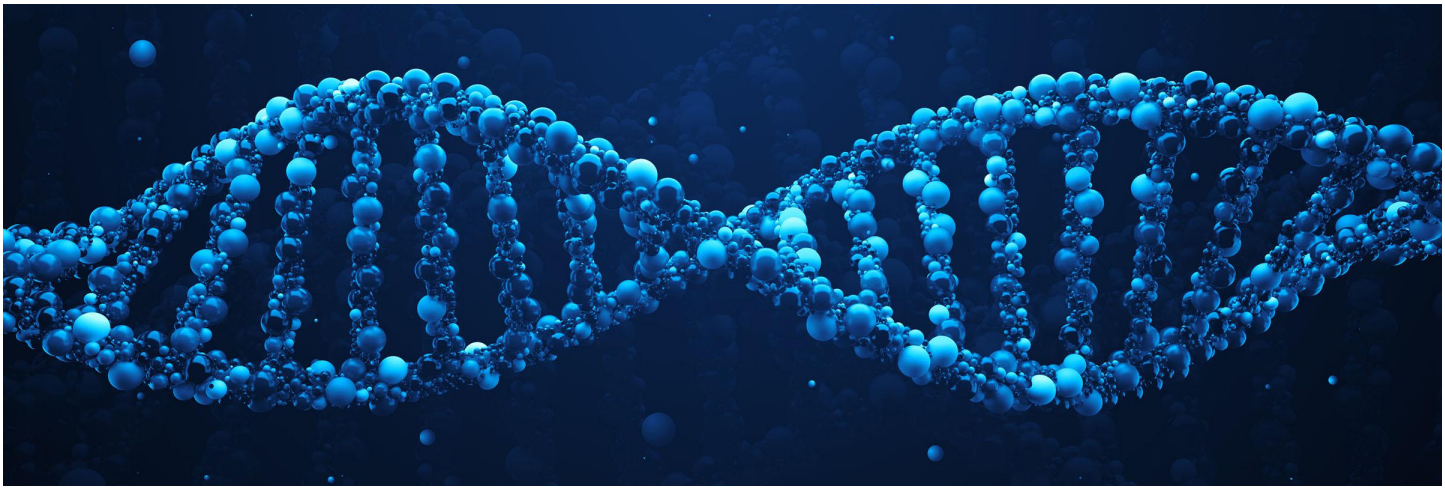

Ultra-customized precision medicine that's tied to your genes

By Vicki Ritterband | June 19, 2019

Genetic testing has been around for decades, but until recently, screenings focused on a very limited number of genes because sequencing the entire genome was prohibitively expensive. Now that the cost has decreased dramatically and the impact of this information on patient care is clear, healthcare companies like Geisinger are broadening their DNA testing programs so doctors can get the jump on genetic variants that put patients at risk for a variety of serious conditions.

Over the past year, Geisinger has ramped up the clinical component of its 12-year-old MyCode Community Health Initiative by expanding the research sequencing program beyond the laboratory and into the exam room. Now, individual patients can benefit from the genetic information that is revealed.

Two primary care clinics are participating in Geisinger's new clinically focused arm, the Population Health Genomic Screening Program. Geisinger plans to roll it out to a third in the next several months. Eventually, the initiative will spread system-wide, with the goal of making genomic sequencing and proactive treatment part of routine care for its 1.5 million patients in Pennsylvania and New Jersey. In addition to its health plan, Geisinger has 13 hospital campuses, a medical school, and a extensive research division overseeing everything from clinical trials to laboratory-based discovery.

Since July 2018, approximately 500 patients have had their exomes — the part of the genome where most disease-causing mutations occur — tested under Geisinger's Population Health Genomic Screening Program. That's in addition to the approximately 145,000 patients who have been tested for purely research reasons since the MyCode initiative began in 2007.

The Population Health Genomic Screening Program informs patients and their physicians of any clinically relevant results of their exome sequencing, including a heightened risk of cardiomyopathy, breast and ovarian cancer, and hypercholesterolemia. The clinical sequencing program tests for these and about two dozen other conditions.

"Over time, we expect to expand the list, but right now we're testing for the slam-dunk genes that are known to cause disease and to be highly actionable," explains Christa Martin, Geisinger's associate chief scientific officer and a board-certified laboratory geneticist. "In each case, something can be done to either detect the disease early or try to prevent it altogether."

How It Works

The Geisinger Population Health Genomic Program works like this: At a regular office visit, patients are asked if they would like to participate in genomic screening. If they consent, their blood is drawn, and a sample is sent to a clinical laboratory for exome sequencing. Of the approximately 500 patients who have been tested under the pilot, about three percent have received a positive result indicating they have a genetic change that puts them at risk.

From there, a genetic counselor will speak with the physician who ordered the test to strategize how to deliver the news; that counselor is usually the provider who discusses the results at the initial meeting with the patient. Geisinger provides physicians with information to assist with follow up, including best practices for monitoring. And because family members may have the same genetic variant, the counselor goes over the family tree and discusses testing for loved ones.

The Impact

The Population Health Genomic Program is still in its infancy, so Geisinger has just begun to understand its impact. But the health system estimates that if physicians relied on clinical indications for genetic testing, they would have missed about half of the people they've identified with concerning DNA changes. Martin expects that as the program increases the number of genes sequenced — now 60 — the impact of the initiative will grow.

Martin adds that success should also be gauged by the patient stories that make the work so compelling, including the 57-year-old grandmother with custody of her four-year-old grandchild who had her ovaries removed after discovering she was at heightened risk for breast and ovarian cancer, and the middle-aged man who stepped up his heart healthy regimen after a genetic change that is a precursor for early heart disease was found. "This information can be life changing," says Martin.

Vicki Ritterband is a frequent contributor to athenaInsight