

What it's like to specialize in medical genetics: Shadowing Dr. Abbott

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Staff News Writer

As a medical student, do you ever wonder what it's like to specialize in medical genetics? Meet Mary-Alice Abbott, MD, PhD, a medical geneticist and a featured physician in the AMA's "Shadow Me" Specialty Series, which offers advice directly from physicians about life in their specialties. Check out her insights to help determine whether a career in medical genetics might be a good fit for you.



"Shadowing" Dr. Abbott

Specialty: Medical genetics.

Practice setting: Hospital.

Employment type: Multispecialty Hospital Group.

Years in practice: 13.

A typical day and week in my practice: In medical genetics, no two days are the same and every day is interesting. Most of what I do is direct patient care, with some supervisory and administrative work, some academic and teaching work, and clinical research.

In my medical genetics practice, I am involved in the care of patients of all ages including preconception (a couple may be interested in understanding reproductive risks related to their personal or family histories of rare conditions or birth defects, or may be seeking screening tests that can inform their risks for having a child with a rare autosomal recessive condition) and prenatal patients (a pregnant patient who has an abnormal test result, such as a prenatal ultrasound that identified that the bones are short and that the fetus may have a skeletal dysplasia).

I am also trained in pediatrics, and more than half of my time is spent with kids—this is particularly fun and gratifying. I evaluate children with challenges including birth defects, growth or neurodevelopmental problems, skeletal abnormalities, metabolic disorders, and family histories of genetic diagnoses, in order to identify possible associations, underlying diagnoses, prognosis and recurrence risks.

Our adult patients are incredibly interesting and complex as well, and are referred from many of our medicine subspecialty colleagues. They may come to see a geneticist because their doctor has diagnosed them with renal failure, hyperparathyroidism, an aortic aneurysm or a myopathy, and suspects that a genetic evaluation will be useful in determining if there is a primary genetic diagnosis (often associated with a more complex or multisystemic problem). Some have genetic syndromes that were diagnosed in childhood. They have grown up and are encountering new syndrome-specific medical needs. Working with our oncology colleagues, we play a crucial role in the Family Cancer Risk program, assessing genetic risks for patients with cancer or with significant cancer family histories.

I care for patients with rare disorders that affect the lysosomes, including Pompe Disease, Fabry Disease and the mucopolysaccharidoses. For these conditions, specific genetic therapeutics have been developed, including enzyme replacement therapies and medical chaperone treatments. Since these diseases are multisystemic, and treatments are necessary throughout life, we are involved in long-term coordination of care for many of these individuals. Other patients who we manage over many years include our pediatric and adult patients who have inborn errors of metabolism, like PKU, and work closely with our metabolic dieticians to maintain metabolic and dietary control of their conditions.

Some of our patients have relatively common conditions like Down syndrome, 22q11 microdeletion syndrome, neurofibromatosis type 1, Marfan syndrome and Fragile X disorder. But many have unknown underlying conditions.

Diagnosing very rare conditions requires knowledge of many of the genetic syndromes and inborn errors of metabolism, dysmorphology skills (using a very careful focused physical examination looking for often subtle findings that may indicate a pattern consistent with a particular diagnosis), family history analysis (taking a three-generation pedigree), and ability to extract and integrate information from databases, online resources, specialized textbooks, and primary medical literature.

Talking with patients and families, and effectively communicating information about general genetics and specific genetic diagnoses requires attention and care. As you can imagine, a clinical genetics consultation can take a lot of time. As genetic specialists, we may spend significantly more time on tasks directly related to the visit compared with primary care physicians or doctors in other specialties.

Delivering excellent medical genetics care requires a team approach. I have the privilege of working directly with genetic counselors, advanced practitioners trained in genetics, metabolic dieticians, a care coordinator and a medical assistant. I work closely with pediatricians and neonatologists, pediatric (and adult) specialists like neurologists and cardiologists, maternal fetal medicine doctors, oncologists, and other referring providers. We have many learners in our area, including medical students, residents and genetic counselor interns.

Every week there are requests for inpatient genetics consultations, most often in the neonatal intensive care unit, but in all areas of the hospital. Colleagues throughout our health system and community contact my colleagues and I seeking information or advice about genetic diagnoses, testing, results interpretation and management recommendations. Genetic testing and treatments are expensive, and careful stewardship of these resources is important, so I am involved in utilization management.

A typical day?

Last Monday, I worked with our nurse practitioner and evaluated a young woman with very unusual skin and veins and other findings concerning for a disorder of the connective ties; I admit I'm still not sure what type of genetic disorder she has. Then I reviewed the dietary needs of one of our adult patients who has Phenylketonuria and is newly pregnant.

I saw a newborn baby in the hospital with abnormal arms whose parents I had met prenatally—she had an associated platelet problem that I didn't expect. I also saw a newborn with a cleft palate that was not seen prenatally.

A woman came to see us on a stretcher, accompanied by her son, because she was suffering from a rapidly progressive form of ALS, like her father and brother had. Later I worked with a genetic counseling student to analyze genotype-phenotype data from a large family with a Marfan-like syndrome. I evaluated a girl with intellectual disability and autism. I met with our reproductive genetic counselors and we reviewed their recent cases together, and did similar rounds with our cancer

genetics group.

I work 40-to-50 hours a week and am available 24 hours, seven days a week by pager. I get calls from the hospital to discuss patients or review labs. There can be emergencies, such as a newborn who presents ill with suspected metabolic disorder, but these occur less than in other specialties.

The most challenging aspect of medical genetics: There are three things. (1) Giving bad news is something that I do a lot, but is never easy. This is particularly difficult when I have diagnosed a condition that is progressive, which could present prenatally, in childhood, or in later life. With experience and practice, you get better at doing this, but it is always hard, and needs to be approached with attention, compassion, and care.

(2) There are many mysteries in medicine, and genetics has unlocked only some of them. So I have to say “I don’t know” a lot. It is difficult, as a scientist and physician, to admit this so frequently—I don’t know the diagnosis, I don’t know what to expect, I don’t understand how or why unexpected things have happened, we don’t know how to treat or cure this. Although there are treatments available or emerging for some conditions, it is rare when I can offer an effective treatment for a genetic condition.

(3) It is overwhelming to keep up to date with the medical genetics literature. This is a very rapidly changing field. Although I read every day, it feels impossible to know everything.

The most rewarding aspects of medical genetics: I have a background in math and science and was seeking a “scientific” specialty. And boy did I find it! I practice at the cutting edge of science during an incredible time in the history of medicine.

(1) I learn something new every day.

(2) Figuring things out, sometimes after many years, is so satisfying. I recently saw a baby who was born with abnormal hips. She was accompanied by her teenage cousin who I first met almost 10 years earlier. Back then, after consulting with an expert in bone disease, we had come to a working diagnosis of a very rare skeletal dysplasia. But at that time, confirmatory testing wasn’t available, and there was barely any information in the medical literature.

Now, so many years later, the baby cousin seemingly had the same condition that I had suspected in the older child. I was able to talk to the family about what we currently understand about the molecular basis of this condition, its autosomal recessive inheritance, the availability of confirmatory testing, and discuss with her orthopedist what is now understood to be the best approach to orthopedic care.

(3) Getting to know patients and families over years, often during challenging times, is a very rewarding aspect of medicine, genetics included.

Three adjectives to describe the typical medical geneticist: Observant, curious and methodical.

How my lifestyle matches, or differs from, what I had envisioned: My husband is also a physician, so we do dedicate a lot of time to work. We have two children, and over the years have relied on family members and others to help us with child care. However, I am able to make it to just about every concert, parent conference and hockey game. I also am able to make time to swim, bike and run.

Skills every physician in training should have for medical genetics but won't be tested for on the board exam: Be able to type well (genetics notes are long!). Be able to ask for help, expertise and advice. You cannot know everything. Be prepared for things to be new, and to change, all the time. Be prepared to cry.

One question physicians in training should ask themselves before pursuing medical genetics: Do you like to find answers? To study, read and think? This is a field for people who love to learn.

Books every medical student interested in medical genetics should be reading:

- Jodi Picoult, *My Sister's Keeper*.
- D.T. Max, *The Family that Couldn't Sleep: A Medical Mystery*.
- Siddhartha Mukherjee, *The Emperor of All Maladies: A Biography of Cancer*.

The online resource students interested in medical genetics should follow:

- GeneReviews.
- Radiolab.
- TED Talks.

Quick insights I would give students who are considering medical genetics: In this specialty, your job satisfaction will be personal and professional.

Mantra or song to describe life in medical genetics: Dr. Seuss: "There's no limit to how much you'll know, depending how far beyond zebra you go."