HiFi Solves Consortium Publishes First Major Study Demonstrating the Clinical Research Power of PacBio HiFi Genomes

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New multi-center study identifies 100% of clinically relevant variants in the study, including those not detected by short-read sequencing technologies, positioning HiFi as the bridge between research and clinical-grade genomics

MENLO PARK, Calif., Nov. 05, 2025 (GLOBE NEWSWIRE) -- PacBio (NASDAQ: PACB), developer of the world's most advanced sequencing technologies, today announced the publication of a new preprint from the HiFi Solves EMEA Consortium, *"HiFi sequencing accurately identifies clinically relevant variants in paralogous genes."* The study shows that PacBio HiFi sequencing combined with Paraphase, a dedicated haplotype-based variant caller, uncovered all known clinically relevant variants present in the study population - even in the hardest-to-sequence regions of the genome - demonstrating its readiness to power the future of clinical discovery.

From five institutions across Europe, in a cohort of 86 individuals carrying 125 known pathogenic variants across 11 complex genomic regions, HiFi sequencing combined with Paraphase detected all 125 clinically relevant variants in the study. The work represents the strongest validation to date of HiFi Solves' founding goal: to show that high-accuracy long reads can bridge today's research with tomorrow's clinical utility.

"When we launched HiFi Solves, we knew that HiFi sequencing could bridge the gap between research and clinical utility," said Christian Henry, President and Chief Executive Officer of PacBio. "The results of this study show that a single HiFi genome can replace multiple separate tests, helping researchers find answers faster and more efficiently while saving time, resources, and significant costs."

Each sample was sequenced on a single SMRT Cell with a median read length of 15.5 kb and accuracy, generating highly accurate HiFi reads with mean per-base accuracy above 99.9%. The multi-center team proved that HiFi sequencing could accurately phase variants, resolve copy-number changes between genes and pseudogenes, and detect complex events such as gene conversions, providing a new a level of full haplotype resolution and copy-number precision that surpasses traditional short-read and targeted approaches.

"This first clinical utility study from the HiFi Solves EMEA Consortium demonstrates the potential of PacBio HiFi long-read sequencing for use in clinical genetics. Across multiple diagnostic laboratories and variant types, HiFi sequencing with Paraphase consistently and accurately identified all known pathogenic variants in the study, including those in regions long considered inaccessible by standard technologies. This multi-center validation provides compelling evidence that HiFi long-read sequencing is robust, reproducible, and capable of addressing some of the most challenging cases in genomic medicine. We believe these results mark a pivotal step toward the widespread adoption of long-read genomes in routine clinical testing and rare disease diagnostics," stated the senior authors of this study, professors Spielmann, Zschocke, Bolz and Hoischen on behalf of the HiFi Solves EMEA Consortium.

Founded in 2023, HiFi Solves unites leading clinical and research institutions worldwide to evaluate the real-world utility of PacBio HiFi sequencing for human health applications. The consortium now includes 23 institutions across 16 countries, spanning Europe, Asia Pacific, and North America.

"I am excited about the convincing outcome of the first multi-center clinical utility study of our HiFi Solves EMEA consortium. What's most remarkable is that this technology performs robustly across multiple centers, with 100% detection rate of very challenging, clinically relevant variants. That reproducibility is key to establishing long-read genomes as part of routine clinical testing," said Prof. Alexander Hoischen, from the Department of Human Genetics in Nijmegen. "Genes that have pseudogene copies – or paralogous sequences with >99% sequence homology – such as *CYP21A2, SMN1/SMN2* and *IKBKG* have long been amongst the toughest to study. HiFi sequencing illuminates them, allowing now to accurately detect pathogenic variants of all variant types including gene conversions that previously posed extreme challenges to genetic laboratories."

By uniting leading research centers and clinicians around the world, the HiFi Solves Consortium is turning data into deeper understanding and showing how complete genomes can fundamentally change what's possible in rare disease research.

Want to learn more about HiFi Solves Consortium? Visit: https://www.pacb.com/hifi-solves/

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 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 advantages, or quality or performance of, or benefits or expected benefits or advantages of using, PacBio products or technologies, such as use in future clinical discovery and utility; changing how rare diseases are understood and studied; powering the future of clinical discovery; widespread adoption in routine testing; accurately detect pathogenic variants of all variant types; 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, challenges inherent in sequencing a large number of genomes, including in a clinical context; regulations regarding, and potential approvals or clearances required for, using, marketing or promoting

products for clinical or diagnostic use; third-party claims alleging infringement of patents and proprietary rights or seeking to invalidate PacBio's patents or proprietary rights; and other risks associated with international operations. 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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