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Celebrated Geneticist Dr. Laird G. Jackson Receives Lifetime Achievement Award from the ACMG Foundation for Genetic and Genomic Medicine

BETHESDA, MD – March 22, 2017 | Laird G. Jackson, MD, FFACMG, is the recipient of the 2017 ACMG Foundation David L. Rimoin Lifetime Achievement Award.

The 86-year-old Dr. Jackson, a professor of obstetrics, gynecology, and medical genetics at Drexel University College of Medicine, was honored for his commitment to teaching, his leadership in the field of prenatal genetic screening, and his decades of work surrounding the characterization and treatment of Cornelia de Lange Syndrome.

"There are many brilliant and accomplished scientists in our field, but Laird Jackson stands out among them for embodiment of the traits that this award seeks to recognize," said Dr. Nancy Spinner, chief of the Division of Genomic Diagnostics at Children's Hospital of Philadelphia. "He shares his ideas freely, encouraging everyone around him, and in all of the time I've known him, I've never witnessed him seeking recognition for the ideas he generates."

Dr. Jackson's work in medical genetics spans more than half a century. He has written more than 50 research articles and book chapters; served as a founding member of the International Society for Prenatal Diagnosis and the American College of Medical Genetics and Genomics; and served on editorial boards for several prominent research journals, including the *American Journal of Medical Genetics*.

In response to the news that he'd received the Lifetime Achievement Award, Dr. Jackson said he was "flabbergasted." Then, he expressed a modesty that so many colleagues had described in their nomination letters, drawing attention instead to the late Dr. David L. Rimoin, for whom the award is named.

"I had the good fortune to meet David Rimoin during the mid-1960s and am honored to have called him a friend," Dr. Jackson said. "He was and is still pivotal to the years-long evolution of the medical specialty that all of us enjoy. The establishment of committees and boards that led to our certification, the movement of our specialty into a professional college, gathering funding from the March of Dimes and the Kaiser Foundation and others, all of this required huge amounts of energy and dedication from David, and everybody should be aware of that."

ACMG Foundation President Dr. Bruce R. Korf said, "The David L. Rimoin Lifetime Achievement Award is intended to recognize individuals who have demonstrated the personal characteristics of empathy, compassion, and mentorship, in addition to having made major scientific contributions to medical genetics. All of these traits were the hallmark of Dr. Rimoin's remarkable life and career, and they are likewise embodied by Dr. Jackson. As is clear from the comments of those who have worked with him, Dr. Jackson is a pre-eminent physician scientist, who places respect for his patients, trainees, and society as foremost priorities. The ACMG Foundation is proud to recognize Dr. Jackson as the recipient of the David L. Rimoin Lifetime Achievement Award."

"There are so many parallels between Rimoin and Jackson," said Dr. Hal Dietz, who is Victor A. McKusick Professor of Genetics at the Johns Hopkins University School of Medicine and has taught alongside Dr. Jackson at the Short Course in Human and Mammalian Genetics and Genomics in Bar Harbor, Maine, for more than 20 years. "They're both consummate physicians and superb educators, but they also have this gentle, generous, approachable demeanor that drew patients and students to them alike. And I think in that context, I really could not think of a better choice for this recognition. They are cut from the same cloth."

Born in Seattle, Washington, Laird Jackson, the son of a successful accountant, respected his father's career but was not interested in following the same path. After graduating high school at 16 years old, Jackson entered Pomona College in California and set his sights on medical school at the suggestion of an advisor who supposed that the student's young age would be advantageous in a profession that required so many years of training. "That was my guidance into medicine, without any relatives or family friends who worked as physicians," Dr. Jackson recalled. "It was serendipitous."

Also serendipitous was Dr. Jackson's introduction to the then-emerging field of clinical genetics. After serving three years with the U.S. Air Force in Montgomery, Alabama, he began his internal medicine residency in 1959 at Jefferson Medical College (now Thomas Jefferson University) in Philadelphia, where a supervising physician drew his attention to a seven-year-old patient who had what appeared to be a tumor in her abdomen. It was, in fact, an enlarged spleen due to chronic myelogenous leukemia (CML). Dr. Jackson and his supervisor collaborated on the case with Dr. David Hungerford and Dr. Peter Nowell, who were making the connection between CML and a cancer-causing mutation that was eventually named the Philadelphia chromosome.

"That experience got me looking into chromosomes," Dr. Jackson said. "I was soon noticed by pediatricians who worked in the newborn nursery, who asked me to come check on a child who appeared to have Down syndrome. And pretty soon I became a geneticist before there were *official* geneticists."

Dr. Jackson completed a series of leukemia research fellowships and taught medical students at Jefferson Medical College between 1969 and 1970; meanwhile, his focus was shifting to pediatric genetics and prenatal genetic testing, where he has played a leading role in the most celebrated advances ever since.

When first-trimester chorionic villus sampling was developed as an alternative to amniocentesis, Dr. Jackson was the one who spearheaded a clinical trial to test it in the United States, leading to a landmark paper published in the *New England Journal of Medicine* in 1989. He generated the proposal for a multi-center trial comparing chromosomal microarray with G-banded karyotype, the results of which were published in the *New England Journal of Medicine* in 2012. Today, he continues this work, collaborating with researchers who are developing tests that will sample fetal cells from the mother's circulating blood—the least invasive prenatal genetic testing option yet.

Along with improvements in genetic testing, Dr. Jackson has long championed genetic counseling as a critical component of effective and ethical patient care. One of his greatest achievements, he recounted, was "insisting that genetic counseling was a responsibility - that we had to explain such things to not only patients but also other caregivers, such as newborn nursery nurses and clinical personnel and that these things were important and not just technical discoveries."

Focusing on the needs of others has become a trademark of Dr. Jackson's style. Dr. Hal Dietz, who teaches at the Jackson Laboratory's annual genetics course in Bar Harbor, Maine, along with Dr. Jackson, described what he has witnessed as Dr. Jackson interacts with families from the surrounding community who visit the course to have their medical histories discussed as part of the curriculum: "It was always absolutely clear through eye contact, body language, through his overt respect for patients and also from their response, that he had provided a superb service that addressed their issues and respected their privacy concerns," said Dr. Dietz. "The admiration that the patients felt for him was quite tangible."

Dr. Jackson is perhaps best known for his work on Cornelia de Lange Syndrome (CDL), a developmental disorder that affects multiple parts of the body and is diagnosed in approximately one out of every 10,000 to 30,000 newborn children. During the course of his own research, Dr. Jackson generously offered CDL patient samples to Dr. Ian Krantz at the Children's Hospital of Philadelphia; Dr. Krantz worked with Jackson to discover one of the first genes linked with the disorder in 2004. Dr. Jackson also organized patients and their families to form the Cornelia de Lange Syndrome Foundation in 1981, and he helped raise funds for the Center for Cornelia del Lange Syndrome and Related Disorders at the Children's Hospital of Philadelphia, the first-ever medical research facility that allows families single-location access to a spectrum of research specialists and caregivers who are familiar with the nuances of CDL.

What has motivated him to work so tirelessly on these issues for more than 50 years? "Working is what counts," Dr. Jackson said. "You've got to use whatever resources you have to try and help out."

He gave the credit for his receipt of this lifetime achievement award to his wife Marie and to the Cornelia de Lange families, whom he described as having "made me a better doctor and a better human."

"With an abundance of critical contributions to the field of medical genetics, along with his personal characteristics of empathy and collaboration, Dr. Jackson is the ideal recipient for the first David L. Rimoin Lifetime Achievement Award," said Dr. Ann Garber, the late Dr. Rimoin's widow. "David would be pleased to know this award was being given to someone he considered both a distinguished physician scientist and a genuine friend."

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 in healthcare. Established in 1992, the ACMG Foundation for Genetic and Genomic Medicine supports the American College of Medical Genetics and Genomics' mission to "translate genes into health" by raising funds to attract the next generation of medical geneticists and genetic counselors, to sponsor important research, to promote information about medical genetics, and much more. To learn more about the important mission and projects of the ACMG Foundation for Genetic and Genomic Medicine and how you, too, can support this great cause, please visit www.acmgfoundation.org or contact us at acmgf@acmgfoundation.org or 301/718-2014.

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

Founded in 1991, ACMG is the only nationally recognized medical society dedicated to improving health through the clinical practice of medical genetics and genomics. The American College of Medical Genetics and Genomics (www.acmg.net) provides education, resources and a voice for nearly 2000 biochemical, clinical, cytogenetic, medical and molecular geneticists, genetic counselors and other healthcare professionals, nearly 80% of whom are board certified in the medical genetics specialties. The College's mission is to develop and sustain genetic initiatives in clinical and laboratory practice, education and advocacy.

Genetics and Genomics are not just in our name, they are part of our mission.

Three guiding pillars underpin ACMG's work: 1) Clinical and Laboratory Practice: Establish the paradigm of genomic medicine by issuing statements and evidence-based or expert clinical and laboratory practice guidelines and through descriptions of best practices for the delivery of genomic medicine. 2) Education: Provide education and tools for medical geneticists, other health professionals and the public and grow the genetics workforce. 3) Advocacy: Work with policymakers and payers to support the responsible application of genomics in medical practice. *Genetics in Medicine*, published monthly, is the official ACMG peer-reviewed journal. ACMG's website (www.acmg.net) offers a variety of resources including Policy Statements, Practice Guidelines, Educational Resources, and a Find a Geneticist tool. The educational and public health programs of the American College of Medical Genetics and Genomics are dependent upon charitable gifts from corporations, foundations, and individuals through the ACMG Foundation for Genetic and Genomic Medicine (www.acmgfoundation.org).

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