

**ACMG Foundation NEWS**  
**For Immediate Release**

ACMG Media Contact: Kathy Beal, MBA ACMG Media Relations Director  
301-238-4582; kbeal@acmg.net

**Anne O'Donnell Luria, MD, PhD, of Boston Children's Hospital, Harvard Medical School, Mari Mori, MD of Duke University, and Laurie Robak MD, PhD of Baylor College of Medicine are Presented with the Pfizer/ACMG Foundation Clinical Genetics Combined Residency for Translational Genomic Scholars Fellowship Award**

BETHESDA, MD—March 25, 2015 | Anne O'Donnell Luria, MD, PhD, of Boston Children's Hospital, Mari Mori, MD of Duke University, and Laurie Robak MD, PhD of Baylor College of Medicine were honored as the 2015-2016 recipients of the Pfizer/ACMG Foundation Clinical Genetics Combined Residency for Translational Genomic Scholars Fellowship Award at the ACMG 2015 Annual Clinical Genetics Meeting in Salt Lake City, Utah.

The objective of this fellowship is to provide an in-depth clinical research training experience at a premier medical center with expertise and significant clinical volume in the area of biochemical genetics, including lysosomal storage diseases, as well as in therapeutics and clinical trials involving patients with these and other metabolic diseases and, thereby, to increase the number of medical geneticists with interest, knowledge, and expertise in this area.

This Award grants \$75,000 per year to the three recipients selected by the ACMG Foundation through a competitive process and will provide for the sponsorship of one year of the trainee's clinical genetics subspecialty in translational genomics following residency.

Dr. O'Donnell Luria received her MD and PhD at Columbia University, New York, and is currently completing a combined residency in Pediatrics and Medical Genetics at Boston Children's Hospital, Boston MA. "I am honored to receive the Pfizer/ACMG Foundation Translational Genomics Scholars Fellowship Award. I appreciate the support from Pfizer and the ACMG Foundation to gain additional training in biochemical genetics. I am grateful for the excellent training environment provided by wonderful clinicians, staff, and families that I have had the pleasure to work and train with at Harvard Medical School, Boston Children's Hospital, and Massachusetts General Hospital. This fellowship supports my efforts to begin a research program looking at transcriptional and epigenetic variation in lysosomal storage disease, with an aim of identifying new biomarkers of disease and potential therapeutic targets." Her research during the award period will focus on diagnosis and management of infants, children and adults with inborn errors of metabolism and the impact of epigenetic alterations.

Dr. Mori received her MD at Nagasaki University School of Medicine, Nagasaki, Japan, and MS in Biomedical Informatics at University of Pittsburgh, Pittsburgh PA. She is currently completing her Medical Biochemical Genetics Fellowship at Duke University Medical Center, Durham, NC,

-more-

after completing a General Genetics Residency at Nationwide Children's Hospital/Ohio State University in Columbus, OH. Her research during the award period will focus on the identification of modifier genes from carefully phenotyped patients with Pompe disease at Duke University Medical Center. "I am deeply honored to be one of the recipients of the Pfizer/ACMG Foundation award. The award allows me to extend my biochemical genetics training to investigate factors that affect variable phenotypes of Pompe disease, under the guidance of Dr. Priya Kishnani, Professor of Pediatrics Division Chief, Medical Genetics at Duke University. The research would lead to a better understanding of prognostication of rare Mendelian diseases, and would have clinical impacts, especially for asymptomatic patients with a lysosomal disease detected by newborn screening."

Dr. Robak received her MD and PhD at University of Rochester, Rochester NY, and is currently completing her combined residency in Pediatrics and Medical Genetics at Baylor College of Medicine. Her research during the award period will focus on exploring the links between Lysosomal Storage Disorders and Parkinson's disease at Baylor College of Medicine. "I am honored to be a recipient of the 2015 Pfizer/ACMG Foundation Fellowship Award. This June, I will be completing my combined Pediatrics/Medical Genetics residency at Baylor College of Medicine. This prestigious award will allow me to continue my research investigating potential links between lysosomal storage disorders and adult-onset neurodegenerative disorders. My project will be under the guidance of Dr. Joshua Shulman, Assistant Professor of Neurology and Molecular & Human Genetics at Baylor College of Medicine. By providing critical support during my transition from residency to junior faculty, this fellowship will promote my successful career development as a physician-scientist."

"With all of the advances in genomics, the Pfizer/ACMG Foundation Clinical Genetics Combined Residency for Translational Genomic Scholars presents an important opportunity to develop new approaches to diagnosis and treatment of genetic disorders. This fellowship is therefore a key component of our initiative to train physician scientists to be leaders in translational research in medical genomics," said Bruce R. Korf, MD, PhD, FACMG, president of the ACMG Foundation.

**Note to editors:** To arrange interviews with experts in medical genetics, contact Kathy Beal, MBA, ACMG Director of Public Relations at [kbeal@acmg.net](mailto:kbeal@acmg.net) or 301-238-4582.

To learn more about the ACMG Foundation for Genetic and Genomic Medicine visit [www.acmgfoundation.org](http://www.acmgfoundation.org)

*-End-*

**The ACMG Foundation for Genetic and Genomic Medicine ([www.acmgfoundation.org](http://www.acmgfoundation.org))**, a 501(c)(3) nonprofit organization, is a community of supporters and contributors who understand the importance of medical genetics and genomics and genetic counseling in healthcare. Established in 1992, the ACMG Foundation supports the American College of Medical Genetics and Genomics' mission to "translate genes into health" by raising funds to promote the profession of medical genetics and genomics to medical students, to fund the training of future medical geneticists, to support best-practices and tools for practicing physicians and laboratory directors, to promote awareness and understanding of our work in the general public, and much more.