



## **KK Women's and Children's Hospital and PacBio Collaborate in Asia's First Study of Unexplained Neurodevelopmental Disorders**

February 16, 2022

### **PacBio HiFi Sequencing Technology Will Be Used to Investigate Effect on Diagnostic Yield of Unresolved Rare Diseases in Children**

SINGAPORE and MENLO PARK, Calif., Feb. 16, 2022 (GLOBE NEWSWIRE) -- [KK Women's and Children's Hospital](#) (KKH) and [PacBio](#) (Nasdaq: PACB), a leading provider of high-quality, highly accurate sequencing platforms, today announced a collaboration to leverage PacBio technology in understanding the genetic causes of neurodevelopmental disorders. As part of the **Bringing Research Innovations for the Diagnosis of GENetic diseases in Singapore (BRIDGES)** program, the collaboration will use HiFi long read sequencing technology to investigate the effect on diagnostic yield in unresolved neurodevelopmental disorder cases, of which only 39 percent were resolved using short read sequencing technologies.

The collaboration brings together two powerhouses within their respective fields. KKH is an academic medical center leading in Obstetrics, Gynecology, Pediatrics and Neonatology. Its 830-bed facility is a tertiary referral center to manage high-risk conditions in women and children. PacBio is a leading provider of long-read sequencing technology, and has extensive rare disease-focused collaborations with many U.S.- and Europe- based organizations.

"Understanding the genetic causes of rare, undiagnosed diseases offers affected families some form of closure, including potential therapy options, and the ability to make informed family planning decisions," said the Lead Principal Investigator of Singapore Undiagnosed Disease Program, Dr. Saumya Jamuar, who is also Senior Consultant, Genetics Service at KKH and Head of SingHealth Duke-NUS Genomic Medicine Center. "As the first hospital in Asia using HiFi long read sequencing technology, our researchers want to better understand the genetic underpinnings or rare diseases in the hopes of ultimately improving the chances of making rare disease diagnoses, which may help us to offer targeted clinical management with improved health outcomes for more families."

PacBio will also work with KKH to build a bioinformatics pipeline optimized for HiFi long read sequencing data analysis, enabling KKH to investigate possible disease-associated variants, including structural variants, in previously unresolved cases in the hopes of increasing diagnostic yield in rare disease cases of suspected but unknown genetic origin.

"Through this collaboration with leading rare disease researchers in Singapore, we hope to bring answers and hope to patients and families plagued by rare, undiagnosed diseases," said Christian Henry, Chief Executive Officer and President of PacBio. "We believe the program at KKH will serve as a model for applying PacBio HiFi sequencing technology to difficult to solve cases across hospitals in Asia."

### **About KK Women's and Children's Hospital**

KK Women's and Children's Hospital (KKH) is Singapore's largest tertiary referral centre for Obstetrics, Gynaecology, Paediatrics and Neonatology. Founded in 1858, the academic medical institution specialises in the management of high-risk conditions in women and children. A team of about 500 specialists adopt a compassionate, multi-disciplinary and holistic approach to treatment, and harness medical innovations and technology to deliver the best medical care possible.

As an Academic Medical Centre, KKH is a major teaching hospital for all three medical schools in Singapore, Duke-NUS Medical School, Yong Loo Lin School of Medicine and Lee Kong Chian School of Medicine. The 830-bed hospital also runs the largest specialist training programme for Obstetrics and Gynaecology and Paediatrics in the country. Both programmes are accredited by the Accreditation Council for Graduate Medical Education International (ACGME-I), and are highly rated for the high quality of clinical teaching and the commitment to translational research.

### **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](http://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 BRIDGES program; potential increases in variant detection and providing answers for rare disease samples, for the BRIDGES program in particular and rare disease research in general; use of PacBio technology in the development of clinical or diagnostic assays; 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.

### **Contacts for PacBio**

Investors:

Todd Friedman

[ir@pacificbiosciences.com](mailto:ir@pacificbiosciences.com)

Media:

Kathy Lynch

[pr@pacificbiosciences.com](mailto:pr@pacificbiosciences.com)

**Contact for KKH**

Charlotte Sam

[media@kkh.com.sg](mailto:media@kkh.com.sg)