MARFAN SYNDROME DIAGNOSTIC ASSESSMENT FOR PATIENT NOTES

Patient Name Hospital Number

Tick features that are present and file completed form in patient's chart

Skeletal System

Major criterion. Presence of at least 4 of the following manifestations.

- ☐ pectus carinatum
- ☐ pectus excavatum requiring surgery
- ☐ reduced upper to lower segment ratio or arm span to height ratio greater than 1.05
- ☐ wrist and thumb signs
- ☐ scoliosis of greater than 20° or spondylolisthesis
- ☐ reduced extension at the elbows (< 170°)
- ☐ medial displacement of the medial malleolus causing pes planus
- ☐ protrusio acetabulae of any degree (ascertained on radiographs)

Minor criteria.

- ☐ pectus excavatum of moderate severity
- ☐ joint hypermobility
- ☐ highly arched palate with crowding of teeth
- ☐ facial appearance (dolichocephaly, malar hypoplasia, enophthalmos, retrognathia, down-Slanting palpebral fissures)

For the skeletal system to be considered involved, at least 2 of the components comprising the major criterion or one component comprising the major criterion plus 2 of the minor criteria must be present.

Ocular System

Major criterion.

- ☐ ectopia lentis

Minor criteria.

- ☐ abnormally flat cornea (as measured by keratometry)
- ☐ increased axial length of globe (as measured by ultrasound)
- ☐ hypoplastic iris or hypoplastic ciliary muscle causing decreased miosis

For the ocular system to be involved, at least 2 of the minor criteria must be present.

Dura

Major criterion

- ☐ lumbosacral dural ectasia by CT or MRI

Minor criteria

- ☐ None

For the dura to be involved the major criterion must be present.

Family/Genetic History

Major criteria.

- ☐ having a parent, child or sib who meets these diagnostic criteria independently;
- ☐ presence of a mutation in *FBN1* known to cause the Marfan syndrome; or
- ☐ presence of a haplotype around *FBN1*, inherited by descent, known to be associated with unequivocally diagnosed Marfan syndrome in the family

For the family/genetic history to be contributory, one of the major criteria must be present.

Cardiovascular System

Major criteria.

- dilatation of the ascending aorta with or without aortic regurgitation and involving at least the sinuses of Valsalva; or
- dissection of the ascending aorta

Minor criteria.

- mitral valve prolapse with or without mitral valve regurgitation;
- dilatation of the main pulmonary artery, in the absence of valvular or peripheral pulmonic stenosis or any other obvious cause, below the age of 40 years;
- calcification of the mitral annulus below the age of 40 years; or
- dilatation or dissection of the descending thoracic or abdominal aorta below the age of 50 years

For the cardiovascular system to be involved a major criterion or only one of the minor criteria must be present.

Pulmonary System

Major criteria.

None

Minor criteria.

- spontaneous pneumothorax or
- apical blebs (ascertained by chest radiography)

For the pulmonary system to be involved one of the minor criteria must be present.

Skin and Integument

Major criteria.

None

Minor criteria.

- striae atrophicae (stretch marks) not associated with marked weight changes, pregnancy or repetitive stress, or
- recurrent or incisional herniae

For the skin and integument to be involved one of the minor criteria must be present.

Requirements for the Diagnosis of Marfan Syndrome

For the index case:

- If the family/genetic history is not contributory, major criteria in at least 2 different organ systems and involvement of a third organ system
- If a mutation known to cause Marfan syndrome in others is detected, one major criterion in an organ system and involvement of a second organ system

For a relative of an index case:

- presence of a major criterion in the family history and one major criterion in an organ system and involvement of a second organ system

Conclusion

Definite Marfan **Possible Marfan** **Not Marfan**

Physician: Date: