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Boxun Zhao, PhD is the Recipient of the 2022 ACMG Foundation/ David L. Rimoin Inspiring Excellence Award

BETHESDA, MD – **March 23, 2022** | The ACMG Foundation for Genetic and Genomic Medicine is proud to present the **ACMG Foundation/David L. Rimoin Inspiring Excellence Award** to Boxun Zhao, PhD for his featured platform presentation at the 2022 ACMG Annual Clinical Genetics Meeting, "Discovery and therapeutic implications of pathogenic retroelements in neurodegenerative diseases."

Dr. Boxun Zhao obtained a PhD in Genetics in 2017 from Peking Union Medical College and Tsinghua University in Beijing, China. He joined the labs of Drs. Timothy W. Yu and E. Alice Lee in the Division of Genetics and Genomics at Boston Children's Hospital in 2018 as a postdoctoral fellow. His research interests include: 1) basic research: somatic transposable element (TE) insertions in the human brain and neurological disorders 2) molecular diagnosis: developing a specialized pipeline to capture the full spectrum of genetic variation to resolve un- or half-diagnosed genetic cases, and 3) genomic medicine: developing individualized medicines for patients with diseases that are considered currently too rare for traditional commercial investment. His strong background in human transposon elements and talents in experimental and computational work give him a solid foundation to pursue the long-term goal—understanding the importance and functional impact of TE insertions in human diseases; and translating scientific discoveries into therapeutics. Boxun was awarded the Pilot Project Award in 2018 and the Rare Disease Research Fellowship by the Manton Center for Orphan Disease Research in 2020.

"Congratulations to Dr. Boxun Zhao, the 2022 recipient of the David L. Rimoin Inspiring Excellence Award! Dr. Zhao's work on diagnosis and individualized treatment for rare genetic disorders is an exciting new direction in the study of rare disorders that were so important to Dr. Rimoin throughout his career," said Bruce R. Korf, MD, PhD, FACMG, president of the ACMG Foundation for Genetic and Genomic Medicine.

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.

"I am thrilled to receive the David L. Rimoin Inspiring Excellence Award. A big thanks to my co-mentors Drs. Tim Yu and Alice Lee for mentoring and supporting my research, and all participating patients and their families. I hope our works from genomic diagnosis to the development of individualized medicines could be helpful in navigating a path that can gather scientists, doctors, regulators, patient families, and charitarians to envision a better future for the treatment of rare diseases," said Dr. Zhao upon receiving the award.

Dr. Ann Garber-Rimoin, Dr. Rimoin's widow, said, "Dr. Zhao, the winner of the Rimoin Inspiring Excellence Award, has earned this award based on his outstanding research that examines transposon insertions and their impact on human disease. The Rimoin family is proud to award Boxun Zhao, PhD with the David L. Rimoin Inspiring Excellence Award for 2022."

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 Senior Director of Communications and Public Relations Kathy Moran, MBA at kmoran@acmg.net.

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