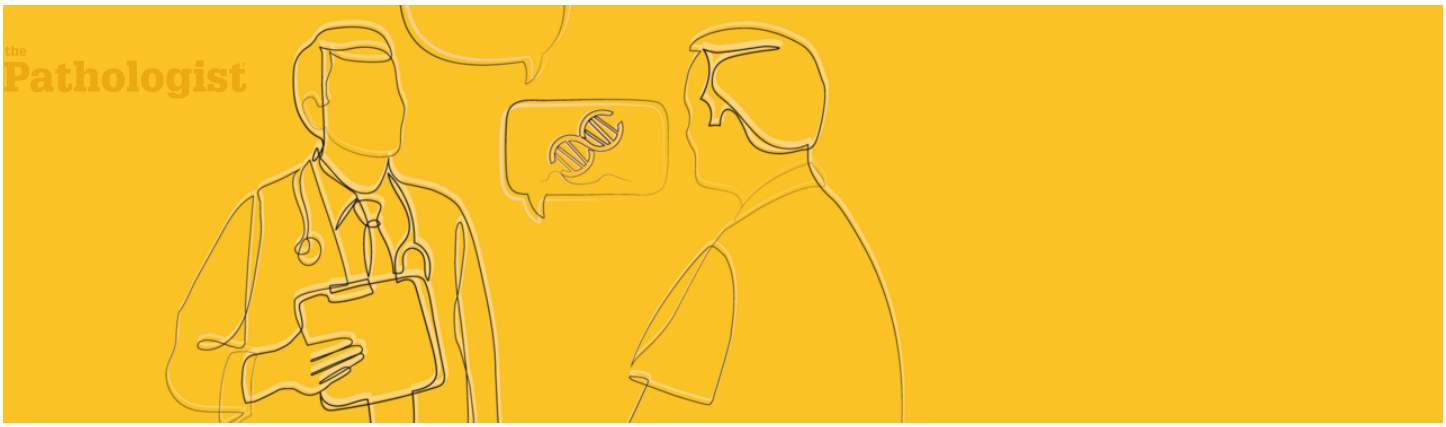

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Preventive Genomics Clinics: The Future of Precision Medicine?

What do preventive genomics clinics mean for patients?

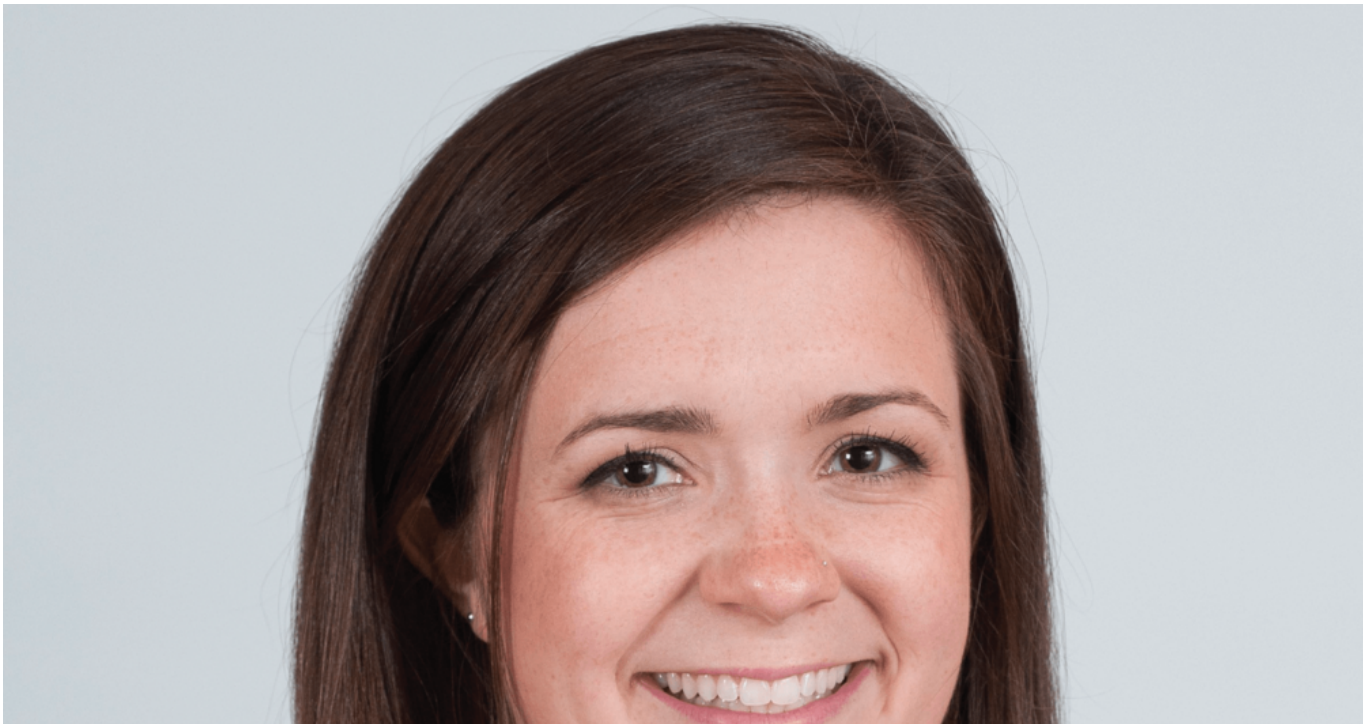
Luke Turner | 12/21/2020 | Longer Read

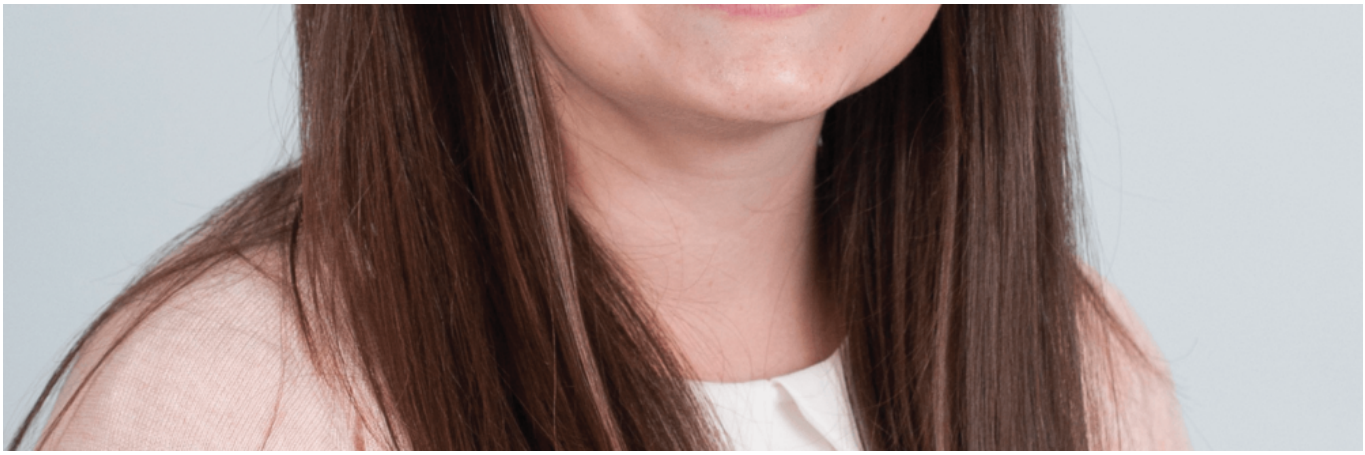
Personal genetic testing is here to stay – so Bethany Zettler and Renée Pelletier recommend preventive genomics clinics as a way to offer the testing patients want without bypassing the guidance they need to fully understand their results.

Since the turn of the millennium, we have made great progress in understanding

how hereditary differences in DNA impact an individual's risk of issues such as cancer, cardiovascular disease, and diabetes. Today, thanks to the recent rise in direct-to-consumer (DTC) genetic testing, it has never been easier to access your own genetic information. It is estimated that, by 2021, 100 million people will have used a DTC genetic test (1), setting the market on track to be worth US$6.36 billion by 2028 (2). But as the number of people who want to discover their own genome increases, so does the need to educate consumers about the implications of their results. This is something that two health systems in Boston are now aiming to address with the introduction of their own preventive gene sequencing clinics.

Brigham and Women's Hospital's Preventive Genomics Clinic provides comprehensive genome sequencing, interpretation, and risk reporting to healthy adults and children. "We discovered that 15–20 percent of apparently healthy people have a strong genetic risk factor for disease – and nearly everyone carries recessive traits that could lead to serious disease in children," says Bethany Zettler, Genetic Counselor and Project Manager. "Preventive genomics is an important milestone in shifting medicine from a reactive, treatment-based model to one where illness can be prevented." More recently, Massachusetts General Hospital (MGH) launched their own Preventive Genomics Clinic, which brings together genetic counselors, clinical geneticists, and physicians to offer personalized testing and treatment plans based on genomic interpretation.





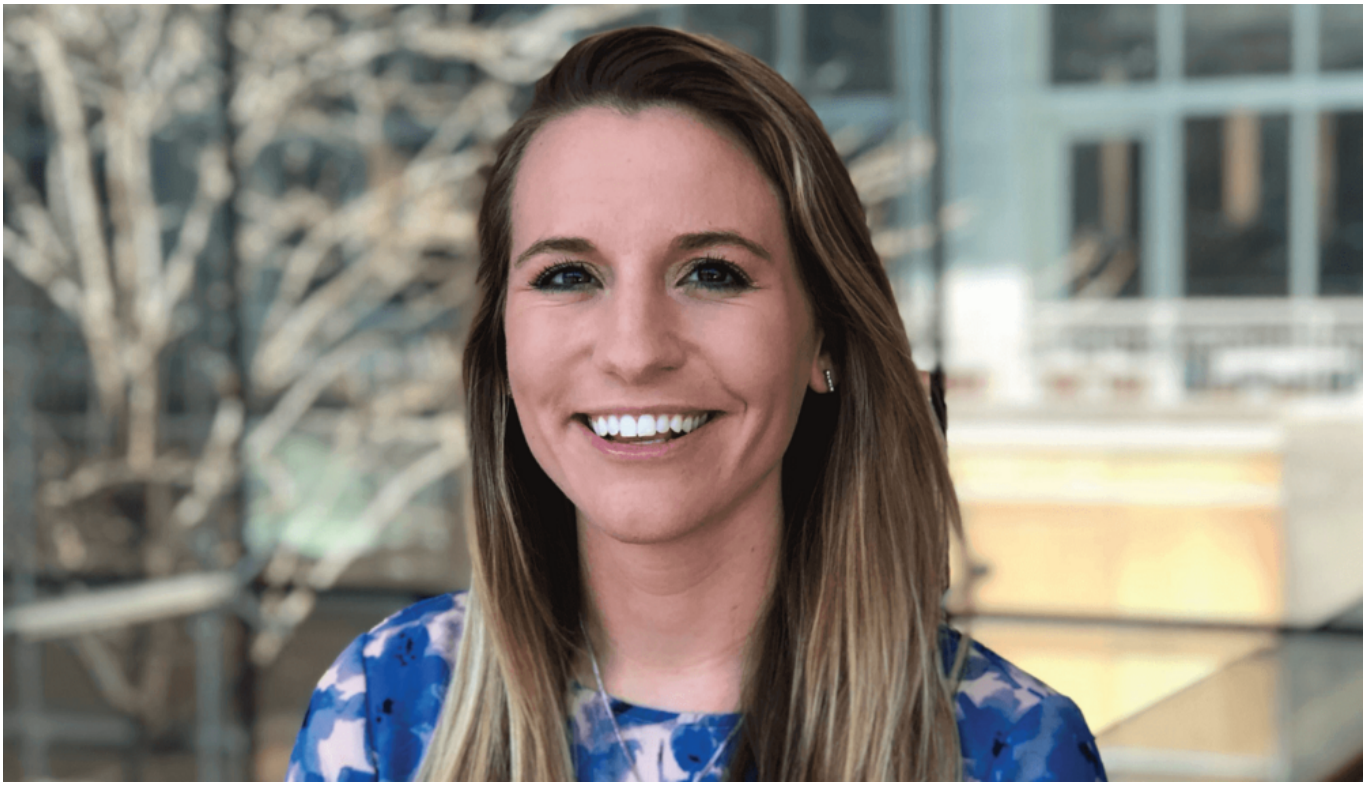
Renée Pelletier

> ***Although genetic information certainly provides another tool in the physician's armory, the best way to distribute genetic information is less clear.***

"Despite a growing interest from MGH patients in medical genetics, there were several key questions for us to first consider," says Renée Pelletier, a Genetic Counselor at MGH. "Which tests are most appropriate and clinically valid? What are the implications of genetic testing for disability and life insurance? How do we best craft individualized care plans? And which patients will benefit most from genetic testing?" Although genetic information certainly provides another tool in the physician's armory, the best way to distribute genetic information is less clear. "Some people advocate for a direct-to-consumer model, but others would prefer a 'consumer-initiated, physician-mediated' approach or even a traditional clinical model," explains Zettler. "Like anything in medicine, it is important to have a range of valid options; what is convenient for one person may be inaccessible for another."

But just how accessible are these options for consumers? "In many cases at MGH, we work with patients' insurance companies or commercial testing companies so that the test is covered at minimal or no cost to the patient," says Pelletier. "We have both genetic counselors and a dedicated genetic testing assistant to help patients navigate the testing options and costs associated." However, when there are no symptoms or family history of disease, genetic testing is not currently the standard of care – and most insurance companies won't cover the service.

Genetic tests can cost as little as $50 or as much as $3,000 – and, for those who cannot afford to shell out, the only option may be to participate in research projects. “Health inequity is a nefarious problem across the entire medical system – and genomics is no different,” says Zettler. “Genomes2People is one of our research studies that strives to make genetic testing more accessible to historically underrepresented populations. For example, we recently began the first effort to provide genetic risk information to a large cohort of African Americans in the Jackson Heart Study.” MGH offers interested patients the opportunity to participate in nationwide programs such as AllofUs, an NIH-funded initiative to enroll and ultimately sequence at least one million Americans. Although the program is committed to returning genetic data to those who are interested, it can take years to receive sequencing results through this route.



Bethany Zettler

> ***The rise of over-the-counter tests has triggered a surge of interest and engagement in personal genetics.***

It’s clear that our DNA can provide insightful and potentially crucial health

information – and the rise of over-the-counter tests has triggered a surge of interest and engagement in personal genetics. “The issue is that many of these tests are not held to the same standard as clinical laboratories to ensure that they are accurate and appropriately addressing the needs of the patient,” explains Pelletier. “What’s more, the results of these tests are typically not integrated into the healthcare system, leaving patients to understand, communicate, and act upon their results alone.” This is where preventive genomics clinics help patients to navigate next steps based on their genetic information, including an in-depth review of results by a clinical laboratory geneticist, genetic counseling, ordering additional testing, and personalized clinical management plans.

One of the classic arguments against DTC testing is that it can cause unnecessary worry for patients who discover potentially harmful genetic risk factors. The Genomes2People research program at Brigham and Women’s Hospital has studied the behavioral and economic outcomes of genome sequencing, finding that even those who receive high-risk information don’t experience psychological distress or incur significantly higher healthcare costs. “Genetic sequencing offers clear potential for precision medicine,” says Zettler. “For example, take cancer or high cholesterol. Someone with a genetic risk factor for colon cancer could have yearly screening colonoscopies to remove polyps, which is 10 times more frequent than population guidelines. Another person might have a genetic risk factor for high cholesterol and start a statin in their 20s to prevent early-onset heart disease.”

As the popularity of DTC testing and the prevalence of preventive genomics clinics grows, genetic information is becoming another tool that patients and doctors can use to make personalized predictions about disease risk. Despite early fears over cost, health equality, and accuracy, it seems clear that the future of precision medicine lies in a more proactive approach to healthcare.

References

About the Author

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While completing my undergraduate degree in Biology, I soon discovered that my passion and strength was for writing about science rather than working in the lab. My master's degree in Science Communication allowed me to develop my science writing skills and I was lucky enough to come to Texere Publishing straight from University. Here I am given the opportunity to write about cutting edge research and engage with leading scientists, while also being part of a fantastic team!

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