



ACMG Foundation for
Genetic and Genomic Medicine
Better Health Through Genetics™

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**The ACMG Foundation for Genetic and Genomic Medicine Announces
the First Nine Recipients of the Shire Training Fellowship Awards**

BETHESDA, MD – March 22, 2017 | The first nine recipients of the **ACMG Foundation/Shire Medical Genetics Residency and Fellowship Training Awards Program** were announced during the American College of Medical Genetics and Genomics (ACMG) 2017 Annual Clinical Genetics Meeting in Phoenix, Arizona. The nine awards were the first given after Shire made a \$1.65 million commitment in 2016 to support the training of future medical geneticists.

Bruce R. Korf, MD, PhD, FACMG, president of the ACMG Foundation said, “This grant represents an important investment in the future of medical genetics and genomics. It will not only help to address the shortage of medical geneticists in the US, but also will help seed the next generation of clinical and laboratory geneticists, who in turn will provide both service and training. The return on investment here will be huge.”

The **ACMG Foundation/Shire Medical Genetics Residency and Fellowship Training Awards Program** provides funding for three different specialties: Clinical Genetics Residencies (2 years), Clinical Laboratory Fellowship (2 years) and Medical Biochemical Genetics Subspecialty Fellowships, (1 year).

Clinical Genetics Residencies:

1. Jonah David Bardos, MD, MBE, University of Miami

Dr. Bardos received his MD from the Albert Einstein College of Medicine in 2013. Since then, he has been participating in an Internship and Residency program in Obstetrics and Gynecology at the Ichan School of Medicine at Mount Sinai in New York. Interested in women's health and better patient care, Dr. Bardos received the Arnold P. Gold Foundation Humanism and Excellence in Teaching Award in 2015 – one of the highest honors given to residents. Each Gold Foundation recipient is awarded for their humanistic medical care within the program. In addition, he received his Master's in Bioethics from Cardozo-Einstein. His research interests include studying causes of miscarriages, pathologic implantation and optimal endometrial preparation for frozen embryo transfer. At the University of Miami, Dr. Bardos will be furthering his work in the area of clinical genetics including prenatal diagnosis, prenatal genetic counseling and gynecologic cancer genetics.

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2. Kerri Bosfield, MD, Children's National Medical Center

After receiving her MBBS from the University of West Indies in Kingston, Jamaica, Dr. Bosfield served as the Senior House Officer at Princess Margaret Hospital in Nassau, Bahamas from 2013-2014 where she served the local population providing healthcare to both children and adults. Afterwards, she began her Pediatric Residency at Rutgers New Jersey Medical School in 2014. As part of the community based program she has participated in various educational activities, such as conference presentations, and community outreach at local schools and adolescent safety houses. Both experiences have given her diverse health experience in pediatrics which has impacted her current future goals. Currently, she is working on a Quality Improvement Project focusing on outpatient pain management in Sickle cell disease with hopes to improve patient's awareness of their condition and provider awareness about managing sickle cell pain as an outpatient. As a recipient of the award Dr. Bosfield will be participating in the Medical Genetics Fellowship at Children's National Medical Center. Her interests include newborn screening, in particular creating a newborn screening program in a developing country, and inspiring medical students to choose a career in medical genetics.

3. Paul Hillman, MD, PhD, University of Texas Health Science Center at Houston (UTHealth)

At a young age, Dr. Hillman aspired to become a medical geneticist. Currently, he is a second year resident physician at The McGovern Medical School of The University of Texas Health Science Center at Houston (UTHealth) where he works in the Division of Medical Genetics, Department of Pediatrics. In 2015, he received his MD and PhD degrees in Medical Sciences from the Texas A&M University Health Science Center, with his dissertation focusing on the genetic regulation of the gene associated with Angelman syndrome. He has experience working in genetic research including studies of: E. Coli, Drosophila, and Mouse; additionally, he has studied the genetic regulation of the developing central nervous system. At UTHealth, he is interested in studying results generated through next generation sequencing technology searching for the genetic underpinnings of the most severe type of neural tube defect compatible with life, myelomeningocele (MM). Dr. Hillman's project will examine the genetics of methylation enzymes and a possible link to development of MM. With this training and education, he hopes to gain valuable and instrumental experience that will allow him to grow as a medical professional and achieve his goal of one day being a medical geneticist.

4. The University of North Carolina at Chapel Hill will name a recipient to begin their Clinical Genetics Residency in 2018.

Clinical Laboratory Fellowship:

1. Chelsea Zimmerman, PhD, University of Alabama at Birmingham

After interning at Aptagen, LLC, Dr. Zimmerman was exposed to medical technologies that sparked her interest in becoming a clinical biochemical geneticist. In 2009, she received her BS in Biology with two minors: Chemistry and Anthropology from Millersville University. Now she is gaining laboratory experience at the University of Alabama at Birmingham (UAB) where she has already participated in three research rotations centered on neurological disorders.

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Her clinical experience at UAB's Department of Neurology has provided her with the foundation to pursue her goal of one day working as a geneticist at an academic institution. From her High Performance Liquid Chromatography (HPLC) techniques to analysis experience and education, she hopes to make an impact on science as a clinical biochemical geneticist. "I am excited to receive this award because it allows me to continue my research in a field that is new and exciting for me. It will not only help UAB out with funding, but it will help others out through the research we will be conducting," said Dr. Zimmerman.

Medical Biochemical Genetics Subspecialty:

1. Erin Cooney, MD, FAAP, Baylor College of Medicine

Dr. Cooney is currently completing her fifth year in the combined Pediatrics-Medical Genetics residency at Baylor College of Medicine. Her career goal is to work as a faculty physician in a university hospital setting diagnosing and managing patients with metabolic and other genetic disorders. She also has a strong interest in clinical research and genetics education. Dr. Cooney has gained lab expertise in molecular and biochemical techniques and has worked on cell culture and animal models. As a recipient of the Shire-ACMG award, Dr. Cooney will continue her training with a one year fellowship in Medical Biochemical Genetics. During this fellowship she will be exposed to the diagnosis, management, and coordination of care of patients with inborn errors of metabolism and will work on a research project under the mentorship of Brett Graham, MD, PhD.

2. Noelle Andrea V. Fabie, MD, Detroit Medical Center

Dr. Fabie has been working on her Combined Pediatrics-Medical Genetics Residency at the Children's Hospital of Michigan/Wayne State University, where she has taken care of patients with inborn errors of metabolism. Having been there the past 4 years, she has had the privilege of being able to follow children with inborn errors of metabolism from the time they were diagnosed as newborns until their toddler years, allowing her to learn more about genetic complications and patient care for children. Dr. Fabie received her MD from the University of the Philippines College of Medicine and hopes to refine her skills as a geneticist and learn from innovative medical techniques and research.

3. Kara Pappas, MD, Detroit Medical Center

Dr. Kara Pappas' primary interests are clinical genetics and biochemical genetics. It was during her undergraduate degree education (Behavioral Biology) at Johns Hopkins University that she became interested in genetics. Eager to learn more about the field, she went on to receive her MD from Wayne State University in 2011, which led her to a pediatrics residency at the Los Angeles County-University of Southern California Medical Center where she became involved in several research projects regarding inborn errors of metabolism. She has continued her interest in biochemical genetics during her Medical Genetics residency at the Detroit Medical Center, participating in research projects involving metabolic diseases such as Carnitine palmitoyltransferase II deficiency (CPT-II). During her biochemical fellowship at the Detroit Medical Center, she plans to study mitochondrial function in patients with 22q11.2 deletion syndrome.

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4. Evgenia Sklirou, MD, University of Pittsburgh

In 1999, Dr. Sklirou received her MD from the National and Kapodistrian University of Athens in Greece. Currently, she is in her second year training in Clinical Genetics at the Children's Hospital of Pittsburgh of the University of Pittsburgh Medical Center (UPMC). Starting July 2017, Dr. Sklirou will begin her Medical Biochemical fellowship where she will participate in a research protocol, which will validate a device for more rigorous monitoring of Phenylalanine (PHE) levels in patients with Phenylketonuria (PKU). The goal of this research is to have a system in place that will allow professionals to monitor PHE levels in patients with PKU, resulting in better metabolic control and fewer health complications for patients.

About the American College of Medical Genetics and Genomics (ACMG) and ACMG Foundation

Founded in 1991, ACMG is the only nationally recognized medical society dedicated to improving health through the clinical practice of medical genetics and genomics. The American College of Medical Genetics and Genomics (www.acmg.net) provides education, resources and a voice for nearly 2000 biochemical, clinical, cytogenetic, medical and molecular geneticists, genetic counselors and other healthcare professionals, nearly 80% of whom are board certified in the medical genetics specialties. The College's mission is to develop and sustain genetic initiatives in clinical and laboratory practice, education and advocacy. Three guiding pillars underpin ACMG's work: 1) Clinical and Laboratory Practice: Establish the paradigm of genomic medicine by issuing statements and evidence-based or expert clinical and laboratory practice guidelines and through descriptions of best practices for the delivery of genomic medicine. 2) Education: Provide education and tools for medical geneticists, other health professionals and the public and grow the genetics workforce. 3) Advocacy: Work with policymakers and payers to support the responsible application of genomics in medical practice. *Genetics in Medicine*, published monthly, is the official ACMG peer-reviewed journal. ACMG's website (www.acmg.net) offers a variety of resources including Policy Statements, Practice Guidelines, Educational Resources, and a Find a Geneticist tool. The educational and public health programs of the American College of Medical Genetics and Genomics are dependent upon charitable gifts from corporations, foundations, and individuals through the ACMG Foundation for Genetic and Genomic Medicine (www.acmgfoundation.org).

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