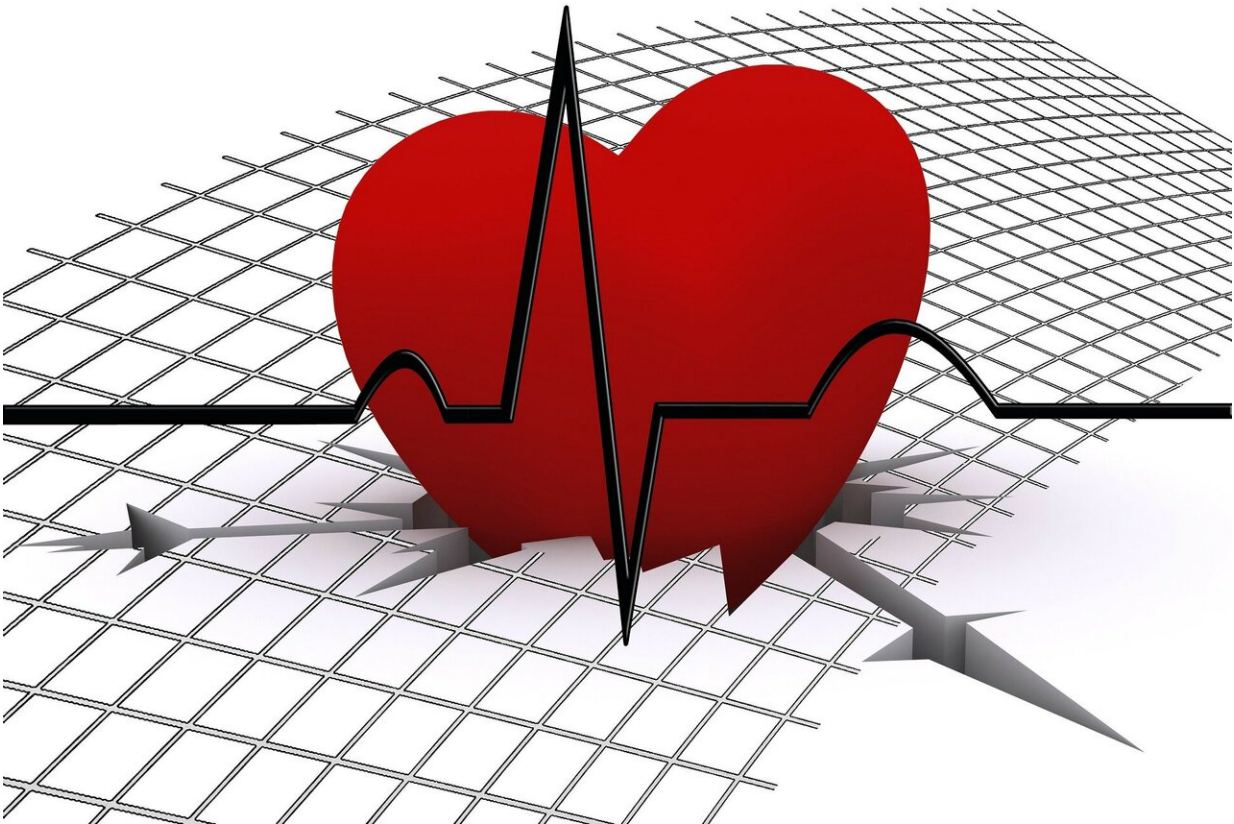


# Researchers uncover more than 1300 genes linked to congenital heart disease

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One in every 100 babies is born with a congenital heart disease (CHD),

and CHD is the major cause of death in newborns—however the genetic cause of these developmental disorders is not fully understood, hindering the development of accurate pre-natal genetic testing.

Now researchers from Monash University in Melbourne, Australia have developed a way to determine which [genes](#) are "in play" in causing these cardiac abnormalities. The technique not only confirmed well-known CHD genes, but also discovered 35 [new genes](#) not previously suspected in the [disease](#). The research opens the way, in the future, for more accurate pre-natal genetic testing for [congenital heart disease](#).

The collaborative study, published in *Genome Biology*, and co-led by Associate Professor Mirana Ramialison from Monash University's Australian Regenerative Medicine Institute and the Murdoch Children's Research Institute, and Dr. Travis Johnson from Monash University's School of Biological Sciences, aimed to improve on the current conventional approach to identifying disease-causing genes "which focuses on screening genes that are present in [heart](#) only— an approach that often overlooks genes that are present in other tissues as well, despite still playing important roles in heart development," Dr. Hieu Nim from Australian Regenerative Medicine Institute, the first author of the study said.

"The trick was to mine genome databases to identify genes that were specifically "switched on" in the heart" added Dr. Nim. The resulting computational pipeline to identify not only genes specific for the heart but genes that may also be associated with other organs such as the liver or kidney "These could comprise many of the missing congenital heart disease genes, but have been, to date, discounted because they are not unique to the heart," Associate Professor Ramialison, said.

The researchers then used the vinegar fly, *Drosophila melanogaster*, as a testing model to determine some of the functional impacts of these novel

genes. Researchers use *Drosophila* because it is a well-established model organism to understand the genetic mechanisms of many human diseases. This is because: about 75% of human disease-causing genes are found in the fly in a similar form, it is easy to work with and breeds quickly, and many tools are available to manipulate any genes in it.

According to Dr. Johnson, the [vinegar-fly](#) studies revealed "a long list of high-quality candidate genes for causing heart abnormalities in humans, giving real insight into just how susceptible this organ is to genetic mutations."

Dr. Johnson cautioned that the identification of dozens of new CHD candidate genes is some time from providing more accurate pre-natal genetic testing for CHD. "We now need to conduct functional studies on all of these genes in animal experiments to determine what they actually do, so its early days, but we now have an excellent starting point."

**More information:** Hieu T. Nim et al, A cis-regulatory-directed pipeline for the identification of genes involved in cardiac development and disease, *Genome Biology* (2021). [DOI: 10.1186/s13059-021-02539-0](https://doi.org/10.1186/s13059-021-02539-0)

Provided by Monash University

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