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**THE WALL STREET JOURNAL**

WSJ.com

HEART BEAT | JULY 13, 2010

## When a Heart Risk Runs in the Family

By [AMY DOCKSER MARCUS](#)

Hundreds of thousands of Americans die of sudden cardiac death each year, an occurrence that can run in families. Now, some doctors are turning to genetic testing to pinpoint the underlying cause of death and help save surviving relatives.

Sudden cardiac death occurs when someone dies unexpectedly, usually within an hour of the appearance of any symptoms, such as dizziness and fainting spells. Often, an autopsy reveals a problem with the heart that hadn't been diagnosed. But sometimes, medical examiners can find no physical explanation for a person's death. In those cases, some doctors have begun to look deeper, running genetic tests to check for mutations that have been connected to cardiac disorders associated with sudden death.



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Michael Rubenstein for the Wall Street Journal

A photo album of Vanessa Vale, who died of sudden cardiac death.

If a gene mutation is identified in a deceased person, doctors suggest that immediate relatives, including siblings, parents, and children, get a cardiac work-up, which may include testing to see if they have the same genetic mutation. Treatment recommendations for relatives carrying the mutation may include lifestyle modifications, such as not exercising vigorously. Medications also may be prescribed, such as beta-blockers, which is commonly used to manage cardiac arrhythmia, or implanting an internal defibrillator that shocks the heart back to a normal rhythm. And regular checkups are advised.

That's what happened after Vanessa Vale suffered sudden cardiac death in her Bronx, N.Y., home nearly three years ago at the age of 27, leaving behind two small children and their father. An autopsy didn't turn up any apparent heart problems. So doctors at the Montefiore-Einstein Center for CardioGenetics ordered a test that showed Ms. Vale had a genetic mutation for a hereditary condition called Long QT syndrome, a malfunction of the heart's electrical

system that can cause people to develop a dangerous and irregular heart rhythm.

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Genetic testing was then performed on the two children, Justin, who was 6 at the time, and Elizabeth, then 3. Both of them tested positive for Long QT syndrome, which puts them at greater-than-normal risk for sudden cardiac death. The children show no other symptoms to indicate serious heart problems; a battery of standard heart tests—an EKG, echocardiogram and 24-hour Holter monitor recording—turned up few irregularities. Still, the children were put on a beta-blocker and they return to the clinic yearly for heart checkups.

"We feel like we have saved these two children," said Robert Marion, a geneticist who is co-director of the Montefiore-Einstein Center for CardioGenetics, which opened in 2008 and focuses on helping families affected by sudden cardiac death. "We are using information from the dead to guide treatment for the living," he said.



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Michael Rubenstein for the Wall Street Journal

Genetic tests showed Elizabeth and Justin Escobar, here with their father Steve Escobar, face an increased risk for sudden cardiac death.

About 350,000 people die from sudden cardiac death in the U.S. every year, according to Michael J. Ackerman, director of the Mayo Clinic Windland Smith Rice Sudden Death Genomics Laboratory. Around 3,000 of these are infants, and another 10,000 are children and young adults between the ages of 1 and 40. Genetic mutations affecting the heart's electrical system, which are passed on from parent to child, account for about 25% of sudden unexplained deaths in children and young adults and about 10% of infant deaths, Dr. Ackerman said.

The best way to find the mutations is by doing genetic testing of tissue from the deceased person's heart or of blood samples. Besides Long QT syndrome, another mutation that doctors might look for is connected with Brugada syndrome, another inherited heart rhythm disorder. Genetic testing for cardiac conditions is expensive—various tests can range from \$1,000 to \$10,000, depending on how many different genetic mutations are tested.

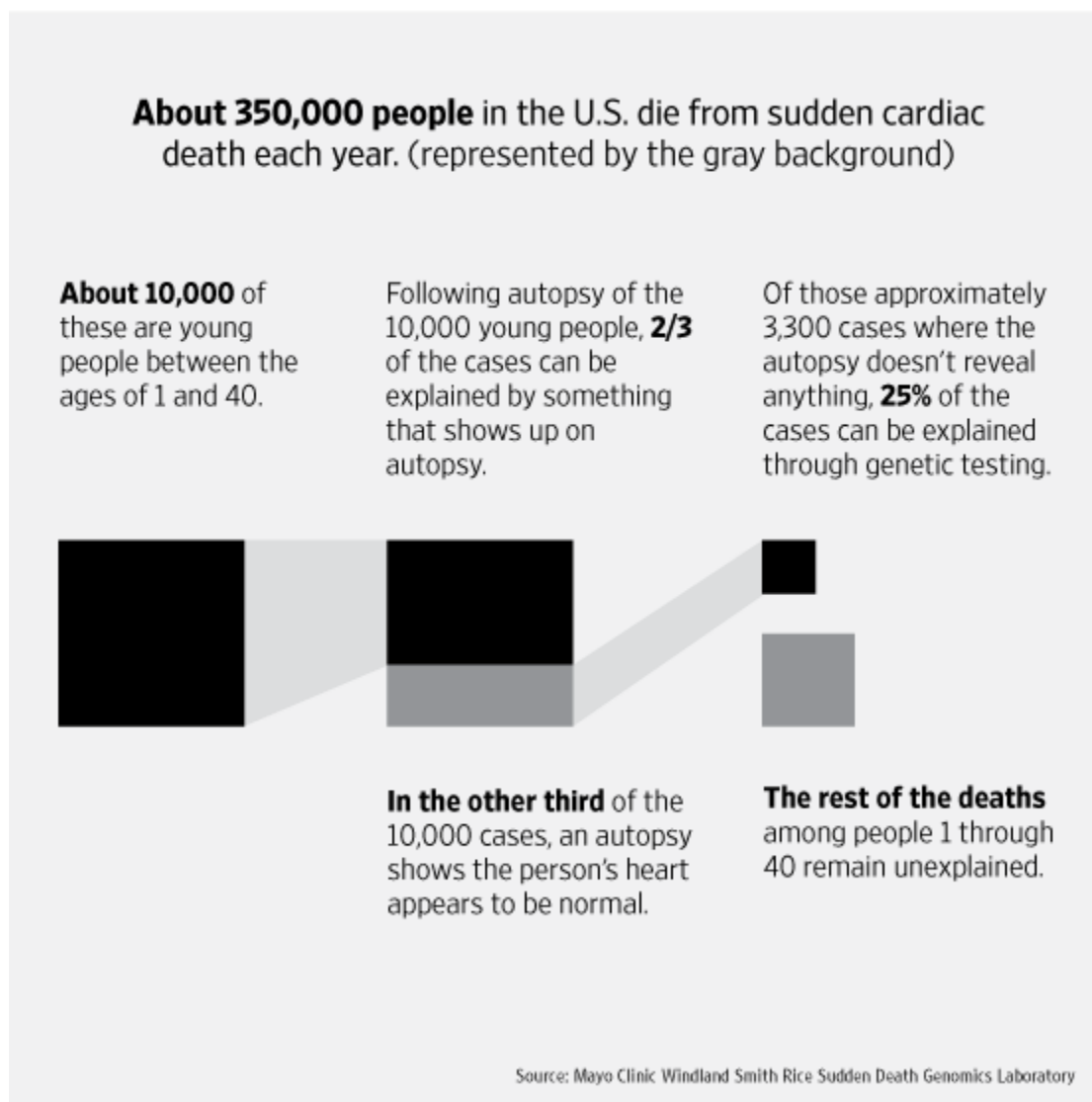
Not every gene mutation linked to hereditary cardiac diseases has been identified. Sometimes genetic tests also find mutations whose role in causing disease isn't well understood. "There are lots of variations in people's DNA. Some are mutations that are dangerous. Some mutations are the things that make us all different, and some mutations are completely irrelevant to your health. Sorting out which ones are which is hard," said Barry London, director of the Cardiovascular Institute at the University of Pittsburgh School of Medicine.

Most doctors consider Long QT syndrome a treatable condition. But people must also exercise caution, such as avoiding taking certain medications that can affect heart rhythm, including antihistamines.

Genetic testing following sudden cardiac death is expanding. Dr. Ackerman said his lab at the Mayo Clinic is raising philanthropic support to help handle the growing number of requests from medical examiner's offices around the country for genetic testing in cases of sudden unexplained death. At the Office of Chief Medical Examiner of the City of New York, when someone dies suddenly and there is no explanation

after an autopsy is performed, genetic testing is now being done in a molecular testing lab set up in the office, said Barbara Sampson, first deputy chief medical examiner.

Since late 2008, genetic tests have been run on 178 people, about half of whom were under the age of 1, Dr. Sampson said. She said mutations known to cause sudden cardiac death were found in 20% of the people tested. "It is important for the accuracy of determining cause of death. It is important to provide accurate statistics on how common these diseases are as a cause of death, and it is important to surviving family members," she said.



When genetic mutations are found, Dr. Sampson said the information is shared with the families, who are encouraged to go to centers like Montefiore-Einstein for evaluation. The medical examiner's office also keeps a database of the DNA so that when new genes are discovered that are associated with sudden cardiac death, the DNA can be retested.

One morning last month, the father of Ms. Vale's children, Steve Escobar, 31, brought Justin and Elizabeth into the Montefiore-Einstein clinic. He said Justin, now 8, is going into the third grade and participates in karate. Elizabeth, now 5, will start kindergarten in the fall. "They know mommy passed away because of her heart," Mr. Escobar said. "They do normal things but they also know that they need to stop if they feel out of breath."

Doctors at the clinic asked Mr. Escobar to tell Ms. Vale's two brothers to come in for testing, and to bring their children in to see if they have the same Long QT Syndrome mutation. So far, neither brother has been tested, Mr. Escobar said.

Edgar Vale, one of Ms. Vale's brothers, said he doesn't plan to come in. He said that he is 34 and in good health, and that he works in

construction so he is active at work. "I've never felt faint. I've never felt weak. I don't think I have it or my kids would have it," he said.

Mr. Vale said he has three children, ages 5, 4, and 11 months. "If I see my children don't look as strong as they should or I hear complaints, I will have them tested," he said.

Dr. Marion said he understands that many people feel uncomfortable about the notion that the dead are guiding doctors in the treatment of the living. "These are diseases that don't just affect an individual but the entire family. Everyone is vulnerable," he said.

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