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Perkin Elmer, Helix collaborate to launch a new genetic screening test

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GenePrism: Actionable Insights is a new genetic screening test, offering the most comprehensive clinical-grade DNA sequencing and interpretation on the market right now, for anyone who wants to learn about underlying disease risks.



In collaboration with Helix, PerkinElmer, Inc., a global leader committed to innovating for a healthier world has launched GenePrism: Actionable Insights, a new genetic screening test, offering the most comprehensive clinical-grade DNA sequencing and interpretation on the market right now, for anyone who wants to learn about underlying disease risks.

Although there are approximately 6,000 genes associated with disease, just a fraction are considered medically actionable or have a therapeutic option. GenePrism: Actionable Insights specifically analyzes a subset of 59 medically actionable genes identified by the American College of Medical Genetics and Genomics (ACMG). Clinically significant changes found in one of the genes greatly increases a person's likelihood of developing the associated health risk, which range from high cholesterol to breast, ovarian or colon cancer. However, with early intervention and proactive health management, these potentially life-threatening conditions are often alleviated or, in some cases, completely prevented.

"With the growing interest in DNA testing, the healthcare community is feeling pressure from an influx of patients looking for health-related insights and answers about their genetic makeup," said Dr. Madhuri Hegde, Ph.D., FACMG, vice president and chief scientific officer, PerkinElmer Genomics. "GenePrism: Actionable Insights is inherently different from other tests on the market because it leverages PerkinElmer's longstanding diagnostics expertise and state-of-the-art clinical genomics program. People will gain insights into potential genetic risks for disease through an interactive results portal and guidance from genetics experts via telemedicine. This allows them to be proactive with their health, and healthcare providers to spend additional time with patients whose results require further action or testing."

Users' DNA is sequenced by Helix, then results are interpreted by board-certified medical geneticists at PerkinElmer Genomics using ODIN (Ordered Data Interpretation Network), PerkinElmer's proprietary high-throughput software platform. While most commercially available tests only look at an extremely limited number of letters in a gene sequence, which represent a very small percentage of a person's overall disease risk and can provide a false sense of reassurance or concern, GenePrism: Actionable Insights analyzes each of the 59 genes in its entirety—including BRCA1 and BRCA2. PerkinElmer Genomics also maintains one of the largest databases of known genetic variations from different ancestries around the world, so GenePrism: Actionable Insights customers receive a more in-depth assessment.

Individuals who purchase GenePrism: Actionable Insights from Helix must first complete a medical questionnaire and receive physician authorization before submitting a DNA sample. Thanks to a collaboration with Genome Medical, customers may do so right from home. Genome Medical will thoroughly vet users' personal and family medical history through its team of physicians, geneticists and genetic counselors. GenePrism: Actionable Insights testing report also includes free genetic counseling sessions from Genome Medical, to ensure customers understand their results fully and in the correct context.

"Typically, clinical genetic testing that detects ACMG 59 genes is restricted to people with a current illness or specific indicators in their family histories. On a population basis, three to four percent of people have a genetic variant that can significantly impact their health, but until now, virtually no one was able to access this critical information even if they request it from their doctor directly," said Justin Kao, co-founder and senior vice president, Business Development & Partnerships, Helix. "Thanks to the affordable clinical-grade sequencing offered by the Helix Exome+ assay, and our ongoing collaboration with PerkinElmer Genomics, we are enabling the average person access to ACMG 59 panel. Now, healthy individuals who were previously ineligible for clinical genetic testing can confidently know if they've inherited a disease-associated variant and take proactive health measures."

PerkinElmer enables scientists, researchers and clinicians to address their most critical challenges across science and healthcare.