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Perspective

Application and Abnormalities of Genetic Disorder of Hypertrophic Cardiomyopathy

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DESCRIPTION

Hypertrophic cardiomyopathy is most often caused by abnormal genes in the heart muscle. These genes make the walls of the ventricle (left ventricle) thicker than normal. Hypertrophic Cardiomyopathy (HCM) is the most common hereditary cardiovascular disease. This disorder is caused by mutations in the genes encoding cardiac sarcomere proteins, resulting in different phenotypic manifestations and clinical courses. HCM is the leading cause of sudden death in young people. The thickened walls stiffen, which can reduce the amount of blood that is taken in and pumped out to the body with each heartbeat. HCM is a chronic disease that can get worse over time. This can lead to decreased function and quality of life, long-term complications, and increased economic and social burden. People with HCM may need to make lifestyle changes.

Hypertension can worsen the symptoms of HCM, as HCM progresses, it can lead to other health problems. People with HCM are at increased risk of developing atrial fibrillation, which can lead to blood clots, stroke, and other heart complications. HCM can also cause heart failure. Sudden cardiac arrest can also occur, but this is rare.

HCM is rare, and it is thought to be a leading cause of sudden cardiac death in adolescents and athletes in North America. Familial hypertrophic cardiomyopathy is inherited in an autosomal dominant manner and results from mutations in one of several genes encoding sarcomere proteins. Mutations in at least one of the nine sarcomere genes have now been identified in approximately 50%-60% of individuals with the high clinical indicators of HCM suspicion. About 40% of these mutations occur in the myosin heavy chain gene on her chromosome 14, and about 40% affect the cardiac myosin-binding protein C gene. HCM is usually an autosomal dominant trait, so a child who has HCM from one of her parents has a 50% chance of inheriting the disease-causing mutation. If such mutations are identified, clinical severity and age of onset cannot be predicted, but family-specific genetic testing can be used to identify relatives at risk for the disease.

Hypertrophic cardiomyopathy most commonly runs in families (hereditary). This is thought to be due to a defect in a gene that controls myocardial growth. Younger people are more likely to have a more severe form of hypertrophic cardiomyopathy. However, this condition occurs in people of all ages. Cardiac hypertrophy may be late-onset and less than 13 mm, the diagnostic cutoff for diagnosis of HCM. Therefore, HCM may be underdiagnosed in such individuals. Conversely, cardiac hypertrophy may result from a phenotypic condition that accounts for 5% to 10% of clinically diagnosed HCM cases in children.

HCM is a classic monogenic disorder with autosomal dominant inheritance, and a single mutation is usually sufficient to cause the disease, although penetrance and severity vary. Phenotypic diversity is due, at least in part, to causative mutations that act in concert with many other genetic and non-genetic influences. Approximately 60% of HCM patients have a recognizable familial condition. Autosomal recessive and X-linked modes of inheritance have been reported but are rare. X-linked inheritance usually increases the likelihood of phenotypic abnormalities such as fabry disease.

CONCLUSION

Gene variants with low phenotypic effect have incomplete penetrance. The penetrance and phenotypic impact of such variants depend on the presence of other genetic and environmental factors. Many genetic variants with low to moderate penetrance are found in sporadic HCM patients and small families with HCM. Factors that complicate causation include human genetic diversity, population-specific frequencies of variants, and the presence of thousands of pathogen-encoding variants in each exome.

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