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**Clinical Geneticist and Pediatrician Elaine Zackai, MD, FACMG Receives
David L. Rimoin Lifetime Achievement Award in Medical Genetics from the ACMG
Foundation for Genetic and Genomic Medicine**

BETHESDA, MD – March 23, 2022 | Revered clinical geneticist and pediatrician Elaine H. Zackai, MD, FACMG has been selected for the 2022 ACMG Foundation for Genetic and Genomic Medicine's **David L. Rimoin Lifetime Achievement Award in Medical Genetics**.

Dr. Zackai, a fellow of the College of Physicians of Philadelphia and a founding fellow of ACMG, directs the Clinical Genetics Program at the Children's Hospital of Philadelphia (CHOP) and holds the Letitia B. and Alice Scott Endowed Chair in Human Genetics and Molecular Biology. She was chosen for this award to recognize her renowned expertise diagnosing birth defects and genetic disorders; her compassionate and resourceful care for patients and families; her prolific research collaboration with colleagues around the world; and her mentorship and teaching of clinicians, researchers, and genetics counselors who have in turn modeled their work on her standards for excellence.

"Dr. Zackai is precisely the person who embodies all the qualities of the David L. Rimoin Lifetime Achievement Award in Medical Genetics," said ACMG President Max Muenke, MD, MBA, FACMG. "Dr. Rimoin influenced Dr. Zackai's early training in genetics. And Dr. Zackai passed on not just knowledge and commitment to patient care but a deep passion for teaching and mentoring to the next generation(s) of trainees. I am personally grateful to Dr. Zackai who trained me and had an imprint on my career."

Dr. Rimoin's widow, Dr. Ann Garber-Rimoin said, "It is with pleasure that the Rimoin family honors Elaine Zackai, MD, FACMG with the 2022 David L. Rimoin Lifetime Achievement Award in Medical Genetics. Dr. Zackai's outstanding career is aligned with the qualities that characterized David's career in medical genetics, including her excellence in teaching and mentoring, her ability to apply lab-based genomics into her clinical practice, and her gift for connecting with patients and their families. We congratulate Dr. Zakai as the recipient of the Rimoin Lifetime Achievement Award for 2022."

With her career now spanning half a century and close to 600 research publications during that time, Dr. Zackai has collaborated with, by one expert's count, at least 4,307 colleagues around the world. Her work, which has elucidated dozens of new genetic conditions, falls into the general categories of craniofacial dysmorphism, neural tube defects, neurofibromatosis, and chromosomal abnormalities.

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Touched and honored by the news that she was selected for the David L. Rimoin Lifetime Achievement Award, she explained the significance of his role in her training, going back to when she was one of his first fellows.

"I have modeled my whole career around what I learned from David Rimoin, always putting the patient first and treasuring the exceptions that don't fit the obvious diagnoses," Dr. Zackai said. "I spent hours xeroxing his entire set of journal articles before he left St. Louis to become an attending in California. This was back in the day before computers! They are still one of my most valuable tools and when I use them, I think of him and what he would have done for the patient I have at hand. I would not be where I am today without the essential foundation that he imparted to me."

President of the ACMG Foundation, Bruce R. Korf, MD, PhD, FACMG said, "Dr. Zackai has trained at least a generation of clinical geneticists and provided care to children and families dealing with genetic disorders for decades. Her commitment, compassion, and expertise are precisely in keeping with the values we seek to recognize in the David L. Rimoin Lifetime Achievement Award in Medical Genetics. Indeed Dr. Zackai began her genetics training with Dr. Rimoin, making this award all the more appropriate."

Dr. Zackai's ability to recognize and recall details that align phenotype with genotype is legendary, and it has earned her the colloquial reputation of master geneticist. In the corridors of CHOP, clinicians regularly use her name as a verb: Has the patient been Zackai'd yet?

"The nonverbal body language Dr. Zackai exudes lets you know that she knows what a patient's diagnosis is just after walking through the door," explained Ian Campbell, MD, PhD, an attending physician at CHOP who completed his fellowship in medical genetics with Dr. Zackai's mentorship. "She has this ability to take in everything about a patient, including the physical exam, and match it in mere seconds to among 5,000 rare genetic diseases. That is an ability I certainly don't have, and I think it shows what an amazing mind she has."

Exceptional thoughtfulness and compassion were also qualities emphasized in Dr. Zackai's nomination letters. "She treats patients and trainees like they are family," more than one colleague said. Small gestures such as arranging transportation, reaching out to colleagues for additional consultation, requesting additional studies in hematology or radiology to shore up possible diagnosis, and coming to the bedside when her particular style of explanation can be the most helpful—these are the standards of care that have left an indelible mark on the people who benefit from Dr. Zackai's expertise and care.

Elaine Zackai was born in Brooklyn, New York in 1943, the older of two children. Her father was a metallurgist who formulated fine-gauge wires, and her mother taught high school science. Her brother studied drama and went on to teach theater, as well as English as a second language, while Elaine was drawn to science and mathematics. "I just liked studying," she recalled. "I liked learning new things, and I was good at it. And that's how it all started."

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She attended New York University expecting to major in math, but once she reached a point in calculus where the course turned toward theory, she was lost and switched to her next-favorite subject: human-focused science. Elaine graduated *magna cum laude* in 1964 with her bachelor of arts in biology and chemistry, earning the Sigma Pi Sigma Prize in Physics, and immediately she commenced medical studies at New York University, completing her medical degree in 1968.

“Medical school was fun,” Dr. Zackai said. “I knew I was in the right place. I understood why I was doing it, the purpose of it. It wasn’t just theory; you could actually touch it, see it, and do it.” In recognition of her performance as a medical student, Dr. Zackai was elected to the Alpha Omega Alpha Honor Society and received its Outstanding Woman Graduate Medical School Alumni Key Pin.

Pediatrics brought her to St. Louis Children's Hospital in St. Louis, Missouri, in 1970. This is where she met David Rimoin, who had arrived just two years earlier to direct the new Medical Genetics Clinic at the Washington University School of Medicine. Medical genetics was an emerging specialty at the time, and in it, Dr. Zackai said, “I saw a field that encompassed many different specialties, and a field where I could use my love of math to figure out the chance of something happening again in a family, the recurrence risk. I also shied away from minute-to-minute management of glucose or fluids or things like this. So, it was clear that this was an ideal spot for me.” She began her fellowship in medical genetics with Rimoin in 1970 and transferred to Yale during her second year to complete her training in 1972.

The next job she took—as an instructor of pediatrics and clinical genetics in Philadelphia—is one she has remained in ever since. She joined the faculty at the Perelman School of Medicine at the University of Pennsylvania in 1972 and then founded the Clinical Genetics Program at CHOP in 1974, overseeing its expansion to include specialty services for neurogenetics, orthogenetics, ophthogenetics, a Craniofacial Clinic, as well as the 22q and You Center, which offers diagnosis and multidisciplinary management of patients who have any number of symptoms associated with chromosome 22q11.2 deletion.

“The environment at CHOP is excellent,” Dr. Zackai said. “We have a wonderful esprit de corps, where the attitude is really about answering the question about colleagues, ‘How can we elevate you?’ The population of patients here is excellent, too. They come from the surrounding neighborhood for general pediatrics, but we also receive patients from a 200-mile catchment area, so cases come in regularly that really challenge us to think.”

Her collaboration with various departments at CHOP is one of the key factors that helped grow the Clinical Genetics Program into what it is today. It also facilitates exceptional care for patients and families who come from around the world. In 2007 CHOP recognized her achievement with the Master Clinician Award.

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“The thing that is most striking about her—and it’s why she can’t retire because nobody will ever be able to replace her—is she really understands both the scientific and emotional implications of our field,” said Alanna Strong, MD, PhD, Instructor of Pediatrics at CHOP who also trained with Dr. Zackai. “She feels the responsibility to share her lifetime of experience with the families she works with and with her trainees. It’s not just a job for her, something she is required to do. She genuinely loves the science, the thinking, the work, the mentoring, and the families. She truly makes a difference in the lives of everyone she meets. I hope she never retires, but if she ever does, it will be a loss to her students, her patients, to the field of Genetics, and frankly to everyone.”

Fortunately, Dr. Zackai said she has no interest in retirement. “It’s daunting to hear the words ‘Lifetime Achievement,’ since I’m still achieving. But I have nine lives, and this is the first one,” she said. “I feel that I often know the answer, and why should people struggle if someone like me knows the answer? I get a lot of satisfaction out of that. It makes me happy.”

The excitement she feels while tracking down a patient’s diagnosis manifests with the expression, “We’re having a moment!” whenever she has a new idea about what should happen next. Two years, 10 years, even 20 years after she has assessed a patient, she will gladly resume the conversation with family if something new in the literature adds to understanding of that child’s particular diagnosis.

“The most important thing she taught me was to be persistent until you find the answer for the patient,” said Sarah Sheppard, MD, PhD, FACMG, who completed her training at CHOP and is now a tenure-track investigator at the *Eunice Kennedy Shriver* National Institute for Child Health and Human Development. “She’s so patient-centered in her approach to everything, and she won’t hesitate to find the right person, whether it’s a different specialist at CHOP or an external researcher or clinician, who can help address the patient’s issue.”

Dr. Zackai has made a conscious effort to pass on this never-give-up approach to her trainees, and for it, she has been recognized with the 2002 Blockley-Osler Award presented by the Blockley Section of the College of Physicians, the American Society of Human Genetic inaugural Mentorship Award in 2016, and the 2019 CHOP Award for Excellence in Mentoring Research Trainees.

“I try to teach them to pay attention to the rarest finding, something they can use as an anchor, because many times our cases are complicated and it can be easy to feel overwhelmed,” Dr. Zackai explained. “Finding an anchor gives us a moment to pause and ask, ‘Ok, if all the patient’s fingerprints are whorls, what does that mean to me?’ Then you look for the next clue, and the next one. I love going to the bedside with them to teach like this, and I think they’re getting it. They have to keep up with my pace. I get a new crop every two or three years, and I feel young with them.”

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.

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About the American College of Medical Genetics and Genomics Foundation and the American College of Medical Genetics and Genomics (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 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.

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.

Note to editors: To arrange interviews with experts in medical genetics, contact ACMG Senior Director of Communications and Public Relations, Kathy Moran, MBA at kmoran@acmg.net.

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