Radboud University Medical Center Increases HiFi Sequencing Capacity with PacBio's Sequel IIe

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Added whole genome sequencing capacity enables the SOLVE-RD research program to significantly increase sample throughput

MENLO PARK, Calif., Jan. 07, 2022 (GLOBE NEWSWIRE) -- PacBio (Nasdaq: PACB), a leading developer of high-quality, highly accurate sequencing platforms, today announced Radboud University Medical Center (Radboudumc) in Nijmegen, the Netherlands will increase their SMRT sequencing capacity with the addition of two new PacBio Sequel IIe Systems.

Radboudumc has been using PacBio long-read technology for the last three years to identify hidden variants that cause rare diseases. "Over time we have adapted our workflows to use our Sequel systems in rare disease cases and with each new system the quality of the test improves while also providing us efficiency and thus cost effectiveness," said Marcel Nelen, Ph.D., Laboratory Specialist Clinical Genetics, Director of Radboud Genomics Technology Center, Radboudumc. "With the contributions of our bioinformatics team, we have been able to develop the infrastructure needed to achieve more routine analysis of long-read SMRT sequencing, enabling us to use it in a diverse array of cases for the detection of variants in regions we know are difficult to explore with short-read sequencing or when using short-read would be laborious and cumbersome."

Among their discoveries, the Radboudumc team has found using PacBio systems makes it easier to identify variants that may cause X-linked color vision deficiencies such as Bornholm Eye Disease and Blue Cone Monochromacy. In addition, they use the Sequel system for mitochondrial DNA (mtDNA) sequencing because it is much more efficient. Unexpected benefits were seen both in efficiency and sensitivity with small amplicon-based sequencing for IGH mutation analysis in CLL (chronic lymphocytic leukemia) cancer samples. Running similar tests on CLL samples using other technology platforms is labor intensive, requiring time-consuming follow-up tests and thus negatively affecting turn-around times.

"Worldwide, approximately 80 percent of rare disease cases are genetic, but more than 50 percent of cases remain unsolved with existing technology. With the accuracy of HiFi sequencing, we are seeing a tremendous interest in PacBio WGS as an important new tool for detecting large or challenging variants missed by short-read technology," said Jonas Korlach, Ph.D., Chief Scientific Officer, PacBio. "PacBio is proud to continue to work closely with Radboudumc, as we have done for the last several years, and to see their growth and be part of their research evolution."

Radboudumc is also involved with several large research projects including the SOLVE-RD European research program – a consortium of more than 20 institutions funded by the European Commission that studies rare diseases. "The SOLVE-RD team knows that long-read HiFi sequencing is essential for discovering the causal elements that have proven elusive with previous approaches and we aim to sequence more than 500 HiFi genomes by the end of 2022 as part of this grant," said Alexander Hoischen, Ph.D., Associate Professor for Genomic Technologies and Immuno-Genomics and a member of the SOLVE-RD team at Radboudumc. "The HiFi genome trios that we have completed with PacBio technology to date are likely the most comprehensive we've ever had at our institute. The increased capacity with these additional systems will enable us to increase our sample throughput so that our research can continue forward and yield additional biological insight that may ultimately make it easier for doctors to diagnose rare disease patients in the future. The value of HiFi genomes in unravelling disease causing, previously hidden, structural variants is becoming apparent now – and in collaboration with several other research groups we hope to start solving unsolved rare disease cases this way." More about Radboudumc's discoveries can be found in their paper on a family with choroideremia and in another on optical genome mapping.

The Sequel IIe System generates HiFi sequencing reads, which provide the accuracy and completeness required for investigating DNA variation in rare genetic diseases and cancer. Short-read WGS can lead to sequence gaps and incomplete coverage of disease-causing gene regions. HiFi WGS produces reads approximately 100 times longer than short-read WGS, enabling haplotype phasing and comprehensive variant detection beyond small variant focused short reads.

To learn more about the benefits of HiFi WGS in rare disease, visit www.pacb.com/rare-disease.

About Pacific Biosciences

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

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