Pacific Biosciences and Rady Children's Institute for Genomic Medicine Announce its First Research Collaboration for Whole Genome Sequencing

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HiFi Sequencing will be used on a cohort of rare disease cases with the aim to identify numerous variants, both small and structural, that are not readily detectable by short-read sequencing

MENLO PARK, Calif., June 23, 2021 (GLOBE NEWSWIRE) -- Pacific Biosciences of California, Inc. (Nasdaq: PACB)("Pacific Biosciences" or "PacBio"), a leading provider of high-quality, long-read sequencing platforms, and Rady Children's Institute for Genomic Medicine (RCIGM), a mission-driven, non-profit seeking to save lives and improve outcomes for patients, clinicians and families, shared today that they are collaborating on a study which aims to identify potential disease-causing genetic variants and increase the solve rates of rare diseases.

The study is focused on long-read whole genome sequencing of rare disease cases for which previous short-read whole genome and exome sequencing yielded no answers. The study, which is currently underway, was able to detect variants that were not identified by short-read sequencing (SRS); of these, an average of 37 were missense mutations in known disease genes.

"PacBio HiFi sequencing can identify numerous variants, both small and structural that are not readily detectable by SRS," said Matthew Bainbridge, Principal Investigator, and Associate Director of Clinical Genomics at RCIGM. "We sequenced this cohort of patients to 10-30X depth of coverage using Pacific Biosciences HiFi long-read technology to assess whether there was an increase in the identification of these variants. We are very pleased by the preliminary results delivered in this collaboration with the team at PacBio."

It is estimated that as many as 25 million Americans — approximately 1 in 13 people — are affected by a rare, and often undiagnosed condition. Ir rare disease studies, conventional techniques for whole-genome and whole-exome analysis based on SRS typically led to identification of a causal variant in less than 50% of cases. Utilizing PacBio's Single Molecule, Real-Time (SMRT®) Sequencing technology to generate highly accurate long-reads, known as HiFi reads, clinical researchers have demonstrated that they can detect disease-causing structural and small variants missed by short-read sequencing platforms. This study is designed to evaluate the rate at which HiFi sequencing identifies overlooked causal variation.

"It is an honor to collaborate with the innovative pediatric translational researchers at RCIGM to bring HiFi Sequencing data to bear on some of their most difficult cases of rare pediatric disease, and hopefully give individuals and families answers regarding potential underlying genetic variants, which may ultimately provide healthcare providers with insights to end their diagnostic odysseys," said Christian Henry, CEO and President at PacBio.

"We've been aware that there's a subset of seriously ill babies and children who don't receive a diagnosis with current sequencing methods, but based on their symptoms, we're fairly certain that they have an underpinning genetic disease," said Stephen Kingsmore, MD, DSc, President and CEO of Rady's Children's Institute for Genomic Medicine. "With this new technology, we are excited to see how many more of these children and families will receive additional insight regarding the identification of potential disease-causing genetic variants."

About Pacific Biosciences
Pacific Biosciences of California, Inc. (NASDAQ: PACB) is empowering life scientists with highly accurate long-read sequencing. 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.

About Rady Children's Institute for Genomic Medicine
We are transforming pediatric critical care by advancing disease-specific healthcare for infants and children with rare disease. Discoveries at the Institute are enabling rapid diagnosis and targeted treatment of critically ill newborns and pediatric patients at Rady Children's Hospital-San Diego and a growing network of more than 60 children's hospitals nationwide. The vision is to expand delivery of this life-changing technology to enable the practice of Rapid Precision Medicine™ at children's hospitals across the nation and the world. RCIGM is a non-profit, research institute of Rady Children's Hospital and Health Center. Learn more at www.RadyGenomics.org. Follow us on Twitter and LinkedIn.

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 the collaboration between PacBio and RCIGM, potential use of SMRT sequencing technology to identify, and increase the rate of identification of, potential disease-causing genetic variants in rare disease, the potential of HiFi data, the applications, insights, and attributes of SMRT sequencing technology, and the benefits of PacBio sequencing. 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.

Contacts

Investors:
Todd Friedman
+1 (650) 521-8450
ir@pacificbiosciences.com

Media:
Jen Carroll
+1 (858) 449-8082
pr@pacificbiosciences.com

Grace Sevilla
+1 858-966-1710 (o); +1 619-855-5135 cell (c)
gsevilla@rchsd.org