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Geneticist and Pediatrician Dr. Ada Hamosh Receives David L. Rimoin Lifetime Achievement Award in Medical Genetics from the ACMG Foundation for Genetic and Genomic Medicine

BETHESDA, MD – April 13, 2021 | Renowned clinical geneticist and pediatrician Ada Hamosh, MD, MPH, FACMG, has received the 2021 ACMG Foundation for Genetic and Genomic Medicine's **David L. Rimoin Lifetime Achievement Award in Medical Genetics**.

Dr. Hamosh, who is the Dr. Frank V. Sutland Professor of Pediatric Genetics and Clinical Director of the McKusick-Nathans Institute of Genetic Medicine at Johns Hopkins University School of Medicine, is being recognized with this award because of her extraordinary commitment to teaching and mentoring, her remarkable energy and dedication while helping patients and their families, and her leadership of online resources—most notably as the Scientific Director of Online Mendelian Inheritance in Man (OMIM®) and co-creator of PhenoDB and GeneMatcher—that have transformed the use of genetic information in mainstream healthcare worldwide.

"Dr. Ada Hamosh embodies all of the qualities of a David L. Rimoin Lifetime Achievement Awardee," said ACMG Chief Executive Officer Dr. Maximilian Muenke, MD, FACMG. "Humility and service to her patients above all! I have used OMIM for over three decades on a daily basis and have had the privilege and pleasure to work with Ada for over a decade on educating the next generation of leaders in genomic medicine. Ada's imprint on medical genetics residents and fellows at Hopkins and through OMIM around the world is priceless."

"The Rimoin family is grateful for Dr. Hamosh's extraordinary achievements in her research on Mendelian disorders as well as her dedication and commitment to teaching and mentoring junior faculty, fellows and residents," said Dr. Rimoin's widow, Dr. Ann Garber-Rimoin. "David would have been particularly impressed with her establishment of PhenoDB and GeneMatcher to advance gene identification, as they share similar features with the International Skeletal Dysplasia Registry, which enhances the ability to diagnose and characterize skeletal dysplasia disorders. Our family offer whole-hearted congratulations to Dr. Hamosh as the recipient of the David L. Rimoin Lifetime Achievement Award."

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Over the course of her career, Dr. Hamosh has published more than 110 papers on topics ranging from genotype-phenotype correlations in cystic fibrosis, to natural history studies in nonketotic hyperglycemia, to novel disease gene discoveries and new treatments for rare Mendelian disorders. In addition, she has given more than 100 invited lectures, including annual participation at the Jackson Laboratory "Human and Mammalian Genetics and Genomics: the McKusick Short Course" in Bar Harbor, Maine, since 1999.

She currently serves on several international committees representing genome-phenome relationships and phenotype ontologies, including the International Rare Disease Research Consortium, the Human Variome Project, the ClinGen Project, the Matchmaker Exchange, the Gene Curation Coalition, the Global Alliance for Genomic Health, and the Human Genome Organization (HUGO). In her role as Clinical Director overseeing genetic specialty services across Johns Hopkins Medicine, she has mentored numerous medical students, residents, and fellows who are now leaders in their respective fields. She also has the privilege of providing acute and continuity care for patients and their families across multiple generations.

The news that she had received the David L. Rimoin Lifetime Achievement Award came to Dr. Hamosh as a tremendous surprise. "You could have knocked me over," she admitted. "I'm a very behind-the-scenes person, so the idea that I would get this award is truly overwhelming, and that it is named after David Rimoin makes it even more meaningful to me. David Rimoin was such a kind, generous, thoughtful person, just a wonderful mentor, doctor and human being."

"Dr. Hamosh is an extraordinarily accomplished medical geneticist and mentor. Aside from the influence she has had on the lives of the patients she has seen and the trainees she has mentored, there are countless others who have benefitted from her contributions to resources such as OMIM and GeneMatcher, which are practically universally used by practitioners in genetics and genomics," said ACMG Foundation President Bruce R. Korf, MD, PhD, FACMG. "Her dedication, knowledge, and compassion make her an ideal recipient of this award."

Those who nominated Dr. Hamosh for this award made it clear she has earned the admiration and gratitude of so many clinicians, researchers, and patient families who recognize and benefit from her exceptional enthusiasm and tireless work ethic.

"I get to witness almost every day how wonderful she is," wrote Dr. Nara Sobreira, Assistant Professor in the McKusick-Nathans Institute of Genetic Medicine at Johns Hopkins University Medical School, who came to the United States from Brazil in 2006 as a visiting medical genetics resident and worked with Dr. Hamosh on the creation of PhenoDB and GeneMatcher. "Dr. Hamosh was born to take care of people, and the ones that benefit the most of it are her patients. She will do anything to ensure that they get the best possible medical care. Her residents get to learn genetics and metabolics from a 'walking encyclopedia,' and even after they finish training, they can and will call her any day and time to discuss difficult cases."

"As a physician and clinical geneticist, she embodies so many of the qualities of Dr. Rimoin, including humility, empathy, integrity, optimism, and assistance to others," wrote Dr. Jill A. Fahrner, Assistant Professor in the Department of Genetic Medicine at the Johns Hopkins School of Medicine. "She is one of the most empathetic physicians I have ever met, and she will do anything

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to make a difficult situation for a patient's family better. She takes the quality of helping others to another level…I have seen her drive to a store in the evening to purchase a nonformulary medical dietary supplement for a patient whose family didn't have transportation. Despite all of her success, she continues to exude humility."

Ada Hamosh was born in Jerusalem, Israel, in 1960 to parents who were both medical scientists. Her mother, a renowned biochemist, studied human milk and neonatal digestion, and her father was a pulmonologist. Their family came to the United States when she was 5 years old for her mother to begin a 2-year fellowship at NIH, and then they stayed.

Ada and her two sisters chose to pursue medical careers, which Dr. Hamosh credits to the mentorship they received from their parents. "I never thought I couldn't do anything because I never heard that I couldn't," she explained. "We had a mailbox at our house in Bethesda that said, 'Paul Hamosh, MD, Margit Hamosh, PhD, and Ada Hamosh, KID.' So, I just knew that I would have to replace the KID with another doctorate after my name."

Ada studied biology as an undergraduate student at Wesleyan University and completed her medical degree at Georgetown University in 1985. Pediatrics was an immediate attraction. "In medical school we rotated through many hospitals in the DC area, and the variability in the quality of care patients received based on their economic status was just too much for me. I couldn't stand it," she recounted. "I knew I wanted to see a wide variety of patients in a hospital where everyone gets the same exceptional treatment, regardless of their ability to pay. That's what brought me to Hopkins, and that's why I haven't left."

After finishing her pediatrics residency at The Johns Hopkins Hospital in 1988 and a MPH from the Johns Hopkins School of Public Health the year after, she applied for a fellowship in Medical and Biochemical Genetics. It was a decision partly motivated by convenience since it allowed her to stay at Johns Hopkins, where her now-husband, renowned pediatrician and physician scientist Hal Dietz, also worked. Fortunately, clinical genetics turned out to be the perfect choice for her intellectual satisfaction, as well.

"It's an absolutely fantastic field. I can't understand why everyone doesn't want to work in genetics because it's just so interesting, often startling" she said. "I've been doing this a long time now, and every two-to-three years there's a complete paradigm shift where we realize that what we thought we knew was at a minimum woefully incomplete and often dead wrong."

Dr. Hamosh's career has been shaped by some of the best mentors in human genetics including Drs. David Valle, Saul Brusilow, and Barton Childs. She began working in genetics research in the laboratory of Dr. Garry Cutting focused on studies of phenotype-genotype correlations in cystic fibrosis, and then switched to research on the metabolic disease nonketotic hyperglycemia. Ultimately, she decided to pursue a different direction when she joined Dr. Victor McKusick's OMIM team as a subject author editor in 1996. She became the Deputy Scientific Director three years later and assumed leadership of OMIM in 2002, and she has remained at the helm ever since.

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"OMIM is an amazing resource! I have been using it for 32 of the 55 years it has been around, and I was fortunate to overlap with Dr. McKusick for 12 years," she explained. "OMIM is used by clinicians and researchers the world over. Because it started as a catalog of Mendelian genetic disorders, it has always had a clinical focus. My commitment to OMIM is based upon the fact that I cannot practice medical genetics without it. I use it for differential diagnosis, to learn quickly about a disorder or gene, and to link to many other useful resources. Being a part of the small team that the maintains and develops OMIM is very gratifying, and their breadth and depth of knowledge is inspirational." According to Google Analytics, OMIM is used by 2.7 million people around the world annually; it gets more than a million page views per week, and it has about 100 thousand users per week. It contains more than 25 thousand entries that describe more than 16 thousand genes and more than 8 thousand phenotypes.

Dr. Hamosh collaborated with Dr. Sobreira to create PhenoDB in 2012 as a tool that allows clinical researchers around the world to store and share phenotypic and genotypic data on suspected Mendelian diseases. In 2013 she led the creation of GeneMatcher, another online tool that facilitates investigation of genes for which the phenotypes have not yet been identified.

These online resources, along with her teaching and patient caseload, represent a tremendous investment of time and responsibility. Dr. Hamosh approaches all of it with energy that many find to be remarkable. She credits her resilience to the example set by her patients who weather the immense challenges of complex diagnoses and uncertain futures with grace, humor and optimism. "They teach me every day," she shared. "The mom of a young man with heart, kidney, intestinal and intellectual issues that I've cared for these past 30 years always replies, 'You know, I'm so blessed, because it could be much worse.' Honestly, I am so blessed to know this woman and her remarkable son," reflected Dr. Hamosh. "It really helps to put my occasional bad days in perspective."

Maintaining long-term relationships with colleagues, trainees and families is one of the most satisfying aspects of her career, Dr. Hamosh said. "I just love people, and always apply the golden rule—treat everyone the way you would like to be treated. I watched Dr. Rimoin in action. His generosity was legendary. He made everyone around him want to pay it forward. I would be thrilled to be remembered in a similar vein."

The David L. Rimoin Lifetime Achievement Award is the most prestigious award given by the ACMG Foundation. A committee of past presidents of the American College of Medical Genetics and Genomics selects the recipient following nominations, which come from the general membership.

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

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 (ACMG) mission to "translate genes into health." Through its work, the ACMG Foundation fosters charitable giving, promotes training opportunities to attract future

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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.

Founded in 1991, the American College of Medical Genetics and Genomics (ACMG) is the only nationally recognized medical society dedicated to improving health through the clinical practice of medical genetics and genomics and the only medical specialty society in the US that represents the full spectrum of medical genetics disciplines in a single organization. The ACMG is the largest membership organization specifically for medical geneticists, providing education, resources and a voice for more than 2,400 clinical and laboratory geneticists, genetic counselors and other healthcare professionals, nearly 80% of whom are board certified in the medical genetics specialties. ACMG's mission is to improve health through the clinical and laboratory practice of medical genetics as well as through advocacy, education and clinical research, and to guide the safe and effective integration of genetics and genomics into all of medicine and healthcare, resulting in improved personal and public health. Four overarching strategies guide ACMG's work: 1) to reinforce and expand ACMG's position as the leader and prominent authority in the field of medical genetics and genomics, including clinical research, while educating the medical community on the significant role that genetics and genomics will continue to play in understanding, preventing, treating and curing disease; 2) to secure and expand the professional workforce for medical genetics and genomics; 3) to advocate for the specialty; and 4) to provide best-in-class education to members and nonmembers. *Genetics in Medicine*, published monthly, is the official ACMG journal. ACMG's website (www.acmg.net) offers resources including policy statements, practice guidelines, educational programs and a 'Find a Genetic Service' tool. The educational and public health programs of the ACMG are dependent upon charitable gifts from corporations, foundations and individuals through the ACMG Foundation for Genetic and Genomic Medicine.

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