PacBio and Boston Children's Hospital Collaborate to Investigate Novel Variants Inaccessible by Short-Read Sequencing

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Researchers to Investigate Genetic Variants Associated with Rare Pediatric Diseases

MENLO PARK, Calif., Dec. 6, 2022 /PRNewswire/ -- PacBio, a leading developer of high-quality, highly accurate sequencing solutions, today announced its HiFi sequencing technology will be used in a pilot project for the **C**hildren's **R**are **D**isease **C**ohorts Initiative (CRDC) at Boston Children's Hospital. Boston Children's Hospital researchers will use HiFi whole-genome sequencing (HiFi WGS) for the purpose of investigating genetic and epigenetic variants associated with rare pediatric diseases. Samples from individuals enrolled in multiple rare disease cohorts that have remained undiagnosed after short-read whole exome sequencing will be examined using HiFi WGS. The study will also explore epigenetic variation using the ability of HiFi WGS to measure DNA methylation in every sequencing run.

"PacBio's technology has been used to help genetic disease researchers explain mysteries where other technologies could not," said Jonas Korlach, Chief Scientific Officer at PacBio. "We are excited to support the CRDC to help uncover the answers they have been seeking. In previous research conducted using PacBio's technology, HiFi sequencing detected more than two-fold structural variants and 200,000 more single-nucleotide variants than short-read sequencing. Structural variants contributed to over twice the number of new explanations."

"HiFi sequencing holds the promise to investigate parts of the genome inaccessible via other technologies," said Piotr Sliz, Vice President and Chief Research Information Officer at Boston Children's Hospital. "We hope that HiFi sequencing will enable us to explore variation in parts of the genome that were not previously detected, potentially enabling us to expand our understanding of causative variation in these patients."

The CRDC supports genomics research around strategically selected disease areas at Boston Children's Hospital and is led by the CRDC Steering Group. As of today, 48 rare disease cohorts have joined the CRDC and nearly 10,000 patients with rare disease and their family members have been sequenced. As causative variants are identified by CRDC researchers, results are returned to participants. Researchers participating in the CRDC have uncovered novel mutations, are developing targeted therapies and are matching patients with innovative clinical trials.

The pilot project will re-sequence a selection of samples from patients who were previously analyzed with short-read sequencing technology that are strongly suspected to have a genetic condition but have not yet been diagnosed. The study is intended to explore the potential benefits of HiFi WGS in identifying genetic mutations and epigenetic variants associated with rare diseases, in particular those that may be associated with a range of conditions, such as brain malformations, predisposition to leukemia, congenital hearing loss, epilepsies, and glomerular disease.

About PacBio
Pacific Biosciences of California, Inc. (NASDAQ: PACB) is a premier life science technology company that is designing, developing and manufacturing advanced sequencing solutions to help scientists and clinical researchers resolve genetically complex problems. Our products and technology under development stem from two highly differentiated core technologies focused on accuracy, quality and completeness which include our existing HiFi long read sequencing and our emerging 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.

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 future availability, uses, accuracy, advantages, quality or performance of, or benefits or expected benefits of using: PacBio products or technologies, including in connection with the CRDC; investigating genetic and epigenetic variants associated with rare pediatric diseases study; and other future events. 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, including, among others, potential product performance and quality issues; and successfully completing a study that has not yet commenced. Readers are strongly encouraged to read the full cautionary statements contained in PacBio'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. PacBio disclaims any obligation to update or revise any forward-looking statements.

Contact:

Investors:
Todd Friedman
ir@pacificbiosciences.com

Media:
Lizelda Lopez
pr@pacificbiosciences.com

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