PacBio and ARUP Laboratories Collaborate in a Study to Improve Rare Disease Diagnosis

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Whole genome HiFi sequencing will be used on unexplained rare disease cases to evaluate if it can identify variants not readily detectable by short-read sequencing

MENLO PARK, Calif., Nov. 18, 2021 (GLOBE NEWSWIRE) -- PacBio (NASDAQ: PACB), a leading provider of high-quality, highly accurate sequencing platforms, and ARUP Laboratories announced today they are collaborating on a study intended to evaluate whether the solve rate for rare disease cases can be increased.

ARUP Laboratories has purchased a PacBio Sequel IIe system for use in the Utah NeoSeq Project. The project, launched in early 2020, leverages an accelerated sequencing and analysis pipeline intended to provide a genetic diagnosis for patients in the neonatal intensive care unit (NICU) in less than a week. By incorporating the Sequel IIe in the study, the team hopes to demonstrate the potential of long-read HiFi sequencing technology to identify variants in areas of the genome that are difficult to sequence with short-read sequencing technologies and, ultimately, increase diagnostic yield for rare disease cases.

Currently, the cause of more than half of rare disease cases worldwide remains unexplained. Using short-read whole genome sequencing (WGS), the lab's current diagnostic yield for rare disease is about 30% to 50%. The study will explore the use of HiFi WGS, which provides more comprehensive, accurate, and high-definition coverage of hard-to-read regions in the human genome, to determine if it can identify variants, both small and structural, that were not readily detectible by short-read WGS. Additionally, ARUP Laboratories is seeking to develop clinical assays for areas that are difficult to sequence with short-read sequencing technologies, such as repeat expansions and pseudogenes.

"Our team at ARUP Laboratories knows that each sample represents a patient, and we want to do everything that we can to provide every patient who comes to us with a diagnosis," said Hunter Best, PhD, Medical and Scientific Director of Genomics, ARUP Laboratories. "With the incorporation of HiFi WGS in the study, we believe that we will be able to obtain more insight into the samples we receive, ultimately helping our clients and their patients find answers."

The initial phase of the study uses HiFi WGS on those samples already sequenced with short-read technology, but without a resulting diagnosis. The goal is to wrap up this phase of the project in the first quarter of 2022. Should the initial phase of the study be successful, the project will explore using HiFi WGS as a frontline diagnostic tool for rare and inherited disorders.

"PacBio continues this march of technology evolution, and we anticipate that ultimately more than half, and perhaps up to two-thirds, of unsolved rare disease cases could be explained through HiFi sequencing," said Jonas Korlach, PhD, Chief Scientific Officer at PacBio. "We are thrilled to be collaborating with ARUP Laboratories to help them with a study designed to find answers for patients with rare diseases. We believe that high-quality WGS is the future of medicine and that this collaboration will be one of many impactful demonstrations."

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

About PacBio
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.

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About ARUP Laboratories
Founded in 1984, ARUP Laboratories is a leading national reference laboratory and a nonprofit enterprise of the University of Utah and its Department of Pathology. ARUP offers more than 3,000 tests and test combinations, ranging from routine screening tests to esoteric molecular and genetic assays. ARUP serves clients across the United States, including many of the nation's top university teaching hospitals and children's hospitals, as well as multihospital groups, major commercial laboratories, group purchasing organizations, military and other government facilities, and major clinics. In addition, ARUP is a worldwide leader in innovative laboratory research and development, led by the efforts of the ARUP Institute for Clinical and Experimental Pathology®. ARUP is ISO 15189 CAP accredited.

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