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**Isabelle B. Cooperstein, BS is the Recipient of
the 2023 ACMG Foundation/David L. Rimoin Inspiring Excellence Award**

BETHESDA, MD – March 15, 2023 | The ACMG Foundation for Genetic and Genomic Medicine is proud to present the **ACMG Foundation/David L. Rimoin Inspiring Excellence Award** to Isabelle B. Cooperstein, BS for her featured platform presentation at the 2023 ACMG Annual Clinical Genetics Meeting, “Phenotypic Patient Matching to Achieve Diagnoses in Rare Disease Cohorts.”

Isabelle B. Cooperstein, BS, is a third-year PhD candidate in the Department of Human Genetics at the University of Utah in Salt Lake City. She obtained a Bachelor of Science in Molecular Biology with a minor in Mathematics from the University of Wisconsin-Madison in 2017. She was awarded the Hilldale Undergraduate Research Fellowship to complete a senior honors thesis with Dr. Gregory Gauthier to study the mechanisms used by fungi to adapt to temperature. Since joining the lab of Dr. Gabor Marth in 2021, her research interests have shifted to developing software tools for use in the diagnostic process of rare disease patients. Her work focuses on combining the properties of the Human Phenotype Ontology with sequencing data to create a visual and interactive representation of the similarity between patients in the Undiagnosed Disease Network. The long-term goal of this research is to create a tool that aids in providing disease solutions without requiring high-level programming or command-line skills, making it accessible to any member of the clinical team.

“Helping the next generation of genetics professionals find their places in our field was a passion and mission for Dr. Rimoin. We are pleased to recognize Isabelle Cooperstein, a graduate student at University of Utah, as this year’s David L. Rimoin Inspiring Excellence Award,” 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.

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“I am thrilled and honored to have been selected to receive the David L. Rimoin Inspiring Excellence Award. I am extremely grateful to the participating families in the Undiagnosed Disease Network as well as those who helped form this collaboration. I would also like to thank Drs. Gabor Marth and Alistair Ward for mentoring my PhD work and supporting me along this path. I hope that our work will be a valuable addition to the diagnostic process and further encourage the collaboration of clinicians, researchers and families with its accessibility,” said Ms. Cooperstein.

Dr. Ann Garber-Rimoin, Dr. Rimoin’s widow, said, “The Rimoin Family is excited to honor Ms. Cooperstein with the Inspiring Excellence Award to recognize her visionary work on combining software and diagnostic criteria to further advance progress in both the Undiagnosed Disease Network and the Global Alliance for Genomics and Health. Her work provides a path towards a greater understanding of patients who lack a clear diagnosis for the patients themselves, their families, and clinicians.”

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 www.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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