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MAYO CLINIC LABORATORIES

Connective Tissue/Cerebrovascular Disease Genetic Testing Patient Information

Instructions: Accurate interpretation and reporting of genetic results is contingent upon the reason for testing, clinical information, ethnic background/ancestry, and family history. To help provide the best possible service, supply the information requested below and **send paperwork with the specimen, or return by fax to Mayo Clinic Laboratories, Attn: Molecular Technologies Laboratory Genetic Counselors at 507-284-1759. Phone: 800-533-1710 / International clients: +1-507-266-5700 or email MLIINT@mayo.edu**

Patient Information

Patient Name *(Last, First, Middle)*	Birth Date *(mm-dd-yyyy)*
Sex Assigned at Birth ☐ Male ☐ Female ☐ Unknown ☐ Choose not to disclose	Legal/Administrative Sex ☐ Male ☐ Female ☐ Nonbinary

Referring Provider Information

Referring Provider Name *(Last, First)*	Phone	Fax*
Genetic Counselor Name *(Last, First)*	Phone	Fax*

**Fax number given must be from a fax machine that complies with applicable HIPAA regulations.*

Is this a postmortem specimen? ☐ Yes ☐ No If "Yes," attach autopsy report if available.

Reason for Testing Check all that apply.

☐ Diagnosis ☐ Family history** ☐ Sudden death

**Genetic testing should be performed on an affected family member first, when possible. FMTT / Familial Mutation Targeted Testing should be ordered when there is a previous positive genetic test result in the family.

Clinical History

Diagnosis/Suspected Diagnosis

☐ Marfan Syndrome ☐ Loeys-Dietz Syndrome ☐ Osteogenesis Imperfecta
☐ Ehlers-Danlos Syndrome ☐ Familial thoracic aortic aneurysm and dissection ☐ Cerebrovascular disease/stroke
☐ Other:

Indicate whether the following are present:

- ☐ Aortic diameter at sinuses of Valsalva Z-score ≥ 2
- ☐ Aortic dissection
- ☐ Ectopia lentis
- ☐ Systemic score ≥ 7 points (see table to the right for calculation)
- ☐ Aortic dilatation/aneurysm (Z-score < 2)
- ☐ Family history of independently diagnosed Marfan syndrome using the revised Ghent criteria
- ☐ Talipes equinovarus
- ☐ Hypertelorism
- ☐ Craniosynostosis
- ☐ Cleft palate
- ☐ Bifid uvula
- ☐ Blue sclerae
- ☐ Arterial tortuosity
- ☐ Patent ductus arteriosus
- ☐ Velvety/translucent skin
- ☐ Easy bruising
- ☐ Widened atrophic scars
- ☐ Spontaneous organ rupture
- ☐ Aortic Dimensions mm, Z-score
- ☐ Fractures; describe:
- ☐ Hearing loss
- ☐ Stroke
- ☐ Other aneurysm; describe:
- ☐ Other:

Ghent Systemic Score Calculation for Marfan Syndrome

Feature	Value	Enter Value if Present
Wrist **and** thumb sign	3	
Wrist **or** thumb sign	1	
Pectus carinatum	2	
Pectus excavatum or chest asymmetry	1	
Hindfoot deformity	2	
Plain flat foot (pes planus)	1	
Pneumothorax	2	
Dural ectasia	2	
Protrusio acetabulae	2	
Reduced upper/lower segment **and** increased armspan/height	1	
Scoliosis or thoracolumbar kyphosis	1	
Reduced elbow extension	1	
3 of 5 facial features: • dolichocephaly • enophthalmos • downslanting palpebral fissures • malar hypoplasia • retrognathia	1	
Skin striae	1	
Myopia > 3 diopters	1	
Mitral valve prolapse	1	
	Total	0.00

List any additional features present:

Connective Tissue/Cerebrovascular Genetic Testing Disease Patient Information (continued)

Patient Name *(Last, First, Middle)*	Birth Date *(mm-dd-yyyy)*

Family History

Are there similarly affected relatives? ☐ Yes ☐ No

If "Yes," indicate relationship and symptoms:

Have any family member had genetic testing? ☐ Yes*** ☐ No ☐ Unknown

*****FMTT / Familial Mutation Targeted Testing should be ordered when there is a previous positive genetic test result in the family. Contact the lab for ordering assistance.**

History of consanguinity: ☐ No ☐ Yes; relationship details:

Ancestry

☐ African/African American	☐ East Asian	☐ Latinx/Latine	☐ South Asian	☐ Unknown
☐ Ashkenazi Jewish	☐ European	☐ Middle Eastern	☐ None of the above	☐ Choose not to disclose

New York State Patients: Informed Consent for Genetic Testing is required. See Informed Consent for Genetic Testing (T576) or Informed Consent for Genetic Testing – Spanish (T826).

Print