

## 2010 Ghent Criteria for Marfan Syndrome Checklist

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Name: \_\_\_\_\_ DOB: \_\_\_\_\_ MRN: \_\_\_\_\_

| Feature                                                                                                                                                                                                                                                          | Points | Score |
|------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|--------|-------|
| Wrist sign AND thumb sign                                                                                                                                                                                                                                        | 3      |       |
| Wrist OR thumb sign                                                                                                                                                                                                                                              | 1      |       |
| Pectus carinatum deformity                                                                                                                                                                                                                                       | 2      |       |
| Pectus excavatum or chest asymmetry                                                                                                                                                                                                                              | 1      |       |
| Hind foot deformity                                                                                                                                                                                                                                              | 2      |       |
| Pes planus                                                                                                                                                                                                                                                       | 1      |       |
| Pneumothorax                                                                                                                                                                                                                                                     | 2      |       |
| Dural ectasia                                                                                                                                                                                                                                                    | 2      |       |
| Protrusio acetabuli                                                                                                                                                                                                                                              | 2      |       |
| Reduced US/LS AND increases arm/height AND no severe scoliosis                                                                                                                                                                                                   | 1      |       |
| Scoliosis or thoracolumbar kyphosis                                                                                                                                                                                                                              | 1      |       |
| Reduced elbow extension                                                                                                                                                                                                                                          | 1      |       |
| Facial features (3 of 5):<br><input type="checkbox"/> Dolichocephaly<br><input type="checkbox"/> Enophthalmos<br><input type="checkbox"/> Down slanting palpebral fissures<br><input type="checkbox"/> Malar hypoplasia<br><input type="checkbox"/> Retrognathia | 1      |       |
| Skin striae                                                                                                                                                                                                                                                      | 1      |       |
| Myopia > 3 diopters                                                                                                                                                                                                                                              | 1      |       |
| Mitral valve prolapse                                                                                                                                                                                                                                            | 1      |       |
| <b>TOTAL (max 20 points)</b>                                                                                                                                                                                                                                     |        |       |

### Diagnosis of Marfan syndrome in the absence of a family history:

- \_\_\_\_\_ Aorta ( $Z \geq 2$ ) AND ectopia lentis
- \_\_\_\_\_ Aorta ( $Z \geq 2$ ) AND FBN1
- \_\_\_\_\_ Aorta ( $Z \geq 2$ ) AND Systemic  $\geq 7$
- \_\_\_\_\_ Ectopia lentis AND FBN1 aortic disease

### Diagnosis of Marfan syndrome in the presence of a family history AND one of:

- \_\_\_\_\_ Ectopia lentis = ELS\*
- \_\_\_\_\_ Systemic score  $\geq 7$  points = MASS\*\*
- \_\_\_\_\_ MVP and Aorta  $Z \geq 2$  (>20yrs) OR  $Z \geq 3$  (<20yrs) = MVPS\*\*\*

\*\* Aorta = aortic diameter at the sinuses of Valsalva or aortic root dissection

### Related Conditions:

**\*ELS - Ectopia lentis syndrome:** Ectopia lentis with or without systemic features AND with an FBN1 mutation not known to be associated with aortic dilation or no FBN1 mutation

**\*\*MASS - Mitral valve prolapse, Aortic, skeleton and skin:** Aorta ( $Z < 2$ ) AND Systemic score ( $\geq 5$  with at least one skeletal feature) without ectopia lentis

**\*\*\* MVP - Mitral valve prolapse syndrome:** Mitral valve prolapse AND Aorta ( $Z < 2$ ) AND Systemic score ( $< 5$ ) without ectopia lentis