PacBio and Radboud UMC Announce Remarkable Study Results Using HiFi Long Read Sequencing to Help Advance Rare Disease Diagnostics

January 14, 2025 at 9:05 AM EST

MENLO PARK, Calif., Jan. 14, 2025 (GLOBE NEWSWIRE) -- PacBio (NASDAQ: PACB), a leading provider of highly accurate long-read sequencing solutions, announced a significant publication from Radboud University Medical Center (Radboudumc) and its research partners in the *American Journal of Human Genetics*. The study demonstrates the impact of PacBio's HiFi long-read sequencing technology in identifying genetic causes of rare diseases, illustrating the feasibility of potentially replacing multiple diagnostic tests with a single, more comprehensive approach.

The study, led by Christian Gilissen and Lisenka Vissers of Radboudumc, used PacBio's Revio platform and HiFi long reads to analyze 100 challenging patient samples where the genetic causes of rare diseases had been difficult to identify in previous investigations using short reads combined with various supplementary tests. HiFi sequencing results from these samples identified an impressive 93% of pathogenic variants, possibly enhancing cost-effective diagnostic implementation. HiFi technology also enabled the detection of genetic variants missed by short-read approaches, including complex structural variants and DNA methylation abnormalities to be identified.

Metrics Demonstrating Momentum Towards Clinical Genomics Implementation

In support of this study and a broader transition to HiFi long-reads, PacBio and Radboudumc are accelerating efforts to study bringing this approach into clinical practice. The collaboration is advancing rapidly, with recent milestones demonstrating measurable progress:

- **Nearly 1,000 samples processed**: As of January 10, 2025, 981 samples have been sequenced, with 862 fully analyzed since August 2024.
- **Operational improvements**: Automated library preparation now enables 24 samples per run, with plans to scale up to 96 samples per run for even greater throughput.
- **Streamlined workflows**: Advanced protocols and a graphical user interface are being optimized to support future diagnostics for all variant types, with the first-tier diagnostic rollout targeted for summer 2025.
- **Commitment to 5,000 diagnostic genomes**: In the fourth quarter of 2024, Radboudumc expanded its Revio instrument fleet to support an order for 5,000 genomes on SPRQ chemistry.

Christian Henry, President and CEO of PacBio, remarked, "The diagnostic capabilities demonstrated in this study represent a watershed moment for the potential of genomic medicine. PacBio is honored to partner with Radboudumc and other leading centers by delivering SPRQ chemistry for 5,000 genomes as we work together to study ways to simplify diagnostics, increase accuracy, and improve patient outcomes. These metrics reflect our shared commitment to advancing clinical genomics with scalable, practical solutions."

Rare Disease Diagnostics Take a Major Step Forward
Dr. Alexander Hoischen, Professor of Genomic Technologies at Radboudumc, emphasizes the significance of these advancements: "For patients and families affected by rare diseases, HiFi sequencing offers a much-needed pathway to answers. This study not only demonstrates the diagnostic power of long reads but also lays the groundwork for their clinical adoption. The scale and efficiency metrics from our collaboration show the potential feasibility of making these advanced diagnostics available to more patients worldwide."

A Defining Moment for Rare Disease Genomics
This study demonstrates the advantages of HiFi long reads in potentially consolidating multiple diagnostic tests into a single, highly accurate, and comprehensive solution. Researchers anticipate that continued advancements and decreasing technology costs will accelerate its potential adoption as the preferred method for rare disease diagnostics.

The study, *HiFi long-read genomes for difficult-to-detect, clinically relevant variants*, is now available in the *American Journal of Human Genetics*.

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 stem from two highly differentiated core technologies focused on accuracy, quality and completeness which include our HiFi long-read sequencing and our SBB® short-read sequencing technologies. Our products 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.

About Radboud UMC:

Radboud university medical center is a university medical center for patient care, research and education, located in Nijmegen. Radboud university medical center strives to be at the forefront of shaping the health and healthcare of the future. This is reflected in our mission: 'to have a significant impact on health and healthcare'. It demands that we innovate and reinvigorate through collaboration within our networks and with the focus on the individual person. We mainly concentrate on prevention, meaningful and prudent healthcare, sustainability, artificial intelligence and data-driven systems, the molecular mechanisms of diseases and new treatments, and training the professionals of tomorrow.

Forward Looking Statements

This press release may contain “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, coverage, advantages, and benefits or expected benefits of using, PacBio products or technologies, including the use of Revio and HiFi sequencing technology in connection with the identification of genetic causes of rare diseases and its related potential use in, and PacBio’s commitment to advancing, clinical genomics; anticipation that continued advancements and decreasing technology costs will accelerate the potential adoption of HiFi long reads as the preferred method for rare disease diagnostics; and other future events. You should not place undue reliance on forward-looking statements because they are subject to assumptions, risks, and uncertainties and could cause actual outcomes and results to differ materially from currently anticipated results, including, the difficulty of generating discoveries in the study of rare diseases and the potential use of PacBio’s long-read and HiFi technology in a clinical context; potential performance, quality and regulatory issues; and third-party claims alleging infringement of patents and proprietary rights or seeking to invalidate PacBio's patents or proprietary rights. 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 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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