# Rapid DNA sequencing transforms care for critically unwell children

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Credit: Murdoch Children's Research Institute

Rapid genomic sequencing for babies and children with suspected genetic conditions improves health outcomes and saves healthcare dollars – transforming rare disease diagnosis in paediatric and neonatal intensive care.

A Melbourne Genomics Health Alliance study, undertaken by a team of experts from Murdoch Children's Research Institute, The Royal Children's Hospital and Monash Children's Hospital, has demonstrated the usefulness of rapid genomic sequencing for intensive care patients with suspected genetic conditions.

More than half the children in the study received a diagnosis; care changed for more than half of those diagnosed.

Parents received their children's results in a median of 16 days (as little as 9 days in some cases): up to 10 times faster than the time usually taken for this kind of test.

"Our study shows good reasons for children in intensive care to receive genomic sequencing results more quickly than the typical four months. Even though the rapid test costs substantially more, we demonstrated greater benefits overall to patient care and reduced hospital costs," said Associate Professor Zornitza Stark from Murdoch Children's Research Institute, the study's lead author.

"Typically, it takes several years and many specialist assessments and tests before an accurate diagnosis of a rare genetic condition is made. We are working to transform this journey for as many children and families as possible."

Of the 40 patients involved in the study, 21 received a diagnosis (53 percent). Medical care changed for 57 percent of diagnosed patients, with more precise treatment able to be provided in light of more accurate diagnosis. Results were provided to 78 percent of patients during their first hospital admission, saving many parents the distress of not knowing the cause or exact nature of their child's illness.

A health economic analysis demonstrated more than half a million dollars in healthcare savings, due to diagnosed patients spending less time in intensive care and due to other (often more invasive) tests being averted.

"Genomics is changing how we work, across many areas of healthcare," said Melbourne Genomics Executive Director, Associate Professor Clara

Gaff. "This study shows the benefits of enabling specialist doctors and laboratory scientists to work together to speed up genomic testing for some of Victoria's most unwell children."

To provide the children's genomic sequencing results up to ten times faster than usual, a highly collaborative, cross-discipline 'rapids team' of specialist doctors and laboratory scientists was developed.

"We continually worked to improve our processes so that results could be delivered in shorter and shorter timeframes," Associate Professor Stark explained. "We've now been able to show that rapid genomic sequencing is highly useful in the acute paediatrics setting and feasible in the Victorian healthcare system. This opens a whole new horizon for the use of genomic sequencing in healthcare."

As a result of this work, a further $2.4 million has been granted to extend the use of rapid sequencing in a national project across multiple paediatric and intensive care units, aiming to deliver results in as little as 5 days.

This study builds on previous findings by Melbourne Genomics Health Alliance, led by the Murdoch Children's Research Institute team, showing diagnosis rates for children with genetic disorders consistently above 50 percent when genomic sequencing is used – four times that of usual care – at half the cost per diagnosis.

Provided by Murdoch Children's Research Institute