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Acclaimed Clinical Geneticist Dr. Bruce Korf Honored with 2024 David L. Rimoin Lifetime Achievement Award in Medical Genetics from the ACMG Foundation for Genetic and Genomic Medicine

BETHESDA, MD — March 13, 2024 | Renowned clinical geneticist Bruce Korf, MD, PhD, FACMG, has been named the recipient of the 2024 ACMG Foundation for Genetic and Genomic Medicine's David L. Rimoin Lifetime Achievement Award in Medical Genetics.

A past president of both the American College of Medical Genetics (ACMG) and the ACMG Foundation, Dr. Korf currently serves as the Associate Dean for Genomic Medicine and the Distinguished Professor of Genetics at the University of Alabama at Birmingham's (UAB) Heersink School of Medicine. A world-leading neurofibromatosis (NF) expert, he is being honored in part for helping to drive its evolution from clinical diagnosis and surveillance to the possibility of treatment. Dr. Korf's contributions span more than four decades of research, clinical practice and service.

"The David L. Rimoin Lifetime Achievement Award recognizes an individual whose career includes a significant contribution to the field of genetics and genomics and whose personal attributes reflect those of Dr. Rimoin," said ACMG Foundation President Nancy J. Mendelsohn, MD, FACMG. "Dr. Korf is a leader in our field, demonstrating true leadership in defining the medical discipline of genomics. He is a dedicated teacher, championing genetics education and training, and an inspiring and devoted educator. His mentorship has impacted countless clinicians who themselves have had great success. He is an international leading authority as a clinician and scientist on the underlying pathophysiology, diagnosis and clinical care of patients with neurofibromatosis. Dr. Korf is an authentic, kind and ethical human being whose life and career in medical genetics are unparalleled."

Dr. Korf is praised by patients and colleagues alike as a caring and thoughtful physician, a warm-hearted colleague and mentor who encourages students to pursue their own unique contributions and a passionate advocate for the continued integration of medical genetics education into mainstream healthcare.

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"Dr. Korf's extraordinary accomplishments in clinical genetics, neurofibromatosis, and genomics, as well as his dedication to education make him a well deserving winner of the David Rimoin Lifetime Achievement Award. Before he died, David appreciated the opportunity to work with Bruce first-hand in the editing of *Principles and Practice of Medical Genetics* as well as on numerous committees for the ACMG. The Rimoin family congratulates Dr. Korf for his remarkable and productive career," said Dr. Ann Garber, the surviving spouse of David Rimoin, for whom the award is named.

"Bruce Korf has been one of the most influential figures guiding my career, and I suspect there is a long line of clinicians and scientists out there who would say the same," said J. Daniel Sharer, PhD, FACMG, a professor and director of UAB's Genetics Research Division. "Bruce has been and remains one of the world's leading authorities — both as a clinician and a scientist — on the underlying pathophysiology, diagnosis and clinical care of patients with neurofibromatosis."

A prolific researcher, Dr. Korf has authored more than 154 peer-reviewed publications, 92 non-peer-reviewed manuscripts, 60 book chapters and 18 books. "He has had an incredible career in genetics and has made countless contributions to our field in the areas of clinical practice, education and research," said Susan D. Klugman, MD, FACOG, FACMG, who is the current ACMG president, director of reproductive and medical genetics at Montefiore Medical Center and a professor at the Albert Einstein College of Medicine.

These publications include the seminal textbook *Emery and Rimoin's Principles and Practice of Medical Genetics*, which he was invited to co-edit by Drs. Rimoin and Reed Pyeritz, last year's winner of this award. Dr. Korf's research has contributed to providing new choices and hope to people with NF. He has been instrumental in coordinating the complex, multi-site research collaborations needed to ensure adequate participants in clinical trials for this rare disorder. He also strongly advocates that research findings be efficiently woven into current medical practice whenever possible.

"If there's one thing that ties the clinic and the lab together, it's what I describe as a sense of urgency that patients feel," Dr. Korf said. "Although it's a time-consuming and laborious process to get from understanding a mechanism behind NF to producing new therapies, there really is an urgency to do this, given the challenges that people and their families who live with these conditions face."

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Dr. Korf's contributions aren't limited to NF. A champion of rare disease research, he also spearheads UAB's Undiagnosed Diseases Program, now part of the National Institutes of Health's (NIH) Undiagnosed Diseases Network, as well as leading two population-level genomics projects: the All of US Southern Network, part of the NIH-led All of Us Research Program, and the Alabama Genomic Health Initiative, a statewide program. He is also the editor-in-chief of the *American Journal of Human Genetics* and serves as principal investigator of the Neurofibromatosis Clinical Trials Consortium, a Department of Defense-funded research program that allows researchers around the country to pool administrative resources, share findings and recruit study participants. The consortium is working to increase treatment options for persons with NF and is a major source of continued hope for the NF patient community.

Along with championing the partnerships needed to advance large-scale studies of NF — a challenge given the relative rarity of the condition — Dr. Korf has also embraced the calling to offer compassionate care to those who may otherwise have few medical options or providers qualified to treat them.

"I have witnessed his compassion; he's always pushing to get meaningful research data back to physicians and patients so patients can get the diagnosis and care that they need," said Dr. Deeann Wallis, PhD, a UAB geneticist and NF researcher who noted that patients "travel the globe" to consult with Dr. Korf.

In the clinic, "It is immediately obvious why they have gone to such great lengths to see him," she added. "He is always calm and collected, even when patients or families are in emotional turmoil and struggling from a diagnostic odyssey, a new diagnosis, or frantically searching for a cure. Dr. Korf offers people with NF not only expertise but clear, concise distillations of complex situations, offering realistic hope and clearly informing patient expectations."

In the classroom and the lab, Dr. Korf is an enthusiastic mentor and warmly regarded colleague. "He readily introduces me to other leading scientists in the field; they are his friends," Dr. Wallis said. "He is a beloved friend, and I have seen him on the national stage recite a rap-style poem for a colleague who was being honored with a prestigious award; he is endearing."

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With characteristic humility, Dr. Korf said he was surprised to receive the award but felt honored. “It certainly is exciting and validating that your peers recognize the effort you've put in over the years,” he said. “Having an award that bears Dr. Rimoin’s name is also a particular honor and very meaningful, given our many years of collaboration on *Principles and Practice of Medical Genetics* and in the American College of Medical Genetics and Genomics.”

Dr. Korf was first exposed to genetics as a teenager working a summer laboratory job in Buffalo, New York, at the Roswell Park Memorial Institute. He came upon a paper that described chromosome analysis in mice and was intrigued to find that its lead researcher worked at the institute. She helped him learn to do chromosomal analysis as part of his summer project. Before that, he’d had little genetics education in school: “Genetics was always at the back of the book, and we never got to the back of the book!” Dr. Korf said.

As an undergraduate student studying genetics at Cornell University, Dr. Korf continued conducting genetics research, working in the Cytogenetics Laboratory at a hospital near his home each summer. This was early in the era of chromosome banding, offering the opportunity to participate in research that resulted in his first publications. This bolstered his successful application to Rockefeller University and Cornell University’s joint MD-PhD programs, from which he graduated in 1979 (PhD) and 1980 (MD).

Dr. Korf completed two years of pediatrics residency at Boston Children’s Hospital, a common pathway to medical genetics at the time. During his residency, he soon observed that many patients being seen by neurologists had genetic disorders yet were not being seen by geneticists. This led him to pursue combined training in medical genetics and child neurology. During his training, when a need arose for a clinician to follow NF patients, Dr. Korf accepted an offer to launch an NF clinic. Soon, the monthly clinic hours became weekly, and patients and their families increasingly traveled to be seen in the NF Clinic at Boston Children’s, which still exists today.

Part of Dr. Korf’s legacy is his empathy for people with NF and their families. “You go to the clinic and see that day’s patients, and then you go back to your office or go home and may think about other things, whereas the patients you're seeing take their condition home with them and live with it every minute,” he explained.

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As the clinic population grew, Dr. Korf still found time for research, leading studies into the diagnosis and multidisciplinary management of NF, correlations among NF and other disorders, gene-targeted therapies, factors influencing gene expression, and more. He completed his training and spent 14 years as clinical director for Boston Children's, sowing the seeds of mentorship that still thrive today and contributing to textbooks and training programs in medical genetics. In 1999, he became the medical director of a then-new genomics center, the Harvard-Partners Center for Genetics and Genomics, before joining the University of Alabama at Birmingham in 2003. He serves there today as a professor of genetics and pediatrics, a clinician at two Birmingham hospitals, and the university's chief genomics officer and associate dean for genomic medicine, among his many roles.

The David L. Rimoin Lifetime Achievement Award is the highest honor given by the ACMG Foundation. A committee of past presidents of the ACMG selects the recipient following nominations from the general membership.

"As a visionary leader, admired teacher and mentor, prolific author and researcher, and caring physician for four decades, Dr. Korf's career perfectly exemplifies the personal qualities embodied by Dr. Rimoin," said Fady M. Mikhail, MD, PhD, FACMG, a UAB genetics professor.

About the ACMG Foundation for Genetic and Genomic Medicine

Founded in 1991, the American College of Medical Genetics and Genomics (ACMG) is a prominent authority in the field of medical genetics and genomics and the only nationally recognized medical professional organization solely dedicated to improving health through the practice of medical genetics and genomics. The only medical specialty society in the US that represents the full spectrum of medical genetics disciplines in a single organization, the ACMG provides education, resources and a voice for more than 2,500 clinical and laboratory geneticists, genetic counselors and other healthcare professionals. 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. *Genetics in Medicine* and the new *Genetics in Medicine Open*, a gold open access journal, are the official ACMG journals. ACMG's website, www.acmg.net, offers resources including policy statements, practice guidelines, and educational programs. The ACMG Foundation for Genetic and Genomic Medicine works to advance ACMG educational and public health programs through charitable gifts from corporations, foundations and individuals.

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