

MEDICAL BREAKTHROUGHS

RESEARCH SUMMARY

TOPIC: GENETIC TESTING: UNRAVELING THE MYSTERY OF SUDDEN CARDIAC DEATH
REPORT: **MB #4947**

BACKGROUND: Sudden cardiac death (SCD) is a sudden, unexpected death caused by loss of heart function. SCD is the largest cause of natural death in the United States, causing about 325,000 adult deaths each year. SCD is responsible for half of all heart disease deaths and occurs most frequently in adults in their mid-30s to mid-40s. It does affect men twice as often as women, and is rare in children, affecting only 1 to 2 per 100,000 children each year. Most SCDs are caused by abnormal heart rhythms called arrhythmias. The most common life-threatening arrhythmia is ventricular fibrillation, which is an erratic, disorganized firing of impulses from the ventricles, or the heart's lower chambers. When this occurs, the heart is unable to pump blood and death will occur within minutes if left untreated.

(Source: <https://my.clevelandclinic.org/health/diseases/17522-sudden-cardiac-death-sudden-cardiac-arrest>)

GENETICS AND SCD RISKS: A patient should be familiar with their medical past and family history. If there is a family member with arrhythmias, heart failure, or amyloidosis and connective tissue disorders, a medical evaluation and possibly an echocardiogram or a cardiac MRI to diagnose whether the patient shares this condition and would benefit from early treatment, may be needed. There can also be an elevated risk for cardiovascular disease if family members have diabetes, high blood pressure, or high cholesterol, which can have genetic predispositions. Lifestyle also plays an important role. If someone is overweight, smokes, has a nutrient-poor diet or doesn't exercise, then no matter how good your family history is, these lifestyle factors will negatively impact health in the long run.

(Source: <https://www.uchicagomedicine.org/forefront/heart-and-vascular-articles/understanding-genetic-risk-for-heart-disease>)

NEW RESEARCH TO PREVENT SCD: Assistant professor of biomedical sciences, Stephen Chelko, with Florida State University College of Medicine, has developed a better understanding of the pathological characteristics behind arrhythmogenic cardiomyopathy (ACM), a disease that leads to SCD. He has also discovered some promising avenues for prevention. Individuals with ACM possess a mutation causing arrhythmias, which ordinarily are non-fatal if managed and treated properly. Studies show that exercise in these individuals not only amplifies those arrhythmias but causes extensive cell death. Chelko says, "This novel study unravels a pathogenic role for exercise-induced, mitochondrial-mediated cell death in ACM hearts." This finding opens avenues for the development of new therapeutic options to prevent myocyte cell death, cardiac dysfunction and pathological progression.

(Source: <https://www.sciencedaily.com/releases/2021/02/210217175153.htm>)

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If this story or any other Ivanhoe story has impacted your life or prompted you or someone you know to seek or change treatments, please let us know by contacting Marjorie Bekaert Thomas at mthomas@ivanhoe.com