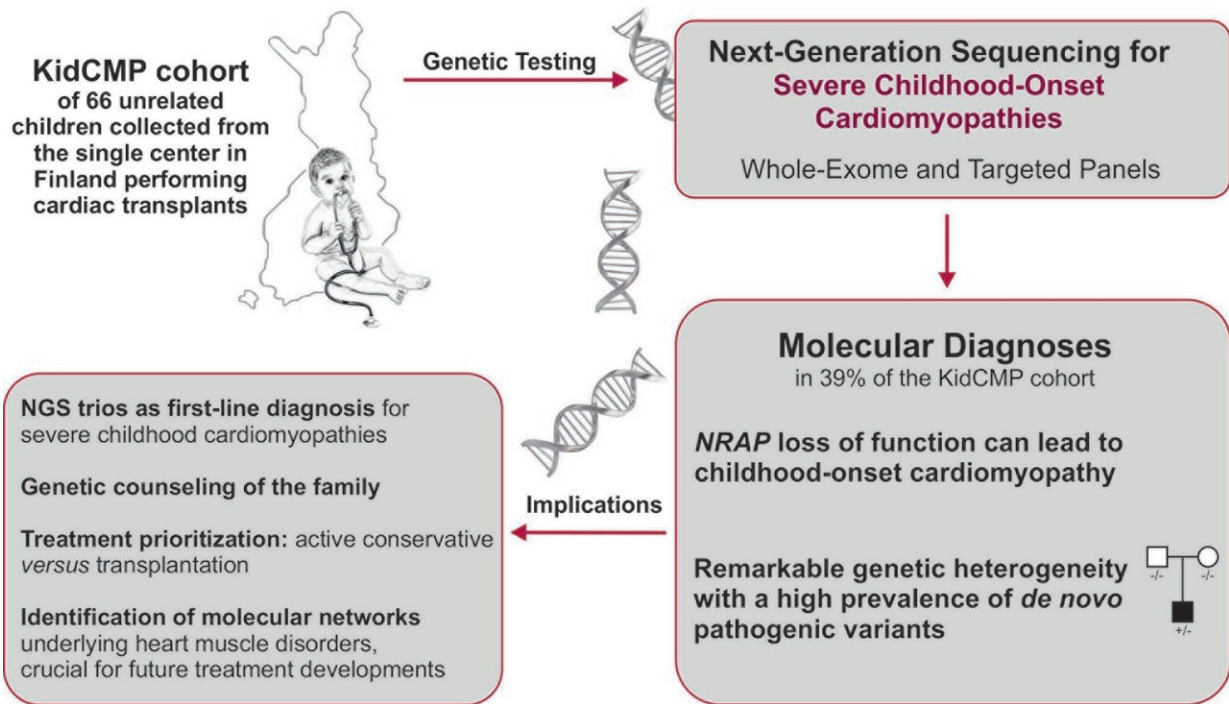


Genetic study clarifies the causes of the most severe heart muscle diseases of children

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Credit: University of Helsinki

Cardiac muscle degeneration—cardiomyopathy—is the most common cause of severe cardiac dysfunction and life-threatening cardiac arrhythmias in children. These severe disorders often lead to consideration of heart transplant. However, their actual cause—the genetic basis, that is—has been poorly characterized.

A collaborative effort of pediatric cardiologists at Helsinki University Hospital and University of Helsinki, led by Docent Tiina Ojala and Academy Professor Anu Suomalainen Wartiovaara, with Catalina Vasilescu, MSc, as the lead author, succeeded in collecting a globally unique KidCMP cohort of children with severe cardiomyopathies from the past 21 years, and analyzed them genetically.

The unique patient collection was enabled by the local cardiology clinic being the only center of cardiac transplantations in Finland. The median age of diagnosis of our patients with severe disease was just four months.

Using next-generation DNA sequencing methodology with rigorous subsequent validation of findings, the investigators uncovered the [genetic causes](#) in 40% of the patients.

"The results highlighted an extremely variable genetic background, each family presenting a different pathogenic variant that often arose during the patient's own embryonal development," Professor Wartiovaara tells.

The authors discovered that the genetic knowledge had direct implications for predicting the disease course and [treatment decisions](#).

"All children had life-threatening diseases early on, and some genetic defects predicted a primarily progressive disorder requiring cardiac transplant. However, if intensively treated, some gene defects predicted a recuperative course, without a transplant," Dr. Ojala says.

Personalized medicine is one of the goals of the current medical research, where the understanding of the genetic cause and [disease](#) mechanism in each individual will promote tailored forms of treatment. The study published by the University of Helsinki investigators makes an important step in this direction by deciphering genetic causes in [children](#)

and their implications for treatment decisions.

More information: Genetic Basis of Severe Childhood-Onset Cardiomyopathies. *Journal of the American College of Cardiology* Volume 72, Issue 19, November 2018; [DOI: 10.1016/j.jacc.2018.08.2171](https://doi.org/10.1016/j.jacc.2018.08.2171)

Provided by University of Helsinki

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