PacBio and Care4Rare Consortium Collaborate on Rare Disease Research Study

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The study will evaluate the impact of high-resolution genomes generated on PacBio technology in studying unsolved rare disease cases

MENLO PARK, Calif., Jan. 05, 2022 (GLOBE NEWSWIRE) -- PacBio, a leading provider of high-quality, long-read sequencing platforms, and the Care4Rare Canada Consortium (Care4Rare) announce today they are collaborating to study some of the most complex unexplained rare disease cases within Canada.

Care4Rare, led out of the CHEO Research Institute affiliated with the University of Ottawa, includes 21 academic sites across Canada and is recognized internationally as a pioneer in the field of genomics and personalized medicine.

“Care4Rare launched the C4R-SOLVE project in 2018 to work to identify the genetic cause of the most difficult to solve rare diseases and to facilitate the introduction of genome-based clinical diagnostic tests into the diagnostic care pathway for all Canadians with a suspected rare disease,” said Kym Boycott, MD, PhD, Senior Scientist, CHEO Research Institute and Chair of the Department of Genetics at CHEO. “Over the years we have used many technologies with great success to provide answers for many families, but there still remain some families for which we are unable to get an answer because their genetic change is hidden to the currently available technologies. We know that PacBio’s HiFi sequencing will allow us to explore new areas of the genome to find answers for these families, thereby advancing diagnostic care as well as providing insights into new areas of biology.”

There are more than 7,000 known rare diseases that impact approximately one million Canadians – two-thirds of the rare diseases cause significant disability, three-quarters affect children, more than half lead to early death, and almost none have any treatment. Further, the cause of more than one-third of these diseases is not yet known.

PacBio’s HiFi long-read whole genome sequencing (WGS) technology will be used to study rare disease samples that have already been sequenced with short-read WGS technology, but for which no genetic variant was identified.

“Our collaboration with Care4Rare is exciting for all of us who want to help find answers in rare disease,” said Jonas Korlach, PhD, Chief Scientific Officer at PacBio. “We believe that high-quality, long read WGS is the future of rare disease research, and we believe studies like this will make a meaningful impact on the medical community’s ability to make that future a reality sooner.”

C4R-SOLVE’s goal is to more than double the capacity to diagnose rare diseases in Canada, thereby improving care for patients by tailoring management to their underlying condition and providing new insights into rare conditions.

About Pacific Biosciences

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.

PacBio products are provided for Research Use Only. Not for use in diagnostic procedures.

About the CHEO Research Institute

The CHEO Research Institute coordinates the research activities of CHEO and is affiliated with the University of Ottawa. The seven programs of research at CHEO RI focus on a full spectrum of pediatric topics. Key themes include cancer, diabetes, obesity, mental health, emergency medicine, musculoskeletal health, electronic health information and privacy, and genetics of rare disease. At the CHEO Research Institute, discoveries inspire the best life for every child and youth. For more information, visit cheoresearch.ca.

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This press release may contain “forward-looking statements” within the meaning of Section 21E of the Securities Exchange Act of 1934, as amended, and the U.S. Private Securities Litigation Reform Act of 1995, including statements relating to future availability, uses, accuracy, advantages, quality or performance of, or benefits or expected benefits of using, PacBio products or technologies; the suitability or utility of such products or technologies for particular applications or projects, including in connection with the Care4Rare Canada and C4R-SOLVE studies; potential increases in variant detection and providing answers for rare disease samples, for the Care4Rare and C4R-SOLVE studies in particular and rare disease research in general; use of PacBio technology in the development of clinical or diagnostic assays; 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 PacBio’s 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 PacBio’s most recent filings with the Securities and Exchange Commission, including PacBio’s most recent reports on Forms 8-K, 10-K, and 10-Q, and include those listed under the caption “Risk Factors.” PacBio 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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