

Facts on Hypertrophic Cardiomyopathy (HCM)

- **HCM is a dominantly inherited disease affecting 1 in 500 individuals.**
[Maron BJ \(2002\) JAMA 287:1308-20.](#)
- **HCM is caused by mutations in any one of at least 12 genes.**
 - Mutations in *TNNT2*, *TNNI3*, *TPM1*, *MYBPC3*, *MYH7*, *MYL2*, *MYL3*, or *ACTC* account for about 60% of familial HCM.
 - Mutations in *PRKAG2* or *LAMP2* account for about 1% of all familial HCM and for about 50% of HCM with ventricular pre-excitation.
[Taylor MR, et al \(2004\) Exp Rev Mol Diagn 4:99-113](#)
[Ho CY, Seidman CE \(2006\) Circulation 113:858-62](#)
[Van Driest SL, et al \(2005\) Mayo Clin Proc 80:463-69](#)
[Arad M, et al \(2005\) N Engl J Med 352:362-72](#)
- **HCM leads to death from severe complications in about 10% of patients.**
 - Of 744 HCM patients, 6% died from SCD (mean age 45), 4% from congestive heart failure (mean age 56), and 2% from stroke (mean age 73).
[Maron BJ, et al \(2000\) Circulation 102:858-64](#)
- **HCM is the most common cause of sudden cardiac death (SCD) in young adults, including trained athletes. Affected individuals are often unaware of their condition.**
 - About a third of SCD in athletes is due to HCM (134 individuals, mean age 17).
[Maron BJ, et al \(1996\) JAMA 276:199-204](#)
- **Extensive cardiac screening at regular intervals can identify patients at high risk for SCD, who may benefit from implantation of a cardioverter-defibrillator.**
 - ICDs corrected potentially lethal arrhythmias in 20% of 506 high-risk HCM patients.
[Maron BJ, et al \(2007\) JAMA 298:405-12](#)
- **Children who are genetically predisposed to HCM may be advised not to participate in certain competitive sports.**
[Maron BJ, et al \(2004\) Circulation 109:2807-19.](#)
- **Genetic testing can confirm a diagnosis of HCM in the index patient for a family and identify family members with a predisposition for HCM at any age.**
[Marian AJ, et al. \(2003\) Circulation 107:2171-4](#)
- **Genetic testing can distinguish metabolic HCM from sarcomeric HCM, informing prognosis and genetic counseling of HCM patients.**
[Arad M, et al \(2005\) N Engl J Med 352:362-72](#)
- **Genetic testing is the only reliable way to identify unaffected family members, who do not need regular cardiac screening for risk factors of SCD.**
 - Physical examination alone cannot rule out HCM since symptoms can develop at any time during life.
[Maron BJ, et al \(2004\) J Am Coll Cardiol 44:2125-32](#)
[Marian AJ, et al. \(2003\) Circulation 107:2171-4](#)