Facts on Hypertrophic Cardiomyopathy (HCM)

- **HCM is a dominantly inherited disease affecting 1 in 500 individuals.**
  

- **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.
  
  
  
  

- **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).
  

- **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).
  

- **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.
  

- **Children who are genetically predisposed to HCM may be advised not to participate in certain competitive sports.**
  

- **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.**
  

- **Genetic testing can distinguish metabolic HCM from sarcomeric HCM, informing prognosis and genetic counseling of HCM patients.**
  

- **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.
  
  