

## **Condition:** Marfan syndrome

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### **Inheritance:**

Autosomal dominant.

### **Genetic etiology:**

Mutation in *FBN1* gene encoding fibrillin 1 protein.

### **Frequency:**

1/5,000 – 1/10,000.

### **Clinical features:**

The Marfan syndrome phenotype affects connective tissue involving multiple organs, including eye, skeleton, and the cardiovascular system. Ocular signs include myopia and dislocation of the lens. Skeletal features include chest wall deformity, scoliosis, flat feet, hypermobile joints, and a tall, lanky habitus with long fingers and toes (arachnodactyly). Cardiovascular manifestations include mitral valve prolapse and dilatation of the aorta and aortic dissection.

### **Management:**

Surveillance for treatable complications, including ophthalmological and orthopedic; monitoring of aortic root and treatment with beta blockers to reduce the risk of dissection. Surgical repair may be indicated in instances of aortic dilatation. Clinical trials underway with losartan, and angiotensin II receptor inhibitor.

### **Genetic counseling:**

Based on autosomal dominant transmission; genetic testing is available.