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Celebrated Clinical Geneticist Dr. Stephen Cederbaum to Receive 2025 David L. Rimoin Lifetime Achievement Award in Medical Genetics from the ACMG Foundation for Genetic and Genomic Medicine

BETHESDA, MD – March 19, 2025 | Celebrated clinical geneticist Stephen Cederbaum, MD, PhD, FACMG, has been named the recipient of the 2025 ACMG Foundation for Genetic and Genomic Medicine's David L. Rimoin Lifetime Achievement Award in Medical Genetics**.**

A founding member of the Society for Inherited Metabolic Disorders and the American College of Medical Genetics, Dr. Cederbaum is now a distinguished professor emeritus of Psychiatry, Pediatrics and Human Genetics at the University of California, Los Angeles (UCLA). During the height of his career at UCLA, he developed a clinical program in inborn errors of metabolism and, alongside it, became a leading expert on urea cycle disorders, most notably arginase deficiency, a disorder in which the body lacks the final enzyme in the urea cycle and is, therefore, unable to rid itself of excess ammonia. Dr. Cederbaum worked on enzyme therapy for the disorder, which remains a focus of ongoing research.

From his first exposure to arginase deficiency in 1972, Dr. Cederbaum devoted decades to outlining the biochemistry and pathology that underlie the disorder, as well as identifying successful therapies to treat it. He developed animal models that enabled the exploration of gene replacement and other early therapies for arginase deficiency.

"As a physician-scientist, [Dr. Cederbaum] identified the genetic basis and mechanistic consequences of arginase deficiency and the role of arginine metabolism in multiple disease conditions," said Brendan Lee, MD, PhD, chair of Molecular and Human Genetics at Baylor College of Medicine. "As such, he traversed classical biochemistry, mouse genetics, human genetics and clinical research/care. I had the privilege of meeting Dr. Cederbaum when I started my independent career at a meeting at the Urea Cycle Disorders research conference. His scientific insight—and wit—stood out to all, and his style of self-deprecating leadership in science and medicine has likewise been appreciated by both colleagues and trainees."

Dr. Cederbaum did not limit his efforts to academic or clinical practice. He was also a keen advocate for public health, sponsoring a bill in California to include adults with inborn errors of metabolism in an existing Genetically Handicapped Persons Program. Dr. Cederbaum's efforts also spurred the broadening of mandated newborn screenings to include mass spectrometry, and

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he founded the California State Genetics Disease Branch Newborn Screening Guidelines Committee. Amid these accomplishments, he also found time to launch a support group for parents, which is now the ARD1-D Foundation; the organization now supports people with arginase deficiency and their families. He also served as an advisor to the National Urea Cycle Disorders Foundation, as well as mentoring and supporting students and colleagues.

“His work ethic and humility stand out. Despite a long list of achievements, Dr. Cederbaum attributes success to his teams and colleagues. His dedication to patient care and advocacy is exemplary, with significant efforts to improve care in underserved regions, particularly in Latin America,” said Marshall L. Summar, MD, ABP, ABMGG, CEO of Uncommon Cures LLC and Professor of Pediatrics at George Washington University.

Dr. Cederbaum sees his connectedness more simply: "People mattered, and I always understood that one of the joys of clinical practice is just meeting people, getting to know people, the one-on-one," he says. "It may be that I'm an extreme example of someone for whom human interaction is very important, but I also always understood that my patients suffered from terrible disorders and that it really mattered how you interacted with them."

A productive investigator, Dr. Cederbaum has authored or co-authored more than 180 peer-reviewed publications and more than 90 book chapters and reviews alongside his duties as a physician. Those publications include a study detailing the development of a mouse model for arginase deficiency (and gene transfer in those mice), cloning and characterizing the human and mouse types I and II arginase genes, and comparing the properties and functions of different arginases.

“His combination of basic science and clinical research is almost unheard of in today’s environment of ultra-specialization and sets him apart as both a pioneer and role model to the field," said Jerry Vockley, MD, PhD, chief of Genetic and Genomic Medicine and director of the Center for Rare Disease Therapy, UPMC Children's Hospital of Pittsburgh.

Dr. Cederbaum’s research has not only defined the diagnosis and treatment of arginase deficiency but also opened new avenues of understanding the role of arginase activity in endothelial and breast cancer cells. Outside arginase investigations, he is responsible for identifying Letterer-Siwe Disease as an immunodeficiency instead of a malignancy and for defining and developing effective therapies for pyruvate dehydrogenase deficiency, a rare disorder sparked by lactic acid buildup.

Dr. Cederbaum built the foundation for his career with an undergraduate chemistry degree from Amherst College in Massachusetts in 1959, followed by a medical degree from New York University in 1963. A yearlong leave of absence before his MD allowed him to pursue bacterial genetics research at Massachusetts General Hospital. With his doctorate in hand, he completed a two-year internal medicine residency at Washington University, St. Louis, and then studied bacterial biochemistry at the National Institutes of Health.

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He returned to medical genetics with a 1970 fellowship at the University of Washington in Seattle. The following year, he joined UCLA, where he was a contemporary of the late Dr. Rimoin, for whom the current award is named. Dr. Cederbaum spent most of his five-decade career at UCLA, where he remains active today as a board member of the university's Faculty Club.

Like many early-career researchers, Dr. Cederbaum knew the importance of choosing his research specialty. A lifelong lover of literature, he instinctively sought what Robert Frost termed "the road less taken." After meeting a few families impacted by arginase deficiency, he recognized that the disorder was poorly understood and began considering this new research path.

"It was a road less traveled and an area in which I could make unique contributions. I started out having no real insight into how viable a long-term problem it was," he said. "It turned out to be one that could sustain a career for 50 years … so it turned out to be very fortunate, though I didn't have that kind of insight at the time." Dr. Cederbaum's mentees continue to pursue these studies.

Before being honored by the ACMG, Dr. Cederbaum received the Sherman Mellinkoff Award, the UCLA School of Medicine's highest honor; the Edward A. Dickson Emeritus Professorship Award, UCLA; the National Organization for Rare Disorders Rare Impact Award; he was the Arno and Gretel Motulsky Lecturer, University of Washington; and was a distinguished visiting professor at the Universidad National Autónoma de México.

"Dr. Stephen Cederbaum's remarkable contributions to the field of inborn errors of metabolism and newborn screening, with an emphasis on disorders of the urea cycle make him a well-qualified winner of the David L. Rimoin Lifetime Achievement Award," said Dr. Ann Garber-Rimoin, Dr. Rimoin's widow. "Using various forms of gene and mRNA therapy, his research has used both patient and animal models to improve clinical care, and to significantly impactnewborn screening policy. More recently, his research interests have been directed to the pre-symptomatic DNA screening for late-onset autosomal dominant disorders. Beyond his outstanding laboratory and clinical achievements, Dr. Cederbaum is known as a dedicated teacher and mentor, as well as a founder and supporter of patient support groups for patients with arginase deficiency. His kindness and genuine care for his patients further enhance his leadership role for his students and trainees. The Rimoin family is proud to recognize Dr. Stephen Cederbaum's outstanding career, and to honor him with the David L. Rimoin Lifetime Achievement Award."

"Congratulations to Dr. Stephen Cederbaum for this well-deserved award. Dr. Cederbaum's life and work epitomizes the objective characteristics of the Rimoin award. His passion for teaching, mentoring, and care for patients and their families is admirable. He has championed the integration of clinical genomics care into the healthcare system. We are honored to be able to recognize his achievements," said Nancy J. Mendelsohn, MD, FACMG, president of the ACMG Foundation.

Even after 50 years in the field, Cederbaum remains engaged with colleagues and community members. "To me, it's very important [to recognize] that you can always be better, and it's important to make yourself better," he said. "You're never finished with your obligations to society."

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About the David L. Rimoin Lifetime Achievement Award

The David L. Rimoin Lifetime Achievement Award is the highest honor given by the ACMG Foundation. It honors professionals who have made career contributions to medical genetics. Its namesake, Dr. David L. Rimoin, was a pioneer in the field of medical genetics who advanced the understanding of skeletal dysplasia, diabetes and Tay-Sachs disease. A committee of past presidents of the ACMG selects the recipient following nominations 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's mission to create "Better Health through Genetics," visit www.acmgfoundation.org

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