Marfan Syndrome Diagnostic History and Physical Form

Patient Sticker

I. INTAKE INFORMATION

Name of Patient:	
Date of Birth:	Current Age:
Referring Physician:	
Reason for Referral: (i.e., signs and symptoms of connective tissue disorder)	

II. PERTINENT PAST MEDICAL HISTORY

CARDIOVASCULAR:	Comments:
☐ Thoracic Aortic Aneurysm	
☐ Abdominal Aortic Aneurysm	
☐ Other Aortic Aneurysm	
☐ Tortuosity of Vessels	
☐ History of Dissection	
☐ Valvular Disease	
EYE, SKIN & PNEUMOTHORAX:	Comments:
☐ Dislocated Lenses	
☐ Retinal Detachment	
☐ Myopia	
☐ Poor Healing	
☐ Abnormal Scars	
☐ Striae	
☐ Pneumothorax	
PAST SURGICAL HISTORY:	Comments:
☐ Cardiovascular	
☐ Ocular	
☐ Skeletal	
☐ Hernia	
☐ Other	

III. FAMILY HISTORY

For 1st degree relatives (parents, children, siblings) or 2nd degree relatives (grandparents, aunts, uncles, half-sibling, etc):

☐ Aortic Conditions (e.g., aneurysms or dissections):

☐ Ectopia Lentis (dislocated lenses):

☐ Marfan Syndrome or Related Disorders:

Name of Patient:

IV. PATIENT'S INITIAL ASSESSMENT FINDINGS

- Ectopia Lentis (dislocated lenses):
  - ☐ Yes:
  - ☐ No
  - ☐ Unknown

- FBN-1 Mutation:
  - ☐ FBN-1 known to be associated with Aortic Aneurysm
  - ☐ FBN-1 not known to be associated with Aortic Aneurysm
  - ☐ Unknown (visit www.geneclinics.org)

- First degree relative with history of Marfan syndrome:
  - ☐ Yes:
  - ☐ No
  - ☐ Unknown

- Calculate Aortic Z-score measurement at the sinuses of Valsalva: *Formula for calculation of aortic root Z-scores are available at marfan.org/dx*

Aortic Root Diameter (mm):

Aortic Root Height (cm):

Aortic Root Weight (kg):

Aortic Z-Score:

Aortic Root Z-Score is:
- ☐ ≥ 2 (age ≥ 20 years old) or
- ☐ ≥ 3 (age < 20 years old)

*Calculate Reduced Upper Segment / Lower Segment and Increased Arm Span / Height Measurements

Height:

Lower Segment:

Upper Segment (Height - Lower Segment):

Upper / Lower Segment:

Arm Span:

Arm Span / Height:

Reduced Upper / Lower Segment (i.e., < 0.85 in whites, < 0.78 in blacks) and Increased Arm Span / Height (i.e., > 1.05) contributes 1 point to the systemic score.

V. SYSTEMIC FEATURE SCORING TABLE

FEATURE	VALUE	ENTER VALUE IF PRESENT
Wrist AND thumb sign	3	
Wrist OR thumb sign	1	
Pectus Carinatum deformity	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 segment / lower segment and increased armspan / height without severe scoliosis*	1	
Scoliosis > 20 degrees or thoracolumbar kyphosis	1	
Reduced elbow extension	1	
3 of 5 facial features: • Dolicholcephaly • Malar Hypoplasia • Enophthalmos • Retrognathia • Down-slanting palpebral fissures	1	
Skin striae	1	
Myopia (-.3 diopters)	1	
Mitral valve prolapse	1	
TOTAL: *Systemic Score ≥ 7 = criteria required for diagnosis as indicated below*		

VI. DIAGNOSIS

To make a diagnosis of Marfan syndrome, complete the systemic score and determine if there a family history of Marfan syndrome. Patient has Marfan syndrome if one of the following (7) scenarios exist.

In the **absence of family history** diagnosis can be made if one of the following 4 scenarios exist.

1. Aortic Z-Score ≥ 2 **AND** Ecoptia Lentis
2. Aortic Z-Score ≥ 2 **AND** FBN-1 mutation associated with aortic aneurysm
3. Aortic Z-Score ≥ 2 **AND** Systemic Score ≥ 7pts
4. Ecoptia Lentis **AND** FBN-1 mutation associated with aortic aneurysm

In the **presence of family history** diagnosis can be made if one of the following 3 scenarios exist.

5. Ecoptia Lentis **AND** Family History of Marfan syndrome: 1st degree relative with Marfan syndrome (as defined in adjacent scenarios)
6. Systemic Score ≥ 7pts **AND** Family History of Marfan syndrome: 1st degree relative with Marfan syndrome (as defined in adjacent scenarios)
7. Aortic Z-Score ≥ 2 **AND** Family History of Marfan syndrome: 1st degree relative with Marfan syndrome (as defined in adjacent scenarios)

VII. IMPRESSIONS AND RECOMMENDATIONS