European Research Initiative Adopts PacBio Sequel II System to Solve Rare Diseases

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SOLVE-RD program will sequence more than 500 human genomes with SMRT Sequencing

MENLO PARK, Calif., Sept. 05, 2019 (GLOBE NEWSWIRE) -- Pacific Biosciences of California, Inc. (Nasdaq:PACB), a leading provider of high-quality sequencing of genomes, transcriptomes and epigenomes, today announced that its Single Molecule, Real-Time (SMRT®) Sequencing technology has been selected by the European research program SOLVE-RD to help reveal the genetic mechanisms responsible for rare genetic diseases. As part of this work, scientists will sequence more than 500 whole human genomes with the PacBio Sequel II System to pinpoint disease-causing variants.

Around the world, some 350 million people have a rare disease[1]. The vast majority of these diseases have a genetic cause, but many have not yet been linked to the specific genetic deleterious element(s) responsible for the disease. The SOLVE-RD research program, a consortium of >20 institutions funded with a five-year, €15 million award from the European Union's Horizon 2020 initiative, aims to improve the diagnosis and treatment of rare diseases by applying novel tools to cases that were not solved with short-read exome sequencing.

"Even with exome sequencing, as many as 50% of rare disease cases remain unsolved," said Alexander Hoischen, Associate Professor for Genomic Technologies and Immuno-Genomics and a member of the SOLVE-RD team, at Radboud University Medical Center. "The SOLVE-RD team believes that long-read SMRT Sequencing will be essential for discovering the causal elements that have proven elusive with previous approaches, and we anticipate that this research will ultimately make it easier for doctors to diagnose other patients with these rare diseases in the future."

Marcel Nelen, Laboratory Specialist in Genome Diagnostics at Radboud University Medical Center, commented: "Our team is eager to deploy PacBio's Sequel II System to generate hundreds of high-quality human genomes for phenotypes very likely to be associated with challenging genomic regions or structural variants including repeat expansions. In our experience, SMRT Sequencing reliably detects far more structural variants — including pathogenic variants — than any other sequencing technology."

Structural variants are increasingly being recognized as the cause of many rare diseases, but these large genomic elements are often undetectable with short-read sequencing tools due to their length and repetitive content. Structural variation affects more base pairs of the human genome than single nucleotide variants and small insertions and deletions combined. With its extraordinarily long reads, SMRT Sequencing detects structural variants with high precision and recall, even at low coverage.

"SMRT Sequencing has already helped scientists untangle previously unsolvable cases of rare disease, and we believe it will provide exciting new answers for many of the medical mysteries targeted by this program," said Jonas Korlach, PhD, Chief Scientific Officer at PacBio. "We congratulate the SOLVE-RD team for securing the funding to perform this important work and look forward to the many new scientific discoveries they will make with PacBio's highly accurate, long-read sequencing technology."

Alexander Hoischen will speak more about this project and long-read sequencing at this week's AGBT Precision Health meeting in La Jolla, Calif. His presentation will take place on Saturday, Sept. 7th, at 10 am PDT.

Reference

1. https://globalgenes.org/rare-facts/

About Pacific Biosciences

Pacific Biosciences of California, Inc. (NASDAQ:PACB) offers sequencing systems to help scientists resolve genetically complex problems. Based on its novel Single Molecule, Real-Time (SMRT®) technology, Pacific Biosciences' products enable: de novo genome assembly to finish genomes in order to more fully identify, annotate and decipher genomic structures; full-length transcript analysis to improve annotations in reference genomes, characterize alternatively spliced isoforms in important gene families, and find novel genes; targeted sequencing to more comprehensively characterize genetic variations; and real-time kinetic information for epigenome characterization. Pacific Biosciences' technology provides high accuracy, ultra-long reads, uniform coverage, and the ability to simultaneously detect epigenetic changes. PacBio® sequencing systems, including consumables and software, provide a simple, fast, end-to-end workflow for SMRT Sequencing. More information is available at www.pacb.com.

Forward-Looking Statements

All statements in this press release that are not historical are forward-looking statements, including, among other things, statements relating to the attributes of the Sequel II System, the future availability, uses, accuracy, quality or performance of, or benefits of using, products or technologies, the suitability or utility of methods, products or technologies for particular applications, studies or projects, the importance of long-read sequencing data, the expected benefits of the SOLVE-RD research program or other sequencing projects, and other future events. You should not place undue reliance on forward-looking statements because they involve known and unknown risks, uncertainties, changes in circumstances and other factors that are, in some cases, beyond Pacific Biosciences' control and could cause actual results to differ materially from the information expressed or implied by forward-looking statements made in this press release. Factors that could materially affect actual results can be found in Pacific Biosciences' most recent filings with the Securities and Exchange Commission, including Pacific Biosciences' most recent reports on Forms 8-K, 10-K and 10-Q, and include those listed under the caption "Risk Factors." Pacific Biosciences undertakes no obligation to revise or update information in this press release to reflect events or circumstances in the future, even if new information becomes available.

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