

Genetic testing helps detect children likely to have heart failure and require a transplant

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Genetic testing can diagnose cardiomyopathy—a disease of the heart muscle—in children and help detect who will have heart failure and require a transplant, a new study has found.

Researchers at the Centenary Institute and the Murdoch Children's Research Institute (MCRI) have shown that [genetic testing](#) for cardiomyopathy in [children](#) and their first degree relatives (a parent or sibling), provides a precise clinical diagnosis that could guide treatment options.

The findings, published in the journal *Circulation: Genomic and Precision Medicine*, evaluated the [genetic causes](#) of infant and childhood-onset cardiomyopathy. A disease that involves a weakened heart muscle, cardiomyopathy makes it hard for the heart to pump blood effectively around the body. Children with cardiomyopathy are more likely than adults to develop life-threatening arrhythmias, severe heart failure and require heart transplantation.

Lead author of the study, the Centenary Institute's Dr. Richard Bagnall, Head of the Bioinformatics and Molecular Genetics Laboratory, said that cardiomyopathy, although rare in infants and children, often resulted in significant health problems and death.

"Genetic testing allows us to look for variations ([mutations](#)) in the genes known to cause cardiomyopathy. We wanted to better understand this disease in children," Dr. Bagnall said.

In the study, 221 children from Melbourne and Sydney, aged 18 years or under, with cardiomyopathy were recruited from pediatric cardiology services or genetic heart diseases clinics. Where possible, family members were also genetically tested. About a third of the participants developed cardiomyopathy at less than a year old and about a third received a [heart transplant](#).

Dr. Bagnall said, as well as the genetic basis, the research team wanted to find out why some infants and children end up with [heart failure](#) and need a transplant, while their parents and siblings do not.

In their analysis, the researchers were able to attribute known gene mutations to 50% of the childhood cardiomyopathy cases investigated.

"We established that cardiomyopathy in children often has an underlying and known [genetic basis](#)," said Dr. Bagnall.

"When a child required a transplant but family members didn't, we often found that the gene mutation causing the disease was not found in their parents or that the child had two separate variants—one from each parent."

The research also uncovered variants in genes that cause multi-organ syndromic diseases, which have not been clinically diagnosed before because the characteristic features are not fully developed yet in children.

Murdoch Children's Professor Robert Weintraub said understanding the cause of these serious and sometimes life-changing conditions was important for patients and their families.

"Knowing that a gene mutation is a cause of their child's condition can provide answers to otherwise unresolved questions," he said. "In addition to the scientific knowledge gained about which mutations cause heart muscle conditions at different ages, unaffected children from the same family who do not have the gene change, no longer require ongoing surveillance."

"Children who have the mutation may not develop the condition but do require close follow-up. There is still a lot to learn however, particularly in those families where no gene mutation has been found."

Dr. Bagnall said that the findings demonstrated that the genetic diagnosis of childhood [cardiomyopathy](#) had important clinical benefits leading to

more precise and quicker diagnosis and identifying recessive and other gene variants.

"Genetic testing can often mean a more accurate diagnosis that can guide therapeutic approaches, inform prognosis and help in better identifying other family members who may also be at risk of developing the disease," he said.

More information: Richard D. Bagnall et al, Genetic Basis of Childhood Cardiomyopathy, *Circulation: Genomic and Precision Medicine* (2022). [DOI: 10.1161/CIRCGEN.121.003686](https://doi.org/10.1161/CIRCGEN.121.003686).
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