

CardioGeneScan Using Genetic Testing to Diagnose Familial Aortopathy

Indications:

- Clinical diagnosis or suspicion of Marfan syndrome (MFS), Loeys Dietz syndrome (LDS), vascular-type Ehlers-Danlos syndrome (vt-EDS), or thoracic aortic aneurysm and dissections (TAAD)
- Family history of MFS, LDS, vt-EDS, or TAAD

Benefits:

Genetic testing for Familial Aortopathy can:

- Confirm a clinical diagnosis of Marfan syndrome, Loeys-Dietz syndrome, vascular-type Ehlers-Danlos syndrome, or TAAD.
- 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 require the same level of intensive cardiac screening at regular intervals.
- In some cases, predict the severity of disease based on the exact mutation found.

Background :

- Aortopathy is characterized by aortic dilation, which can lead to life threatening aneurysm and/or dissections.¹⁻⁵
- About 30% of thoracic aortic aneurysm and dissections (TAAD) is familial and typically associated with more aggressive disease progression than non-familial forms.⁶
- Familial TAAD is seen in the context of several multi-system syndromes with overlapping symptoms, such as Marfan syndrome,¹ Loeys-Dietz syndrome,² and vascular-type Ehlers-Danlos syndromes,³ but can also occur in isolation.^{4,6}
- Identifying the underlying genetic cause of TAAD is very important, since therapy and prognosis may vary considerably.¹⁻⁵
- Genetic testing can assist in the differential diagnosis of aortopathies.¹⁻⁴
- Genetic causes of aortopathy tested for in the Aortopathy sequencing evaluation account for about 30% of familial cases overall and about 90% of syndromic cases.^{1-4,6}

For References and Ordering Information: please see other side

Ordering Information for Familial Aortopathy Testing

Indications for Testing

- Clinical diagnosis or suspicion of Marfan syndrome, Loeys Dietz syndrome, vascular-type Ehlers-Danlos syndrome, or TAAD
- Family history of Marfan syndrome, Loeys Dietz syndrome, vascular-type Ehlers-Danlos syndrome, or TAAD

Ordering Information

Test Code 190660

Test Methodology

- Protein coding regions are selectively enriched by hybridization to specific nucleic-acid probes or by PCR and sequenced using next-generation sequencing technology. All variants known or predicted to be pathogenic are confirmed by uni-directional Sanger sequencing.

Sample Requirements

- 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 DNA samples:
 - Minimum requirement of 15 µg DNA, preferably in 10 mM TE (if different, please specify)
 - DNA concentration should be clearly indicated
- All sample types should be shipped overnight at room temperature
- For all sample types, a minimum of 2 sample identifiers are required on every sample sent to Correlagen
- To request a sample shipping kit, please call 1-617-851-5798

References

1. Dietz H. Marfan syndrome. GeneReviews. <http://www.ncbi.nlm.nih.gov/bookshelf/br.fcgi?book=gene&part=marfan>. Accessed 022110
2. Loeys BL, Dietz HC. Loeys-Dietz syndrome. GeneReviews. <http://www.ncbi.nlm.nih.gov/bookshelf/br.fcgi?book=gene&part=loeys-dietz>. Accessed 022110
3. Pepin MG, Byers PH. Ehlers-Danlos syndrome, Vascular Type. GeneReviews. <http://www.ncbi.nlm.nih.gov/bookshelf/br.fcgi?book=gene&part=eds4>. Accessed 022110.
4. Milewicz DM, Tran-Fadulu V. Thoracic aortic aneurysms and aortic dissections. GeneReviews. <http://www.ncbi.nlm.nih.gov/bookshelf/br.fcgi?book=gene&part=taa>. Accessed 022110.
5. Judge DP, Dietz HC (2008) Therapy of Marfan syndrome. Annu Rev Med 59:43-59.
6. Albornoz G, Coady MA, Roberts M, Davies RR, Tranquilli M, Rizzo JA, and Elefteriades JA (2006) Familial thoracic aortic aneurysms and dissections – incidence, modes of inheritance, and phenotypic patterns. Ann Thorac Surg 82:1400-1405.

**For more information, please contact Correlagen Diagnostics, Inc., at 1-617-577-0152
or visit us on the web at www.correlagen.com.**