MAYO CLINIC
Mayo Medical Laboratories

Marfan and Related Disorders Patient Information

T G F B R 2

Reset Form

Instructions: The accurate interpretation and reporting of the genetic results is contingent upon the reason for referral, clinical information, ethnic background, and family history. Supply the information requested below and **send this paperwork with the specimen.**

Patient Information

Patient Name *(Last, First, Middle)*	Birth Date *(Month DD, YYYY)*	Sex ☐ Male ☐ Female
Referring Physician Name	Phone	Fax
Other Contact	Phone	Fax

Clinical History

Patient's Diagnosis/Suspected Diagnosis: ☐ Marfan syndrome ☐ Ehlers-Danlos Syndrome Type IV (Vascular Type)
☐ 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 _____

See the diagnostic criteria at the bottom of this page and the following website for specific information about using the revised Ghent criteria: http://www.marfan.org

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

Ethnic Background and Family History - *Attach pedigree if available*

☐ European Caucasian ☐ African American ☐ Hispanic ☐ Asian ☐ Other *(specify)* _____
Are other relatives known to be affected? ☐ Yes ☐ No If Yes, indicate their relationship to the patient _____
Have other relatives had molecular genetic testing? ☐ Yes ☐ No
For Known Mutation orders, a familial mutation must be provided.
Indicate: Gene _____ Exon _____ Amino Acid _____ Nucleotide _____ OR Intron _____ Nucleotide _____
If relative was tested at the Mayo Clinic, name and relationship of relative or Family Number from relative's report _____
Attach a copy of the genetic test lab report if available

For the diagnosis of Marfan syndrome:

In the absence of family history

1. Ao (Z ≥2) and EL=MFS
2. Ao (Z ≥2) and FBN1=MFS
3. Ao (Z ≥2) and Syst (≥7 pts)=MFS
4. EL and with known Ao=MFS

In the presence of family history

5. EL and FH of MFS=MFS
6. Syst (≥7 pts) and FH of MFS=MFS
7. Ao (Z ≥2 above 20 years old, ≥3 below 20 years old) + FH of MFS=MFS

Please complete, print and submit. Print