



PacBio Supports SickKids to Investigate Use of HiFi Sequencing in Undiagnosed Genetic Conditions

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SickKids will use HiFi Sequencing to explore potential genetic causes of a range of medical and developmental conditions

MENLO PARK, Calif., Feb. 28, 2022 (GLOBE NEWSWIRE) -- [PacBio](#), a leading provider of high-quality, highly accurate sequencing platforms, today announced it will be supporting The Hospital for Sick Children (SickKids) in Toronto, Canada in using HiFi whole genome sequencing (HiFi WGS) to potentially identify genetic variants that may be associated with medical and developmental conditions. Samples that will be examined using HiFi WGS were previously sequenced using short-read DNA sequencing technology, but still lack the identification of a disease-causing variant.

Even though more than 70 percent of rare disease, autism and intellectual disability have genetic causes, more than 50 percent lack an identified causative genetic alteration despite the use of microarrays, whole-exome or short-read whole-genome sequencing (srWGS).

SickKids will use HiFi WGS to analyze samples from research participants who are highly suspected to have a genetic condition but have not yet received a diagnosis despite previous genetic testing, including srWGS. The team will explore whether HiFi WGS can detect potential genetic causes for a range of conditions, such as autism spectrum disorder and congenital diseases.

"HiFi sequencing allows us to investigate the entire genome in a way that is not accessible with other technologies," said Dr. Christian Marshall, Clinical Laboratory Director, Genome Diagnostics and Associate Director at The Centre for Applied Genomics (TCAG) at SickKids. "We hope HiFi sequencing will enable us to take an exploratory look at classes of variation and large sections of the genome that were not detected previously, potentially showing unidentified causative genomic variants in these samples."

"PacBio's technology has been used to help genetic disease researchers explain mysteries where other technologies could not," said Edd Lee, Director of Human Genomics at PacBio. "We are excited to support the SickKids team to hopefully uncover the answers they have been seeking."

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

About PacBio

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](https://twitter.com/PacBio).

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

Forward-Looking Statements

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 SickKids' collaboration; potential increases in variant detection and providing answers for rare disease samples, in the SickKids' collaboration in particular and rare disease research in general; and other future events. Readers are cautioned not to place undue reliance on these forward-looking statements and any such forward-looking statements are qualified in their entirety by reference to the following cautionary statements. All forward-looking statements speak only as of the date of this press release and are based on current expectations and involve a number of assumptions, risks and uncertainties that could cause the actual results to differ materially from such forward-looking statements. Readers are strongly encouraged to read the full cautionary statements contained in the Company's filings with the Securities and Exchange Commission, including the risks set forth in the company's Forms 8-K, 10-K, and 10-Q. The Company disclaims any obligation to update or revise any forward-looking statements.

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