UNDER THE MICROSCOPE

Zoom In on the Ins, Outs, and Abouts of Genetic Screening and Testing

By Brian Justice

The National Human Genome Research Institute defines *genetic screening* as a process used to identify a smaller group of people from within a population. The identified subgroups may have a higher risk of having a disease, developing that disease, or having children who develop that disease. Genetic testing, however, is focused on the individual.[1]

"The best thing to understand is that screening and testing are two different words, because they are different things," says Pamela Trapane, MD, chief of the pediatric genetics division at the University of Florida College of Medicine and medical director of the Duran Genetics Center at Wolfson Children's Hospital, both in Jacksonville, Florida. "A screening is not a diagnostic test. A breast cancer screening is not diagnostic, for instance. If you find something [during the screening], you go in, get a sample, test, and *then* make a diagnosis."

"Many people get this confused," agrees Christine Hricak, CMA (AAMA), a genetic counseling assistant with the Lehigh Valley Health Network in Allentown, Pennsylvania. "Screening [tells] you risk factors. [A patient] may be at risk for a condition, but it doesn't necessarily mean [the patient has] it. Testing is more definite with results."

Notably, screenings are not necessarily based on preexisting conditions.

"Common preventive screenings can be based on age or gender—like colonoscopies, for example," explains Nicole Koelling, CMA (AAMA), senior population health coordinator at St. Luke's Hospital in Chesterfield, Missouri. "Genetic testing is usually recommended by a physician based on a significant family history for a certain condition."

Examine Health Cues

"The benefits of screening and testing include knowledge," says Danielle Jalbert, CMA (AAMA), lead medical assistant and preceptor at Swedish Mercer Island Primary Care in Washington. "A big part of my work is helping patients overcome barriers of all kinds—financial, logistical, emotional. So, if a patient is hesitant, I want to know why. Then we can focus our energy and efforts on learning more and taking action."

Genetic screening can improve both population health and health equity by identifying high-risk populations and prompting targeted treatments that drive better outcomes.[2]

Nevertheless, both pros and cons are present in screening and testing, observes Hricak.

"Genetic counselors [must] consider the patient's anxiety level or desire to have more information," she says. "It varies from patient to patient, and a discussion with their medical provider can help them decide what testing [will] work best."

Up to Code?

Another reason for clarifying the capabili-

ties of genetic screening and testing is the popularity of recreational genetics, such as 23andMe and AncestryDNA. The market for these services is projected to be almost $2 billion by 2026.[2]

"I think genetic screening or testing is becoming more mainstream," says Peter Hulick, MD, medical director of the Mark R. Neaman Center for Personalized Medicine and division head of the Center for Medical Genetics with NorthShore University HealthSystem in Evanston, Illinois. "[Many] of the ancestry-related direct consumer offerings out there started to get genomics and genetics into the [common] nomenclature."

However, a 2021 study published in *JAMA Cardiology* revealed how direct-to-consumer testing could mislead users. Researchers compared the results of genetic tests ordered by clinicians versus commercial genetic tests for familial hypercholesterolemia—a condition in which variants of several genes cause markedly high cholesterol and an increased risk of heart attack, stroke, and more. They found that commercial genetic tests missed crucial variants for almost 70% of participants, and the impact was even greater—and potential consequences even more dire—for people of color.[3]

A separate investigation by the *New York Times* revealed that commercial prenatal screening produced false results 85% of the time.[4]

The significance of accurate testing cannot be overstated. "Genetic testing can only tell you [whether] you have a specific gene variant or mutation, not [whether] you will get cancer," explains the American Cancer Society.[5] "So, the test can tell what *might* happen, but it cannot tell what *will* happen. A positive test result does not always mean you will get the disease. And a negative result does not mean you have no risk of getting the cancer."[5]

"[Recognizing] that a positive result isn't destiny [is crucial]," agrees Dr. Hulick. "[A result] doesn't guarantee that something will happen, but it allows patients to … get information [before] being diagnosed so they can be put on a plan to reduce that risk."

Tried and Tested

Privacy concerns can also inform patients' opinions about screening and testing. The Genetic Information Nondiscrimination Act of 2008 prohibits employers and insurers from making decisions based on genetic information.[5] Regardless of federal and state protections and the thoroughness of clinician-conducted testing and review, how frontline professionals interact with apprehensive patients impacts how patients will react to their testing or screening results.

"I always joke that our profession is one of the most highly technical and, at the same time, one of the lowest [technological]," says Dr. Trapane. "A lot of it is counseling and psychiatry. [We help] people by educating them about what their test is and is not."

"I encourage patients to realize that results give us a starting point for action," says Jalbert. "After all, knowledge is power!" ✦

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SPECIMEN

Through the Lens of DNA

Researchers envision a day when an app knows its user's risk for certain diseases, the odds of passing diseases on, and what medicines would best treat the ailments. Genomics, the study of genomes, will make these advances possible. Genes compose DNA, and a genome is a complete picture of an individual's DNA and how it functions and interacts in an individual's body.[6]

Humans are 99.9% identical in their genetic makeup, so understanding that minuscule—but crucial—0.1% holds the key to detecting, treating, and even preventing certain diseases. One of the most intriguing developments is the rapid growth of pharmacogenomics, the study of how genes affect an individual's response to drugs. Increasingly, one's DNA can predict what medications *and* dosages would be most effective.[6]

"There are so many things on the horizon," says Pamela Trapane, MD. "It's kind of like all our [smartphone] updates. It just keeps going faster and faster."