

# Hypertrophic Cardiomyopathy Mechanisms

Hypertrophic cardiomyopathy is a familial disease leading to ventricular hypertrophy. It is a cause of sudden death in young athletes, and is typically asymptomatic until death, though patients are sometimes mildly symptomatic.



**PLAY PICMONIC** 

# **Pathophysiology**

#### 2/3 Autosomal-Dominant

(2) Tutu / (3) Tree and Domino

Approximately 60-70% of hypertrophic cardiomyopathy cases are familial, which follows an autosomal dominant inheritance pattern. Furthermore, as this is mostly an autosomal dominant disease, children of a parent with HCM have a 50% chance of inheriting the disease.

# 1/3 Sporadic Type

(1) Wand / (3) Tree and Sporadic-spear

The majority of cases of hypertrophic cardiomyopathy are familial, displaying autosomal dominant inheritance, but up to ? of cases result from sporadic, <i>de novo</i> mutations in any one of several sarcomere protein genes that influence the cell contractility.

## **Mutation in Gene for Sarcomere Protein**

Mutant Shark-mirror

Hypertrophic cardiomyopathy (HCM) results from a mutation in one of the many sarcomere protein genes.

# Cardiac Myosin Binding Protein C

Mayo-sun and Protein-ribbon Cat

The majority of hypertrophic cardiomyopathy cases result from a mutation in cardiac myosin binding protein-C. However, it is important to note that many sarcomere mutations may result in HCM with varying levels of penetrance, heterogeneity, age of onset, and severity. Another causative mutation of note occurs in the ?-myosin heavy chain which occurs on chromosome 14 and results in higher penetrance, earlier age of onset, and more severe disease.

## Asymmetric Septal Hypertrophy

Asymmetrical Scepter Hiker-trophy

Along with left ventricular hypertrophy, roughly 2/3 of patients with HCM develop asymmetric septal hypertrophy (towards the left ventricular outflow tract).

### **Outflow Tract Obstruction**

Heart Outflow Obstructed

Left ventricular hypertrophy (common with HCM), along with asymmetric septal hypertrophy leads to left ventricular outflow obstruction. The asymmetric septa bulges towards the left ventricle, causing a large part of this obstruction. This occurs in 25% of patients, but 70% of patients show symptoms of outflow obstruction if provoked under specific conditions.

# **Diastolic Dysfunction**

Dysfunctional Dice Won't Fill Heart

Diastolic dysfunciton occurs due to hypertrophy. The primary abnormality is reduced stroke volume due to impaired diastolic filling, as a result of increased left ventricular stiffness. Paired with increased demand (as a result of outflow obstruction), this increase in diastolic pressure (decreased blood volume), symptoms such as angina and arrhythmias arise.