

## 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)	Test Code
<b><i>TNNT2</i></b>	190501
<b><i>TNNI3</i></b>	190502
<b><i>TPM1</i></b>	190503
<b><i>MYBPC3</i></b>	190504
<b><i>MYH7</i></b>	190505
<b><i>ACTC</i></b>	190506
<b><i>LMNA</i></b>	190507

### Multi-Gene Panels

<b><i>TNNT2, TPM1, MYBPC3, MYH7, ACTC, LMNA</i></b> <b><i>(dominantly inherited causes of DCM)</i></b>	190599
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### 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](http://www.correlagen.com).**