MARFAN SYNDROME: WHAT EVERY HEALTHCARE PROVIDER NEEDS TO KNOW
Objectives

 Increase awareness of clinical presentations of Marfan syndrome
 Understand the diagnosis of Marfan syndrome based on clinical findings
 Initiate the work-up of Marfan syndrome
Case 1

- 14 y/o previously healthy male presents with sudden onset shortness of breathing
- Spontaneous Pneumothorax
- Chest tube inserted
- Labs WNL.
On Examination:

- General: tall, thin male with long fingers
- Craniofacial: facial features of MFS
- Ocular signs: myopia
- Vascular/Cardiac: Thoracic: PTX
- Musculoskeletal/Neurological:
  - Wrist and thumb sign
  - Pectus excavatum
  - Scoliosis
  - Reduced upper/lower segment ratio
  - Increased arm span
Hospital Course

- Pt. underwent VATS x 2, with thoracostomy tube
- After resolution of symptoms, pt. discharged home with f/u to PCP and Pediatric Surgery Clinic
- 2 variants in FBN 1 gene
  - FBN1 Exon 32 c.3890A>G; heterozygous
  - FBN1 Exon 32 c.3896C>T
Case 2:

Tall, thin, previously healthy teenage male with recurrent wound dehiscence x 3 after abdominal surgery

- General: tall, thin male with long fingers
- Craniofacial: long face, retrognathia, crowded teeth, high arched palate
- Ocular signs: myopia
- Vascular/Cardiac: Thoracic: no heart murmur

Musculoskeletal/Neurological:
- Wrist sign
- Scoliosis & Pectus excavatum
- Reduced upper/lower segment ratio
- Increased arm span

Dx: Marfan Syndrome based on clinical features and FBN1 gene testing
Case 3:

14 y/o previously healthy male with abdominal pain x 2 days, diagnosed with acute perforated appendicitis

- General: tall, thin male with long fingers
- Craniofacial: long face, retrognathia, crowded teeth, high arched palate
- Ocular signs: myopia
- Vascular/Cardiac: Thoracic: no heart murmur

Musculoskeletal/Neurological:
- Wrist sign
- Scoliosis & Pectus excavatum
- Reduced upper/lower segment ratio
- Increased arm span
Case 3: Hospital Course

- Prolonged in-hospital course due to poor wound healing
- ECHO: WNL
- FBN 1 gene testing: negative, however, other variants of Marfan syndrome exist for which we do not yet have genetic tests
What Are the Signs and Symptoms of Marfan Syndrome?

- Marfan syndrome often affects the long bones of the body. Can lead to:
  - A tall, thin build.
  - Long arms, legs, fingers, and toes and flexible joints.
  - Scoliosis.
  - Pectus excavatum and pectus carinatum.
  - Teeth that are too crowded.
  - Flat feet.
  - Stretch marks on the skin also are a common trait, usually appear on the lower back, buttocks, shoulders, breasts, thighs, and abdomen.

- Some of these traits also are signs of other connective tissue disorders.

Differential Diagnosis
- Congenital contractural arachnodactyly
- Ehlers–Danlos syndrome
- Homocystinuria
- Loeys–Dietz syndrome
- MASS phenotype
- Shprintzen-Goldberg syndrome
- Stickler syndrome
- Multiple endocrine neoplasia, type 2B
Complications of Marfan Syndrome

Heart and Blood Vessel Complications

- Aortic dilation, aortic aneurysm, aortic dissection
- Aortic dissection can cause severe pain in either the front or back of the chest or abdomen.
- Marfan syndrome also can cause mitral valve prolapse.
- MVP can cause shortness of breath, palpitations, chest pain.

Eye Complications

- Dislocated lens in one or both of the eyes. A dislocated lens often is the first sign that someone has Marfan syndrome.
- Other eye complications include nearsightedness, early glaucoma, and early cataracts. A detached retina also can occur.
Complications of Marfan Syndrome

Nervous System Complications

- **Dural ectasia** can occur in people who have Marfan syndrome as they grow older. Eventually, the bones of the spine may wear away.

- Symptoms of this condition are lower back pain, abdominal pain, headache, and numbness in the legs.

Lung Complications

- Marfan syndrome can cause sudden **pneumothorax**
- Scoliosis and pectus excavatum can prevent the lungs from expanding fully.
- Marfan syndrome also can cause changes in the lung tissue, and it can lead to early **emphysema**.
- Marfan syndrome also has been linked to **sleep apnea**.
Diagnosis

- Marfan syndrome is caused by a defect in the fibrillin 1 gene on chromosome 15.
- There is no simple blood test that can conclusively diagnose Marfan syndrome.
- Therefore, diagnosis is made through a clinical evaluation.
The diagnostic criteria for Marfan syndrome have been revised by medical specialists. The new publication explaining the 2010 Revised Diagnostic Criteria is now available at:

http://jmg.bmj.com/content47/7/476.abstract.
Marfan syndrome features are most often found in the heart, blood vessels, bones, joints, and eyes. Sometimes the lungs and skin are also affected.

Marfan syndrome does not affect intelligence.

With timely diagnosis, appropriate specialist care for eyes, teeth, heart, & lifestyle modifications

~1 in 5,000 people have Marfan syndrome. Includes men and women of all races and ethnic groups; we encounter this disease and must keep it on our differential