"Study of Sudden Cardiac Death Exposes Limits of Genetic Testing"

A synopsis by Amy Hu

Sudden Cardiac Death (SCD) occurs when there is an unexpected loss of heart function affecting breathing and consciousness. The causes range from coronary artery disease to hypertrophic cardiomyopathy. Diseases and habits such as diabetes mellitus and smoking are associated with an increased risk in SCD. Experts are now saying that it results from a "complex interplay of structural, metabolic, and genetic determinants".

Scientists are now investigating the heritability of one specific case: hypertrophic cardiomyopathy (HCM). It is believed that this is passed on as a genetic trait, affecting about one in 500 Americans. If a parent has HCM, the children have a 50:50 chance of developing it. Thus, many experts have turned to genetic testing in an attempt to prevent further cases of HCM.

In previous genetic testing samples, they had discovered that the risky gene variants were affecting more people of African descent. Their previous tests had consisted of comparing genetics of affected families and healthy people. However, many of the participants in the healthy group were white and thus, there was a possibility that the "risky variant genes" that were being flagged represented racial differences and not HCM. Furthermore, Dr. Barry Maron, from Tufts Medical Center evaluated a large number of athletes and the tests suggested they might have a heart condition. There is a possibility that these are false positives as a result of the test. Many experts are also saying that diseases such as HCM are complicated and may require much more than just a test of simple genetics. Genetic testing seems to imply that many of our questions are binary with a simple yes or no answer. But it seems that this is not the case.

For the time being, genetic testing is still yielding positive results. Vanessa Vale, from Bronx, NY, passed away from SCD caused by Long QT Syndrome. This was discovered when doctors at the Montefiore-Einstein Center for CardioGenetics ordered a test following an autopsy that did not indicate any heart problems. Vale's two children were then tested for Long QT syndrome. The children have not shown any symptoms of SCD but are still regularly checking into the clinic for yearly check-ups. Although many are still wary of genetic testing, including Vale's brother, this is a key example of how we can use this to give the next generation a chance. "'We feel like we have saved these two children,'" said Robert Marion, a geneticist from the Montefiore-Einstein Center for CardioGenetics.

References:

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