ACMG Foundation for Genetic and Genomic Medicine
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Barry Eisenberg, MS
beisenberg@acmg.net

Ali H. Bereshneh, PhD is the Recipient of the 2025 ACMG Foundation David L. Rimoin Inspiring Excellence Award

BETHESDA, MD – March 19, 2025| The ACMG Foundation for Genetic and Genomic Medicine is proud to present the **ACMG Foundation David L. Rimoin Inspiring Excellence Award** to Ali H. Bereshneh, PhD for his featured platform presentation, "Heterozygous *De novo* variants in *CDKL1* and *CDKL2* cause neuroregressive phenotypes in Human and *Drosophila* and are dominant negative alleles," at the 2025 ACMG Annual Clinical Genetics Meeting in Los Angeles, CA.

Dr. Bereshneh obtained an MSc in Human Genetics from Tehran University of Medical Sciences (TMUS) in 2016 and a PhD in Medical Genetics, with a focus on molecular neurogenetics and rare genetic disorders, from the Faculty of Medical Sciences, TMU, Iran, in 2020. He then served as the Director of the Prenatal Diagnosis and Medical Genetics Lab at the Prenatal Diagnosis and Genetic Research Center at Shiraz University of Medical Sciences in Shiraz, Iran until October 2022. His efforts, under the supervision of distinguished Principal Investigators, led to the establishment of a comprehensive Rare Neurogenetic Disorders Registry for Iran and led to the identification of causative genes/variants in individuals with diverse ultra-rare neurodevelopmental and neuromuscular conditions. Dr. Bereshneh is currently a postdoctoral fellow in the Department of Molecular and Human Genetics at Baylor College of Medicine. He joined the laboratories of Drs. Hugo Bellen and Oguz Kanca in November 2022 and has been studying rare and undiagnosed disorders using sequencing data, bioinformatic analyses, molecular dynamic simulations and especially animal modeling. Their research focuses on demonstrating that variants observed in individuals with rare neurological diseases are at the basis of their phenotypes in order to help end the diagnostic odyssey of these individuals.

"It is a real honor to accept the ACMG Foundation for Genetic and Genomic Medicine David L. Rimoin Inspiring Excellence Award and I am extremely grateful. I would like to express my deepest appreciation to my mentors, Dr. Hugo Bellen and Dr. Oguz Kanca, our collaborators, and the patients and families who participated in our research. Their support has been instrumental in the discovery of unknown human neurological diseases and identification of novel causative variants. I am also grateful for the support I receive from my spouse and our darling one-year-old, Diana. This award highlights the significance of our work, and I am optimistic that the characterization of this new neurodevelopmental syndrome will encourage further collaborative efforts among clinicians, scientists and families to develop personalized therapies for the '*CDKL2* Deficiency Disorder' and related neurodevelopmental syndromes," said Dr. Bereshneh.

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"Ali H. Bereshneh, PhD is an exemplar individual for the 2025 ACMG Foundation David L. Rimoin Inspiring Excellence Award. Dr. Bereshneh's work teaches us more about the importance of clinical, research and family collaborators in expanding our understanding of new disorders," said Nancy J. Mendelsohn, MD, FACMG, president of the ACMG Foundation.

The David L. Rimoin Inspiring Excellence Award was created in memory of the late Dr. David L. Rimoin, one of the founders of the American College of Medical Genetics and Genomics (ACMG) who died in 2012. Dr. Rimoin touched the lives of generations of patients as well as trainees and colleagues. The award is a cash award given to a selected student, trainee or junior faculty ACMG member whose abstract submission was chosen for a platform presentation to be given at the ACMG Annual Clinical Genetics Meeting.

"The Rimoin Family is delighted to present this year's winner of the Inspiring Excellence Award to Dr. Bereshneh, based on his outstanding abstract on the expression of dominant negative alleles in overlapping neurological phenotypes. The quality of his work makes us proud to be able to honor him and wish him continued success in Clinical Genetics and Genomics," said Dr. Ann Garber-Rimoin, Dr. Rimoin's widow.

About the ACMG Foundation for Genetic and Genomic Medicine

The ACMG Foundation for Genetic and Genomic Medicine, a 501(c)(3) nonprofit organization, is a community of supporters and contributors who understand the importance of medical genetics and genomics in healthcare. Established in 1992, the ACMG Foundation supports the American College of Medical Genetics and Genomics (ACMG) mission to "translate genes into health." Through its work, the ACMG Foundation fosters charitable giving, promotes training opportunities to attract future medical geneticists and genetic counselors to the field, shares information about medical genetics and genomics, and sponsors important research. To learn more and support the ACMG Foundation mission to create "Better Health through Genetics" visit acmgfoundation.org.

Note to editors*:* To arrange interviews with experts in medical genetics, contact ACMG Communications Manager, Barry Eisenberg, MS at beisenberg@acmg.net.

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