

Genetic heart diseases cause fewer SIDS deaths than previously thought, study finds

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Genetic mutations linked to heart disease have been considered a leading cause of sudden infant death syndrome, but a new study by Mayo Clinic, British and Danish researchers finds they are to blame for far fewer SIDS deaths than previously thought. The findings are opening new lines of inquiry into possible causes of the syndrome and may help prevent unnecessary genetic testing of surviving family members. The study results appear in the *Journal of the American College of Cardiology*.

The research, believed to be the largest genetic investigation into SIDS, found that [heart disease](#)-associated genetic mutations account for roughly 5 percent of SIDS deaths. In the study, whole-exome sequencing and a targeted analysis of 90 genes implicated in heart disease were applied to 419 unrelated SIDS cases.

Previous smaller studies of up to 170 cases suggested that heart disease-related genetic mutations caused up to 20 percent of SIDS deaths.

SIDS is the [sudden unexpected death](#) of an infant under 1 that remains unexplained despite thorough clinical and pathological investigations. SIDS represents up to 80 percent of all sudden unexpected infant deaths, with an incidence of 0.4 in 1,000 live births in the U.K. and 0.5 in 1,000 live births in the U.S.

The peak incidence occurs between 2 months and 4 months of age; the average age of infants in the new study was 2.7 months old. The [syndrome](#) is more common in boys.

"Through this research, we now know that the vast majority of SIDS cases do not stem from genetic heart diseases," says co-senior author Michael Ackerman, M.D., Ph.D., director of Mayo Clinic's Long QT Syndrome/Genetic Heart Rhythm Clinic and the Windland Smith Rice Sudden Death Genomics Laboratory. "We now are turning our attention to the genes implicated in other organ systems, like the brain, to determine their potential contribution. In addition, we are now exploring other genetic contributions to SIDS, because it is now abundantly clear that most SIDS cases are not due to a single genetic cause."

In 2001, Dr. Ackerman and Mayo colleagues discovered [genetic mutations](#) causing the heart disease long QT syndrome in a small number of SIDS cases derived from the first population-based study of the syndrome. Long QT is an inherited heart rhythm condition that can cause fast, chaotic heartbeats. Though estimates suggest that LQTS occurs in only 1 of every 2,000 people, it is a common cause of sudden cardiac [death](#) before age 40.

Now, 17 years later, researchers have analyzed every gene implicated in [sudden cardiac death](#) from genetic [heart](#) muscle and rhythm abnormalities, Dr. Ackerman says.

"Our results also highlight the crucial importance of accurate variant classification to enable proper counseling of surviving family members," Dr. Ackerman says.

Erroneously or prematurely labeling a genetic variant as pathogenic has the potential to harm patients and their families."

Provided by Mayo Clinic

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