PacBio Enters High-Throughput Carrier Screening Market with Standalone HiFi Sequencing Assay for Challenging Genes

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Enhanced PureTarget portfolio replaces the need for multiple tests with single scalable solution for inherited disease screening

MENLO PARK, Calif., Sept. 22, 2025 (GLOBE NEWSWIRE) -- PacBio (NASDAQ: PACB), a leading developer of high-quality, highly accurate sequencing solutions, today announced its entry into the high-throughput carrier screening market with a significantly expanded and enhanced suite of PureTarget products. The updated solutions leverage PacBio's highly accurate HiFi sequencing technology to allow clinical laboratories to consolidate multiple specialized assays into a single, scalable test capable of resolving some of the most challenging genes associated with inherited conditions.

Recent research shows that up to 71% of individuals carry at least one pathogenic variant, highlighting the growing importance of carrier screening in family planning.[1] The use of carrier screening is rapidly expanding across commercial labs, health systems, and government-funded programs worldwide. The analysis of technically difficult hereditary genes – such as those linked to fragile X syndrome (*FMR1*), spinal muscular atrophy (*SMN1*), and Friedreich Ataxia (*FXN*) – has historically required multiple technologies and specialized workflows. This fragmentation slowed adoption of comprehensive carrier screening, increased costs, and limited global access.

PacBio's expanded PureTarget portfolio now provides laboratories with broad, accurate carrier screening solutions, including coverage of all challenging tier 3 genes identified in the American College of Medical Genetics technical standard.[2] The panels are available in 24- and 96-sample kit formats, with three complementary configurations designed to meet a range of laboratory needs:

- A carrier screening panel for inherited reproductive conditions
- A repeat expansion disorder panel for neurological diseases
- A control panel to support custom assay design and validation

Together, these kits allow laboratories to replace multiple specialized assays with a single streamlined workflow, adaptable from targeted clinical programs to national population-screening initiatives.

"PureTarget is a breakthrough because it focuses PacBio's outstanding long-read sequencing quality on clinically relevant, hard-to-sequence regions across many samples at once," said Dale Muzzey, Chief Scientific Officer of Myriad Genomics. "This approach not only streamlines lab workflows by reducing the need for multiple specialized assays, but it can also enhance both the sensitivity and specificity of results. It's a win for labs, clinicians, and patients."

The upgraded kits support throughput of up to 100,000 samples per year on a single Revio system, making them ideal for population-scale screening initiatives, reproductive health clinics, and large health systems.

"Clinical laboratories need scalable, accurate, and easy-to-use carrier screening tools," said Christian Henry, President and Chief Executive Officer of PacBio. "Our carrier screening products consolidate fragmented workflows into one test, enabling laboratories everywhere to offer comprehensive carrier screening at scale, with high confidence in clinical outcomes and operational efficiency. All of this is possible at price points required to enable laboratories to make a profit given the reimbursement rates available for this type of testing."

With the addition of a carrier screening panel, expanded disease gene content, automation support, and custom panel design, PureTarget gives laboratories the flexibility to scale programs securely and efficiently. PacBio's entry into this market shows a clear commitment to simplifying complex genomic testing and expanding access to comprehensive screening worldwide.

For more information, visit: https://www.pacb.com/technology/puretarget.

Notes

[1] Guo, D., et al. (2025). Regional patterns of genetic variants in expanded carrier screening. *Frontiers in Genetics*. https://doi.org/10.3389/fgene.2025.1527228

[2] Guha, S., Aarabi, M., DiStefano, M., Wakeling, E., et al. (2024). *Laboratory testing for preconception/prenatal carrier screening: ACMG technical standard. Genetics in Medicine*. https://doi.org/10.1016/j.gim.2024.101137

About PacBio

PacBio (NASDAQ: PACB) is a premier life science technology company that designs, develops, and manufactures advanced sequencing solutions to help scientists and clinical researchers resolve genetically complex problems. Our products and technologies, which include our HiFi long-read sequencing, address solutions across a broad set of research applications including human germline sequencing, plant and animal sciences, infectious disease and microbiology, oncology, and other emerging applications. For more information, please visit www.pacb.com and follow @PacBio.
PacBio products are provided for Research Use Only. Not for use in diagnostic procedures.

Forward-Looking Statements

This press release contains "forward-looking statements" within the meaning of Section 21E of the Securities Exchange Act of 1934, as amended, and the U.S. Private Securities Litigation Reform Act of 1995. All statements other than statements of historical fact are forward-looking statements, including statements relating to the uses, advantages, quality or performance of, or the benefits or expected benefits of using, PacBio products or technologies, including in connection with PureTarget, consolidating multiple assays into a single broad and accurate panel assay, streamlined

workflows, interrogating difficult genes, operational efficiency, throughput, scalability, customization, automation, accuracy, ease of use, potential use cases, and other future events. You should not place undue reliance on forward-looking statements because they are subject to assumptions, risks, and uncertainties that could cause actual outcomes and results to differ materially from currently anticipated results. These risks include, but are not limited to, risks inherent in developing and commercializing new technologies; rapidly changing technologies and extensive competition in genomic sequencing; unanticipated increases in tariffs, costs or expenses; interruptions or delays in the supply of components or materials for, or manufacturing of, PacBio products and products under development; third-party claims alleging infringement of patents and proprietary rights or seeking to invalidate PacBio's patents or proprietary rights; diagnostic product promotion regulations; and other risks associated with general macroeconomic conditions and geopolitical instability. Additional factors that could materially affect actual results can be found in PacBio's most recent filings with the Securities and Exchange Commission, including PacBio's most recent reports on Forms 8-K, 10-K, and 10-Q, and include those listed under the caption “Risk Factors.” These forward-looking statements, including PacBio’s preliminary unaudited financial information and PacBio’s financial guidance, are based on current expectations and speak only as of the date hereof; except as required by law, PacBio disclaims any obligation to revise or update these forward-looking statements to reflect events or circumstances in the future, even if new information becomes available.

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