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Genetic testing for inherited heart diseases: Impacting clinical care

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Inherited heart diseases include a broad spectrum of cardiomyopathies and primary arrhythmia syndromes that are associated with an increased risk of sudden cardiac death. Hypertrophic cardiomyopathy (HCM), dilated cardiomyopathy (DCM), long QT syndrome (LQTS), Brugada syndrome, catecholaminergic polymorphic ventricular tachycardia (CPVT), arrythmogenic right ventricular cardiomyopathy (ARVC), short QT syndrome (SQTS) and idiopathic ventricular fibrillation (IVF) are some of the important conditions included in this category. These range from being relatively common in the general population, as is the case with HCM (~1/500), to very rare as with CPVT (~1/10000) and may account for 13-50% of sudden deaths, particularly in previously healthy young people. Numerous genes have been reported to be associated with inherited heart conditions. These typically follow an autosomal dominant inheritance pattern and first-degree relatives of an affected individual generally have a 50% risk of being affected and at risk for associated arrhythmias. There are important exceptions of autosomal recessive or X-linked subtypes which are often, but not exclusively, identified in the context of syndromic or extra-cardiac features. Technology has evolved in the past few years and genetic testing has become increasingly accessible. However, it"s important to understand and appreciate the value of genetic testing and also be aware of the associated challenges and limitations. Diagnosing an inherited heart disease is hence very important not only for medical management of index cases but also for the identification of other at-risk relatives in order to reduce the risk of arrhythmic events and sudden deaths.