



Genetics Diagnostic Laboratory Hereditary Thoracic Aortic Aneurysms & Aortic Dissections Requisition

**Blood Work Instructions
For Non-CHEO Patients:**
Bring requisition to local community
lab for blood collection

Sample to be shipped to:
Genetics Diagnostics Laboratory
401 Smyth Road, Rm W3401
Ottawa, ON, K1H 8L1
Tel: (613) 738-3230
Fax: (613) 738-4814
Website: www.cheo.on.ca/GDL

Patient Name:

Last First Initial

Health Card Number:

DOB (yy/mm/dd):

Address:

Telephone:

Legal Sex:

☐ Male ☐ Female ☐ Unknown

**Optional - Sex
assigned at birth:**

☐ Male ☐ Female ☐ Unknown ☐ Uncertain ☐ Not recorded

**Optional - Patient
Consent to share
results:**

Sharing results for family testing: By checking the box below, the patient authorizes CHEO to use their results to guide testing or interpretation for relatives within CHEO's Genetic Diagnostic Lab. Results are used only to inform family member testing decisions. Consent can be changed at any time by contacting the lab.

☐ Patient consents to sharing results with family members

Priority of Testing

- ☐ Routine
Expedited:
☐ Patient/Partner Pregnant
☐ Prenatal Diagnosis
☐ Newborn (≤ 3 months)
☐ Clinical management
contingent upon STAT results

Sample Type (for prenatal/cord blood: separate MCC requisition required)

- ☐ Blood 2x6 mL EDTA ☐ Cultured Amniocytes 2xT25
☐ Blood 3 mL EDTA ☐ Cultured CVS 2xT25
☐ Blood 2x3 mL EDTA ☐ Direct amniotic fluid 20 mL
☐ DNA _____ μ g ☐ Direct CVS 10–20 mg
source: _____ ☐ Cultured cells
source: _____

For Lab Use Only

Collection Date:

Collection Centre:

Collected By:

Billing (Non-OHIP / Institutional Only- *Attach Ministry of Health or IFHP approval documentation)

- ☐ Ministry of Health Approved (e.g. RAMQ)*
☐ Ministry of Health Pending Approval
☐ IFHP Approved*
☐ Non-insured (i.e. self-pay)

☐ Institutional Billing (complete details below)

Institution Name:

City: _____ **Province:** _____ **Postal Code:** _____

Tel: _____ **Fax:** _____

Authorizing Health Care Provider(s) (testing will be accepted from Cardiology or Genetics Clinic referrals only)

Provider Name:

Registration #:

Address:

Telephone: _____ **Fax:** _____

COPY TO Name:

Registration #:

Address:

Telephone: _____ **Fax:** _____

Clinical Indication (required—testing will not proceed unless completed)

- ☐ Confirmed or suspected diagnosis of disease
☐ Not affected ☐ Family history
☐ Other clinical features (specify): _____

☐ Familial variant testing (attach family report)

Specify family relationship: _____

Privacy statement: By ordering this test, I attest that my patient/patient's guardian has obtained express consent or I have obtained express consent from the family member for CHEO to use the family member's report for the purpose of genetic testing and evaluation.

Test/Panel Requested

☐ Thoracic aortic aneurysms and aortic dissection

Includes genes found in Loeys-Dietz, Marfan and Arterial Tortuosity panels (see page 2)

☐ Loeys-Dietz syndrome

☐ Marfan syndrome

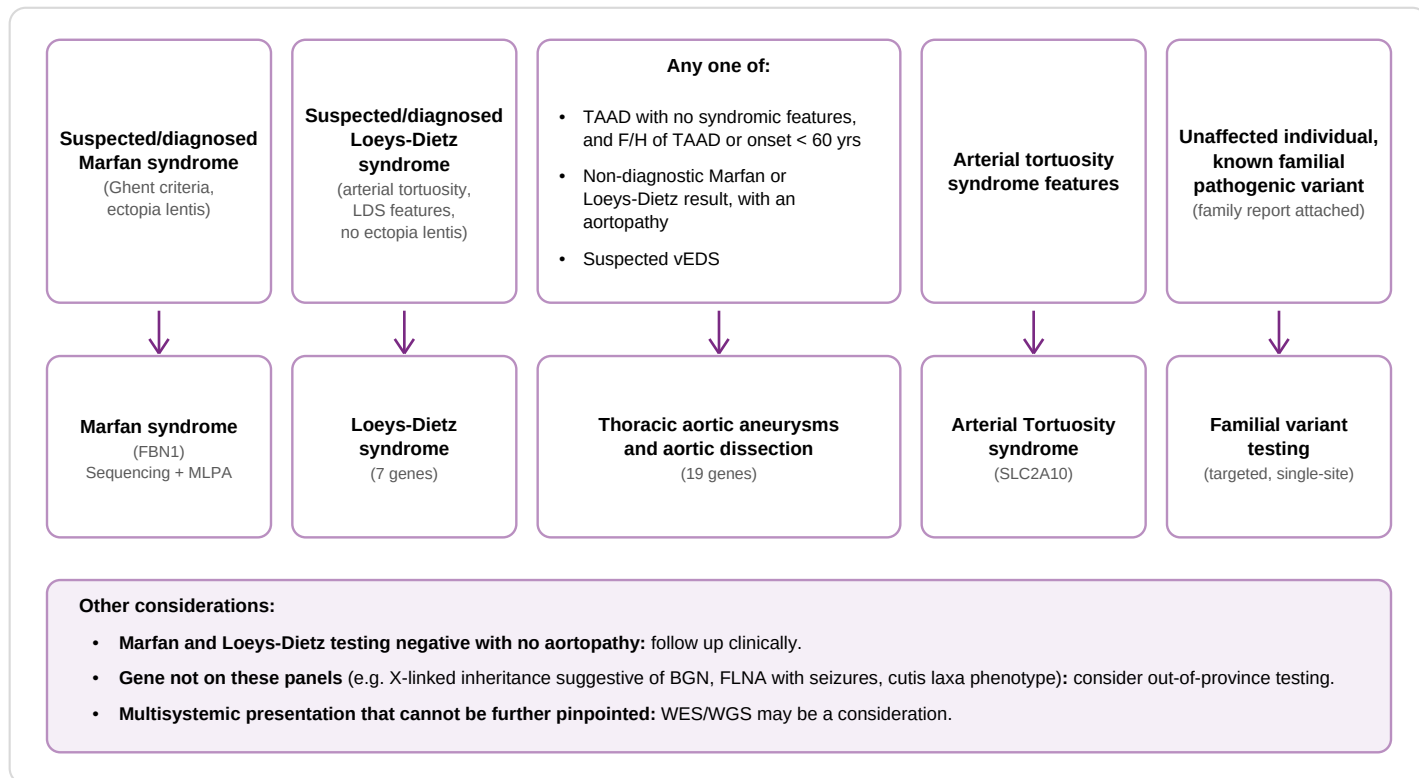
☐ Arterial Tortuosity syndrome

☐ Familial Variant Testing gene(s) and variant(s): _____ ☐ Single gene testing specify: _____

☐ Maternal Cell Contamination (MCC)

☐ DNA Storage (2-year retention)

Vascular Connective Tissue Disorders / TAAD Panels - Genetic Testing Utilization Considerations



TAAD- thoracic aortic aneurysm and dissection. **F/H**- family history. **LDS**- Loeys-Dietz syndrome. **MFS**- Marfan syndrome. **vEDS**- vascular Ehlers-Danlos syndrome. **HTN**- hypertension. **MLPA**- multiplex ligation-dependent probe amplification. **WES/WGS**- whole exome / whole genome sequencing. Per CHEO Connective Tissue Clinic referral criteria: personal history of thoracic/brain aneurysm, vascular dissection, or hollow organ rupture; F/H of TAAD, peripheral or intracranial aneurysm, or unexplained sudden death at a young age in a first- or second-degree relative; F/H of MFS, LDS, or vEDS warrants referral.

Gene content for the Vascular Connective Tissue / TAAD Panels

Panel	# Genes	Genes Included
Marfan Syndrome	1	FBN1 (sequencing and MLPA)
Loeys-Dietz Syndrome	7	SLC2A10, SMAD2, SMAD3, TGFB2, TGFB3, TGFB1, TGFB2 (sequencing; MLPA of TGFB1 and TGFB2)
Thoracic Aortic Aneurysms and Aortic Dissection	19	ACTA2, ARIH1, COL3A1, EFEMP2, FBN1, FOXE3, LOX, MYH11, MYLK, PRKG1, ROBO4, SLC2A10, SMAD2, SMAD3, TGFB2, TGFB3, TGFB1, TGFB2, THSD4 (sequencing; MLPA of COL3A1, FBN1, TGFB1 and TGFB2)
Arterial Tortuosity Syndrome	1	SLC2A10 (sequencing)