Complete and print.

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MAYO CLINIC LABORATORIES

Marfan and Related Disorders Patient Information

M F R G P

Instructions: The accurate interpretation and reporting of the genetic results is contingent upon the reason for referral, clinical information, ethnic background, 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: Personalized Genomics Laboratory Genetic Counselors at 507-284-1759. Phone: 507-266-5700 / International clients: +1-507-266-5700 or email mclglobal@mayo.edu**

Patient Information

Patient Name *(Last, First, Middle)*	Birth Date *(mm-dd-yyyy)*	Sex ☐ Male ☐ Female
Referring Provider Name *(Last, First)*	Phone	Fax*
Other Contact 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.

Clinical History

Reason for Testing (Check all that apply.)

☐ Diagnosis ☐ Carrier testing ☐ Presymptomatic diagnosis ☐ Family history ☐ Sudden death

Note: Genetic testing should always be initiated on an affected family member first, if possible, in order to be most informative for at-risk relatives. See Ethnic Background and Family History section for more information.

Diagnosis/Suspected Diagnosis

☐ Marfan Syndrome ☐ Ehlers-Danlos Syndrome ☐ Loeys-Dietz Syndrome ☐ Familial thoracic aortic aneurysm and dissection

☐ 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

Additional Features

- ☐ 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 _____
- ☐ Other: _____

List any additional features present:

Systemic Score Calculation

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

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Marfan and Related Disorders Patient Information (continued)

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

Ethnic Background and Family History

☐ European Caucasian ☐ African American ☐ Hispanic ☐ Asian ☐ Middle Eastern ☐ Other (specify): _____

Are other relatives known to be affected? ☐ Yes ☐ No

If yes, indicate their diagnosis and relationship to the patient: _____

Have other relatives had molecular genetic testing? ☐ Yes ☐ No

For known mutation test requests, order known variant analysis:

KVAR1 / Known Variant Analysis-1 Variant, Varies KVAR2 / Known Variant Analysis-2 Variants, Varies

KVAR3 / Known Variant Analysis-3+ Variants, Varies

New York State Patients: Informed Consent for Genetic Testing is required. See Informed Consent for Genetic Testing (T576), Informed Consent for Genetic Testing – Spanish (T826), or Informed Consent for Genetic Testing for Deceased Individuals (T782).

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