



PacBio Announces Collaboration with Genomics England to Sequence Biobanked Rare Disease Samples Using HiFi Sequencing Technology

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Pilot project designed to demonstrate the power of HiFi sequencing to effectively and efficiently identify novel variants following short-read genome sequencing

MENLO PARK, Calif., Jan. 11, 2022 (GLOBE NEWSWIRE) -- [PacBio](#) (NASDAQ: PACB), a leading provider of high-quality, highly accurate sequencing platforms, today announced a collaboration with [Genomics England](#) to study PacBio's technology for identifying genetic variants in unexplained rare disease cases.

Genomics England was initially created to deliver the 100,000 Genomes Project – the largest whole genome sequencing disease cohort of cancer and rare disease participants in the National Health Service (NHS). The study will re-sequence a selection of samples collected during Genomics England's 100,000 Genomes Project, which were previously analyzed with short read sequencing technology. The study is intended to reveal potential operational and clinical benefits of long-read sequencing in identifying genetic mutations associated with rare diseases.

"PacBio HiFi sequencing is a powerful tool for understanding the genetic underpinnings of rare disease, cancer, and other applications as it provides clinical researchers with the ability to view the genome more completely than other sequencing technologies," said Christian Henry, President and Chief Executive Officer of PacBio. "Our collaboration with Genomics England is part of a broader strategy to demonstrate the benefits of HiFi sequencing when attempting to identify rare diseases, and follows recent announcements of rare disease-focused research collaborations with [Radboud University Medical Center](#), [Care4Rare Canada Consortium](#), [ARUP Laboratories](#), [UCLA Health](#), [Rady Children's Institute for Genomic Medicine](#), and [Children's Mercy Kansas City](#)."

"The 100,000 Genomes Project was able to find actionable mutations in around 25 percent of patients with rare disease," said Parker Moss, Chief Ecosystem & Partnership Officer at Genomics England. "We are excited to discover whether new technologies like PacBio's HiFi, can identify additional genomic variants in regions of the genome that are not readily accessible by short-read sequencing technologies. We are hopeful that additional insight gained during the study may, ultimately, lead to new therapeutic or clinical trial options for patients with rare disease. This study represents our continued commitment to the 100,000 Genomes Project participants, and also to our quest to seek out the benefits of new disruptive technologies."

About PacBio

Pacific Biosciences of California, Inc. (NASDAQ: PACB) is empowering life scientists with highly accurate sequencing platforms. The company's innovative instruments are based on Single Molecule, Real-Time (SMRT®) Sequencing technology, which delivers a comprehensive view of genomes, transcriptomes, and epigenomes, enabling access to the full spectrum of genetic variation in any organism. Cited in thousands of peer-reviewed publications, PacBio® sequencing systems are in use by scientists around the world to drive discovery in human biomedical research, plant and animal sciences, and microbiology. 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, including statements relating to PacBio's collaboration with Genomics England in connection with the 100,000 Genomes Project and efforts to further identify causes of rare disease; anticipated efforts and outcomes in connection with such collaboration; and interest in and anticipated capabilities or benefits of PacBio's products and technology, including in connection with the detection of genomic variants and providing information that may potentially lead to new therapeutic or clinical trial options, or increase diagnostic yield, for individuals with rare diseases. Readers are cautioned not to place undue reliance on these forward-looking statements and any such forward-looking statements are qualified in their entirety by reference to the following cautionary statements. All forward-looking statements speak only as of the date of this press release and are based on current expectations and involve a number of assumptions, risks and uncertainties that could cause the actual results to differ materially from such forward-looking statements. Readers are strongly encouraged to read the full cautionary statements contained in the Company's filings with the Securities and Exchange Commission, including the risks set forth in the company's Forms 8-K, 10-K, and 10-Q. The Company disclaims any obligation to update or revise any forward-looking statements.

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