

# Genetics of hypertrophic cardiomyopathy

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Hypertrophic cardiomyopathy (HCM) is the most common familiar heart disease with worst hereditary seen over the past 25 years. Mutations in 11 genes encoding proteins in the cardiac sarcomere and more than 1400 variants have been identified. Offspring of an affected individual has a 50% probability of inheriting a mutation and risk for disease, rarely it may be due to sporadic mutations in the proband but absent in parents. A mutation can be pathogenic or could be a variant of uncertain significance. Screening of family members to identify those at risk for developing disease will have an important role for genetic testing to play. However the power of

HCM mutation analysis has a more limited than initially envisioned. Genetic testing also allows expansion of the broad HCM spectrum and diagnosis of HCM phenocopies with different natural history and treatment option. However it is not a reliable strategy for predicting prognosis. Genotyping in HCM can be a powerful tool for diagnosis and family screening, however wider adoption and future success of genetic testing depends on a standardized approach to mutation interpretation and bridging the communication gap between basic scientists and clinicians.