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Family tree may clarify death risk for inherited heart rhythm disorders

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Reconstructing family trees dating back to 1811, Dutch researchers have estimated the death risk for people with inherited heart rhythm disorders, according to a study in *Circulation: Cardiovascular Genetics*, a journal of the American Heart Association.

Heart rhythm disorders can result in sudden cardiac death in apparently healthy people because of severe disturbances in the rhythm of the heart. The risk is high for people who carry one of these rare genes and have symptoms such as fainting.

Before the study, the risk in people without symptoms was less certain. Thus, physicians developed preventive measures — including medications and implanted devices that can cause side effects — without a clear idea of the death risk.

With greater use of genetic testing for the relatives of people with one of the disorders, questions about how and when to treat people without symptoms now arises frequently.

Using Dutch archives, the researchers reconstructed family trees for patients with different conditions and compared death statistics in 266 people who carried a mutation in a gene linked with arrhythmia, 904 family members with a 50 percent chance of having the mutation, and the general population.

Researchers identified age ranges during which the risk of death

increased for people who had the mutations (and their family members) but had not yet been diagnosed or treated for the condition.

"This information can help doctors know when to treat and screen families with arrhythmia syndromes caused by different gene mutations," said Eline A. Nannenberg, M.D., lead researcher of the study and a clinical geneticist at the Academic Medical Centre in Amsterdam, The Netherlands.

The mutations studied are rare in the population; however, if a person carries one, their children have a 50 percent chance of inheriting it.

Among the researchers' findings:

- In long QT syndrome, a condition related to sudden infant death syndrome, death risk was high throughout childhood (1-19 years old), but particularly in the first 10 years of life for one mutation (LQTS1). For two other mutations that cause this syndrome (LQTS2 and LQTS3), the risk of death doesn't become significantly increased until the teen years or adulthood.
- For SCN5a-overlap syndrome, in which the SCN5A gene mutation affects the way heart cells respond to the heart's internal electrical current, deaths start to increase at age 5 and become a significant risk between ages 10-59, with a peak between ages 20-39.
- For catecholaminergic polymorphic ventricular tachycardia (CPVT), a condition that causes fainting often during exercise or strong emotions, death risk was highest from ages 20-39.
- In Brugada syndrome, which causes fainting and a rapid heart rhythm that can lead to sudden cardiac death, excess deaths occurred between ages 40-59 and were primarily in males.

"We have to be careful not to draw conclusions for families with arrhythmias caused by different mutations," Nannenberg said. "However, this new data can guide screening. In LQTS1, we advise starting genetic and heart screening of first-degree family members (children, siblings, parents) at a very young age."

In contrast, in Brugada syndrome, females without symptoms may not need invasive treatment for the first three decades, but should follow standard prevention measures, such as fighting fever.

In SCN5a-overlap syndrome, implantation of a pacemaker or ICD may be postponed until after age 5, researchers said.

Provided by American Heart Association