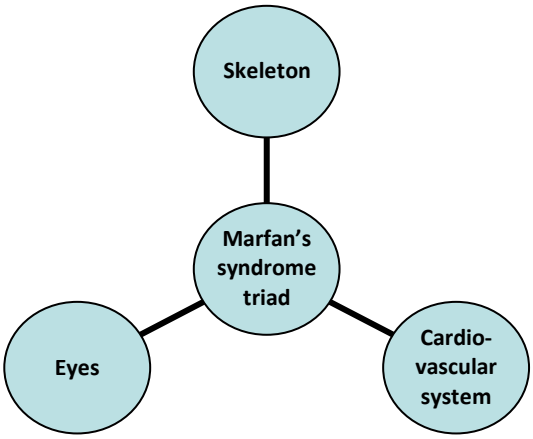
Marfan's Syndrome

Genetic disorder of connective tissue primarily affecting the skeleton, eyes and cardiovascular system.



Marfan's Examination Findings

- General inspection
  - Tall and thin
  - Scoliosis
- Hands and arms
  - Arachnodactyly (ask them to wrap their fingers around their wrist)
  - Wide arm span
  - Flexible joints
- Face
  - Long thin face
  - Myopia/ lens dislocation
  - High arched palate
- Chest and abdomen
  - Pectus carinatum/excavatum
  - Stretch marks
  - Auscultate heart for murmurs (especially aortic regurgitation)
- Legs and feet
  - Long legs
  - Flat feet
- To complete
  - Full cardiovascular exam

Cause

- Autosomal dominant FBN-1 gene mutation which transcribes Fibrillin-1, a major protein of elastin in connective tissues.

Clinical Features

- Skeleton
  - Generally
    - Tall and slim
    - Long limbs
    - Flexible joints
  - Chest
    - Pectus excavatum/carinatum
  - Back
    - Scoliosis
    - Spondylolisthesis
    - Dural ectasia (dura weak and expands outwards causing headache/backache)
- Eyes
  - Lens dislocation (50%)
  - Myopia
  - Others
    - Cataract
    - Retinal detachment
- Cardiovascular system
  - Weak thoracic aorta
    - Aneurysms
    - Dissection
  - Valves
    - Aortic regurgitation
    - Mitral/tricuspid prolapse

Investigations

- Genetic tests

- Eye exam
- Echocardiogram

Management

- No cure
- Symptomatic treatment
- Surveillance for complications
    - MDT regular check ups
    - Beta blockers (for weak aorta)