S-24 Imaging in the Diagnosis of Marfan Syndrome

Objective

➢How Marfan Syndrome is diagnosed
➢Imaging modalities used to diagnose Marfan Syndrome

Thesis Statement

➢Recognize and determine how imaging modalities diagnose Marfan Syndrome

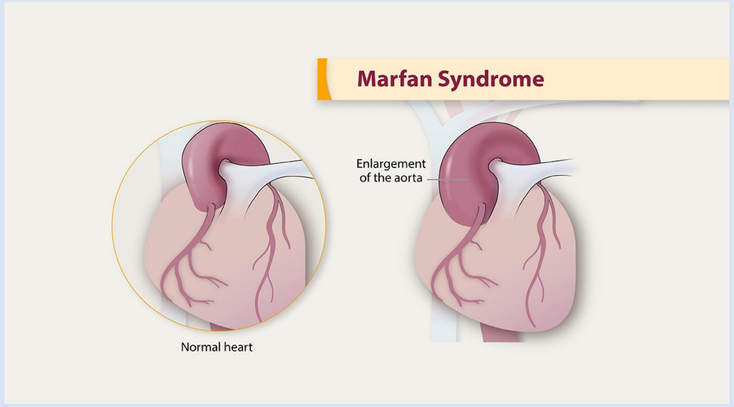
Defining Marfan Syndrome

➢The Center for Disease Control defines Marfan syndrome as "a genetic condition that affects connective tissue, which provides support for the body and organs" (CDC, 2024)
➢It is caused via a mutation of the gene referred to as FBN1
➢Marfan Syndrome happens to roughly 1 in 5,000 people making it a rare condition and can effect anyone regardless of gender, race or ethnic background

Manifestations of Marfan Syndrome

➢Symptoms of Marfan Syndrome range in severity as it can affect varying aspects of the human anatomy such as:
- **Cardiovascular**
  - Aortic Aneurysms
  - Aortic Dissections
  - Valve Malformations
- **Ocular**
  - Lens dislocation
  - Retinal Issues
  - Glaucoma/Cataracts
- **Skeletal**
  - Long arms, legs and fingers
  - Scoliosis/Kyphosis of the Spine
  - Sinking or Protruding Chest

➢"The most dangerous complications of Marfan Syndrome involve the heart and blood vessels." (Mayo Clinic 2023)



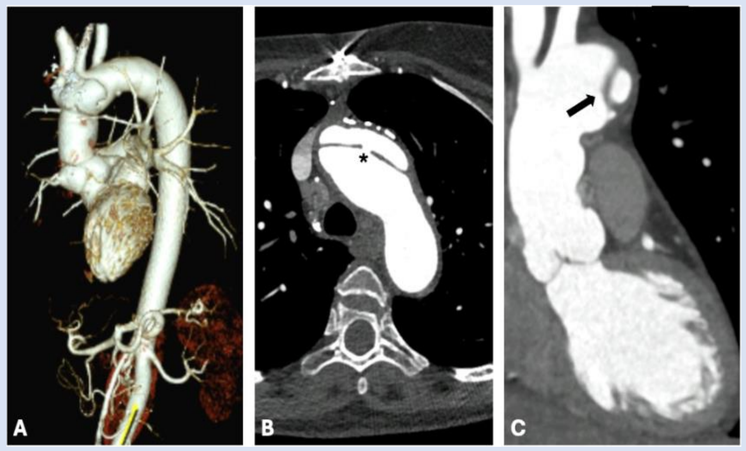
CDC (2024) Illustration
https://www.cdc.gov/heart-disease/about/marfan-syndrome.html
The above image illustrates how someone with Marfan Syndrome heart can be affected because of the disease.

How Marfan Syndrome is Diagnosed

➢Marfan Syndrome suspicion is generally deduced by presence of its physical markers (Shirley et al 2009)
- Tall stature and thin habitus
- Long slender limbs
- Ligamentous laxity

➢Echocardiograms
- Look for problems with the aorta and heart valves (CDC 2024)

➢Genetic Testing



Perrone et al (2025) Figure 5. [Photograph] Diagnostics (Basel) https://research-ebsco-com.ezproxy.lib.uwm.edu/c/vkjmfn/viewer/pdf/ejpohxuxkr?route=details
The images above are of a 34 year old woman presenting with Marfan Syndrome that needed an emergency CT on suspicion of an Aortic Dissection. Panels B & C demonstrate the breach and false lumen while Panel A demonstrates the 3-D reconstruction.

Imaging Modalities and their uses in Care of MFS

➢Multiple Modalities are used in the care of Marfan Syndrome
- **Echocardiography**
  - Screens for Mitral Valve Prolapse and Mitral Annular Disjunction (MAD) (Perrone et al, 2025)
  - Evaluation of dimensions of arteries and aortas (Perrone et al, 2025)
- **Computed Tomography**
  - Detailed evaluation of Aorta in emergent situations (Perrone et al, 2025)
  - Administration of contrast specific to constraints and needs of patient (Perrone et al, 2025)
  - Utilize 3-Dimensional Reconstruction for pursuant monitoring and surgical evaluation (Perrone et al, 2025)
- **Nuclear Medicine**
  - Pertinent tracers alongside myocardial SPECT to evaluate for cardiac pump function, perfusion and viability (Perrone et al, 2025)
  - PET/CT for inflammation of the wall of the aorta and other complications such as infection of grafts and chronic peri-aortitis (Perrone et al, 2025)
- **Cardiovascular Magnetic Resonance**
  - Evaluation of the aorta in its entirety to monitor for aneurysms, dissections, and hemodynamic properties (Perrone et al, 2025)
  - Incorporated into routine monitoring to detect progression of MFS
  - Evaluates the impact of skeletal deformities on cardiac function and detection of early myocardial dysfunction (Perrone et al, 2025)

Conclusion

➢Imaging in the Diagnosis of Marfan Syndrome is paramount to managing and evaluating the progress in each individual patient
➢Imaging is one of the most powerful tools in managing and assessing the advancement and severity of the disease that is Marfan Syndrome