Pudlak syndrome type-5. *J Invest Derm* 127:1471-1478, 2007.
10. Huizing M, Parkes JM, **Helip-Wooley A**, White JGM, Gahl WA. Platelet alpha granules in BLOC-2 and BLOC-3 subtypes of Hermansky-Pudlak syndrome. *Platelets* 18:150-157, 2007.
11. Schreyer-Shafir N, Huizing M, Anikster Y, Nusinker Z, Bejarano-Achache I, Maftzir G, Resnik L, **Helip-Wooley A**, Westbroek W, Gradstein L, Rosenmann A, Blumenfeld A. A new genetic isolate with a unique phenotype of syndromic oculocutaneous albinism: clinical, molecular, and cellular characteristics. *Hum Mutat* 27:1158, 2006.
12. **Helip-Wooley A**, Westbroek W, Dorward H, Mommaas M, Boissy RE, Gahl WA, Huizing M. Association of the Hermansky-Pudlak syndrome type-3 protein with clathrin. *BMC Cell Biol* 6:33, 2005.
13. Griffin AE, Cobb BR, Anderson PD, Claassen DA, **Helip-Wooley A**, Huizing M, Gahl WA. Detection of hemizygosity in Hermansky-Pudlak syndrome by quantitative real-time PCR. *Clin Genet* 68:23-30, 2005.
14. Boissy RE, Richmond B, Huizing M, **Helip-Wooley A**, Zhao Y, Koshoffer A, Gahl WA. Melanocyte-specific proteins are aberrantly trafficked in melanocytes of Hermansky-Pudlak syndrome-type 3. *Am J Pathol* 166:231-240, 2005.
15. Gwynn B, Martina JA, Bonifacino JS, Sviderskaya EV, Lamoreux ML, Bennett DC, Moriyama K, Huizing M, **Helip-Wooley A**, Gahl WA, Webb LS, Lambert AJ, Peters LL. Reduced Pigmentation (rp), a Mouse Model of Hermansky-Pudlak Syndrome, Encodes a Novel Component of the BLOC-1 Complex. *Blood* 104:3181-3189, 2004.
16. Kleta R, Romeo E, Ristic Z, Ohura T, Stuart C, Arcos-Burgos M, Dave MH, Wagner CA, Camargo S, Inoue S, Matsuura N, **Helip-Wooley A**, Bockenhauer D, Warth R, Bernardini I, Visser G, Eggermann T, Lee P, Chairoungdua A, Jutabha P, Babu E, Nilwarangkoon S, Anzai N, Kanai Y, Verrey F, Gahl WA, Koizumi A. Mutations in *SLC6A19*, encoding B^0AT1, cause Hartnup disorder. *Nat Genet* 36:999-1002, 2004.
17. Huizing M, Hess R, Dorward H, Claassen DA, **Helip-Wooley A**, Kleta R, Kaiser-Kupfer MI, White JG, Gahl WA. Cellular, Molecular and Clinical Characterization of Patients with Hermansky-Pudlak Syndrome Type 5. *Traffic* 5:711-722, 2004.
18. **Helip-Wooley A**, Thoene JG. Sucrose-induced vacuolation results in increased expression of cholesterol biosynthesis and lysosomal genes. *Exp Cell Res* 292:89-100, 2004.
19. Park MA, **Helip-Wooley A**, Thoene JG. Lysosomal cystine storage augments apoptosis in cultured cultured human fibroblasts and renal tubular epithelial cells. *J Am Soc Nephrol* 12:2878-2887, 2002.
20. **Helip-Wooley A**, Park MA, Lemons RM, Thoene JG. Expression of CTNS Alleles: Subcellular localization and aminoglycoside correction *in vitro. Mol Genet Metab* 75:128-133, 2002.

Book Chapters

21. **Helip-Wooley A**, Kleta R, Gahl WA. Lysosomal Free Sialic Acid Storage Disorders: Salla Disease and ISSD, in: *Lysosomal Storage Disorders* (eds. Barranger J and Cabrera M), Springer, New York, NY, 2007.
22. Kleta R, **Helip-Wooley A**, Gahl WA. Cystinosis, in: *Lysosomal Storage Disorders* (eds. Barranger J and Cabrera M), Springer, New York, NY, 2007.

Invited Reviews

23. Huizing M, **Helip-Wooley A**, Westbroek W, Gunay-Aygun M, Gahl WA. Disorders of lysosome-related organelle biogenesis: clinical and molecular genetics. *Annu Rev Genomics Hum Genet* 9: 359-386, 2008.

Platform Presentations

24. **Helip-Wooley A**, Westbroek W, Dorward H, Held P, Ayub M, Boissy R, Huizing M, Gahl WA. The function of BLOC-2 in lysosome-related organelle biogenesis. American Society of Human Genetics Meeting. October 12, 2006; New Orleans, LA.

25. **Helip-Wooley A**, Westbroek W, Dorward H, Held P, Ayub M, Boissy R, Huizing M, Gahl WA. Improper trafficking of melanocyte-specific proteins in Hermansky-Pudlak syndrome type-5. PanAmerican Society for Pigment Cell Research. September 9, 2006; Cincinnati, OH.

26. **Helip-Wooley A**, Dorward H, Westbroek W, Stanescu H, Hess R, Boissy R, Huizing M, Gahl WA. Unraveling lysosome-related organelle biogenesis through the cell biology of Hermansky-Pudlak syndrome. Society for Inherited Metabolic Disorders. March 7, 2005; Asilomar, CA.

27. **Helip-Wooley A**, Westbroek W, Dorward H, Boissy R, Gahl WA, Huizing M. Localization and clathrin binding of the Hermansky-Pudlak syndrome type 3 protein. PanAmerican Society for Pigment Cell Research. June 25, 2004; Newport Beach, CA