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Increasing the Ranks of Medical Geneticists

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—By Jen Uscher, special to the *Reporter*

Christian Schaaf, M.D., Ph.D., discovered he enjoyed solving medical mysteries during a pediatrics rotation as a medical student. In particular, he savored the detective work of searching the literature to find out if there could be an underlying genetic cause to a patient's disease.

"During that rotation, I diagnosed a young patient with Rett syndrome—a rare genetic neurodevelopmental disorder," said Schaaf, now an assistant professor in the Department of Molecular and Human Genetics at Baylor College of Medicine. "That case fascinated me and shaped my career direction."

By pursuing a residency in medical genetics, Schaaf joined a field that experts say needs more new recruits. According to the American College of Medical Genetics and Genomics (ACMG), only about 50 percent of the available clinical genetics training slots in the United States are filled. In addition, the number of trainees who pass the American Board of Medical Genetics exam to become board certified as clinical geneticists has more or less plateaued since 1999.

"Many jobs that are available in the field for qualified people are going unfilled. There aren't enough physicians being trained to fill those positions," said Jerry Feldman, M.D., Ph.D., professor of molecular medicine and genetics, pediatrics, and pathology, and director of clinical genetics services at Wayne State University School of Medicine. "There's also an expectation that the number of individuals needed to be trained in genetics is only going to increase as genetic information is utilized more and more in everyday medical practice."

Clinical genetics trainees typically complete a two-year residency in pediatrics, obstetrics and gynecology, or internal medicine, followed by an additional residency in medical genetics. Some opt to pursue a combined residency program in pediatrics/medical genetics, maternal-fetal medicine/medical genetics, or internal medicine/medical genetics. The majority of clinical geneticists go on to work at academic medical centers, where they diagnose, counsel, and treat patients who are at risk for or affected by genetically influenced disorders. Many also are involved with teaching and conduct research on genetic diseases and treatments.

The shortage of medical geneticists can translate into challenges for both patients and physicians. In some areas of the country, there are long wait times—sometimes months—to see a medical geneticist, noted Bruce R. Korf, M.D., Ph.D., president of the ACMG Foundation for Genetic and Genomic Medicine, and professor and chair of the genetics department at the University of Alabama at Birmingham School of Medicine. "Many physicians don't have colleagues who are medical geneticists to keep them up to date on new tests and treatment options. There are new medications that can be used to treat certain genetic conditions now—for example, in the last year the Food and Drug Administration approved a drug for treating some of the brain and kidney tumors in patients with tuberous sclerosis," Korf said.

One key reason for the shortage of geneticists may be a lack of awareness among medical students that clinical genetics is a career option. "In medical school, the people who advise students on careers tend to talk about the big primary specialties like pediatrics and internal medicine," said Mira Irons, M.D., the Park Gerald Chair in Genetics and associate chief in the division of genetics at Boston Children's Hospital. "There are so few medical geneticists at most medical schools and teaching hospitals that there aren't as many role models for students compared with other specialties."

Korf explained that students mostly learn about genetics in basic science courses during their first two years of medical school. The faculty members who teach these courses often are basic scientists rather than medical geneticists. "The students learn about principles of genetics but don't necessarily hear that genetics is a medical specialty, so they may get the perception that genetics doesn't have immediate clinical relevance," Korf said.


In 2011, the ACMG joined with several other genetics organizations, including the Association of Professors of Human and Medical Genetics and the American Society of Human Genetics, to form the Task Force on Medical Genetics and Genomics Education and Training. This task force, which Korf chairs, is working on a number of outreach

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—Mira Irons, M.D.

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initiatives to attract more college and medical students to pursue careers in medical genetics and genomics. In 2012, for example, the ACMG released a video, available on YouTube, that features interviews with medical genetics trainees and encourages medical students to consider a residency in medical genetics. Medical genetics residency training programs across the country can customize the video with information about their own programs and use it as a recruitment tool.

Another initiative involves helping medical students form ACMG medical student interest groups at their schools. These groups host speakers who discuss career paths in medical genetics and offer other educational activities, such as shadowing experiences and trips to genetics labs and clinics. Each group has a faculty advisor to mentor students who are interested in medical genetics but might not otherwise have opportunities to connect with people working in the field.

Schaaf, who completed his medical genetics residency at Baylor in 2010, believes that faculty mentors have an important role to play in drawing new recruits to the specialty. “I might not be in the field if it weren’t for the head of the genetics department at my medical school. He inspired me as a role model,” Schaaf said.

Now that he is a clinical geneticist, Schaaf said one of the things he appreciates most is the variety of cases he encounters. “It’s never boring. Every day I go to work at the clinic, there is a patient who has a disorder I’ve never seen before, or there are two or three cases where I’m trying to make a diagnosis and it’s a mystery at first. I learn something new every day because the disease presentation made me go back to the literature and read about it,” he said.

As new genetic tests, including whole-genome sequencing, become more widely available, geneticists are likely to be increasingly in demand for the expertise they can offer to patients, as well as to their colleagues. “We’ll need more medical geneticists to educate all health care providers in the basics so that they can use the tools of genetics and genomics in the care of their patients,” Irons said.