



HEREDITARY THORACIC AORTIC ANEURYSMS & AORTIC DISSECTIONS Requisition

Ship to:
CHEO Genetics Diagnostic Laboratory
401 Smyth Road, Rm w3401
Ottawa, ON, K1H 8L1
Tel: (613) 737-7600 ext. 1230 Fax: (613) 738-4814
<https://www.cheo.on.ca/en/clinics-services-programs/genetics-diagnostic-laboratory.aspx>

Patient Name: _____
Last First Initial
Health Card Number: _____
DOB: (yy/mm/dd) _____
Address: _____

Telephone: _____
Sex (check one): Male Female

ALL SECTIONS MUST BE COMPLETED

Collection Date: _____
Collection Centre: _____
CHEO Pedigree Number: _____

Sample Requirements

Blood: Blood 2x 6 mL EDTA Blood 2x 3 mL EDTA (child) Blood 3 mL EDTA (infant ≤1 year)
For any other sample types, please contact the laboratory directly.

Health Care Provider Requesting Test

Name: _____ Copy to: Name: _____
Registration Number: _____ Registration Number: _____
Address: _____ Address: _____

Telephone: _____ Telephone: _____
FAX: _____ FAX: _____

Test Requested (see next page for the clinical testing criteria and a list of the genes included in each panel)

- Arterial Tortuosity syndrome (*SLC2A10*)
- Marfan syndrome (*FBN1*)
- Loews-Dietz syndrome panel (7 genes; see page 2)
- Thoracic aortic aneurysms and aortic dissection panel (19 genes; see page 2)
- Single Gene Testing (Specify Gene): _____
- Store DNA for future testing (DNA will be stored for 2 years, then discarded)
- Family Variant Specific Test
(Include a copy of the family member's genetic test report.)
Gene(s) _____
Variant(s) _____
Proband name: _____
Proband date of birth: _____
Relationship to proband: _____

Clinical Information

Thoracic aortopathy Yes (specify below) No Unknown

Type: Aneurysm
 Dissection

Location: Root (Z score: ____)
 Ascending
 Arch
 Descending

- Ghent systemic score (following geneticist examination): _____
- Ectopia lentis
- Hypertension (treated with medication)
- Mitral valve prolapse
- Bicuspid aortic valve
- Hypertrophic cardiomyopathy

Other clinical features: _____

Positive Family History (1st and 2nd degree relatives only) Yes (specify below) No Unknown

Ethnicity: _____

HEREDITARY THORACIC AORTIC ANEURYSMS & AORTIC DISSECTIONS TEST DETAILS

Methodology of genetic testing:

- 1) Sequencing: next-generation sequencing analysis of coding sequences of the relevant genes and 10 base pairs immediately adjacent to each exon. In addition, several deep intronic regions are analyzed for the presence of specific clinically relevant variants.
- 2) MLPA: to detect large genomic deletions and duplications, multiplex ligation-dependent probe amplification (MLPA) is performed for certain genes.

Arterial Tortuosity Syndrome

Gene tested: *SLC2A10* (sequencing)

Marfan Syndrome

Gene tested: *FBN1* (sequencing and MLPA)

Loeys-Dietz Syndrome Panel

Genes included in panel: *SLC2A10*, *SMAD2*, *SMAD3*, *TGFB2*, *TGFB3*, *TGFBR1*, and *TGFBR2*

Analysis includes sequencing as described above, and MLPA of *TGFBR1* and *TGFBR2*

Familial Thoracic Aortic Aneurysms and Aortic Dissections Panel

Genes included in panel: *ACTA2*, *ARIH1*, *COL3A1*, *EFEMP2*, *FBN1*, *FOXE3*, *LOX*, *MYH11*, *MYLK*, *PRKG1*, *ROBO4*, *SLC2A10*, *SMAD2*, *SMAD3*, *TGFB2*, *TGFB3*, *TGFBR1*, *TGFBR2*, and *THSD4*

Analysis includes sequencing as described above, and MLPA of *COL3A1*, *FBN1*, *TGFBR2*, and *TGFBR1*

Selecting a CHEO panel

