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Clinical Geneticist Dr. Reed Pyeritz Receives
2023 David L. Rimoin Lifetime Achievement Award in Medical Genetics
from the ACMG Foundation for Genetic and Genomic Medicine

BETHESDA, MD — March 15, 2023 | Internationally acclaimed clinical geneticist Reed Pyeritz, MD, PhD, FACMG, is the recipient of the 2023 ACMG Foundation for Genetic and Genomic Medicine's David L. Rimoin Lifetime Achievement Award in Medical Genetics.

Dr. Pyeritz, the William Smilow Professor of Medicine *emeritus* at the University of Pennsylvania Perelman School of Medicine and one of the founders of the American College of Medical Genetics and Genomics (ACMG), is being recognized for his decades of pioneering work on Marfan Syndrome, his commitment to helping patients and their families through outreach and support groups, as well as his extraordinary contributions to furthering the field of medical genetics — including serving as the second president of ACMG.

Over the course of his career, Dr. Pyeritz led many of the seminal studies and clinical trials responsible for extending the lifespan of people with Marfan Syndrome from 40 to 70 years. He has published more than 700 research articles and reviews, helped found the Marfan Foundation, and is the co-editor of the standard textbook in medical genetics, *Principles and Practice of Medical Genetics and Genomics*.

"Reed Pyeritz has been involved with the College since its inception, including serving a term as president. He is a distinguished physician-scientist, and was a close associate of Dr. Rimoin's, especially as one of the co-editors of *Emery and Rimoin's Principles and Practice of Medical Genetics*. His recognition as this year's recipient of the Rimoin Lifetime Achievement Award is therefore especially poignant and highly deserved," said ACMG Foundation President Bruce R. Korf, MD, PhD, FACMG.

"My late husband, David, considered Dr. Pyeritz a trusted friend and colleague, and an exemplary clinician, teacher and researcher," said Dr. Ann Garber, the surviving spouse of David Rimoin, for whom the award is named. "Reed projects genuine honesty, intelligence, and a deep inspiration for Medical Genetics. I still remember observing Dr. Pyeritz teach at a Marfan clinic he held at Johns Hopkins in the mid-1980s. He combined warmth and respect for the patient, while pointing out important clinical features for the residents and fellows."

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Dr. Pyeritz said he is incredibly honored to receive the David L. Rimoin Lifetime Achievement Award.

“David Rimoin was both a role model and a mentor for me from the very beginning of my career,” he said. “To receive this award named after him is absolutely an honor.”

Reed Pyeritz was born in Pittsburgh, Pennsylvania in 1947. In high school, he was drawn to chemistry and, during college at the University of Delaware, he became interested in biochemistry. With the intent of becoming an academic scientist, Pyeritz enrolled in a graduate biochemistry program at Harvard University. His graduate research focused on repetitive sequences of DNA eventually found in many organisms’ genomes.

Then, his graduate advisor invited him across the street to the Boston Lying-in Hospital — now part of Brigham and Women's Hospital — where an obstetrician was about to perform one of the very first amniocenteses ever done in Boston. The experience of watching the procedure, which involved removing a small sample of fetal cells from the fluid surrounding a growing fetus, changed Dr. Pyeritz’s career path.

“I was absolutely fascinated by this idea that you could use what were then very high-tech genetic methods to diagnose an unborn baby,” said Dr. Pyeritz. “I began to wonder how else genetics could be applied to medicine.”

At the time, he lived with five medical students, and when he returned to the house that evening gushing with sudden excitement about medicine, they encouraged him to apply to medical school. So, in 1972, he finished his PhD in biological chemistry and stayed put in Boston — enrolled now at Harvard Medical School and intent on becoming a medical geneticist.

On the first day of his fellowship at the Johns Hopkins Hospital in 1977, Dr. Pyeritz’s career evolved on an even more focused path. He met Victor McKusick, MD, widely considered to be one of the founding fathers of medical genetics. Dr. McKusick quickly steered Dr. Pyeritz down the hallway to the room of a patient diagnosed with Marfan Syndrome, a genetic disease known to affect the body’s connective tissues.

“Victor said, ‘I want you to take care of this patient and his family from now on,’ and so literally from day one of my training, I became obsessed with wanting to learn everything I could about Marfan Syndrome,” remembers Dr. Pyeritz. “I saw all the gaps in knowledge and decided this was an area that was really in need of further research.”

Over the coming years, as a faculty member at the Johns Hopkins University School of Medicine from 1978 to 1993, Dr. Pyeritz co-led the research that associated pathogenic variants in the gene fibrillin-1 (*FBN1*) with Marfan Syndrome. He also led the first trial to show how beta-blockers, a medicine commonly used to reduce blood pressure, can benefit patients with the syndrome, which often leads to aortic dissection and sudden death. And he collaborated with cardiologists and cardiothoracic surgeons to develop prophylactic methods to reduce early deaths from Marfan Syndrome.

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In 1979, Dr. Pyeritz began to wonder how else he could help support the patients he treated, many of whom faced a plethora of medical challenges related to their diagnoses. He had seen the success of patient and family support groups for other genetic diseases and began organizing an informal group for people with Marfan Syndrome. "Dr. Pyeritz gathered several dozen Marfan patients, genetic counselors and a social worker in Dr. Victor McKusick's living room," said Dr. Jacqueline Hecht, FACMG, a clinical geneticist who has long collaborated with Dr. Pyeritz. "Over the course of a year, that meeting resulted in the incorporation of the National Marfan Foundation. Today, this patient support organization has thousands of members and an annual meeting that attracts 600 to 800 people."

His career took Dr. Pyeritz to the Medical College of Pennsylvania from 1993 to 2001 and then the University of Pennsylvania School of Medicine from 2001 until his retirement in 2020. In each role, he balanced research with the clinical care of patients with genetic diseases including not only Marfan Syndrome but other related inherited diseases.

"Dr. Pyeritz is an extremely thoughtful researcher and clinician, and a phenomenal translator of genetic services into practical benefits for patients," said R. Rodney Howell, MD, FACMG, who has known Dr. Pyeritz since they first crossed paths at Johns Hopkins in the 1970s. "He has a very deep understanding and appreciation of medical genetics and has been very intimately and personally involved with people affected by these conditions."

In 1991, Dr. Pyeritz and David Rimoin helped organize a small group of medical geneticists to discuss the possibility of a new professional group. Their efforts led to the founding of ACMG, with David Rimoin serving as the first president. Dr. Pyeritz became a member of the Board of Directors, chaired the Publication Committee that created the journal *Genetics in Medicine*, and was selected in 1994 as the second president of the College. More recently, Dr. Pyeritz served on the Finance Committee and as the ACMG representative to the American Medical Association.

"My work helping establish ACMG and lead the College through its infancy is one of the most rewarding and important roles of my career," said Dr. Pyeritz.

Those who know him know that Dr. Pyeritz has, over the decades, not only flourished in his career but also his personal life. He and his wife Jane Tumpson have been married for 50 years and raised two daughters. During Operation Desert Storm, he served as a Lieutenant Colonel in the Army Reserve Medical Corps. He competed in road races for many years, was a nationally ranked triathlete, and co-held a world record for the 100-man, 100-mile relay. When asked what his secret to balancing everything has been, Dr. Pyeritz laughed.

"I learned to live with very little sleep," he said. "And I'm great at multi-tasking. I remember driving my daughter to elementary school while dictating clinic notes, helping her with her spelling and eating bagels at the same time."

Even in retirement from his official clinical duties, Dr. Pyeritz remains involved in Marfan Syndrome research, training and outreach. He explained that the field has never gotten boring or stopped providing new questions for him to answer.

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"What keeps me stimulated now is that as people with Marfan Syndrome live longer, they're developing new problems we never saw before," said Dr. Pyeritz. "When I have patients that I've followed for thirty years, I want to keep helping them deal with new challenges and issues they may confront."

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 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.

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