

# Study identifies new gene variations associated with heart rate

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Through a collaborative genome-wide study on individuals, researchers have discovered 14 new genetic variations that are associated with heart rate. Since heart rate is a marker of cardiovascular health, these findings could provide a better understanding of genetic regulation of heart beat and is a first step towards identifying targets for new drugs to treat cardiovascular disease.

The study, titled, "Identification of Heart Rate-Associated Loci and Their Effects on Cardiac Conduction and Rhythm Disorders," was published online this week in the April issue of *Nature Genetics*. Led by researchers at the Icahn School of Medicine at Mount Sinai and the Medical Research Council Epidemiology Unit in Cambridge, UK, the collaboration involved 268 researchers from 211 institutions, as well as six large research consortia joined forces.

In order to gain new insights into the genetic regulation of heart rate, Dr. Ruth Loos, Director of the Genetics of Obesity and Related Metabolic Traits Program at the Charles Bronfman Institute for Personalized Medicine at Mount Sinai and honorary investigator at the Medical Research Council Epidemiology Unit and her team, spent three years working on a genome-wide association study using data from 181,171 participants from 65 studies during 2009-2012. "Without any prior hypothesis, we studied the entire human genome hoping to identify new genetic variations that no one before had even imagined would play a role in the regulation of heart rate," said Dr. Loos, senior author of the study. "This discovery is just the beginning of something new and

exciting and can hopefully be used to identify new drugs that can be used for the treatment of [heart rhythm disorders](#)."

In a follow-up study, experimental down-[regulation of gene expression](#) was then conducted on fruit flies and [zebra fish](#), to better understand how genetic variations might affect heart rate. These experiments identified 20 genes with a role in heart rate regulation, signal transmission, embryonic development of the heart, as well as cardiac disorders, such as dilated cardiomyopathy, congenital heart failure and sudden heart failure. "Our findings in humans as well as in [fruit flies](#) and zebrafish provide new insights into mechanisms that regulate heart rate," said Dr. Marcel den Hoed, post-doctoral fellow at the Medical Research Council Epidemiology Unit and lead author of the study.

The follow-up study also showed that a genetic susceptibility for higher heart rate is associated with altered [cardiac conduction](#) and a reduced risk of sick sinus syndrome, a common indicator for pacemaker implantation. "Our study tripled the number of genetic variations that are known to be associated with heart rate, some of which are also associated with other cardiovascular risk factors and with heart rhythm disorder," said Dr. Loos.

**More information:** Identification of heart rate–associated loci and their effects on cardiac conduction and rhythm disorders, [DOI: 10.1038/ng.2610](#)

Provided by The Mount Sinai Hospital

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