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Recent Projects Highlight Success of SMRT Sequencing for Characterizing Structural Variation in Human Genomes

PacBio Sequencing Reveals More Functionally Important Variants; Complete Solution for Structural Variant Detection to be Featured at ASHG Annual Meeting

MENLO PARK, Calif., Oct. 16, 2017 (GLOBE NEWSWIRE) -- A series of publications and initiatives from the genomics community demonstrates the rapid adoption of Single Molecule, Real-Time (SMRT®) Sequencing for discovering structural variants throughout the human genome. As these previously overlooked structural variants have been elucidated, there is increasing recognition among scientists of the important role many of them play in human disease. PacBio recently released new analysis software, SMRT Link Structural Variant Calling, to support routine structural variant identification in low-coverage, whole genome SMRT Sequencing data.

Most of the DNA sequence that is different between any two people is not contained in single nucleotide variants (SNVs), but rather in structural variants representing genetic differences of 50 base pairs or longer. Structural variants can involve the deletion and insertion of hundreds of base pairs, making them difficult or impossible to detect with short-read DNA sequencers. SMRT Sequencing produces read lengths averaging 10-18 kb, thus effectively identifying structural variants in a genome, even at low coverage.

"Discovering and identifying structural variants is essential for a deeper understanding of human biology and, increasingly, for characterizing and ultimately diagnosing disease," said Jonas Korlach, Ph.D., Chief Scientific Officer for PacBio. "After years of overlooking structural variants due to technology limitations, scientists are now making enormous leaps in our understanding of these genetic elements with SMRT Sequencing. We expect that this trend will accelerate with continued adoption of the Sequel® System and our new Structural Variant Calling software."

Recent structural variation publications and projects include:

Long-read genome sequencing identifies causal structural variation in a Mendelian disease, *Genetics in Medicine*

> Stanford scientists used whole genome SMRT Sequencing to detect a disease-causing structural variant in an individual left undiagnosed for nearly twenty years, finally confirming Carney complex as the underlying syndrome.

Multi-platform discovery of haplotype-resolved structural variation in human genomes, bioRxiv preprint

> This consortium effort analyzed structural variation in three family trios with several technology platforms, including SMRT Sequencing, and report seven times more structural variation than was previously found with short-read sequencing. The resulting variant data sets can be used as gold standards throughout the genomics community.

Novogene to build a comprehensive Chinese genome database

> To improve precision medicine for the Chinese population, Novogene announced that it will use SMRT Sequencing to develop a structural variant database from genome sequences of 1,000 Chinese individuals.

Discovery and genotyping of structural variation from long-read haploid genome sequence data, *Genome Research*

> Scientists developed the SMRT-SV approach to identify structural variants in two haploid human genomes. They conclude that PacBio long-read sequencing is five times more sensitive to detecting structural variants than short-read sequencers.

Accurate detection of complex structural variations using single molecule sequencing, bioRxiv preprint

> Scientists present two new computational tools, NGMLR and Sniffles, designed to enhance sensitivity and precision for calling structural variants from SMRT Sequencing data.

Complete Solution for Structural Variant Detection Featured at ASHG Annual Meeting

Geneticists and genetics researchers can learn about PacBio's complete solution for structural variant detection, featuring the Sequel System and Structural Variant Calling software, at the American Society for Human Genetics 2017 Annual Meeting in Orlando. Attendees can also visit PacBio in booth #722 or attend the company's workshop on Wednesday, October 18th, at 12:30 pm. More information about PacBio's activities at ASHG is available at www.pacb.com/ASHG2017.

For more information about PacBio's complete solution for structural variation detection, please visit http://www.pacb.com/sv.

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 is the only DNA sequencing technology that provides 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 future availability, uses, quality or performance of, or benefits of using, products or technologies, the suitability of the company's products for particular applications 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."

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