October 3, 2017

Structural Variation Detection a Key Focus of PacBio Sequencing Projects Presented at ASHG Annual Meeting

More than 30 presentations demonstrate utility of SMRT Sequencing for human biomedical applications

MENLO PARK, Calif., Oct. 03, 2017 (GLOBE NEWSWIRE) -- Pacific Biosciences of California, Inc. (Nasdaq:PACB), today announced that the human genetics community is demonstrating increasing recognition of the value of Single Molecule, Real-Time (SMRT®) Sequencing technology for structural variant detection and other key genetics applications, as evidenced by the more than 30 presentations to be featured at the upcoming American Society of Human Genetics (ASHG) annual meeting in Orlando. Significant scientific advances will be presented in structural variation, amplification-free targeted sequencing, and the Iso-Seq® method for transcriptome analysis, among others.

"We are looking forward to updating the community on our recently launched solution for structural variation detection, including our new pbsv software that boosts sensitivity for identifying large insertions and deletions while dramatically lowering project costs for Sequel® System users," said Jonas Korlach, Chief Scientific Officer of PacBio. "We are also excited to share an update on our amplification-free targeted sequencing application in development, which will advance the study of disease-causing loci that are difficult to target and sequence, such as the genes responsible for Huntington's disease and Fragile X syndrome." This method was recently featured in a *Nature* publication focused on Parkinson's disease from scientists at Houston Methodist Research Institute and collaborating institutions.

Scientific Program Highlights

Highlights from the scientific presentations featuring SMRT Sequencing include:

- "Advances in the Genetics of Autoimmune Disease," a Concurrent Platform Session on October 19th from 11:00 am-12:30 pm featuring a talk titled "The MHC Diversity in Africa Resource: A roadmap to understanding HLA diversity in Africa," by Martin Pollard of the Wellcome Trust Sanger Institute.

- "Analysis of Cancer Genome Variation Using Long-read Sequencing," a Concurrent Invited Session on October 19th from 4:15-6:15 pm featuring presentations from Fritz Sedlazeck of Baylor College of Medicine and Jacques Banchereau of Jackson Labs for Genomic Medicine.

- Xufeng Zhao and Mark Chaisson, on behalf of the Human Genome Structural Variation Consortium, will present a poster titled "Comprehensive Discovery of Structural Genomic Variants Through Integration of Multiple Sequencing Platforms," October 18th from 2:00-3:00 pm (Poster #1501). This team recently released a preprint on bioRxiv detailing the structural variant analysis of three family trios, finding 10 times more variants than previous 1000 Genomes Project efforts with short-read technology.

- A "Reviewer's Choice Poster" by PacBio scientist Tyson Clark titled "Targeted Enrichment Without Amplification and SMRT Sequencing of Repeat-expansion Disease Causative Genomic Regions," October 18th from 3:00-4:00 pm (Poster #1480).

- Genome Reference Consortium (GRC) & Genome in a Bottle consortium (GIAB) Workshop on October 17th from 1:00-4:00 pm, which will discuss the latest updates and improvements to the human reference genome assembly.

SMRT Sequencing Workshop

PacBio will hold a workshop entitled "Population and Clinical Genetics Studies Using Long-Read SMRT Sequencing" on Wednesday October 18th between 12:30-1:45 pm. The workshop will be hosted Dr. Korlach, who will be sharing updates on the company's recent progress and technology roadmap.

Speakers at the workshop include: Han Brunner, Head Clinical Genetics, Radboud University Medical Center who will discuss his work using SMRT Sequencing to find genetic variants underlying intellectual disabilities; Karen McFarland,

Professor, University of Florida, College of Medicine, Dept. of Neurology who will discuss using a new PacBio method for amplicon-free enrichment using CRISPR/Cas9 on patient samples with ataxia (SCA10) and Parkinson's disease; and Charles Lee, Scientific Director and Professor at The Jackson Laboratory for Genomic Medicine and Co-PI for the 1000 Genomes Project, SV Working Group, who will discuss recent progress on the 1000 Genomes Project and his work with the Human Genome Structural Variation Consortium.

Attendees interested in reserving a seat at the workshop can register here.

CoLab Session

ASHG meeting attendess will also have the opportunity to hear a talk on the Exhibit Hall floor as part of a CoLab Session on Friday, October 20th, titled "CRISPR/Cas9 enrichment and long-read WGS for structural variant discovery." PacBio and its collaborators have employed this new method to target and sequence multi-kilobase long repetitive elements that cause repeat expansion disorders in genes such as *HTT*, *FMR1*, *C9orf72*, and *ATXN10.* PacBio scientists will also discuss a new low-fold, long-read whole-genome sequencing method that can sensitively detect upwards of 20,000 structural variants in a human genome with a very low 5% false-discovery rate.

PacBio will also be exhibiting at ASHG in booth #722.

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 future 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."

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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