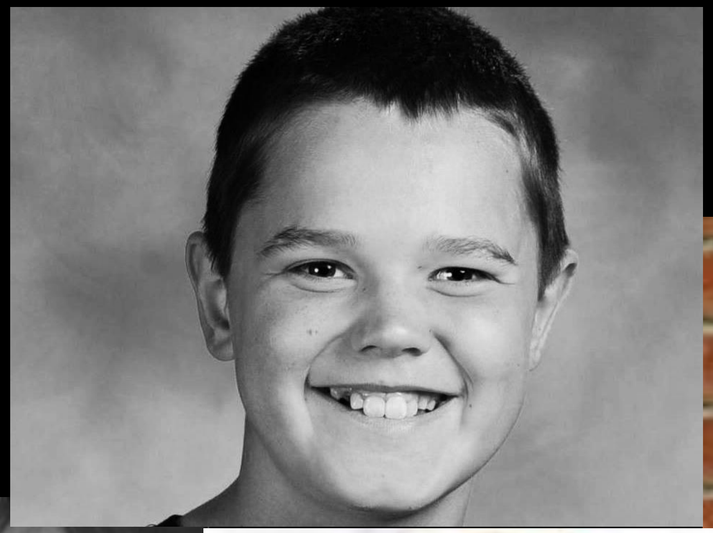
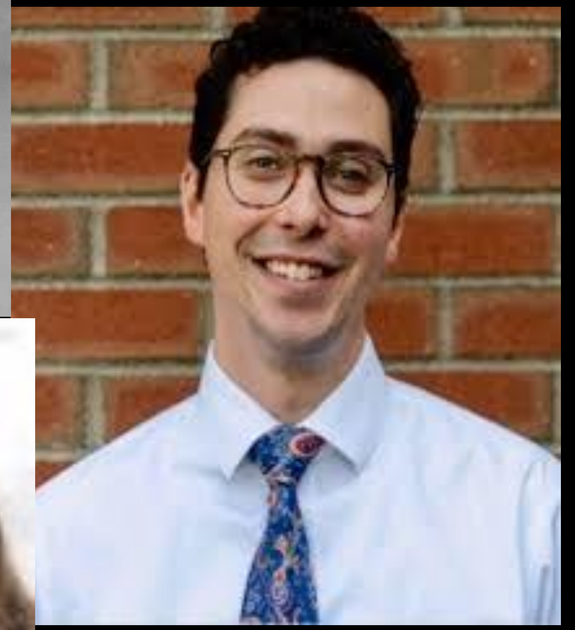
“When you hear hoofs, think horse, not zebra.”

Approximately 6,000 Rare Diseases

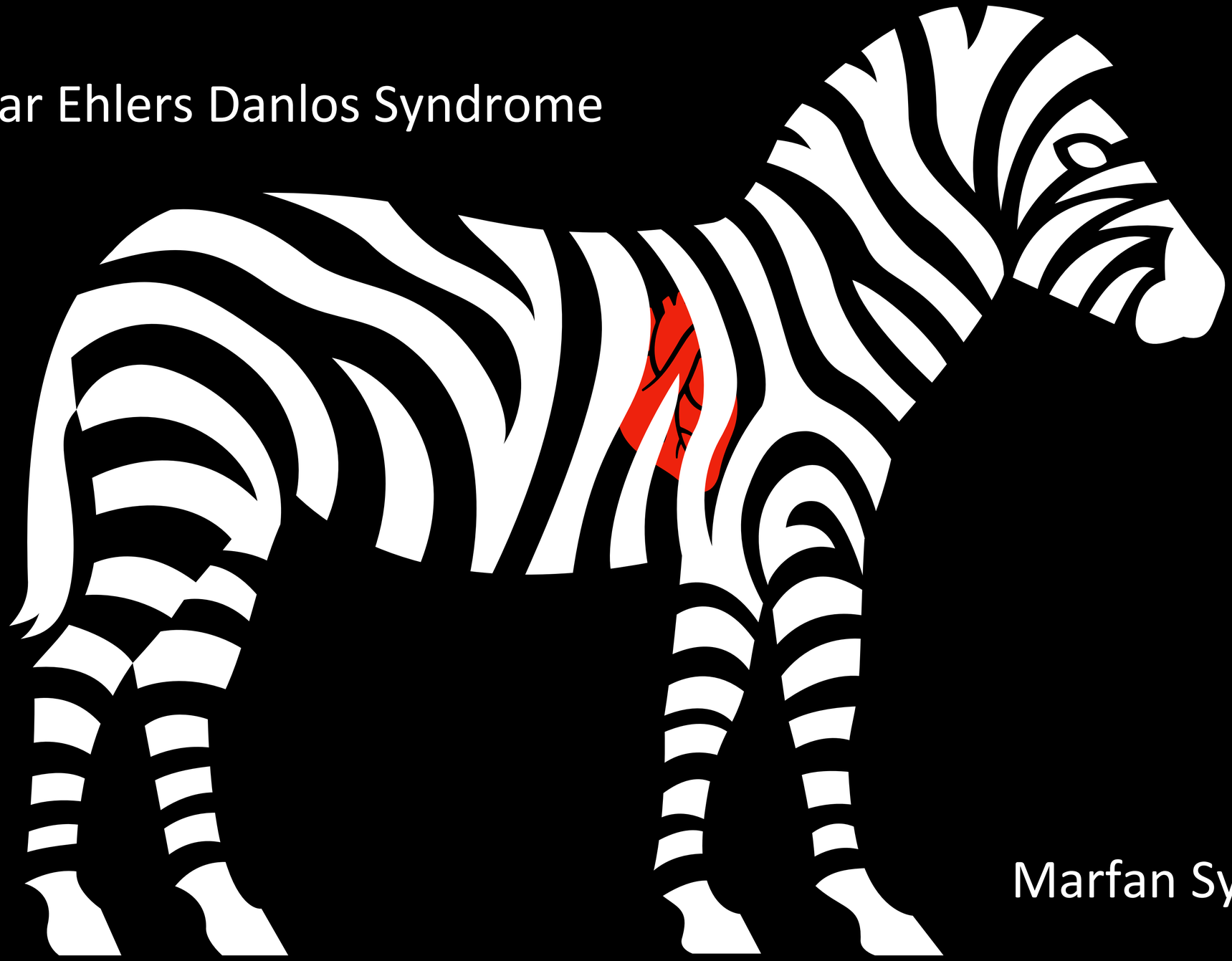
impact

Approximately 300 Million People Worldwide



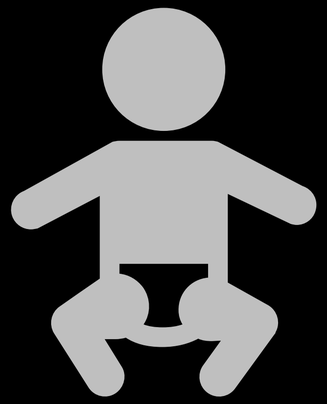


Vascular Ehlers Danlos Syndrome



Marfan Syndrome

Possible Signs at Birth



congenital clubfoot

hip dislocation

Signs May Not Come Until Puberty

easy or unexplained bruising

some develop features after puberty

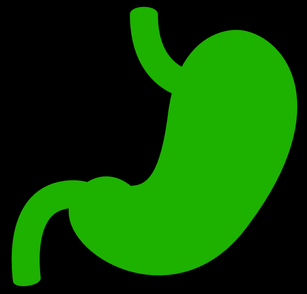
usually only tested if known family Hx

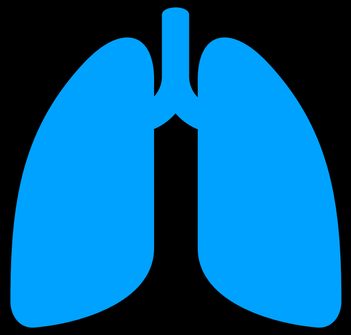
major complications rare, death <10 less common

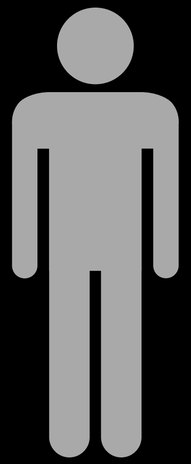
What is Vascular Ehlers Danlos Syndrome (VEDS)?

Genetic disorder that affects the body's connective tissue.

Connective tissue is made up of proteins and the protein that plays a role in VEDS, is called collagen III. VEDS is caused by a mutation of the COL3A1 gene, which is the gene that tells the body how to make part pf type III collagen.

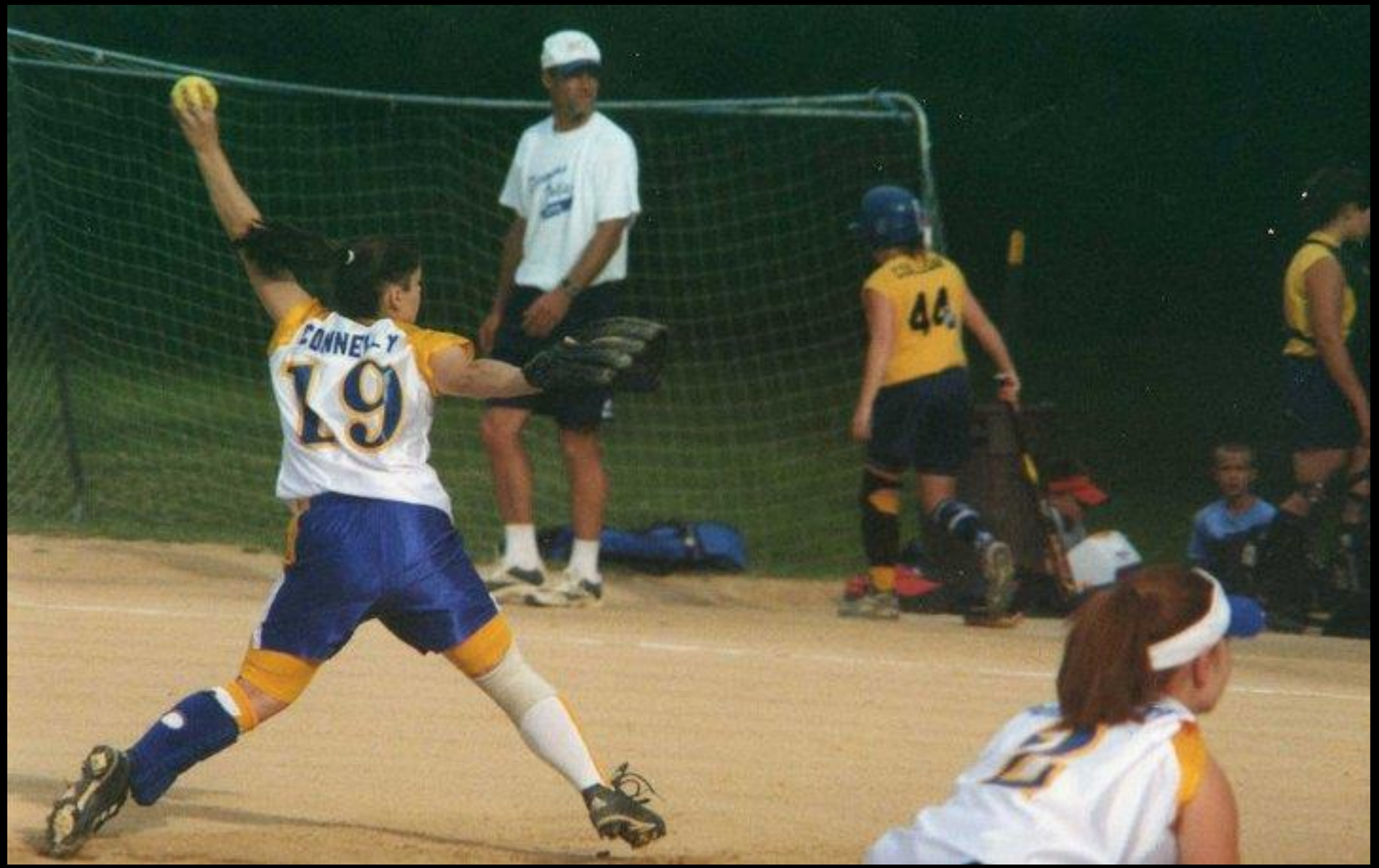






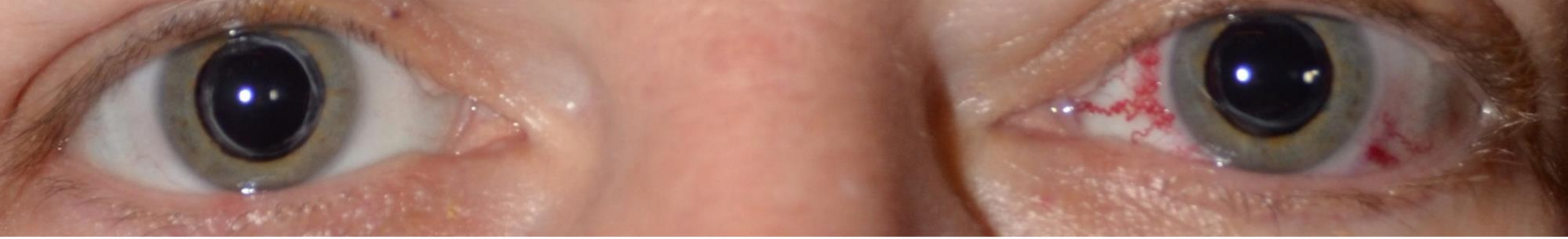
Approximately 8,000

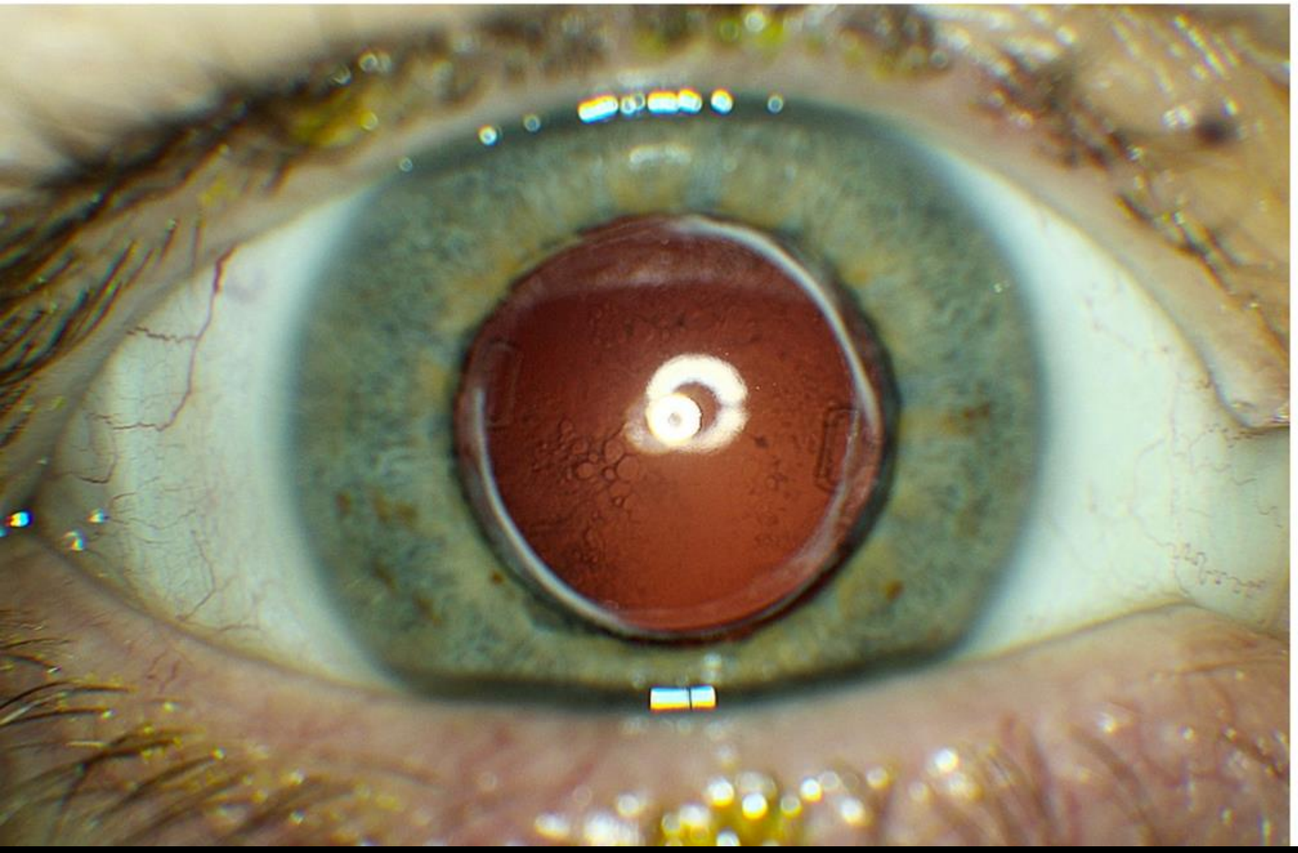
1: 40,000

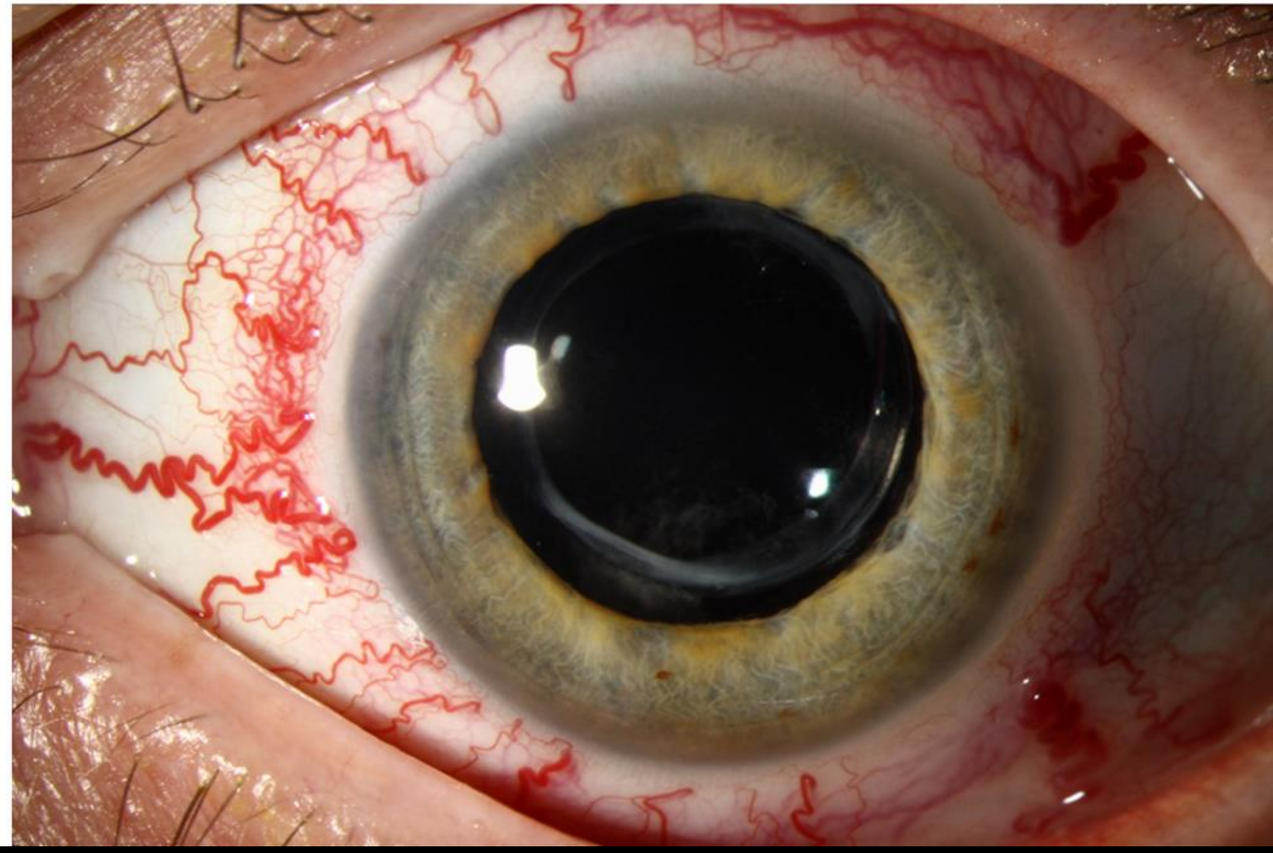

CONNELLY
19
44

1/4 have had a significant complication, such as bowel rupture, arterial rupture, or spontaneous pneumothorax.

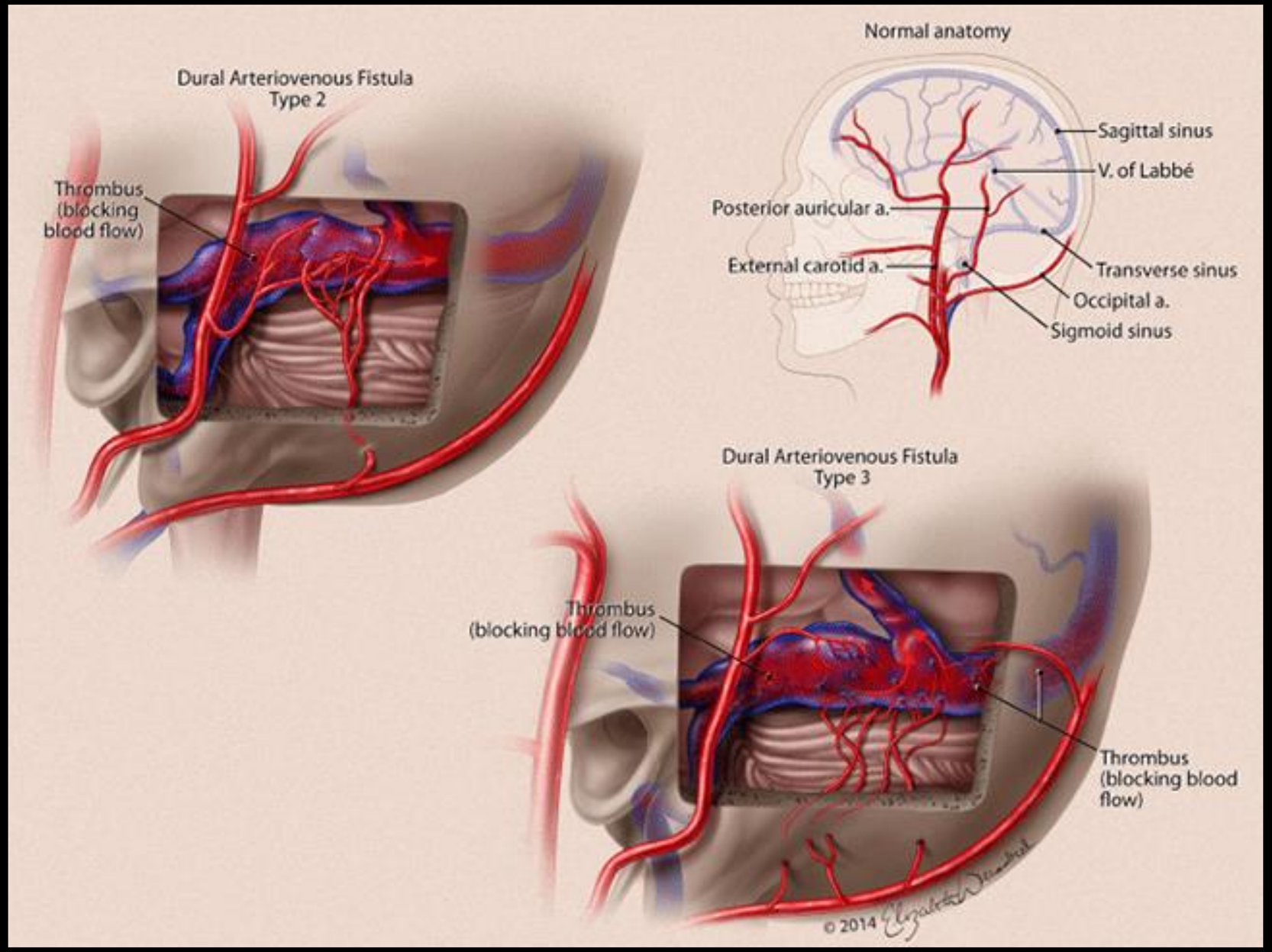
Some have developed a prematurely aged appearance or have thin translucent skin. Even with such features, many do not get diagnosed due to clinicians not knowing about VEDS.







Carotid Cavernous Fistula



Blurry Vision

Headache

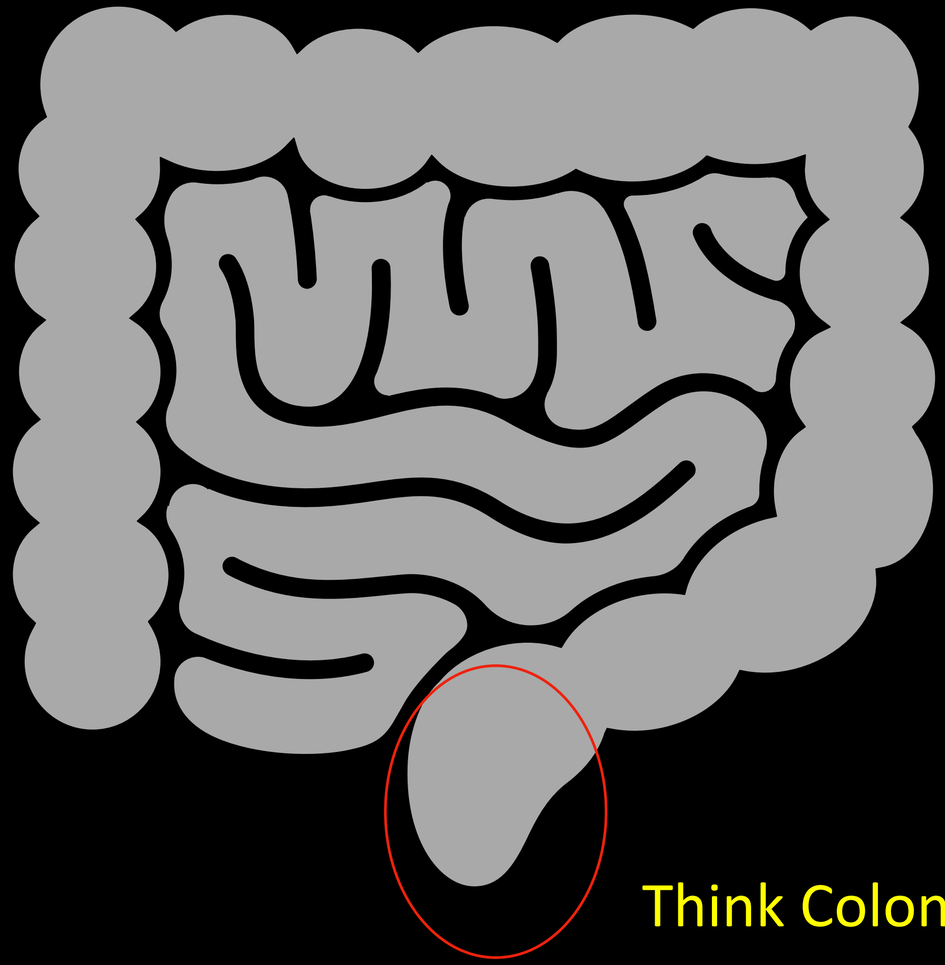
Pulsatile Tinnitus

Bloodshot Eye

In Absence of Trauma?

Bowel Rupture/Perforation

Nausea

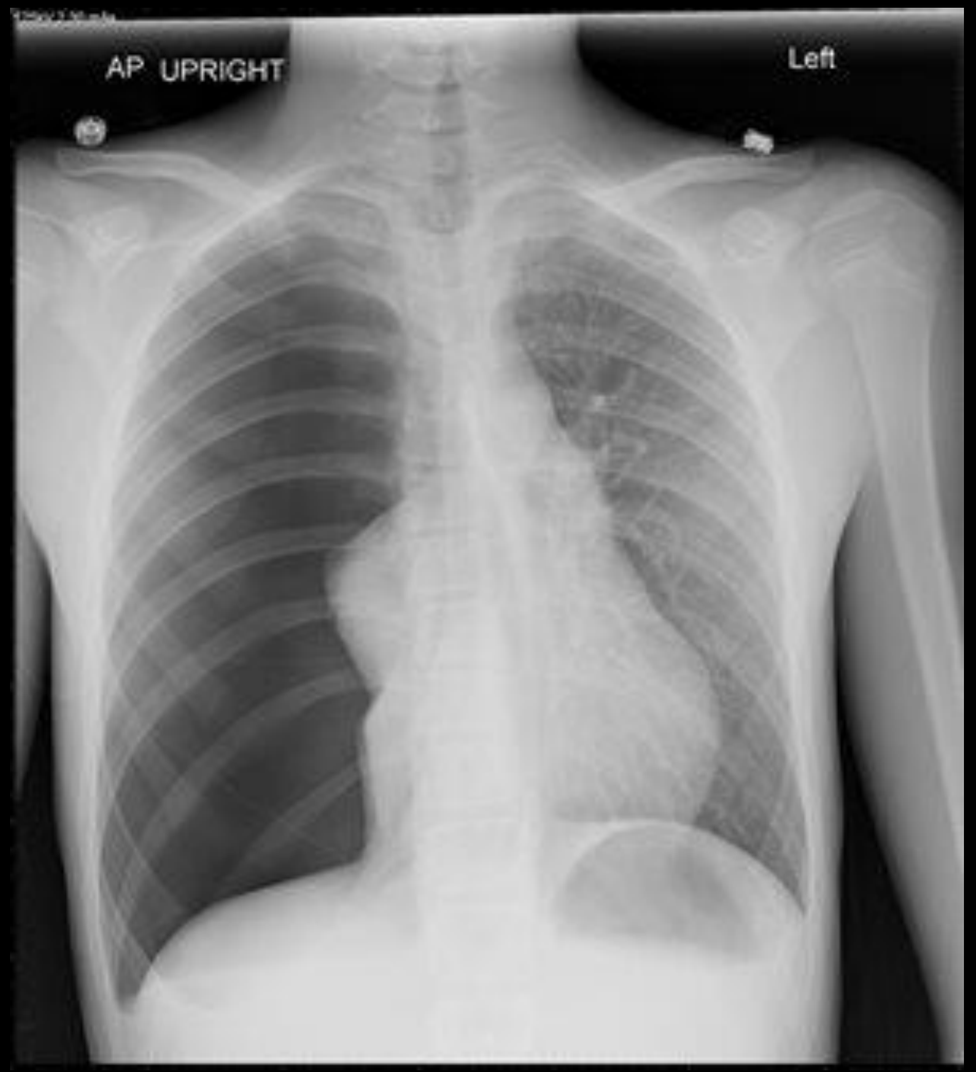


Abdominal Pain

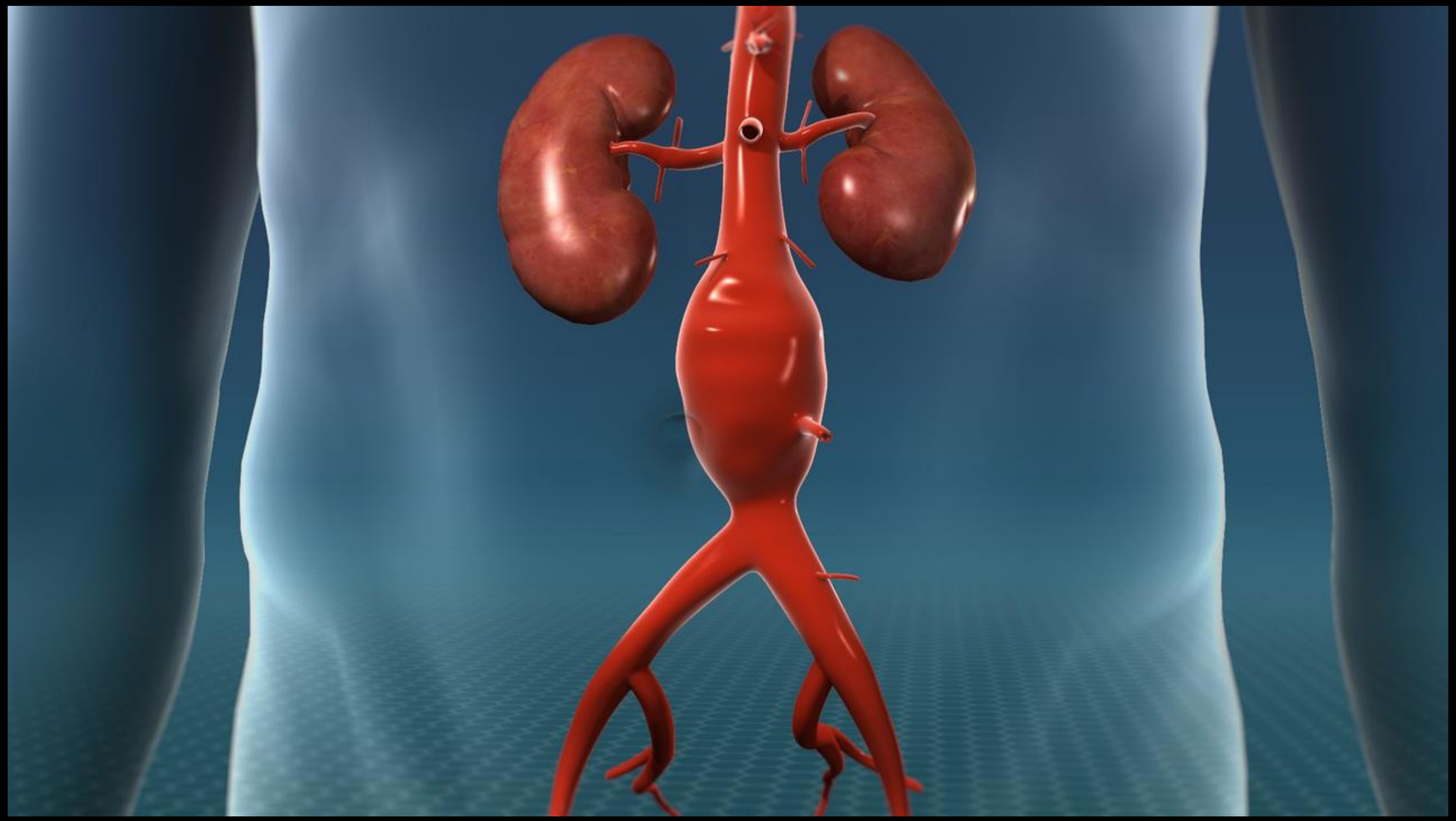
Vomiting

Think Colon

Spontaneous Pneumothorax



Aneurysms, Dissections, Ruptures



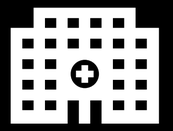
General Assessment, Transport and Treatment Considerations

Zebras have hoofs too. 

Check for Medical ID/Hx. 

Listen to the caregiver/patient. 

Think in terms of Trauma. 

Appropriate Facility - Think Vascular and Trauma Surgery. 

Long transports = 

Likely on a Beta Blocker. 

Considerations Cont.

IV Cannulation (fragile, fragile, fragile) and fluids - May get ultrasound guided central line later.

POCUS (future?)

ET Intubation/Airway Placement (fragile)

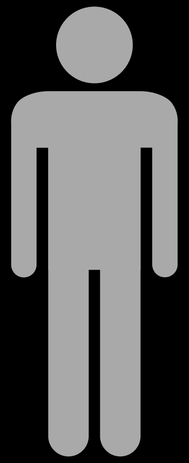
Less is More


TYPE 1
DIABETES

Marfan Syndrome

Marfan syndrome is a genetic disorder that affects the body's connective tissue.

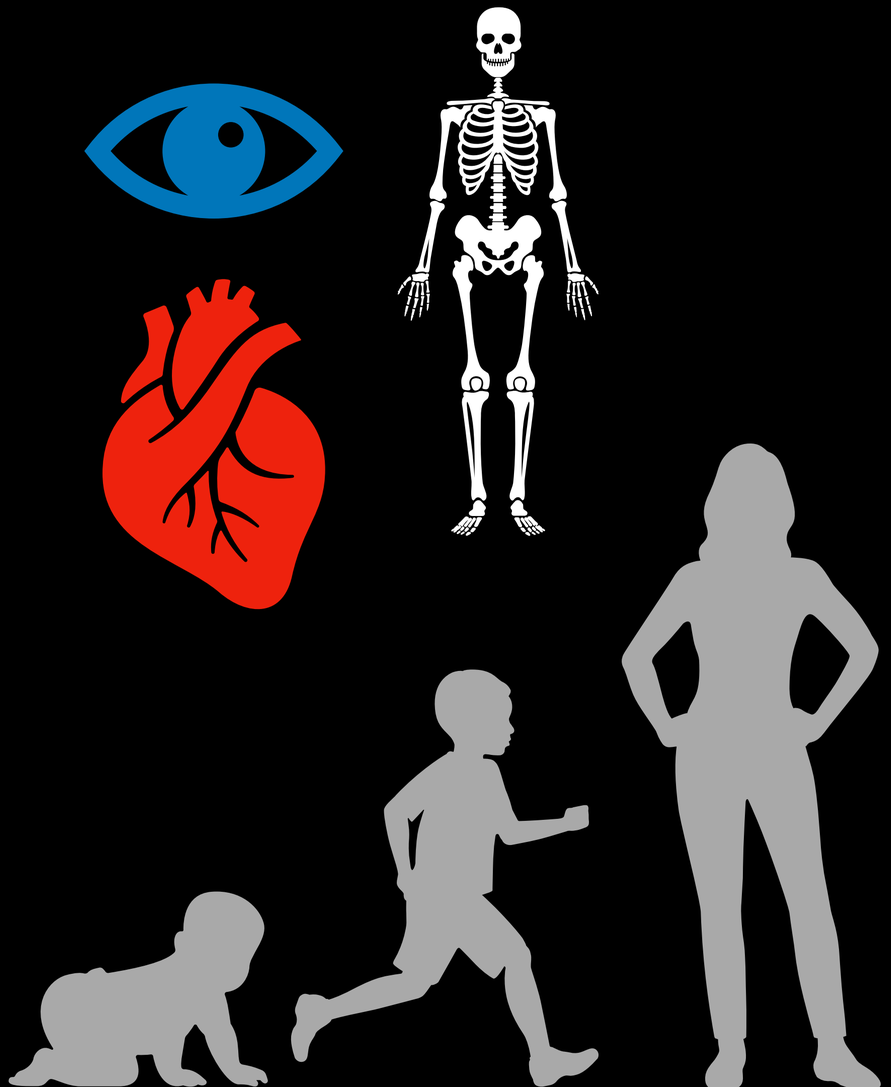
The protein that plays a role in Marfan syndrome is called fibrillin-1. Marfan syndrome is caused by a defect (or mutation) in the gene that tells the body how to make fibrillin-1. This mutation results in an increase in a protein called transforming growth factor beta, or TGF-β. The increase in TGF-β causes problems in connective tissues throughout the body, which in turn creates the features and medical problems associated with Marfan syndrome and some related conditions.



1: 5,000

3/4 of Marfan Patient Inherited the disease, while the other 1/4 are the first in their family to have it, resulting from a spontaneous mutation. Those with Marfan have a 50% chance of it to their children.

Marfan Syndrome is not always obvious. It may present itself at birth, during childhood or adulthood.



heart murmurs

aorta enlargement/aneurysm

spontaneous pneumothorax

eye problems (detatched retina and early glaucoma)

chest wall sticks out

tall and slender physique

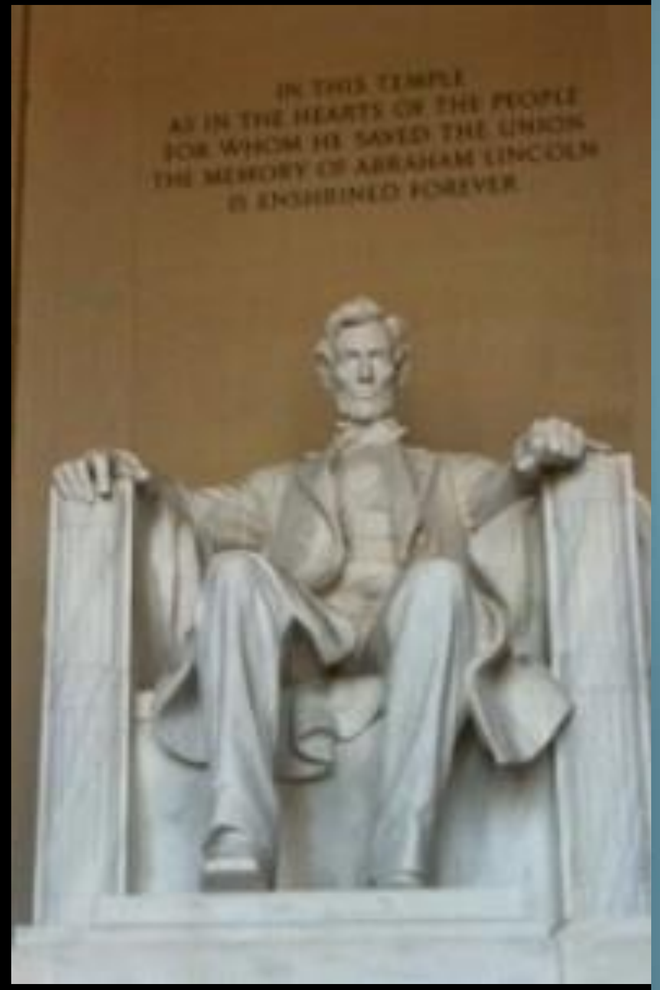
long arms, legs and fingers

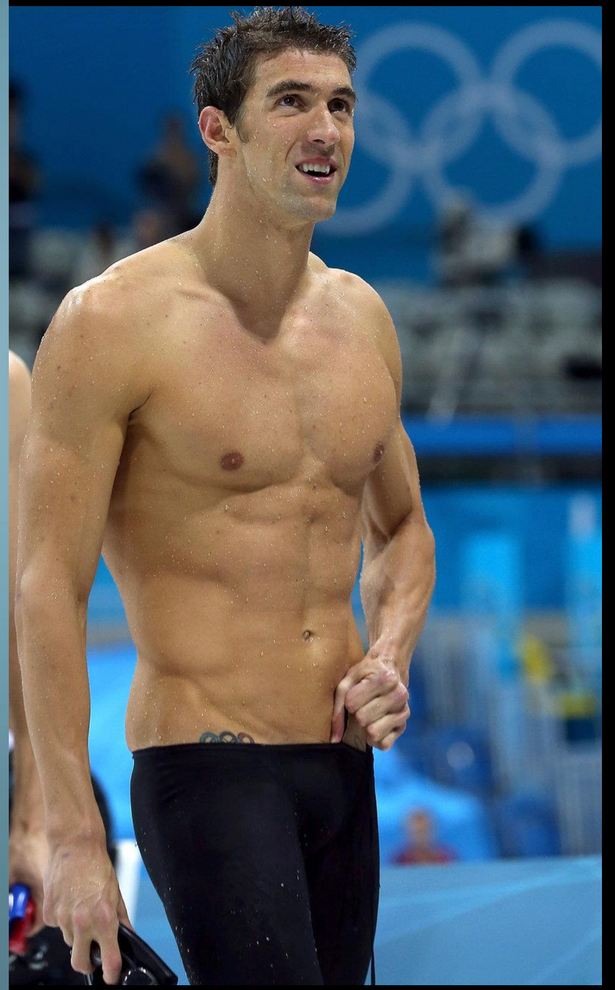
curved spine

flexibility

crowded teeth


NMF

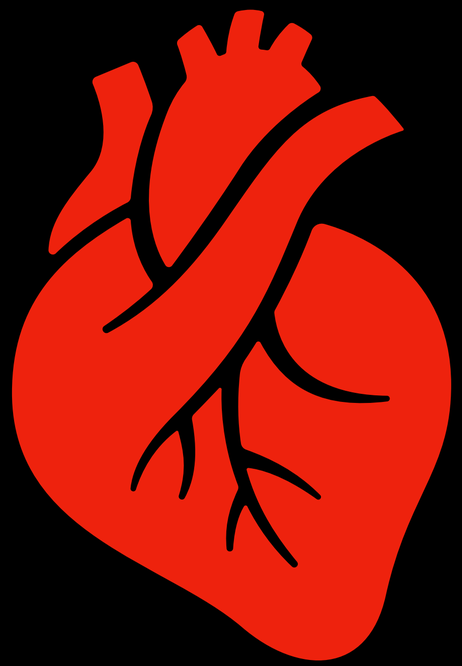

IN THIS TEMPLE
AS IN THE HEARTS OF THE PEOPLE
FOR WHOM HE SAVED THE UNION
THE MEMORY OF ABRAHAM LINCOLN
IS ENSHRINED FOREVER



Cardiovascular Complications
account for 90% of Marfan Deaths

Common dysrhythmias come from prolonged AV conduction, creating a delay in conduction from the atria to the ventricles, shown on the ECG as first, second or third-degree heart blocks and a prolonged PQ interval.

Some have issues related to ventricular repolarization, with ST segment abnormalities, prolonged QT intervals, the presence of U waves, and other ventricular dysrhythmias. Twenty-one percent of Marfan patients will experience a ventricular dysrhythmia during their lifetimes, with 4% experiencing sudden cardiac arrest due to this dysrhythmia.



Sudden and Severe Abdominal Pain

Syncope

Loss of Consciousness

often ascending aorta

AORTIC DISSECTION / RUPTURE

80%

Leg Paralysis

Shortness of Breath

Difficulty Walking

Severe Chest or Back Pain (tearing or shearing and may radiate to neck)

General Assessment, Transport and Treatment Considerations

Check for Medical ID/Hx. and take with you.

Listen to the caregiver/patient.

Appropriate Facility - Think Vascular and Trauma Surgery.

Long transports =

Immediate or pending life threat = Cardiovascular Issues.

Immediate or pending life threat = Pneumothorax.

May be on Beta Blocker or Ace Inhibitor.

Considerations Cont.

- 12 Lead EKG! (Ventricular Dysrhythmias - BLS can acquire and transmit)
- POCUS (future?)
- ET Intubation/Airway Placement (Deviated Septum?)
- Medical Control/Advanced Notice


TYPE 1
DIABETES

www.thevedsmovement.org

www.fightveds.org

www.marfan.org

THE VEDS MOVEMENT

Charging forward. Saving lives.

facebook.com/VEDSMovement

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THE MARFAN FOUNDATION

Marfan.org

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EMERGENCY

MANAGEMENT MATTERS, LLC 🚨