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 2×6 mL EDTA
☐ Blood 3 mL EDTA
☐ Blood 2×3 mL EDTA
☐ DNA _____ µg source:_____
☐ Cultured Amniocytes 2×T25
☐ Cultured CVS 2×T25
☐ Direct amniotic fluid 20 mL
☐ Direct CVS 10–20 mg
☐ 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**

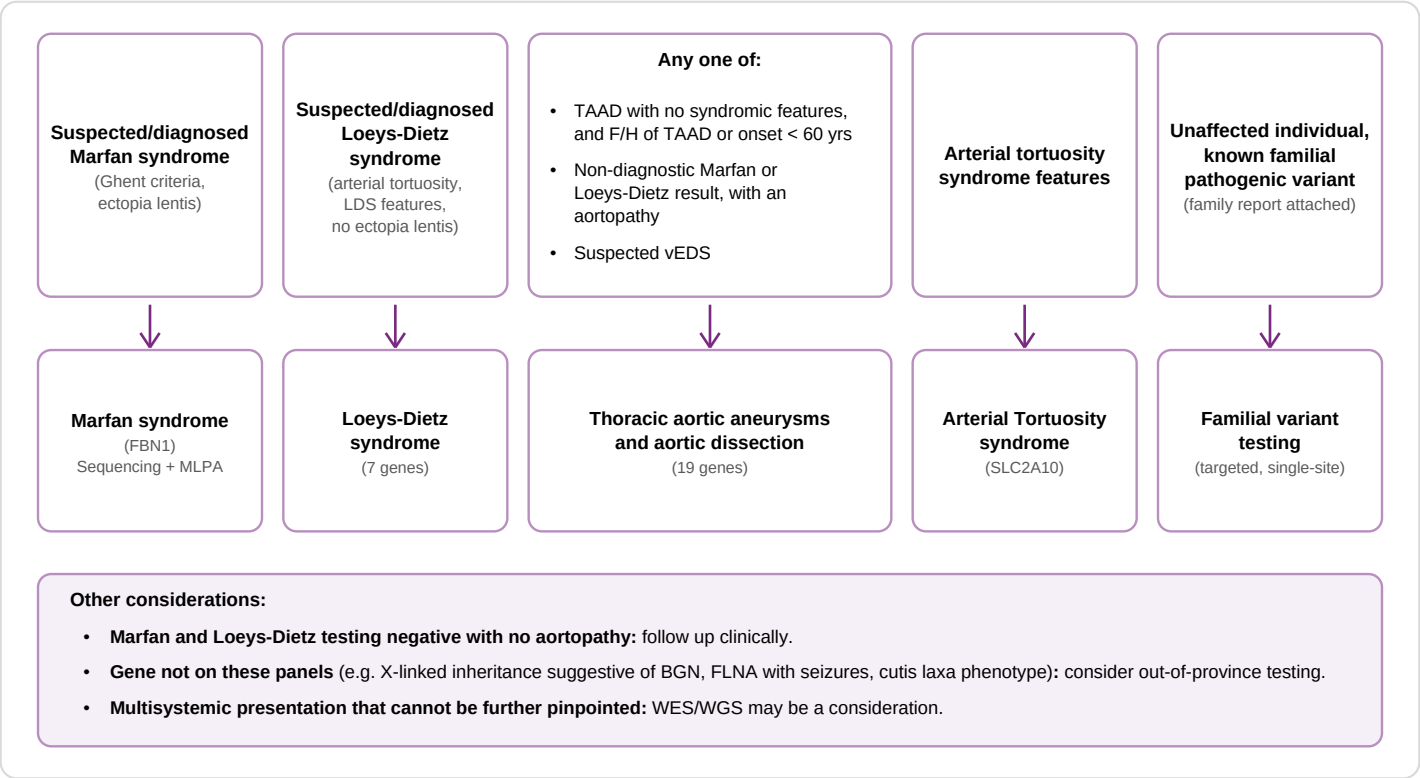
Includes genes found in Loeys-Dietz, Marfan and Arterial Tortuosity panels (see page 2)

☐ **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, TGFBR1, TGFBR2 (sequencing; MLPA of TGFBR1 and TGFBR2)
Thoracic Aortic Aneurysms and Aortic Dissection	19	ACTA2, ARIH1, COL3A1, EFEMP2, FBN1, FOXE3, LOX, MYH11, MYLK, PRKG1, ROBO4, SLC2A10, SMAD2, SMAD3, TGFB2, TGFB3, TGFBR1, TGFBR2, THSD4 (sequencing; MLPA of COL3A1, FBN1, TGFBR1 and TGFBR2)
Arterial Tortuosity Syndrome	1	SLC2A10 (sequencing)