

## 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:

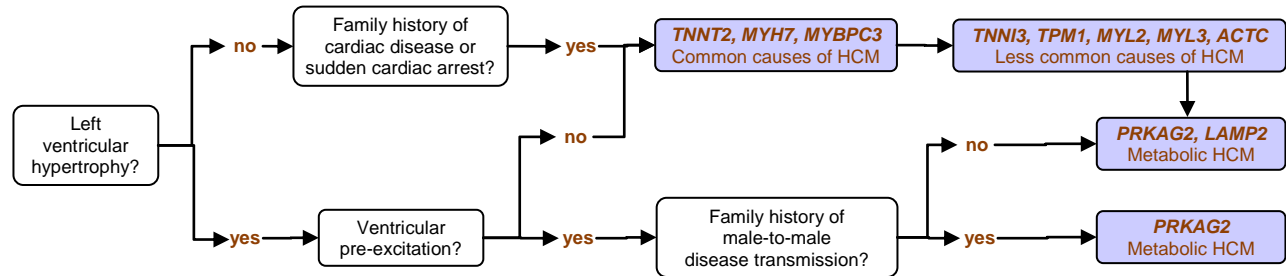
- Hypertrophic cardiomyopathy (HCM), which is characterized by a thickening of the heart muscle, occurs at a prevalence of 1 in 500 (0.2%) and typically shows dominant mode of inheritance.<sup>1,2</sup>
- HCM has a benign prognosis in most patients, but leads to severe complications including heart failure,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

**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)	CPT Codes	Test Code
<b>TNNT2</b>	83891(1) 83892(1) 83898(14) 83904(28) 83909(28) 83912(1)	190301
<b>TNNI3</b>	83891(1) 83892(1) 83898(7) 83904(14) 83909(14) 83912(1)	190302
<b>TPM1</b>	83891(1) 83892(1) 83898(10) 83904(20) 83909(20) 83912(1)	190303
<b>MYBPC3</b>	83891(1) 83892(1) 83898(30) 83904(60) 83909(60) 83912(1)	190304
<b>MYH7</b>	83891(1) 83892(1) 83898(36) 83904(72) 83909(72) 83912(1)	190305
<b>MYL2</b>	83891(1) 83892(1) 83898(6) 83904(12) 83909(12) 83912(1)	190306
<b>MYL3</b>	83891(1) 83892(1) 83898(4) 83904(8) 83909(8) 83912(1)	190307
<b>ACTC</b>	83891(1) 83892(1) 83898(6) 83904(12) 83909(12) 83912(1)	190308
<b>PRKAG2</b>	83891(1) 83892(1) 83898(16) 83904(32) 83909(32) 83912(1)	190309
<b>LAMP2</b>	83891(1) 83892(1) 83898(9) 83904(18) 83909(18) 83912(1)	190310

### Ordering Information for Multi-Gene Panels\*

<b>TNNT2, TNNI3, TPM1, MYBPC3, MYH7, MYL2, MYL3, ACTC</b>	83891(1) 83892(1) 83898(113) 83904(226) 83909(226) 83912(9)	190396
<b>TNNT2, MYH7, MYBPC3</b>	83891(1) 83892(1) 83898(80) 83904(160) 83909(160) 83912(4)	190398
<b>TPM1, TNNI3, MYL2, MYL3, ACTC</b>	83891(1) 83892(1) 83898(33) 83904(66) 83909(66) 83912(6)	190397
<b>PRKAG2, LAMP2</b>	83891(1) 83892(1) 83898(25) 83904(50) 83909(50) 83912(3)	190395

### Reflexive testing option:

**TNNT2, MYH7, MYBPC3** → **TPM1, TNNI3, MYL2, MYL3, ACTC** (reflexing occurs if no (probable) disease variant is found) 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)

(applies to all genes) 83891(1) 83892(1) 83898(1) 83904(2) 83909(2) 83912(1) use single-gene test code

### Test Methodology and Sample Requirements

- Amplification by polymerase chain reaction (PCR); sequencing of entire protein-coding region
- For blood samples:
  - 2mL whole blood in EDTA tube (lavender top)
  - Samples can be stored briefly at 4°C, but should be shipped on day of collection.
- For buccal swab samples: (only accepted for family testing)
  - Please contact client services at 1-866-647-0735 for instructions.
- All sample types should be shipped overnight at room temperature.
- To request a sample shipping kit, please call 1-866-647-0735.

### Turn-around Times

Turn-around times typically range from 7 to 21 days of receipt of sample and all required forms, but may vary depending on test volume and test-specific technical difficulties. Current TATs are posted on our website. Please schedule patient follow-up appointments for discussion of test results conservatively at 6 weeks.

**For more information, please contact Correlagen Diagnostics, Inc., at 1-866-647-0735 or visit us on the web at [www.correlagen.com](http://www.correlagen.com).**