

## Using Genetic Testing to Diagnose Familial Dilated Cardiomyopathy

### Indications:

- Clinical diagnosis of DCM
- Documented dilation and/or dysfunction of the left ventricle
- Family history of DCM

### Benefits:

Genetic testing for DCM can:

- confirm a diagnosis of familial DCM, even in absence of family history.
- identify family members of an index patient who harbor the familial mutation and should undergo extensive cardiac screening at regular intervals.
- identify family members of an index patient who do not harbor the familial mutation and do not need to undergo extensive cardiac screening at regular intervals.

### Background:

- Dilated cardiomyopathy (DCM) is characterized by left ventricular dilatation and systolic dysfunction leading to progressive heart failure.<sup>1</sup>
- DCM occurs at a prevalence of at least 1 in 2500 and is familial in 30-60% of cases.<sup>2</sup>
- Mutations in *TNNT2*, *TNNI3*, *TPM1*, *MYBPC3*, *MYH7*, *ACTC*, and *LMNA* account for about 15-40% of familial DCM.<sup>3</sup>
- Untreated DCM is associated with a five-year mortality rate of up to 50%.<sup>4</sup>
- The most common manifestation of DCM is congestive heart failure; sudden cardiac death or stroke can also occur.<sup>1,2,4</sup>
- Due to the progressive nature of DCM, absence of symptoms cannot guarantee a favorable prognosis at any age.<sup>5</sup>

**References:** 1. Mestroni L, et al. (1999) Eur. Heart J 20:93-102. 2. Colombo MG, et al. (2008) Cardiovasc Ultrasound 6: 62. 3. Burkett EL, et al. (2005) J. Am. Coll. Cardiol. 45:969-981. 4. Elliott P (2000) Heart 84:106-112. 5. Crispell KA, et al. (2002) J. Am. Coll. Cardiol. 39:1503-1507.

**Ordering Information:** please see other side

## Ordering Information for Familial Dilated Cardiomyopathy (DCM) Testing

### Indications for Testing

- Clinical diagnosis of DCM
- Documented dilation and/or dysfunction of the left ventricle
- Family history of DCM

### Ordering Information

Gene(s)	CPT Codes	Test Code
<b>TNNT2</b>	83891(1) 83892(1) 83898(14) 83904(28) 83909(28) 83912(1)	190501
<b>TNNI3</b>	83891(1) 83892(1) 83898(7) 83904(14) 83909(14) 83912(1)	190502
<b>TPM1</b>	83891(1) 83892(1) 83898(10) 83904(20) 83909(20) 83912(1)	190503
<b>MYBPC3</b>	83891(1) 83892(1) 83898(30) 83904(60) 83909(60) 83912(1)	190504
<b>MYH7</b>	83891(1) 83892(1) 83898(36) 83904(72) 83909(72) 83912(1)	190505
<b>ACTC</b>	83891(1) 83892(1) 83898(6) 83904(12) 83909(12) 83912(1)	190506
<b>LMNA</b>	83891(1) 83892(1) 83898(12) 83904(24) 83909(24) 83912(1)	190507

### Multi-Gene Panels\*

<b>TNNT2, TPM1, MYBPC3, MYH7, ACTC, LMNA (dominantly inherited causes of DCM)</b>	83891(1) 83892(1) 83898(108) 83904(216) 83909(216) 83912(6)	190599
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### Family Testing (single amplicon)

<b>(applies to all genes)</b>	83891(1) 83892(1) 83898(1) 83904(2) 83909(2) 83912(1)	Use single-gene test code
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### Test Methodology and Sample Requirements

- Amplification by polymerase chain reaction (PCR); sequencing of entire protein-coding region
- For blood samples:
  - 2 mL 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.

**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)**