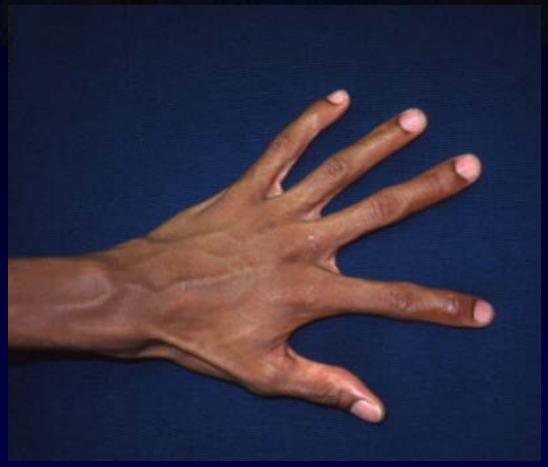
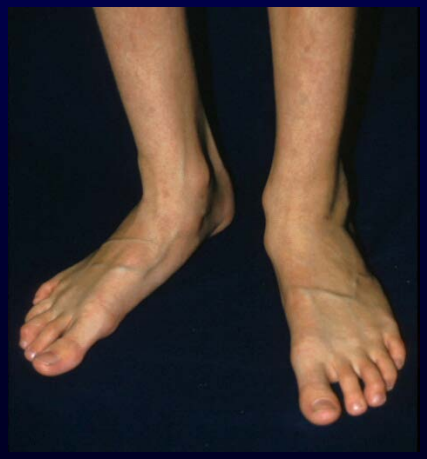
Marfan Syndrome

- **Musculo-skeletal**
  - arachnodactyly
  - increased arm span; tall, thin habitus
  - pectus changes, scoliosis
  - flat feet
- **Heart**: dilated aortic root; mitral valve prolapse
- **Eye**: dislocated lens; myopia
- **Other**
  - stretch marks, recurrent herniae, spontaneous pneumothorax, high arched palate
- **Gene**: Fibrillin-1; its protein is crucial to function of the microfibrils of connective tissue.
- **Inheritance**: autosomal dominant

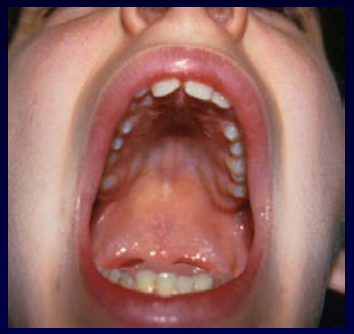
Features of Marfan Syndrome



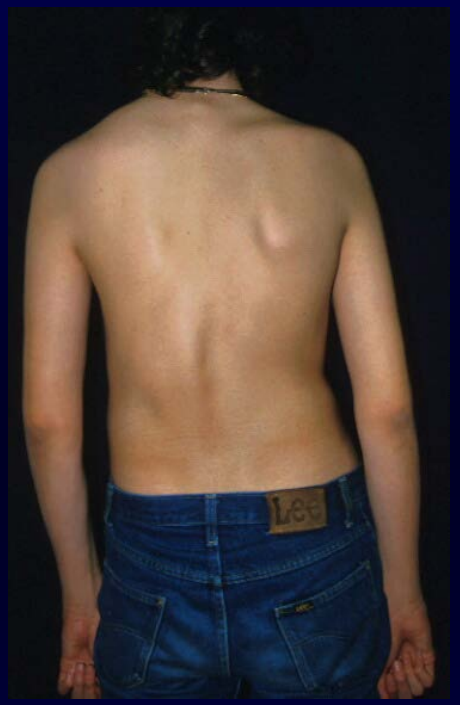
Arachnodactyly



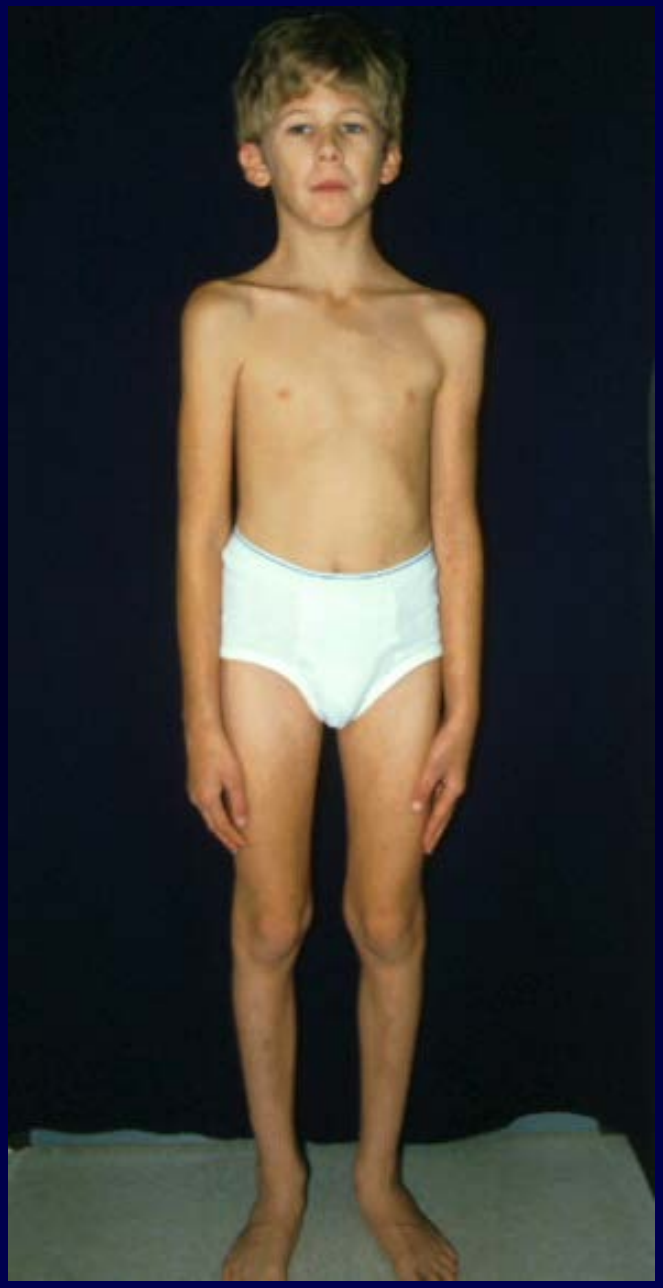
Pes planus



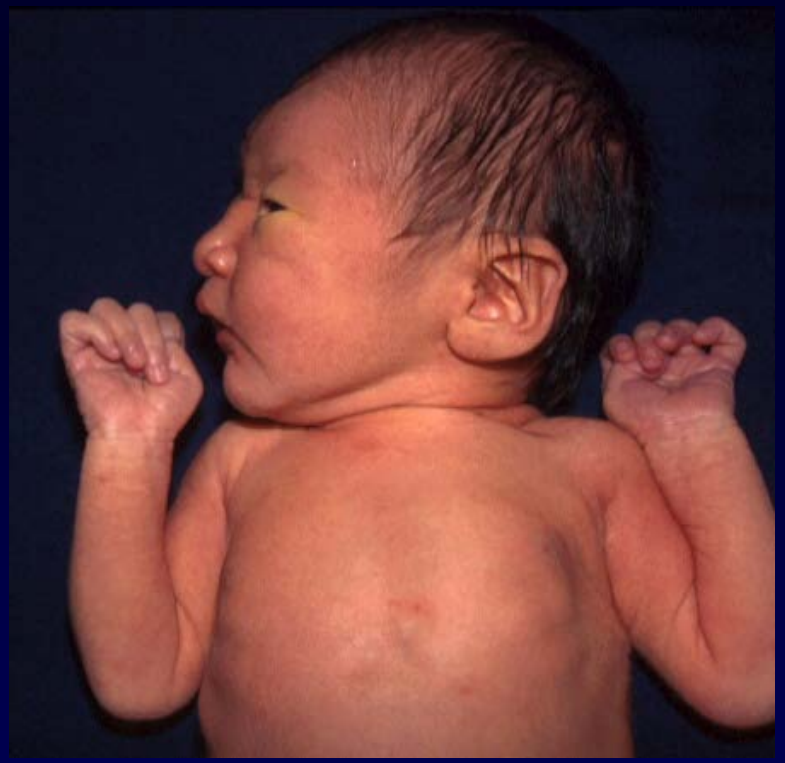
High arched palate



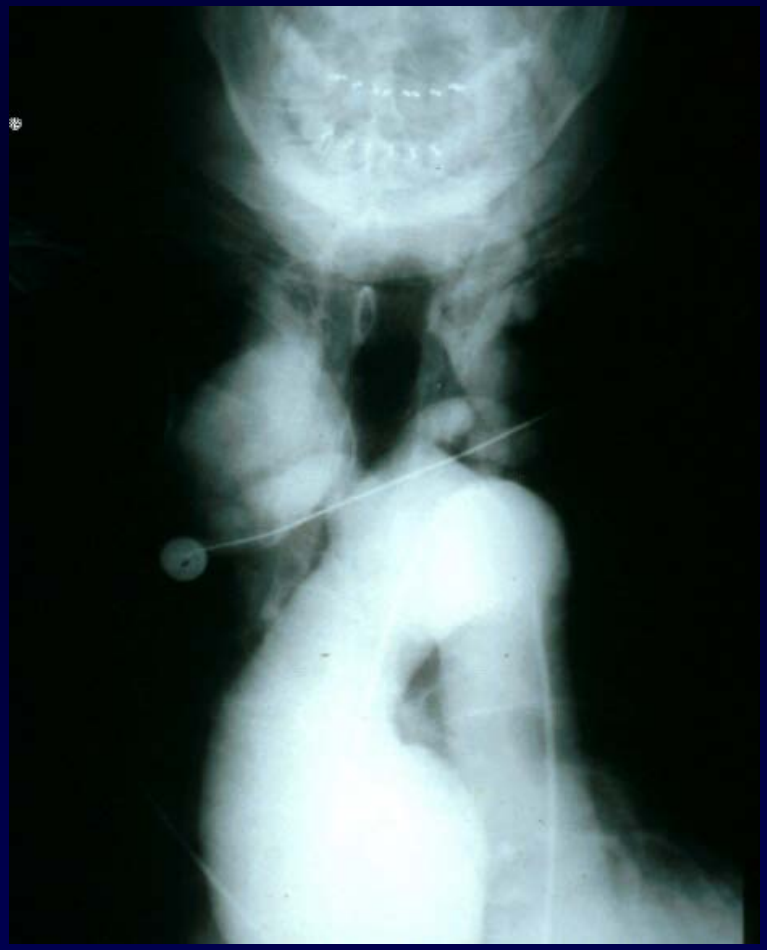
Scoliosis

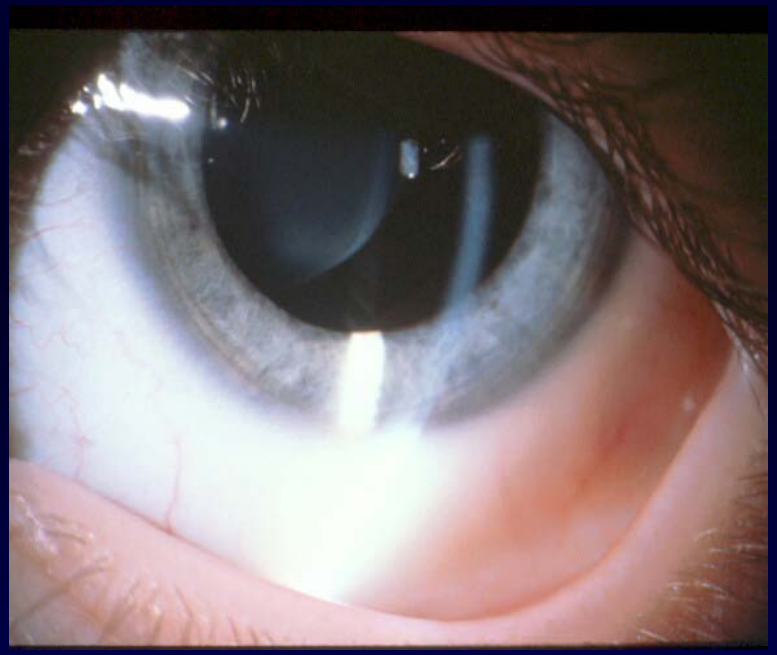


Relatively long fingers, arms and legs in older children. Overlapping fingers, long narrow feet in infants.



Marfan syndrome complications: aortic root dilation and lens dislocation





Other Complications

- Scoliosis
- Pectus Excavatum
- Mitral Valve Prolapse
- Abdominal Aortic Aneurysm
- Severe Myopia, Retinal Detachment
- Spontaneous pneumothorax

Differential Diagnosis

- Homocystinuria
- Congenital Contractural Arachnodactyly
- Familial Marfan-like Habitus
- MASS (myopia, mitral valve prolapse, mild aortic dilation, skin and skeletal)
- Familial mitral valve prolapse syndrome

Skeletal Criteria Used in Evaluating for the Marfanoid Habitus

Severe

- Pectus carinatum
- Pectus excavatum requiring surgery
- Reduced U/L ratio or AS to height ratio $>$ 1.05
- Wrist and thumb signs
- Scoliosis $>$ 20^{o} or spondylolisthesis
- Elbow extension ($<$ 170^{o})
- Pes planus
- Protrusio acetabulae (x-ray)

Mild

- Pectus excavatum of moderate
- Joint hypermobility
- High arched palate with dental crowding
- Facial appearance (dolichocephaly, malar hypoplasia, enophthalmos retrognathia, down slanting palpebral fissures)

Major involvement: 4 severe features
Minor involvement: 2 major and 2 minor

De Paepe et al., Am J Med Genet 62: 1996