Ultra-rapid testing a game changer for children in intensive care

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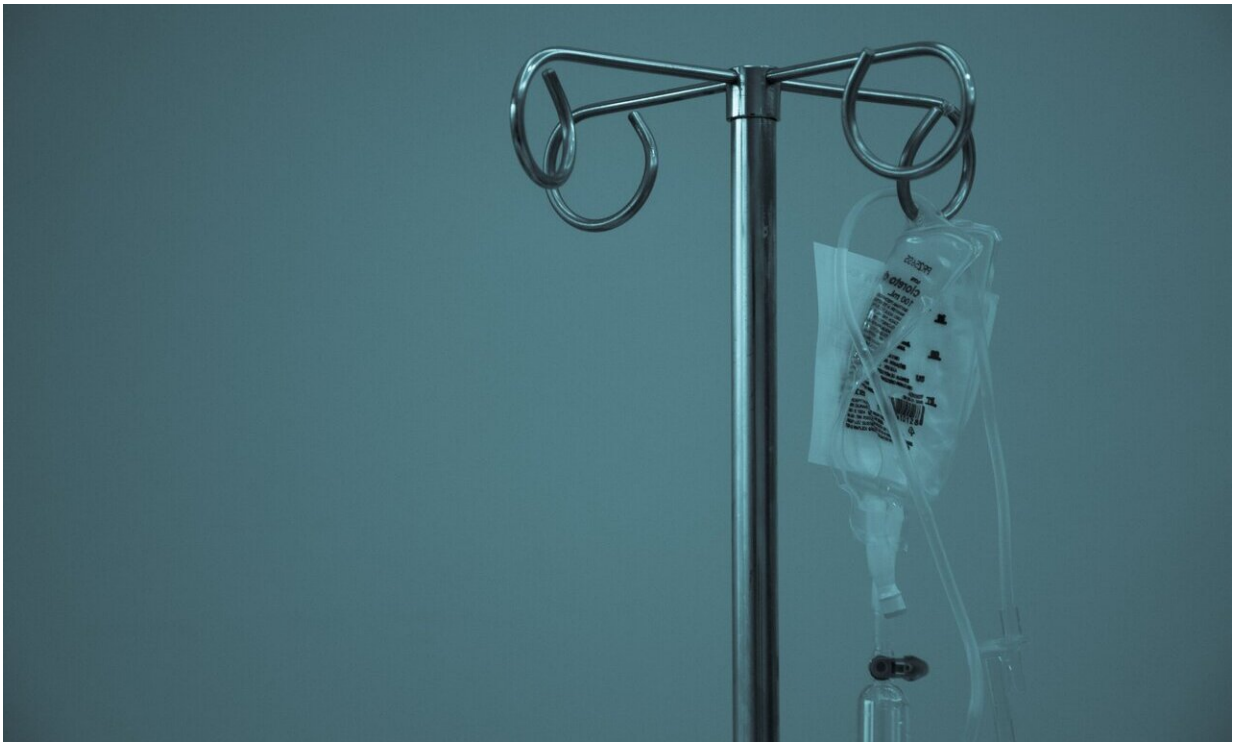

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Ultra-rapid genetic testing can transform the diagnosis and treatment of critically ill children with suspected genetic conditions, according to an Australian Genomics study.

The study tested 108 critically ill babies and children with genetic

conditions admitted to intensive care units in 12 Australian hospitals.

University of Queensland researchers involved in the study received genomic test results within less than five days, compared to traditional tests which can take three to six months.

As a result of using the new procedure, they were able to make a diagnosis in 51% of cases and change clinical care for 75% of children in ICU.

UQ's Associate Professor Luregn Schlapbach said ultra-rapid genomic results in ICU were a game changer for children where a genetic condition is suspected.

"Thanks to now genomic diagnostics, this rapid turnaround time provides more certainty to families and doctors who previously often had to wait months for results," Dr. Schlapbach said.

"It enables parents to better understand their child's illness, and how to manage the condition and plan for the future.

"For a number of children, timely diagnosis permits precision treatments which otherwise may have been missed."

Improved technology and treatment capacity enables doctors to better respond to urgent timeframes associated with children in intensive care.

Associate Professor Luregn Schlapbach said ultra-rapid testing has enabled sequencing of the entire genome of a patient.

"While genomic testing is currently provided within research studies, these results are very encouraging in view of the need for wider implementation in clinical practice," he said.

The next phase of the study will test more than 240 critically ill children across Australia over three years using whole genome sequencing as a first-tier test in diagnosing rare diseases, as well as increasing rapid testing in all states and territories.

The Acute Care Genomics study was published in *JAMA*.

More information: undefined undefined et al. Feasibility of Ultra-Rapid Exome Sequencing in Critically Ill Infants and Children With Suspected Monogenic Conditions in the Australian Public Health Care System, *JAMA* (2020). DOI: 10.1001/jama.2020.7671

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