

Marfan Syndrome

Eileen Wright

BioC 118Q

13 January 2011



6 ft 3.75 in

Elongated, gaunt face and neck

Pectus excavatum

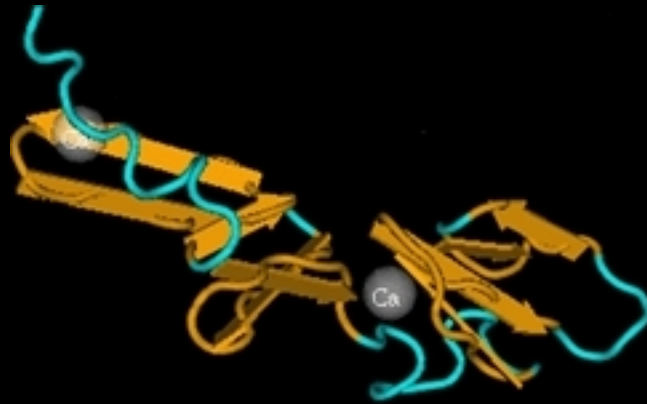
Disproportionate long arms

Long, thin fingers

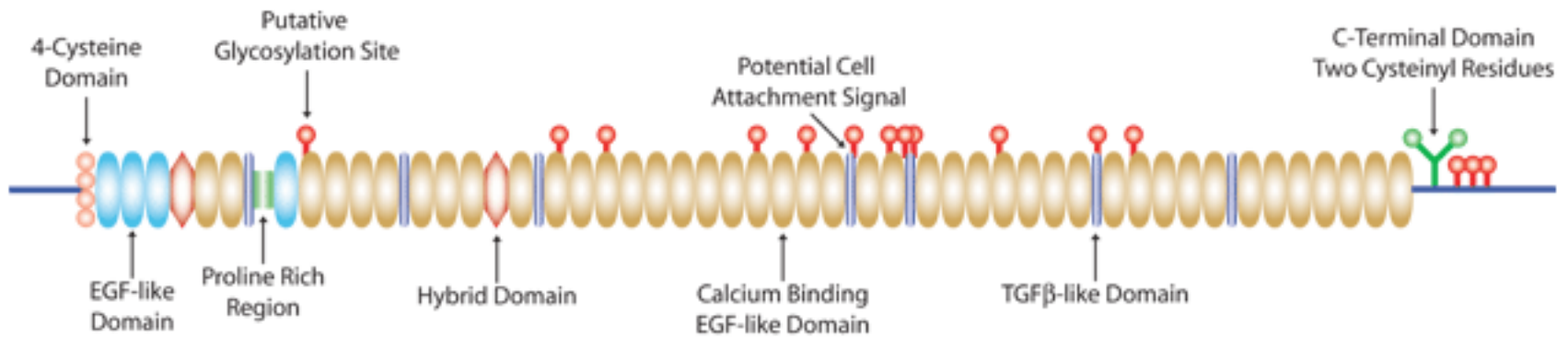
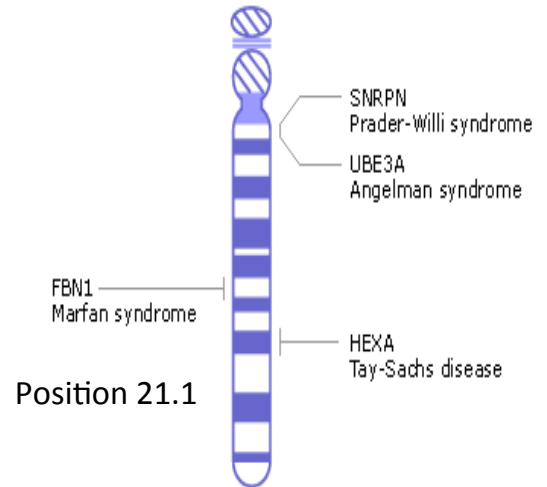
Disproportionate long legs & feet



Pathogenesis

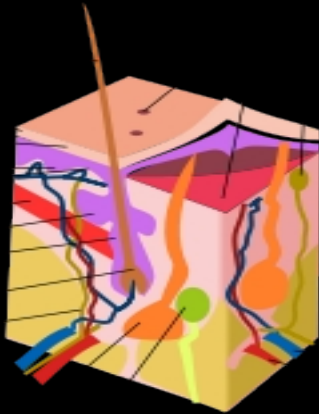


Chromosome 15

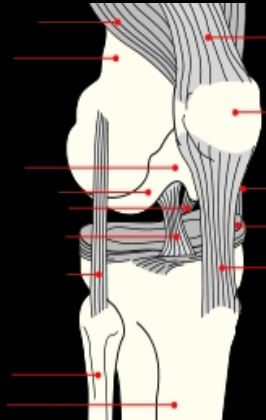


Fibrillin-1 Primary Structure

Elastic Fibers



Skin

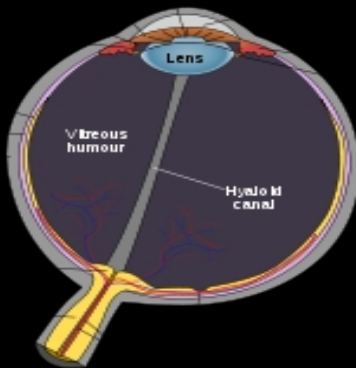


Ligaments

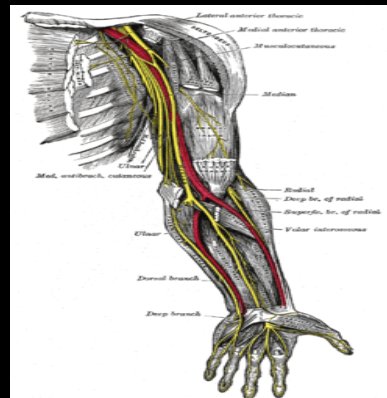


Blood Vessels

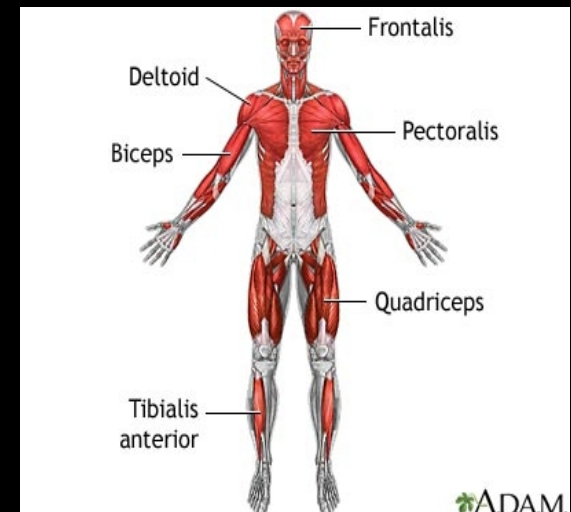
Rigid Tissue



Eyes



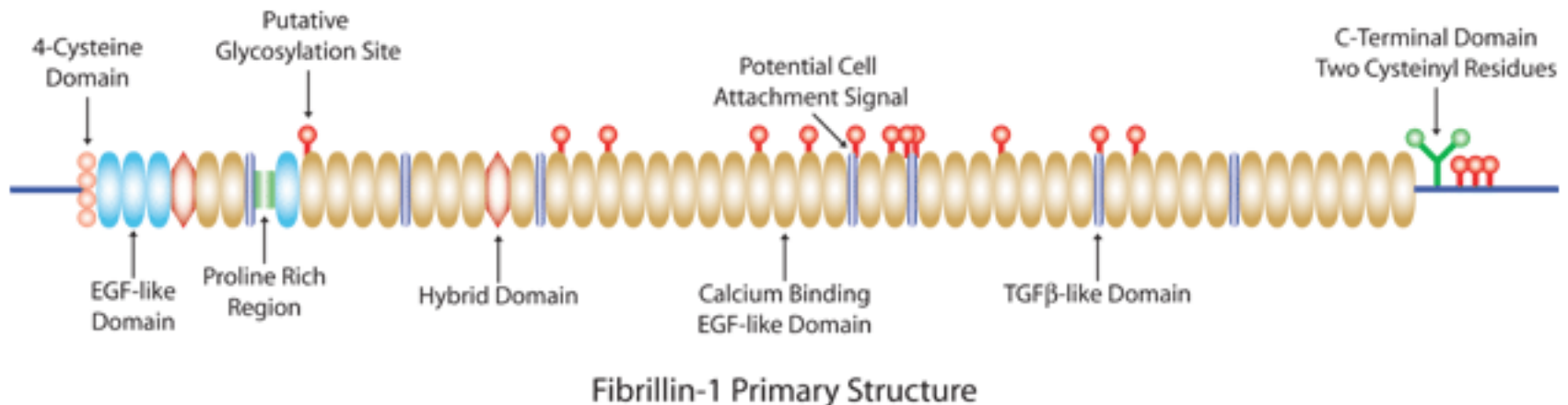
Nerves



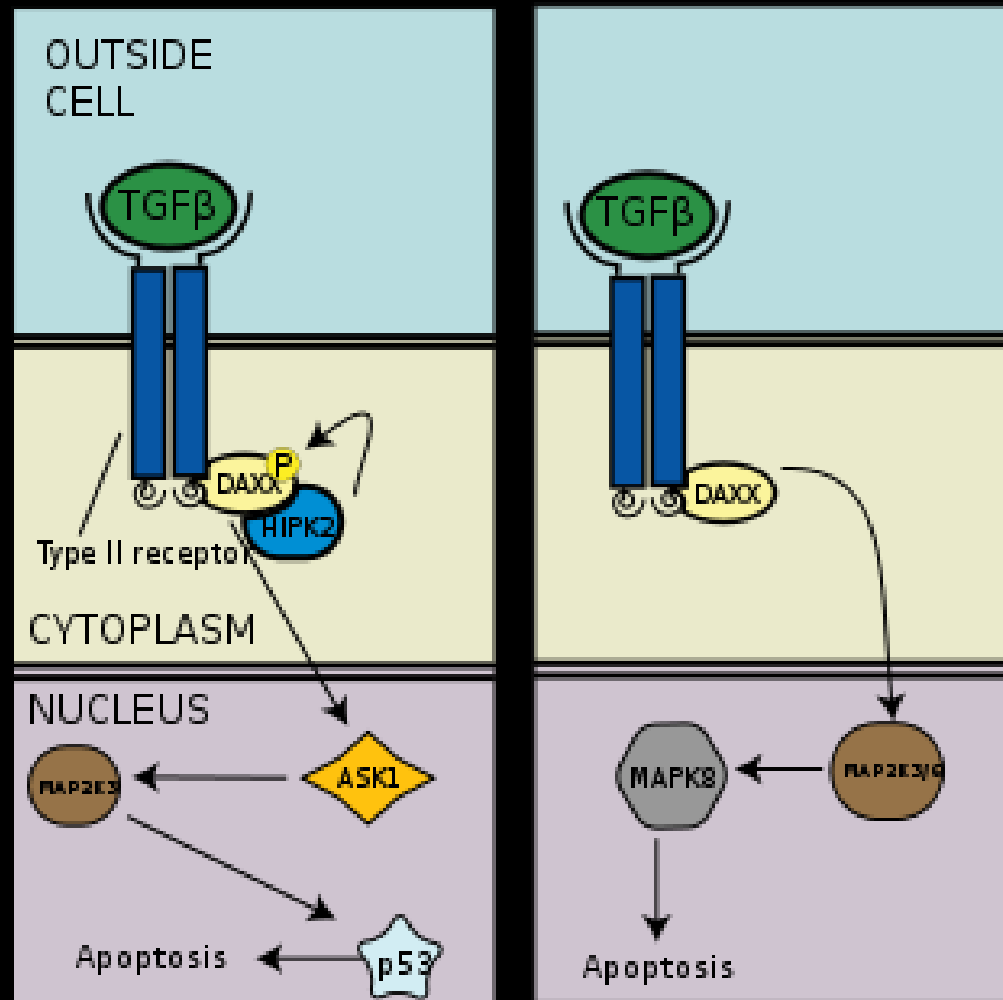
Muscles

FBN1 Mutations

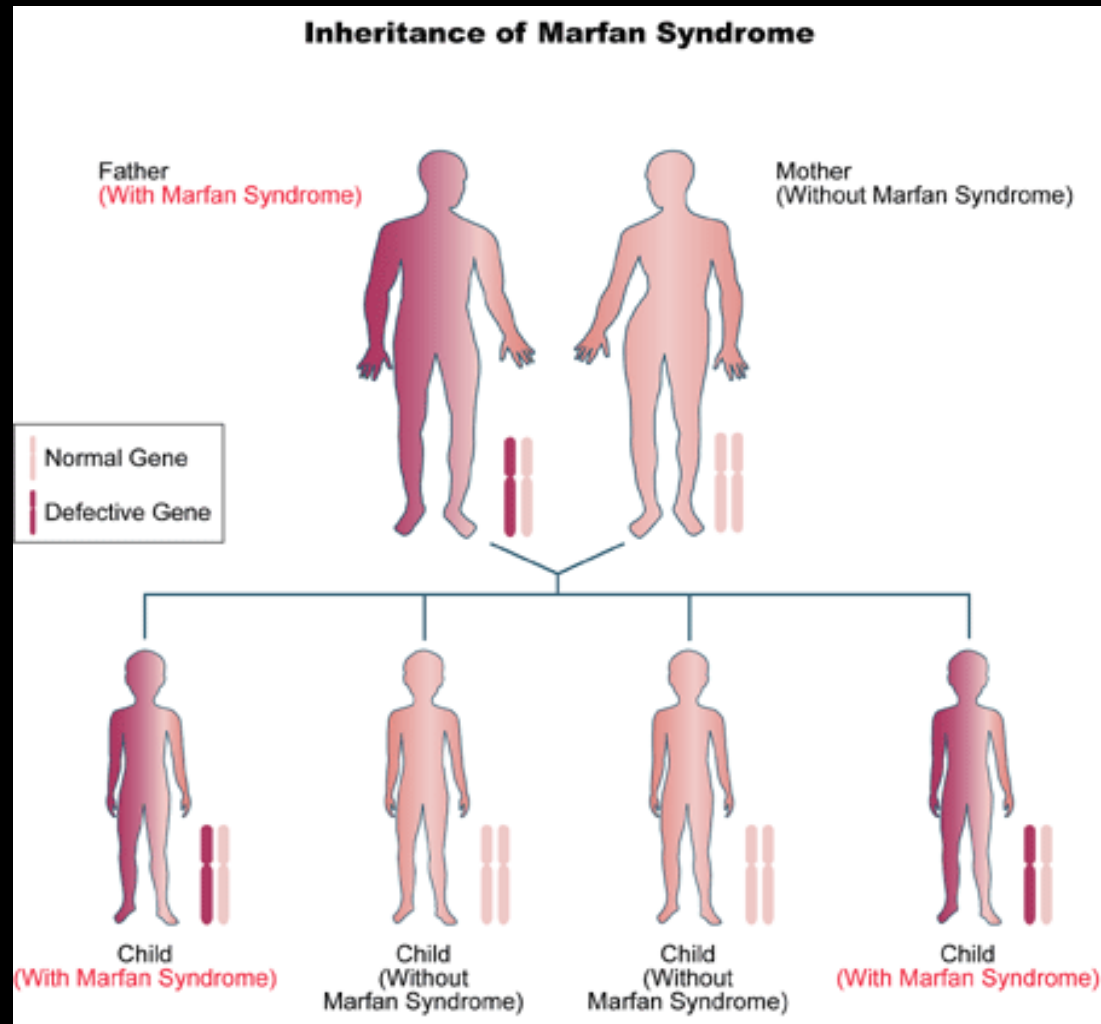
- 600 FBN1 Mutations
- 60% are amino acid changes
- 40% produce abnormally small Fibrillin-1
- All reduce, alter structure, or impair transport
- Result: Amount of Fibrillin-1 available to form microfibrils reduced → weakens elastic fibers and over activates TGF-beta growth factors.



Transforming Growth Factor Beta (TGF beta)

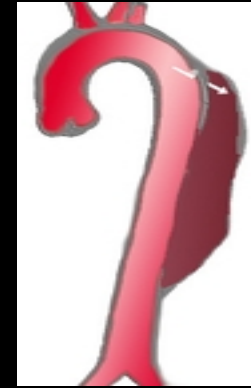


Autosomal Dominant Disorder

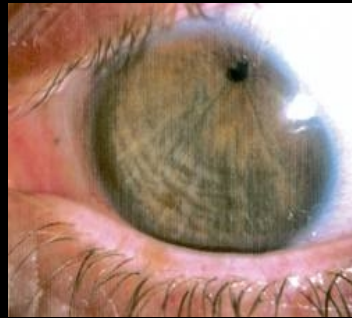


Diagnostic Methods

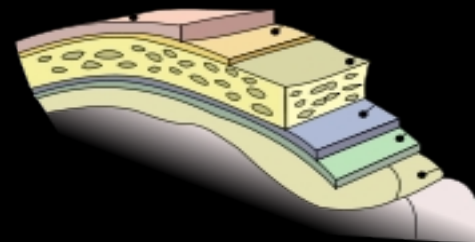
- Aortic dilation or dissection



- Ectopia lentis

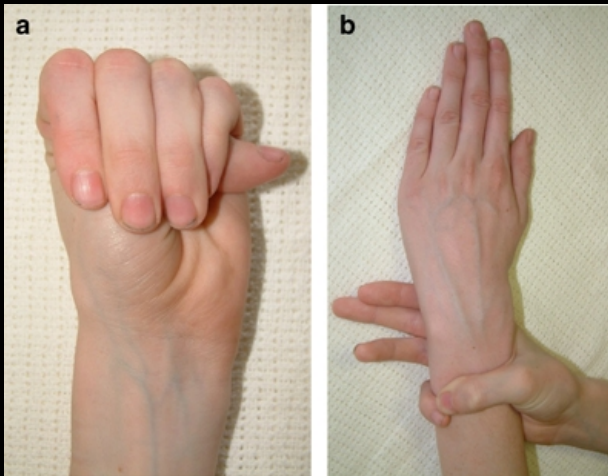
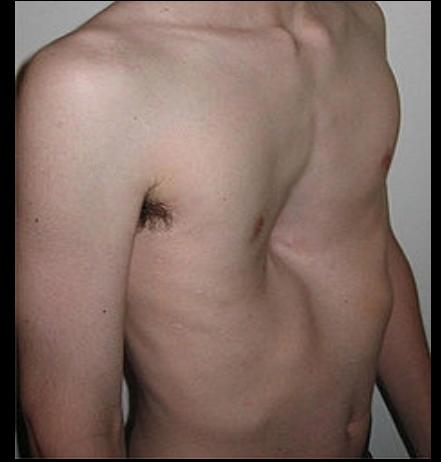


- Dural ectasia

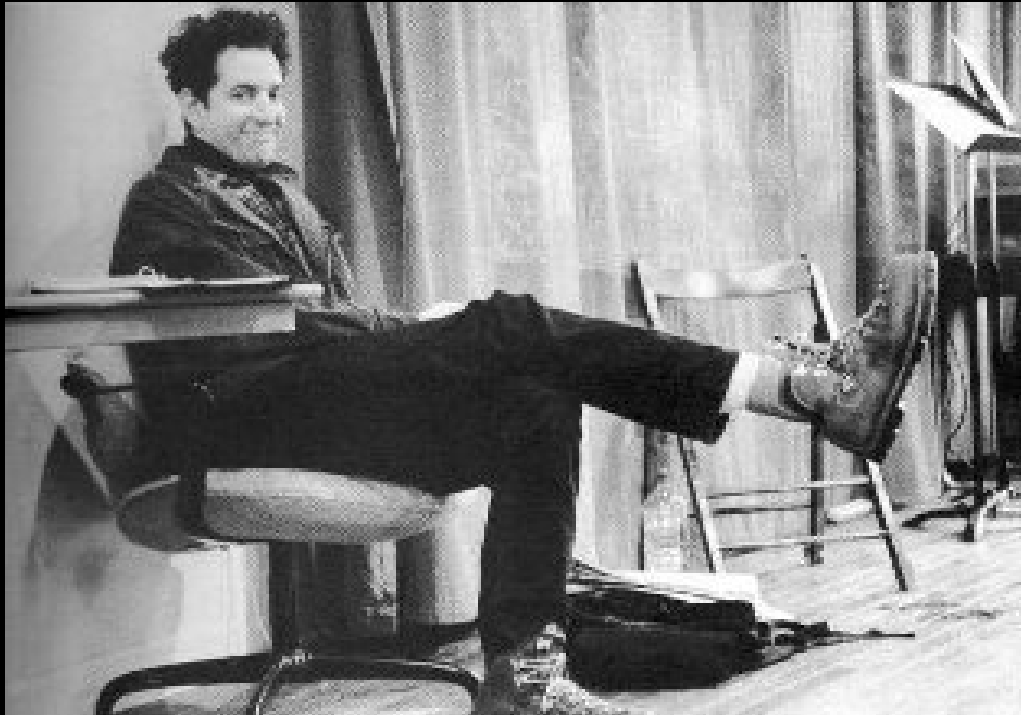


At least four skeletal features:

- Pectus carinatum or excavatum
- Joint Hypermobility
- Scoliosis
- Highly arched palate with tooth crowding
- Arm span to height ratio > 1.05
- Dolichocephaly

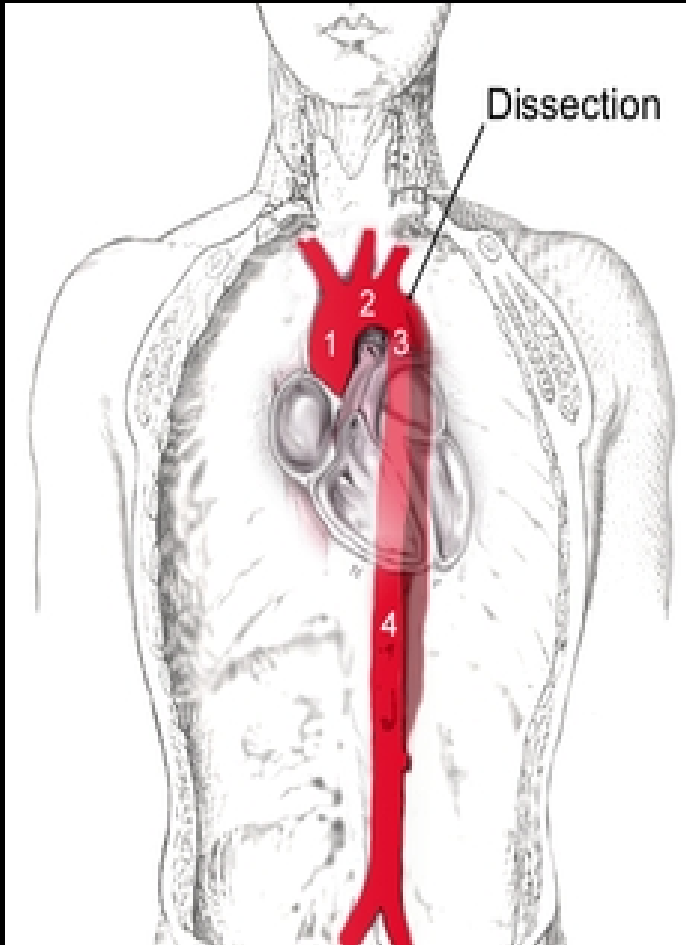


Rent playwright Jonathon Larson



“excruciating chest pain”

Aortic Dissection



Blood first penetrates the *intima* layer, which is composed of a special type of epithelial tissue.

It moves to the *tunica media*, which is made up of elastic fibers and muscle cells.

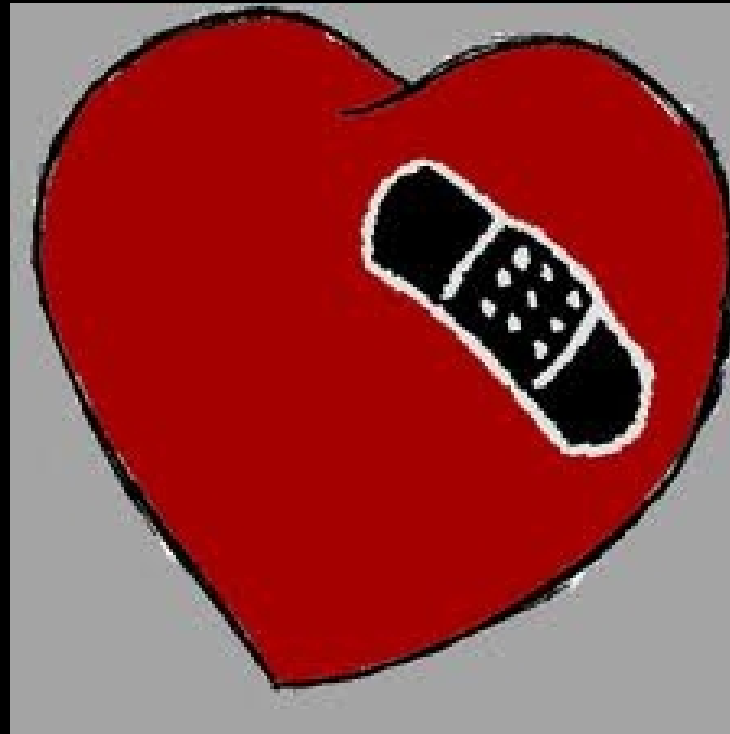
Finally, it penetrates the outermost layer, the *tunica adventitia*, which is composed of connective tissue.

Surgery necessary at 6 cm

Management

- ✓ Pectus surgery
- ✓ Medication for dural ectasia
- ✓ Eyeglasses
- ✓ No sports!
- ✓ Aortic repair
- ✓ Genetic Counseling & Prenatal Testing





90%



70s vs. 40s



DID YOU KNOW:

Jonathan Larson, Creator of RENT,
died suddenly of a misdiagnosed
aortic dissection, thought to have
been caused by Marfan syndrome.

His death could have been prevented.
Marfan syndrome: Learn about it.
Recognize it. Save a life.

