Rapid genomic sequencing to diagnose critically ill children with rare diseases

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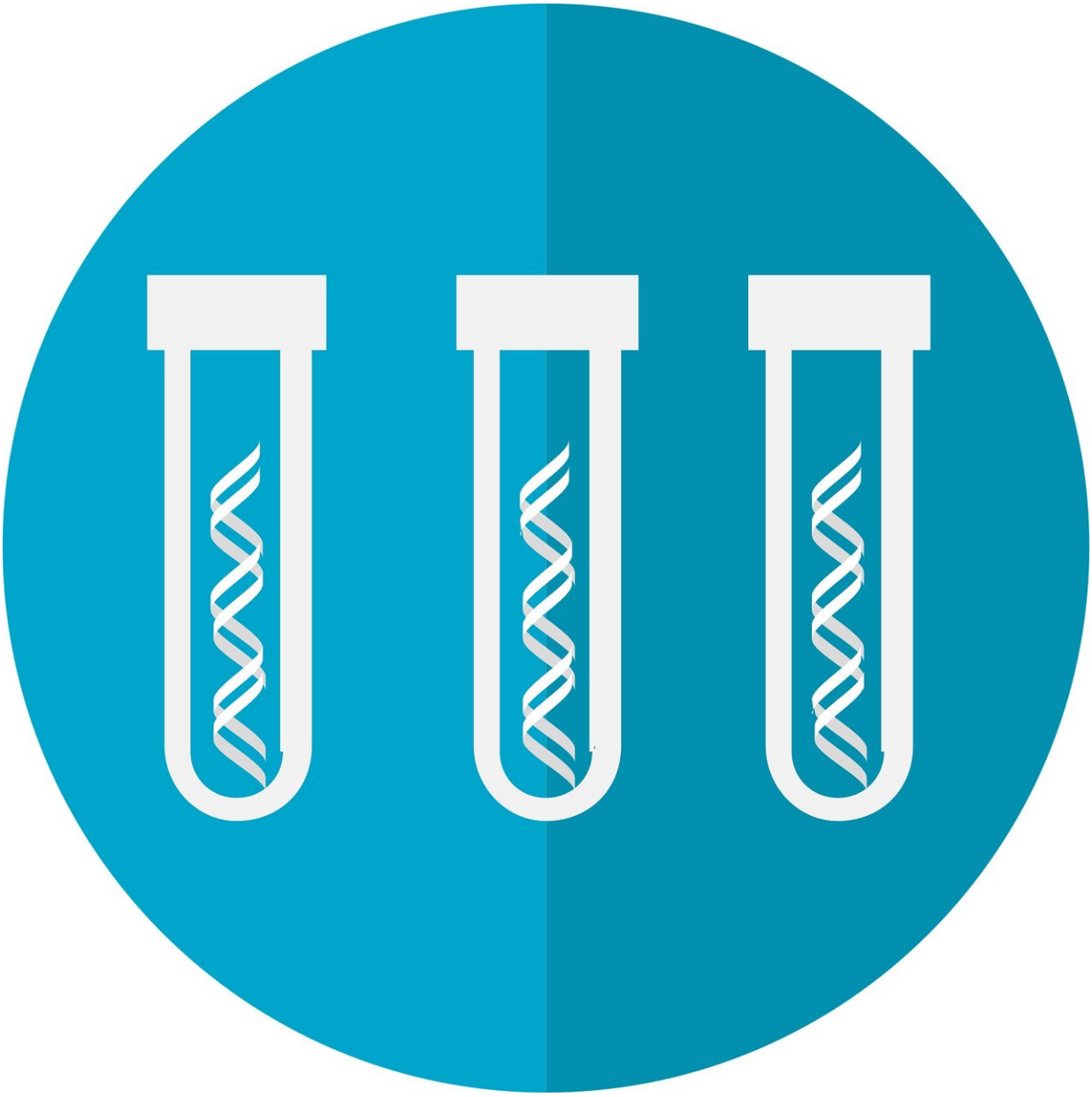

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A multidisciplinary team at KK Women's and Children's Hospital (KKH) has developed a test to enable faster diagnosis of rare diseases to help critically ill children. Rapid genomic sequencing or RapidSeq of critically ill children in the neonatal and children's intensive care units is a test, the first of its kind in Singapore, to provide information on the underlying genetic diagnosis of these critically ill children.

It is estimated that, globally, 2 percent of all fetuses have a genetic disorder. About a quarter of the disorders present at birth or soon after with significant medical issues, some of which require admission to the intensive care unit. These hospital admissions are often emotionally stressful for parents and families, who do not know what the future holds for them. There is also the stress of possible financial implications.

Developed by an expert team from the genetics service and the DNA Laboratory And Research Centre of KKH, the rapid genomic sequencing test is a collaborative effort aimed at directly helping parents and families by giving timely diagnosis that can be used to guide the child's clinical care management. Genomic sequencing is a powerful tool for the diagnostic evaluation of critically ill children with suspected genetic disorders in the neonatal and children's intensive care units.

Timely diagnosis of genetic diseases in severely ill children can be difficult and lengthy, yet definitive diagnoses are often critical. Using RapidSeq in intensive care units will help clinicians to give better treatment to critically ill children suspected of genetic disorders. Rapid sequencing further helps to improve clinical care and replace time-consuming and/ or invasive diagnostic testing, allowing these children to avoid unnecessary tests and treatments. In comparison with conventional

genetic tests, it can also highlight the need for other specialists to get involved with the clinical care of the child. For the families, the information from the test results sheds light on the genetic disease and helps them make decisions about their care.

The team at KKH has used advancements in genomic sequencing to reduce the turnaround time to get the results for diagnosis from months to 10 working days. This much shorter time frame expedites identification of the underlying genetic problem. Comparatively the typical turnaround time for standard sequencing reports range from three months to years. Rapid genomic sequencing provides a diagnosis with unmatched speed, thereby allowing critically ill children to get the treatment they need.

The RapidSeq test is being launched under the first phase of the BRIDGES programme. In 2014, the team collaborated with genomic research institutes (including Duke-NUS and A*Star) to create an integrated approach, and developed a programme known as BRIDGES (Bringing Research Innovations for the Diagnosis of GEnetic diseases in Singapore). Tapping innovations in genomic technologies, BRIDGES incorporated the sequencing service into routine clinical care to directly benefit patients and families and improve their health outcomes. Since 2014, the programme has included over 380 families from KKH and, using the sequencing technique, has completed analysis for 303 families, and made a diagnosis in 120 families (39 per cent). The results led to a successful diagnosis in one out of three families.

"This is a testament of our concerted commitment to improve delivery and quality of care for our patients through innovative approaches. We are truly excited by the promise of this new test and hope that this new initiative strengthens care and brings hope to many more families in the near future. Knowing the diagnosis brings clarity and closure to the families of these patients," said Dr. Tan Ee Shien, Head of Genetics

Service, at KKH."

Provided by KK Women's and Children's Hospital