



Children's Mercy Kansas City Expands Investment in Pacific Biosciences Sequel Ii Systems to Scale Up Whole Genome Research Initiatives Focused on Rare Disease Diagnosis

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PacBio's highly accurate HiFi sequencing will help clinical researchers identify disease-causing genetic variants and increase our understanding of rare diseases

KANSAS CITY, Mo. and MENLO PARK, Calif., Feb. 24, 2021 (GLOBE NEWSWIRE) -- Pacific Biosciences of California, Inc. (Nasdaq: PACB), a leading provider of high-quality, long-read sequencing platforms, today announced that Children's Mercy Kansas City, one of the nation's top pediatric medical centers, has increased its investment in highly accurate HiFi sequencing with four new Sequel Ii Systems to add to its two existing Sequel Ii Systems. These additional systems will significantly increase Children's Mercy's large-scale whole genome sequencing capacity and help accelerate its initiatives focused on identifying potential disease-causing genetic variants and increasing solve rates for families and children living with undiagnosed rare diseases.

In October 2020, [Children's Mercy and PacBio announced a research collaboration](#) focused on long-read whole genome sequencing of rare disease cases for which previous short-read whole genome and exome sequencing studies yielded no answers. The study, which is currently underway, has already resulted in the detection of multiple new diagnoses, including a novel expansion disorder, precise definition of breakpoints and orientation of structural and copy number variants, and the identification of novel inversions. Based on the success of that collaborative effort, Children's Mercy is scaling their sequencing capacity with the goal of completing PacBio HiFi whole genome sequencing for approximately 1,000 exome-negative cases over the next 12 months.

"Thousands of children do not have a clear explanation for what are likely genetic-driven diseases. We are committed to addressing this challenge by refining genomic sequencing to achieve a higher-resolution view of all DNA variation using technologies such as the Sequel Ii System," said Tomi Pastinen, MD, PhD, Director of the Center for Pediatric Genomic Medicine at Children's Mercy. "The accuracy and completeness of genomes provided by PacBio's HiFi reads from the Sequel Ii System have already helped us explain seemingly intractable cases. We expect HiFi whole genome sequencing to help us shorten the diagnostic journey for families not helped by current clinical genome or exome sequencing. Studying the genetics of thousands of children will help us make rapid progress against unknown genetic diseases and lead the way for more children and families in need."

The [PacBio Sequel Ii System](#) generates HiFi sequencing reads, which provide the accuracy and completeness required for investigating DNA variation in rare genetic diseases. Short-read whole genome sequencing can lead to sequence gaps and incomplete coverage of disease-causing gene regions. HiFi whole genome sequencing produces reads approximately 100 times longer than short-read whole genome sequencing, providing high-quality mapping across a genome for comprehensive variant detection. As part of their collaboration, scientists at Children's Mercy and PacBio will take advantage of the ability of HiFi sequencing data to reproduce all variants previously detected by short-read whole exome and whole genome methods and to identify novel variants that could explain disease phenotypes.

"At least half of known rare disease cases have not been explained with existing short-read sequencing or other techniques. As a result, we are seeing tremendous interest in HiFi sequencing as an important new tool for detecting large or challenging variants missed by short reads, with early successes showing the enormous promise of this exciting technology," said Christian Henry, President and Chief Executive Officer of Pacific Biosciences. "We are proud to support Children's Mercy, which for more than 120 years has been committed to the mission of creating a world of well-being for children. It is important to remember that each of these explained cases represents a family that is now closer to the end of their diagnostic odyssey. We look forward to continuing our collaboration and working every day to bring advanced genomic technologies to help people live longer, healthier lives."

The six PacBio Sequel Ii Systems will be installed in the new Children's Mercy Research Institute (CMRI) building, which was opened on February 18, 2021. The Institute's new nine-story, 375,000-square-foot building features state-of-the-art laboratories that will support a collaborative approach to research, fostering a partnership among healthcare professionals, researchers, patients, and families. For more information about CMRI, please visit: www.childrensmercy.org/childrens-mercy-research-institute.

Findings from the PacBio and Children's Mercy research collaboration will be delivered in a platform presentation at the ACMG Annual Clinical Genetics Meeting, April 15, 2021. The presentation, titled, "Clinical Application of Long-Read Sequencing in Unsolved Rare Disease," will be given by Emily Farrow, PhD, CGC, Director, Laboratory Operations, Genomic Medicine Center, Children's Mercy Kansas City. More information can be found online at www.acmgmeeting.net.

About Children's Mercy Kansas City

Founded in 1897, Children's Mercy is one of the nation's top pediatric medical centers. With not-for-profit hospitals in Missouri and Kansas, and numerous specialty clinics in both states, Children's Mercy provides the highest level of care for children from birth through the age of 21. U.S. News & World Report has repeatedly ranked Children's Mercy as one of "America's Best Children's Hospitals." For the fifth time in a row, Children's Mercy has achieved Magnet nursing designation, awarded to only about eight percent of all hospitals nationally, for excellence in quality care. Its faculty of more than 800 pediatric subspecialists and researchers across more than 40 subspecialties are actively involved in clinical care, pediatric research, and educating the next generation of pediatric subspecialists. Thanks to generous philanthropic and volunteer support, Children's Mercy provides medical care to every child who passes through its doors, regardless of a family's ability to pay. For more information about Children's Mercy and its research, visit childrensmercy.org. For breaking news and videos, follow us on [Twitter](#), [YouTube](#) and [Facebook](#).

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

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

Forward-Looking Statements

All statements in this press release that are not historical are forward-looking statements, including, among other things, statements relating to market leadership and uses, accuracy, quality or performance of, or benefits of using, our products or technologies, including HiFi and SMRT technology, the suitability or utility of our methods, products or technologies for particular applications or projects, including in connection with rare disease research, the ability of the Company to be successful in reaching its technological and commercial potential, 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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