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# Is it Time for Health Systems to Offer Preventive Genomic Screening?

*We know this screening saves lives. Why aren't health systems adopting it?*



By Carrie Blout Zawatsky

Preventive genomic screening (aka elective testing) has been offered in research settings and directly to consumers for nearly a decade. Direct to consumer (DTC) testing has reached over 100 million individuals. Large research initiatives such as health system biobanks and NIH's All of Us Research Program are offering preventive genetic testing results (unrelated to a medical indication) to millions of people. And some cutting-edge health centers are starting to offer preventive genomic screening clinically to their patients.

The Preventive Genomics Clinic at Brigham and Women's Hospital launched in 2019, and was the first acad

genomic screening to healthy adults who are seeking to understand and mitigate their risk of genetic disease in themselves and their family members.

At this point, we can safely say that genomic screening can identify genetic risk factors that have direct medical screening and management implications. There is even preliminary evidence that for some of the most common conditions, like Hereditary Breast and Ovarian Cancer, preventive screening may be cost effective—on top of saving lives.

Professional genetics organizations have developed consensus documents surrounding this topic; the National Society of Genetic Counselors just published a practice resource on elective genetic testing and the American College of Medical Genetics and Genomics (ACMG) has also published multiple documents on this topic. In fact the ACMG set the stage for this in 2013 when they published a list of genes that they deemed to have enough medical actionability to be returned if found incidentally (unexpectedly) when testing for other genetic conditions. Though not its intended purpose, this list, which has been updated four times since, is often used or adapted by health centers when offering this type of screening on a research or clinical basis to their patients.

So what is holding health systems back from offering this type of testing clinically? Admittedly, there is still clinical utility data being collected to better understand all the implications and, yes, there is still the question of balancing sacred health care resources in a climate prone to health care provider burnout and overspending. But is there something more to the holdup? As with any new(ish) technology, especially a genetic technology, health insurers are waiting to have more comprehensive clinical utility data before providing coverage. But is this a big enough reason to restrict offering this potentially life saving technology to patients who want it? Especially when research studies are showing no increases in negative health outcomes, like increased patient anxiety/depression or medical mismanagement?

Is genomic screening cost prohibitive? Maybe, but some might argue that for a few hundred dollars, cost is becoming less of an issue (the first BRCA single gene testing was over \$3,000). In fact we know that many individuals typically underrepresented in medical research are very interested in this type of testing, and that the labs that give them access to genetic screening, either via a DTC test or a physician mediated test (ordered by a physician connected to the lab), are actually increasing their access because they just are not getting it through their medical providers. Other groups at medical risk, like those who are adopted, have no other option to learn this health information, and many don't know how to get it other than ordering DTC testing, which offers limited health information due to stricter FDA

regulations. Patients don't even realize that they could be getting a more medically informative test if it was ordered by their healthcare provider.

Though all of these components are part of the big picture in explaining why population based preventive genomic testing is not clinically available at the health system level, we argue that the real reason it is not available is due to implementation challenges. Health systems can see the potential in this type of testing, but aside from getting millions of dollars in grants to help pay for the infrastructure to offer the test, they just don't see a path forward.

Implementation of healthcare innovations can be considered the "last mile" in getting new technologies in the hands of clinicians, but the devil is in the details, and this last mile often proves to be the hardest in launching emerging forms of care. And when you consider the breadth of changes required to prepare a health system to offer genetic screening, these changes often become a huge barrier to entry.

But what if a simple scalable path forward could be designed to make preventive genomic testing a reality, one that considers the needs of the patients, the providers and the leadership? That is exactly what the Precision Population Health (PPH) initiative led by Ariadne Labs and Genomes2People is doing. We are working with health systems to design a pathway to successful implementation and to offer sustainable preventive genomic screening. So far our early projects have been developed for adults in primary care, but we also know that newborn genomic screening is not far behind, as more groups around the world are tackling this issue. We are starting to explore what that implementation might look like. This unique group is made up of genetic counselors and experts in implementation, human centered design, and outcomes assessment. The PPH team is creating implementation pathways, patient and provider tools, and guides that work across various clinical contexts so that they may be accessible to any health system interested in practicing precision medicine. We are doing this to advance our mission to improve population health equitably by creating innovations and solutions that lead to widespread integration of precision medicine in healthcare and everyday life.

So, is it time for health systems to offer preventive genomic screening? We think so and we are working to help them do it!

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