

Using Genetic Testing to Diagnose Hypertrophic Cardiomyopathy

Indications:

- Clinical diagnosis of hypertrophic cardiomyopathy (HCM)
- Unexplained cardiovascular symptoms such as chest pain, dyspnea and/or history of syncopal episodes in young adults, especially athletes
- Family history of sudden cardiac death (SCD) in individuals under age 45

Benefits:

Genetic testing for HCM can:

- confirm a clinical diagnosis of HCM.
- distinguish between different forms of HCM
- identify at-risk family members who should undergo regular cardiac screening
- identify family members who do not need to undergo regular cardiac screening

Background:

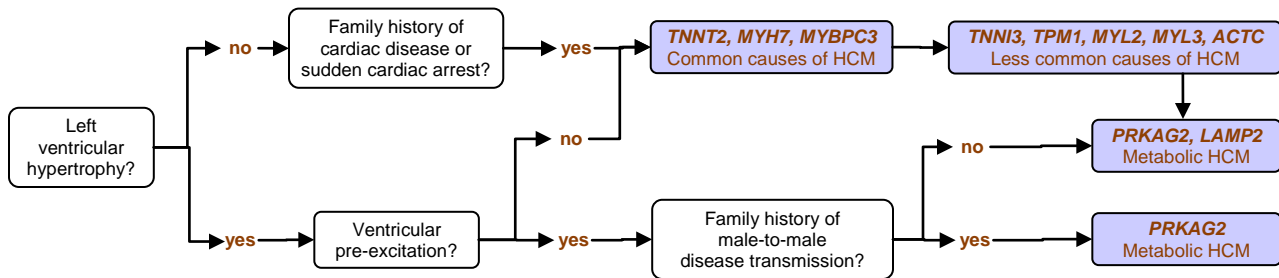
- Hypertrophic cardiomyopathy (HCM), which is characterized by a thickening of the heart muscle, occurs at a prevalence of 1 in 500 (0.2%) and typically shows dominant mode of inheritance.^{1,2}
- HCM has a benign prognosis in most patients, but leads to severe complications including heart failure, stroke, and cardiac arrest in about 10% of patients.^{1,2}
- HCM the most common cause of SCD in young adults, including competitive athletes.³
- Extensive cardiac screening at regular intervals can identify patients at high risk for SCD, who may benefit from implantation of a cardioverter-defibrillator.⁴
- About 60% of all familial HCM is associated with mutations in any one of at least 10 different components of the sarcomer.^{5,6}
- About 1% of HCH and up to 50% of HCM with ventricular pre-excitation is due to cytoplasmic or lysosomal glycogen accumulation in cardiac myocytes (metabolic HCM).^{7,8}
- Metabolic HCM may differ from sarcomeric HCM with regard to prognosis and mode of inheritance.^{7,8}

References: 1. Maron BJ (2002) JAMA 287:1308-20. 2. Taylor MRG, et al. (2004) Expert Rev. Mol. Diagn. 4:99-113. 3. Maron BJ, et al. (2000) Circulation. 858-64. 4. Maron BJ, et al (2007) JAMA 298:405-12. 5. Van Driest SL, et al. (2005) Mayo Clin. Proc. 80(4):463-9. 6. Ho CY, Seidman CE (2006) Circulation 113 22:858-62. 7. Arad M, et al (2005) N Engl J Med 352:362-72. 8. Gollob MH, et al (2002) Curr Opin Cardiol 17:229-34.

Ordering Information: Please see other side.

Ordering Information for Hypertrophic Cardiomyopathy Testing

Guidelines for Test Selection



Ordering Information for Single Gene Tests

Gene(s)	Test Code
TNNT2	190301
TNNI3	190302
TPM1	190303
MYBPC3	190304
MYH7	190305
MYL2	190306
MYL3	190307
ACTC	190308
PRKAG2	190309
LAMP2	190310

Ordering Information for Multi-Gene Panels*

TNNT2, TNNI3, TPM1, MYBPC3, MYH7, MYL2, MYL3, ACTC	190396
TNNT2, MYH7, MYBPC3	190398
TPM1, TNNI3, MYL2, MYL3, ACTC	190397
PRKAG2, LAMP2	190395
Reflexive testing option: TNNT2, MYH7, MYBPC3 → TPM1, TNNI3, MYL2, MYL3, ACTC <i>(reflexive occurs if no (probable) disease variant is found)</i>	190350

*For multi-gene panels, a summary report will be issued in addition to an abbreviated report for each individual gene.

Family Testing (single amplicon)

Family Testing is available for all genes. Please contact Client Services at 1-866-647-0735 for requirements.

Test Methodology

- Amplification by polymerase chain reaction (PCR); sequencing of entire protein-coding region

NOTE: Specimens must be accompanied by a completed consent form. In the case of family tests (ie, known mutations), a copy of the result of the first patient tested in the family (the index case) must be submitted unless that test was performed at Correlagen. Other family members are subsequently tested for the specific mutation found in the first patient tested.

For test information, sample requirements, or to request a sample shipping kit, please contact Client Services at 1-866-647-0735 or visit us on the web at www.correlagen.com.