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Genetics of sudden cardiac death

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Cardiovascular diseases or disorders of the heart and blood vessels are the leading cause of death and disability in the world today. While coronary heart disease, myocardial infarction and post-infarction heart failure are the main disease groups in individuals above the age of 40 years, congenital heart disease and inherited heart disease play a part in the young. The aim of this presentation is to understand the role of genetic aberrations in sudden cardiac death. The genetic underpinnings of inherited arrhythmias and familial cardiomyopathies have come to light through extensive research in the last 3-4 decades, leading to the emergence of a new branch of medicine called cardiogenetics. Familial ca rdiomyopathies, typically causing abnormal cardiac structure and function, are further classified based on their pathological outcome into hypertrophic, dilated, restrictive, arrhythmogenic and non-compaction cardiomyopathies. A wide spectrum of structural genes is implicated in these diseases. Inherited arrhythmias, on the other hand, are characterized by potentially fatal arrhythmias in young individuals with structurally normal hearts. Congenital long QT syndrome, Brugada syndrome, catecholaminergic polymorphic ventricular tachycardia and short QT syndrome are some of the inherited arrhythmias. As a clinician or a geneticist managing individuals and families with these life-threatening disorders, it is pertinent to work as a team, to know the genotype-phenotype correlations and the diagnostic, therapeutic and prognostic significance of genetic tests. The etiology of coronary and congenital heart disease is truly multifactorial; however, at times they have a genetic basis that gets turned on or off by external factors.