BETHESDA, MD - April 3, 2019 | The ACMG Foundation for Genetic and Genomic Medicine is proud to announce that Sarah Brnich, MD-PhD in-training from the University of North Carolina at Chapel Hill is the recipient of the ACMG Foundation/David L. Rimoin Inspiring Excellence Award.

The David L. Rimoin Inspiring Excellence Award was created in memory of the late Dr. David L. Rimoin, one of the founders of ACMG who passed away in 2012. Dr. Rimoin touched the lives of generations of patients as well as trainees and colleagues. This award is a cash award given to a selected student, trainee, or junior faculty ACMG member whose abstract submission is chosen as a platform presentation during the ACMG Annual Clinical Genetics Meeting.

Ms. Brnich was selected to receive this award for her platform presentation, “Making sense of missense: a pathway-based approach to functional assays for improved clinical variant interpretation.” She is currently an MD-PhD student at the University of North Carolina at Chapel Hill in the laboratory of Dr. Jonathan Berg, MD, PhD within the Department of Genetics. Ms. Brnich is interested in developing methods of classifying germline genetic variants by their functional consequence, with the long-term goal of improving the clinical interpretation of variants of uncertain significance. Her research lies at the intersection of wet lab and clinical applications, with special focus on the incorporation of functional data into the ACMG/AMP guidelines for sequence variant interpretation. This work has resulted in two publications, including a first-author paper in Human Mutation. Ms. Brnich is a contributing member of ClinGen’s Sequence Variant Interpretation (SVI) Workgroup.

“It is such an incredible honor to receive the David L. Rimoin Inspiring Excellence Award. Dr. Rimoin was a giant in the field of medical genetics and a consummate physician-scientist. As an MD/PhD in-training, I am inspired by his example and grateful for the doors he opened for trainees in medical genetics. His legacy will guide me on my path to becoming a better physician, researcher, mentor, and leader. Thank you to the selection committee for this prestigious award,” said Ms. Brnich upon receiving the award.

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“Training remains crucial to the advancement of our field, with increasing opportunities for a new generation of medical geneticists. We recognize in this award Dr. Rimoin’s commitment to training and mentorship and congratulate Ms. Brnich,” said Bruce R. Korf, MD, PhD, FACMG, President of the ACMG Foundation for Genetic and Genomic Medicine.

Ann Garber, Dr. Rimoin’s widow said, “The Rimoin family is delighted to congratulate Ms. Brnich on her exciting research that opens the door to the clinical interpretation of germline genetic variants. We are proud to recognize and support her valuable contributions to medical genetics with the David L. Rimoin Inspiring Excellence Award.”

The ACMG Foundation for Genetic and Genomic Medicine (ACMGF) is a community of supporters and contributors who understand the importance of medical genetics and genomics in healthcare. A nonprofit organization established in 1992, the Foundation supports the American College of Medical Genetics and Genomics (ACMG) mission to “translate genes into health”; to foster charitable giving, promote training opportunities to attract future medical geneticists and genetic counselors, to share information about medical genetics, to sponsor important research and much more.

To learn more and support the ACMGF mission to create “Better Health through Genetics”, please contact Nicole O. Bell, ACMG Foundation Manager, at nbell@acmg.net or 301-718-9604 or visit acmfgfoundation.org.

Note to editors: To arrange interviews with experts in medical genetics, contact Kathy Moran, MBA, ACMG Senior Director of Communications and Public Relations at kmoran@acmg.net.

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