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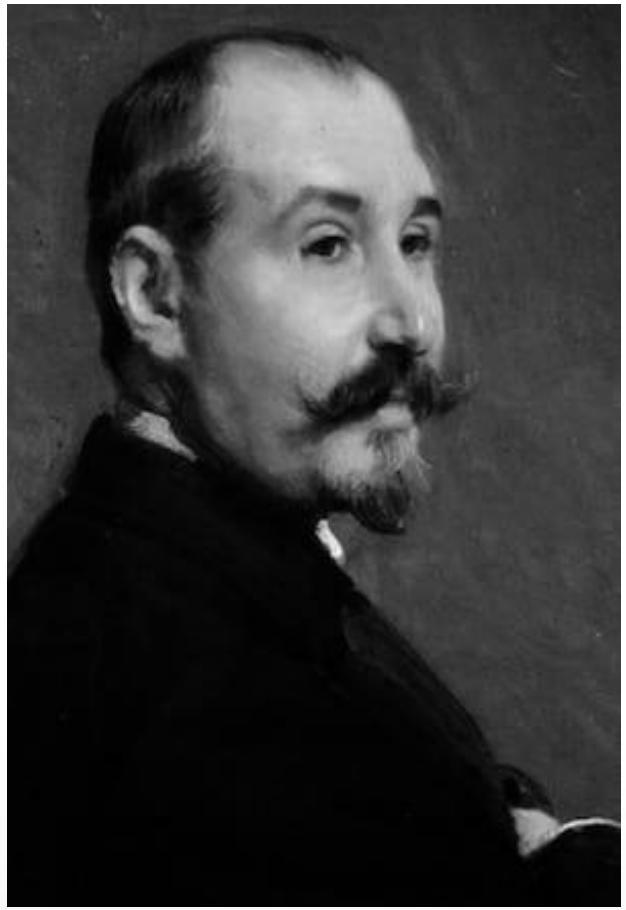
Department of Propaedeutics of Internal Medicine and Physical Rehabilitation

MARFAN SYNDROME

CLINICAL CASE WITH AORTIC ROOT DISSECTION

Assistant professor Shmidt Olena Y.

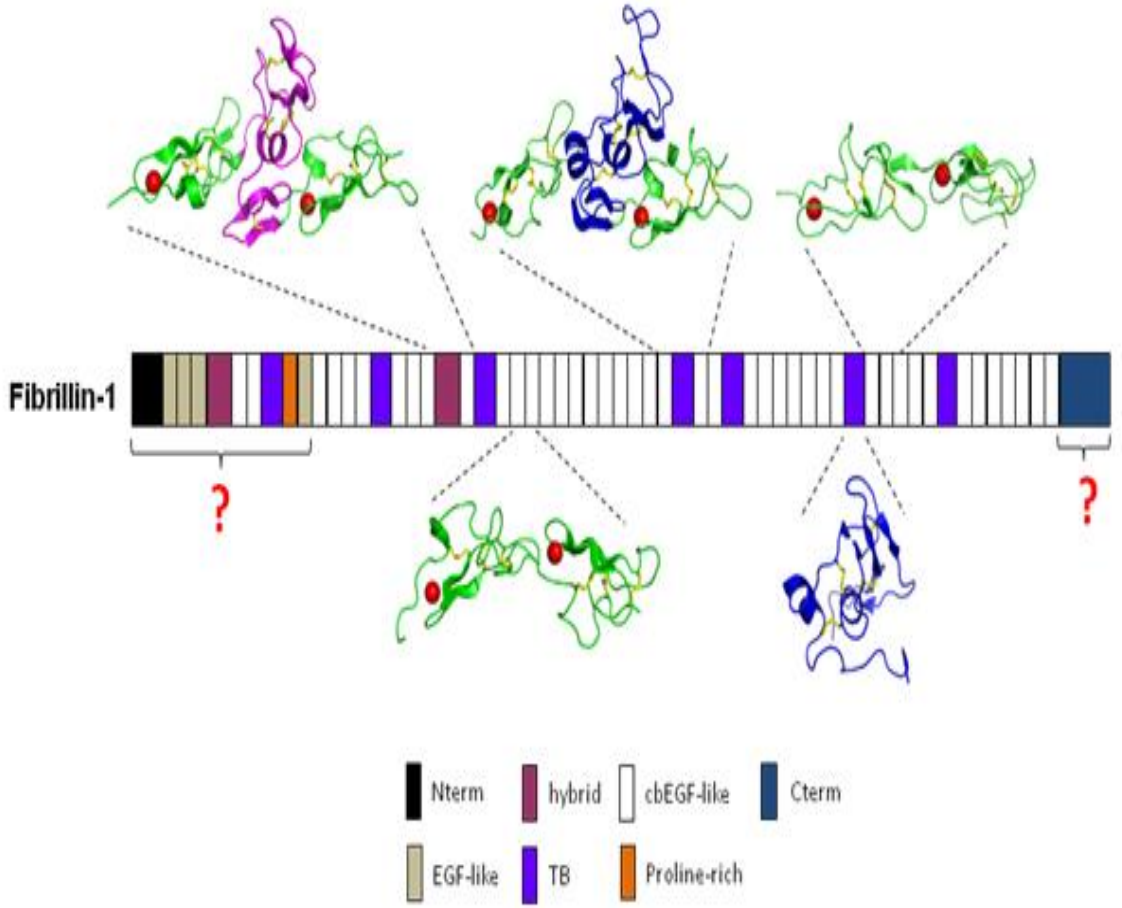
OVERVIEW



Marfan’s syndrome is a systemic disorder of connective tissue, first described more than 100 years ago by a Parisian professor of paediatrics, Antoine-Bernard Marfan, who reported the association of long slender digits and other skeletal abnormalities in a 5-year-old girl

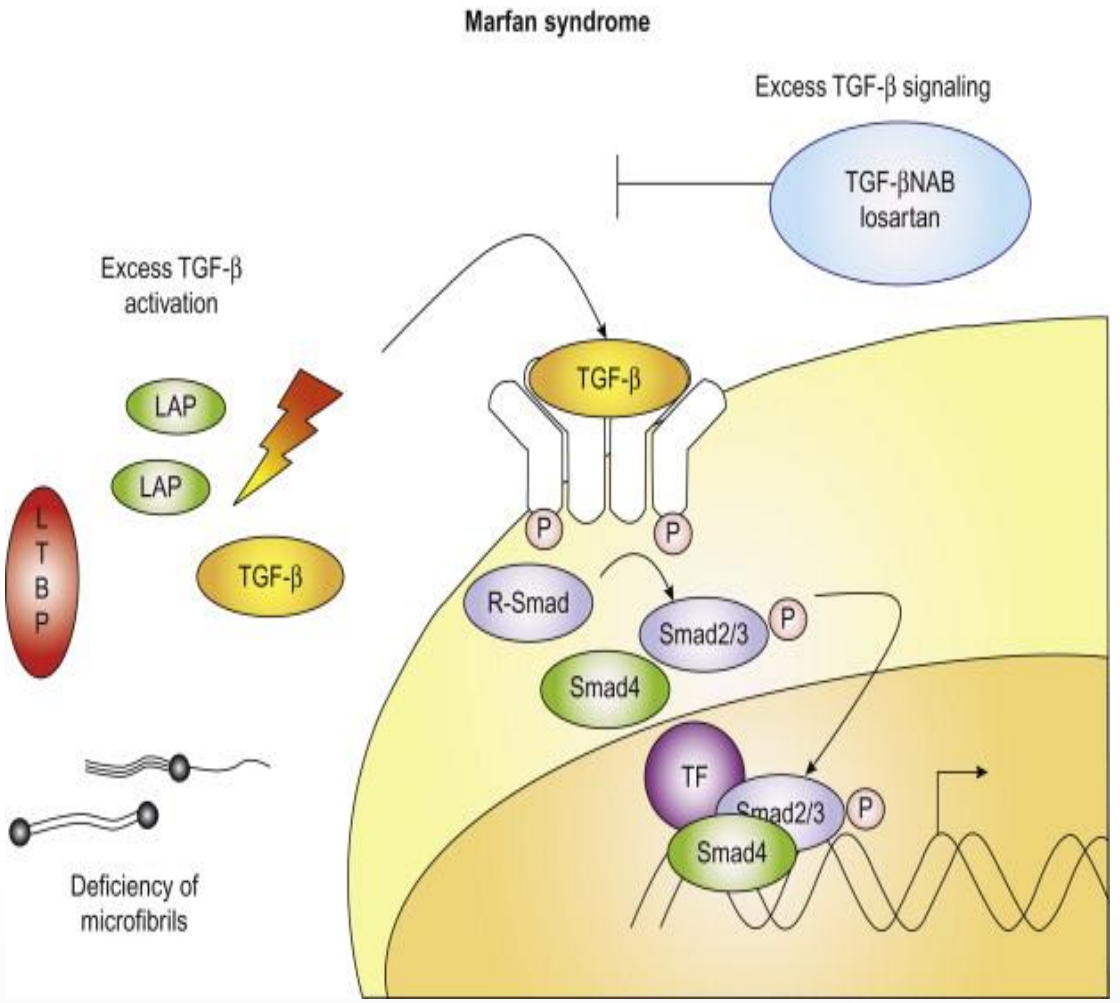
OVERVIEW

- **Marfan syndrome** (MS) is a genetic connective tissue disorder with an autosomal dominant mode of inheritance caused by mutations in the gene coding for fibrillin-1 (FBN1) on chromosome 15

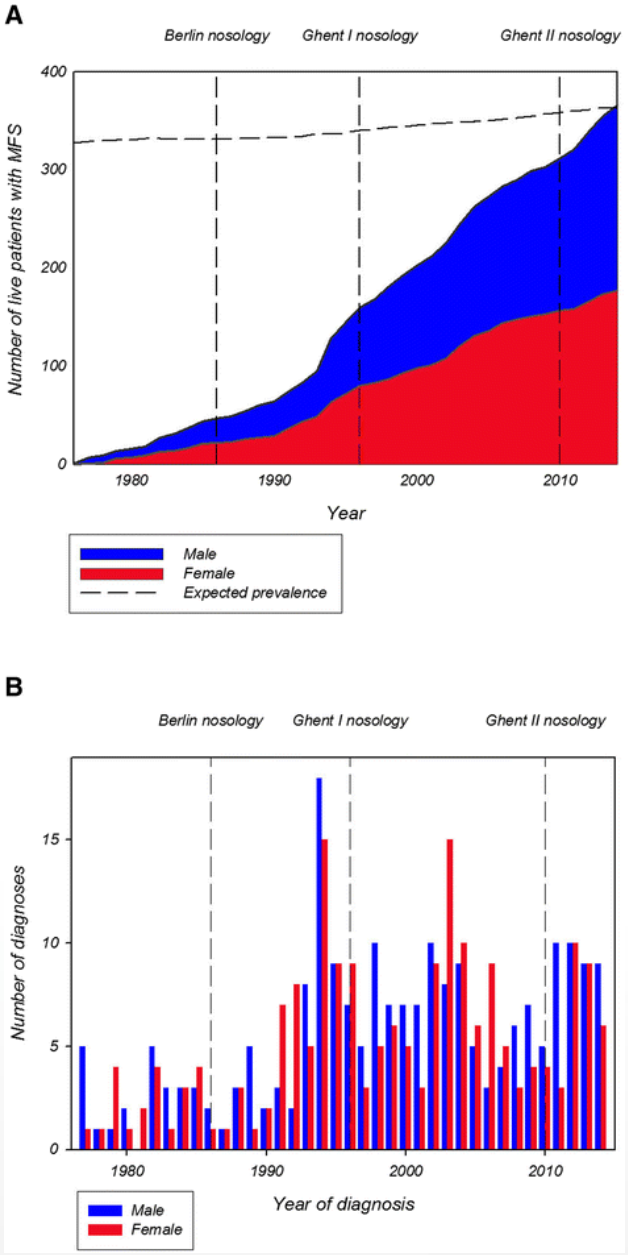


OVERVIEW

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.



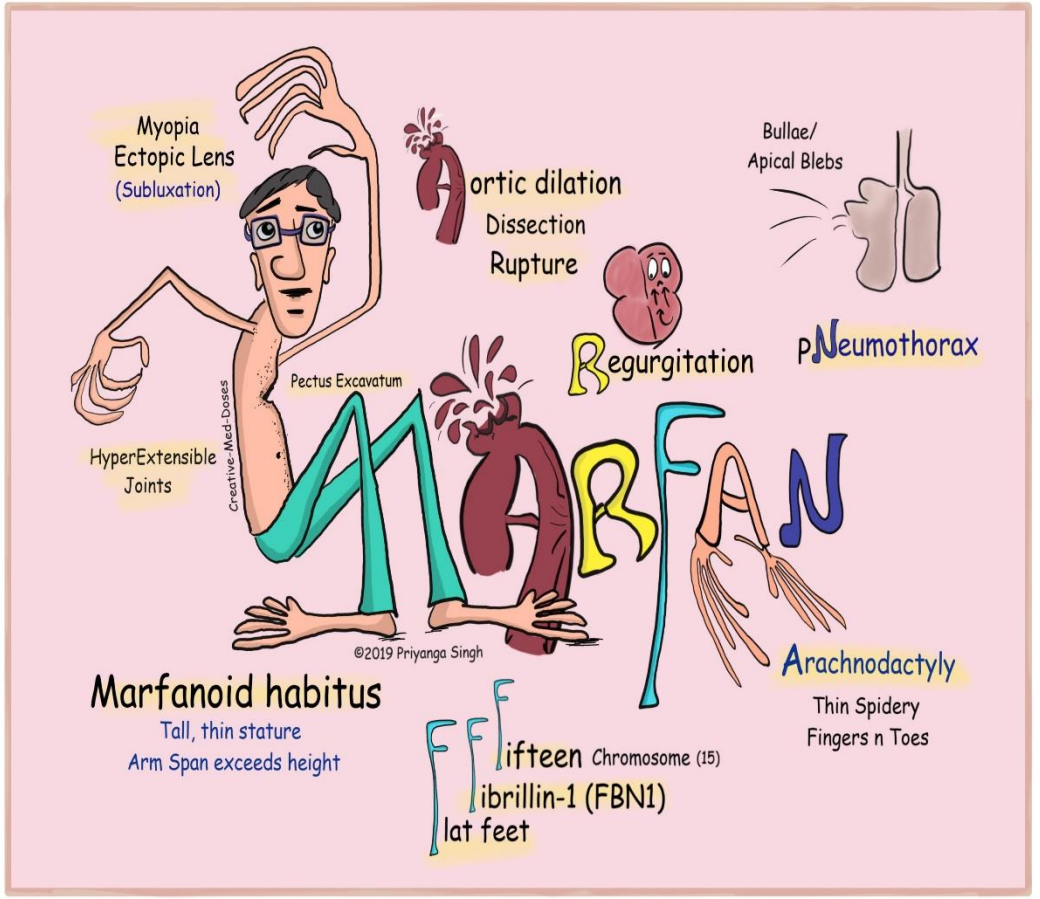
EPIDEMIOLOGY



- The incidence of classic Marfan's syndrome is about 2-3 per 10000 individuals
- The disease occurs worldwide, with no predilection for either sex.

myriad of distinct clinical problems manifestations

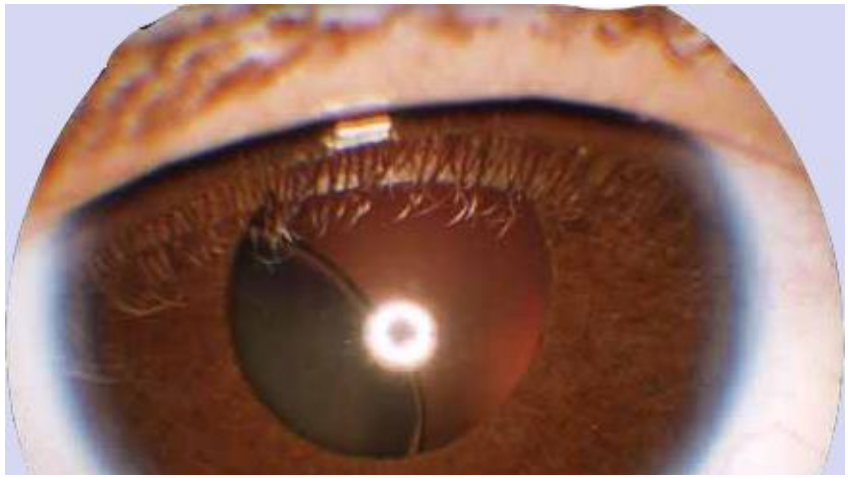
Clinical manifestations of which the musculoskeletal, cardiac, and ocular system problems predominate. Thus, it cause pleiotropic effects. In most patients, it leads to abnormalities of the aortic wall, causing



DIAGNOSTIC CRITERIA

- Relies on a set of defined criteria (Ghent nosology) developed to facilitate patient management and counseling.
- The new diagnostic criteria puts more weight on the cardiovascular manifestations of the disorder. Mainly Aortic Dilation and MPV.

DIAGNOSTIC CRITERIA
Ocular system

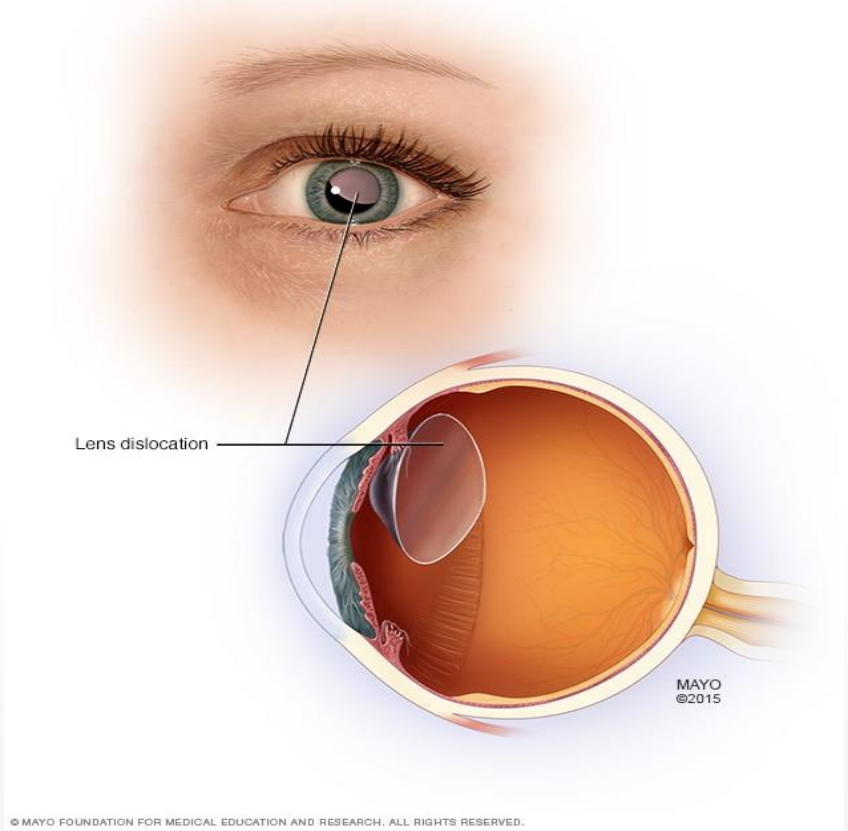


Major criterion

- Ectopia lentis

Minor criteria

- Abnormally flat cornea
- Increased axial length of globe
- Hypoplastic iris or hypoplastic ciliary muscle, causing decreased miosis



DIAGNOSTIC CRIT

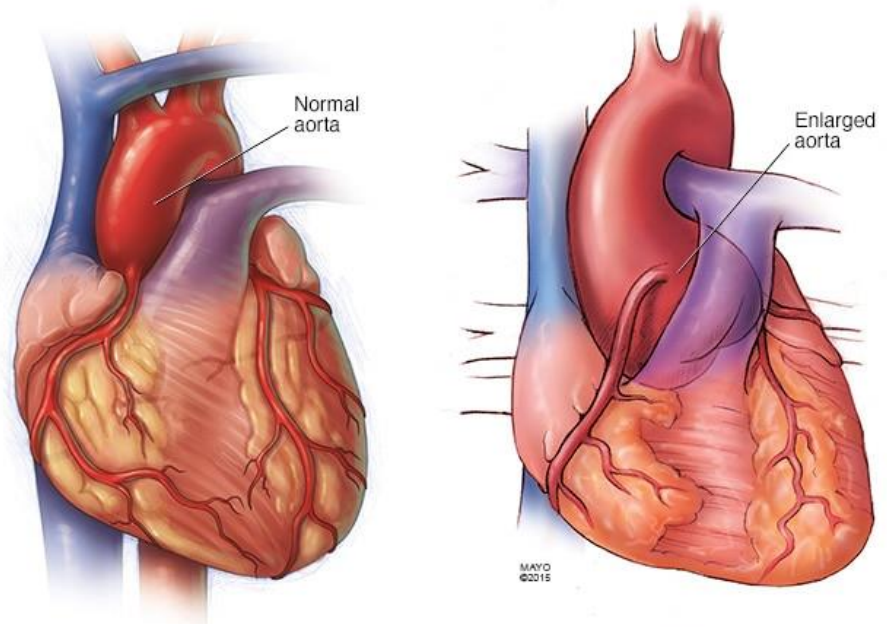
Cardiovascular system

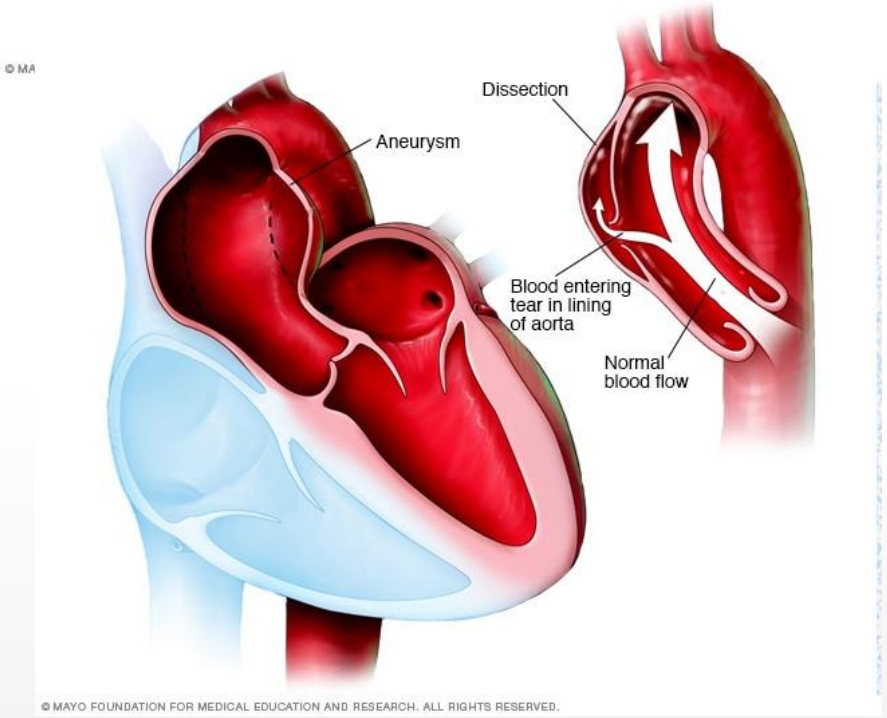
Major criteria (either of the following)

- Dilatation of the ascending aorta, with or without aortic regurgitation, and involving at least the sinuses of Valsalva
- Dissection of the ascending aorta

Minor criteria

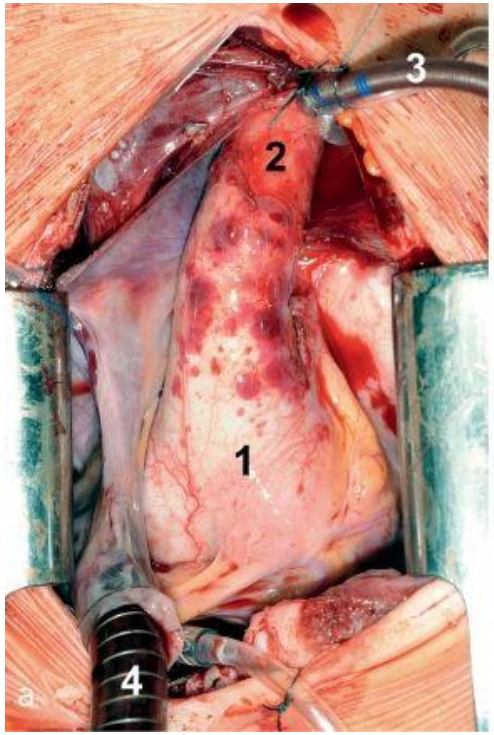
- Mitral valve prolapse with or without mitral valve regurgitation
- Dilatation of the main pulmonary artery, in the absence of valvular or peripheral pulmoni stenosis or any other obvious cause, younger than age 40 years
- Calcification of the mitral annulus younger than age 40 years
- Dilatation or dissection of the descending thoracic or abdominal aorta younger than age 50 years

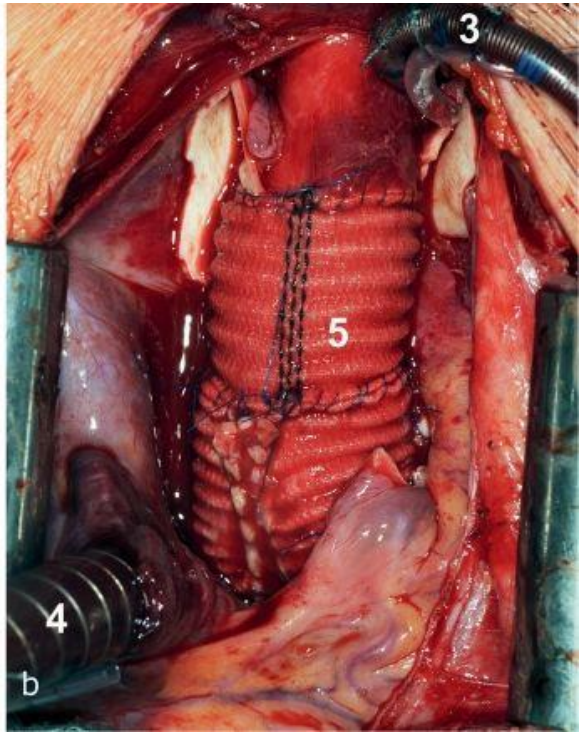


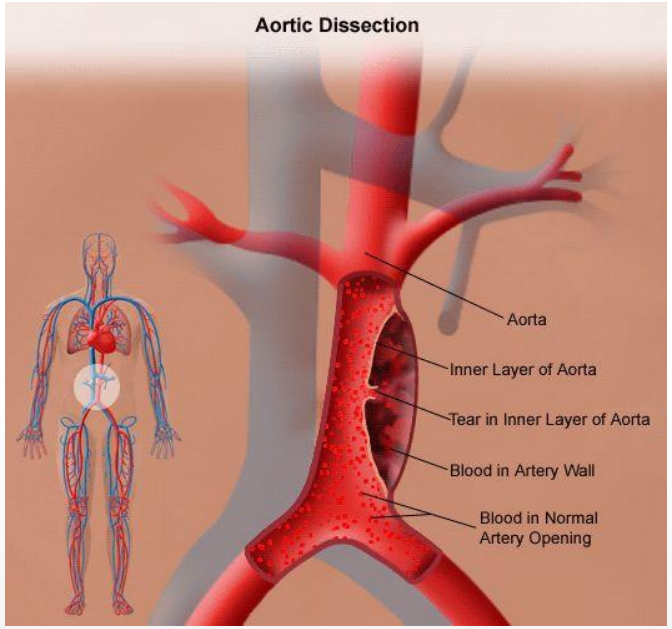


CVS COMPLICATIONS

- Aortic aneurysm
- Aortic dissection
- Valve malformations



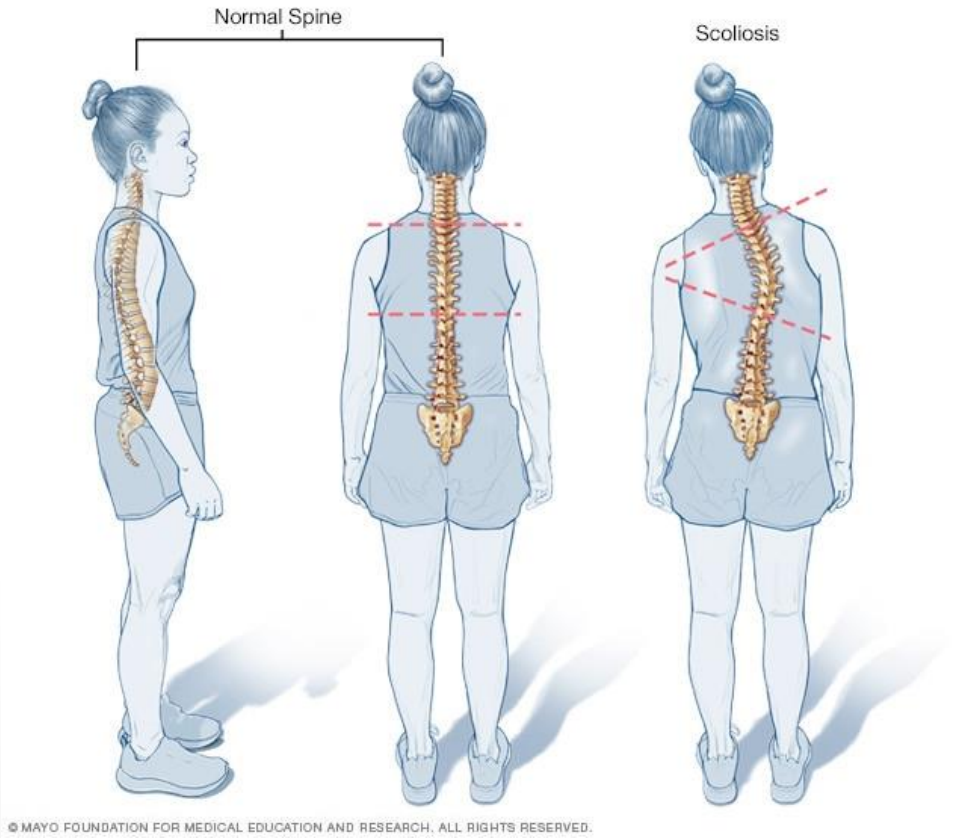




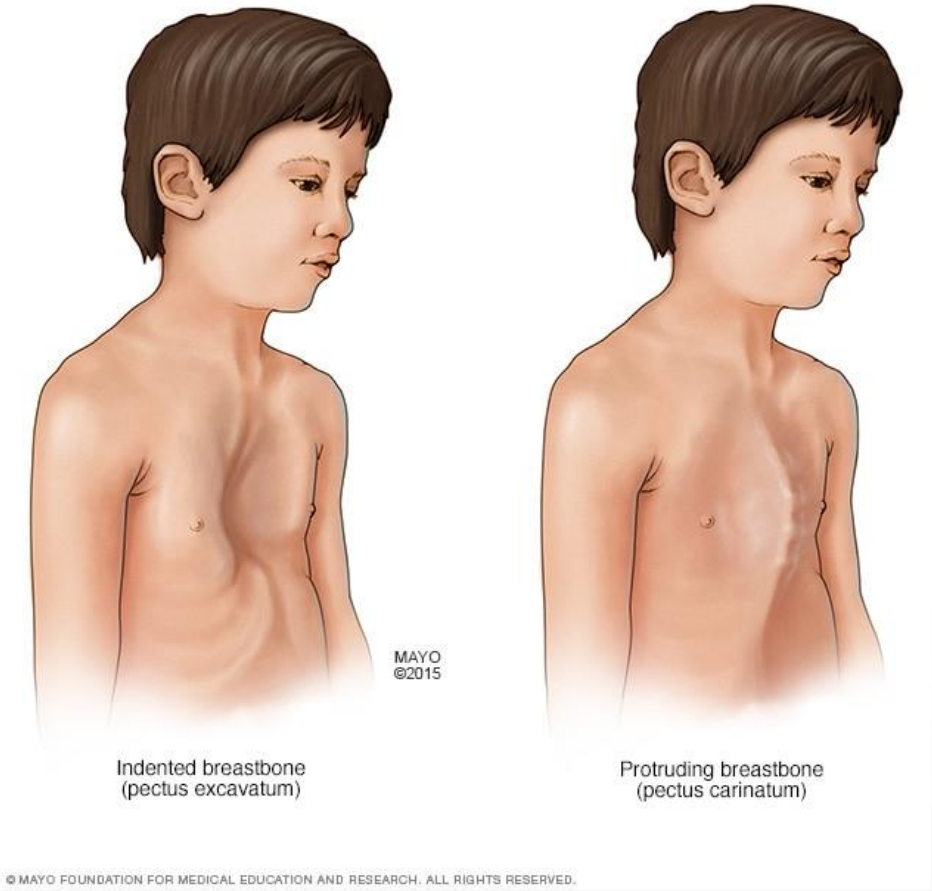
DIAGNOSTIC CRITERIA

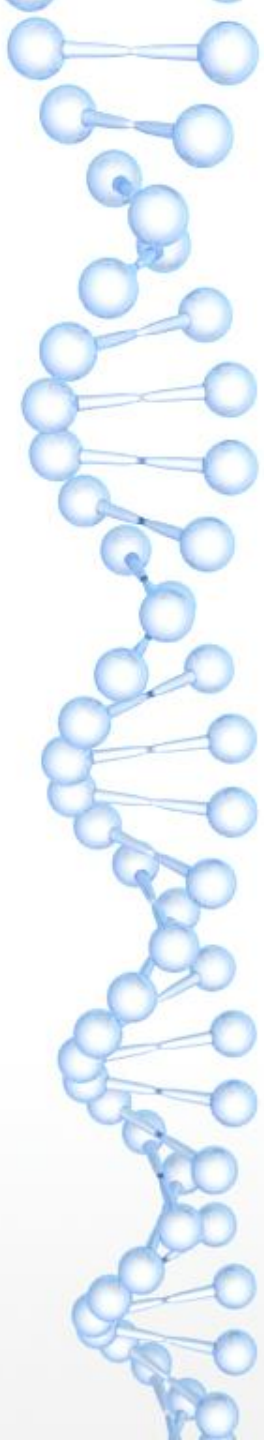
Skeletal system

- Scoliosis



- Chest abnormalities





Major criteria

Pectus carinatum

Pectus excavatum, needing surgery

Reduced upper-segment to lower-segment ratio or arm span to height ratio >1.05

Wrist and thumb signs

Scoliosis of >20° or spondylolisthesis

Reduced extension at the elbows (<170°)

Medial displacement of the medial malleolus, causing pes planus

Minor criteria

Pectus excavatum of moderate severity

Joint hypermobility

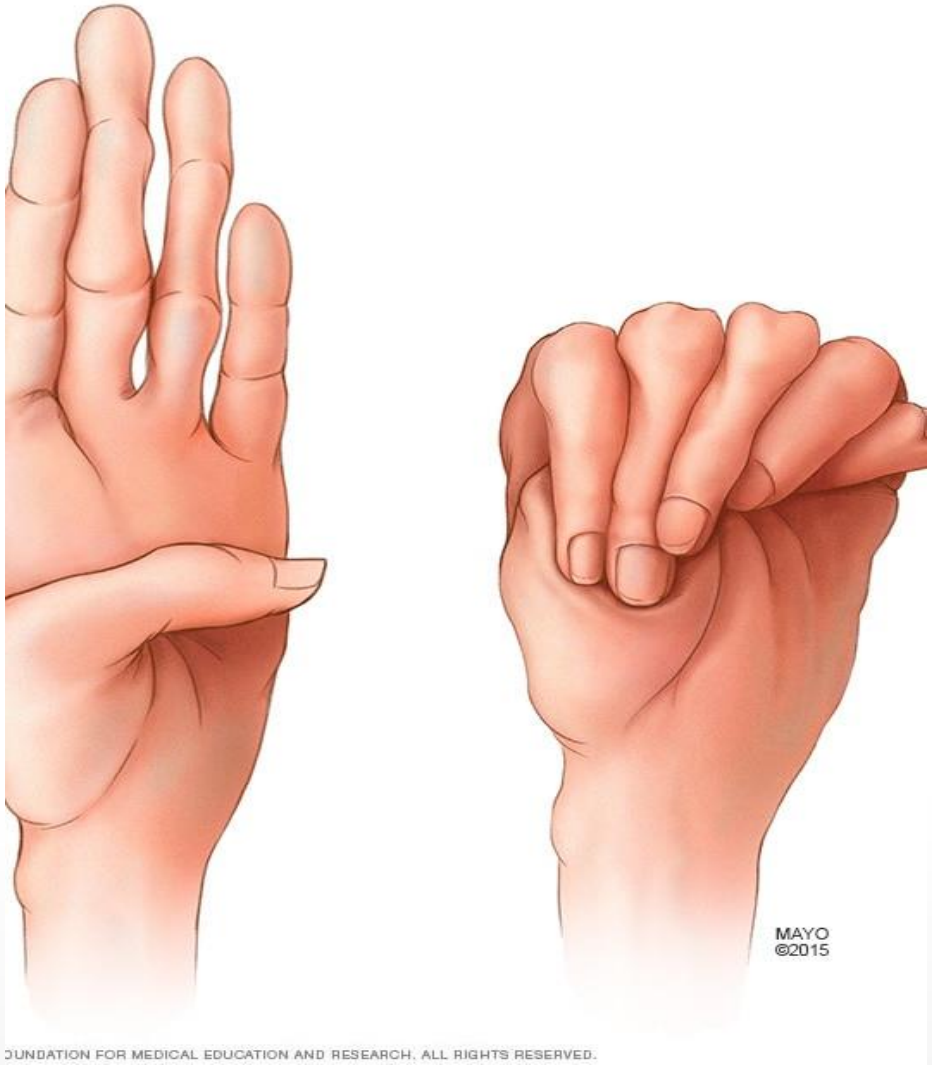
Highly arched palate with crowding of teeth

Facial appearance (dolichocephaly, malar hypoplasia, enophthalmos, retrognathia, down-slanting palpebral fissures)

FINGER LENGTH IN MARFAN SYNDROME

Long fingers.

It's common for their thumbs to extend far beyond the edge of their hands when they make a fist.

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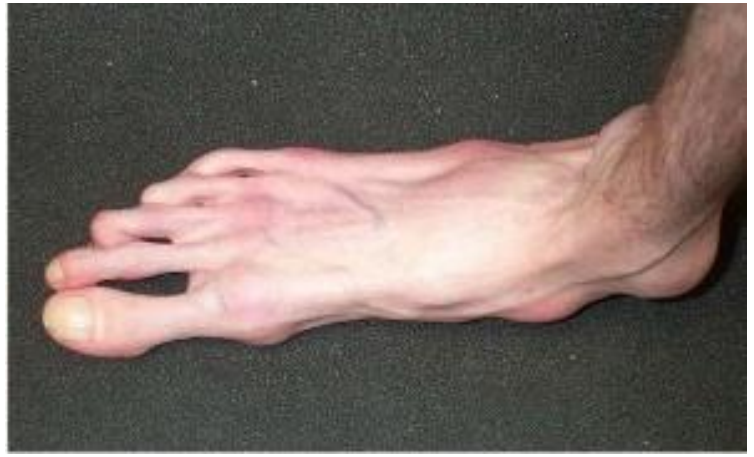
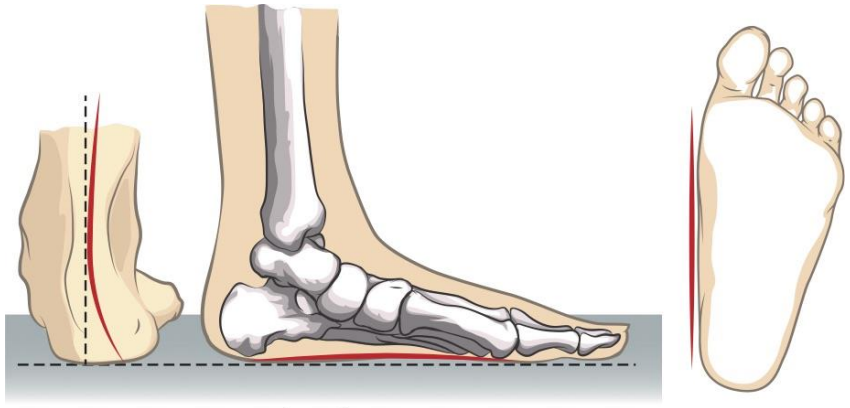
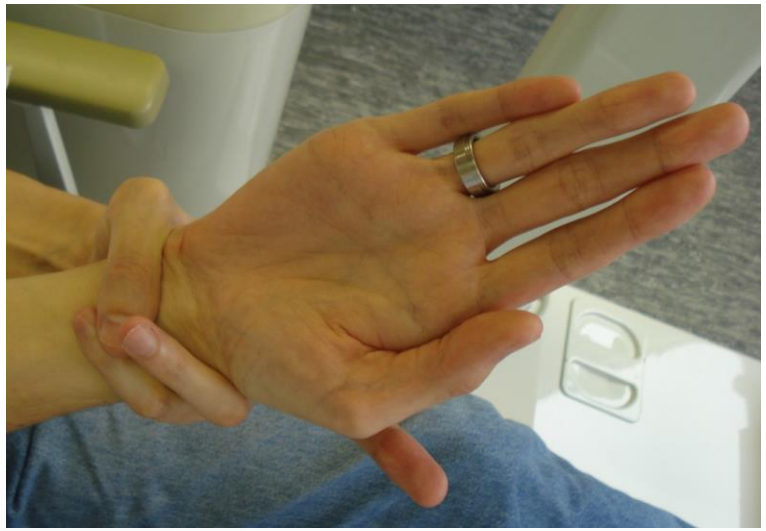
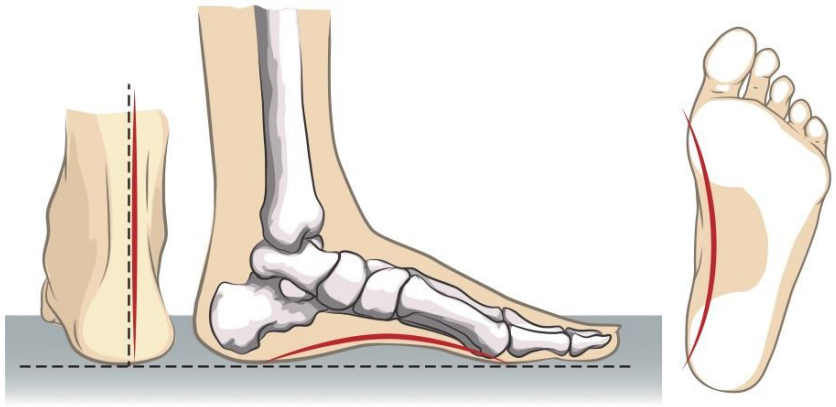




Flat feet





Normal feet

Other symptoms that can have patients with MfS

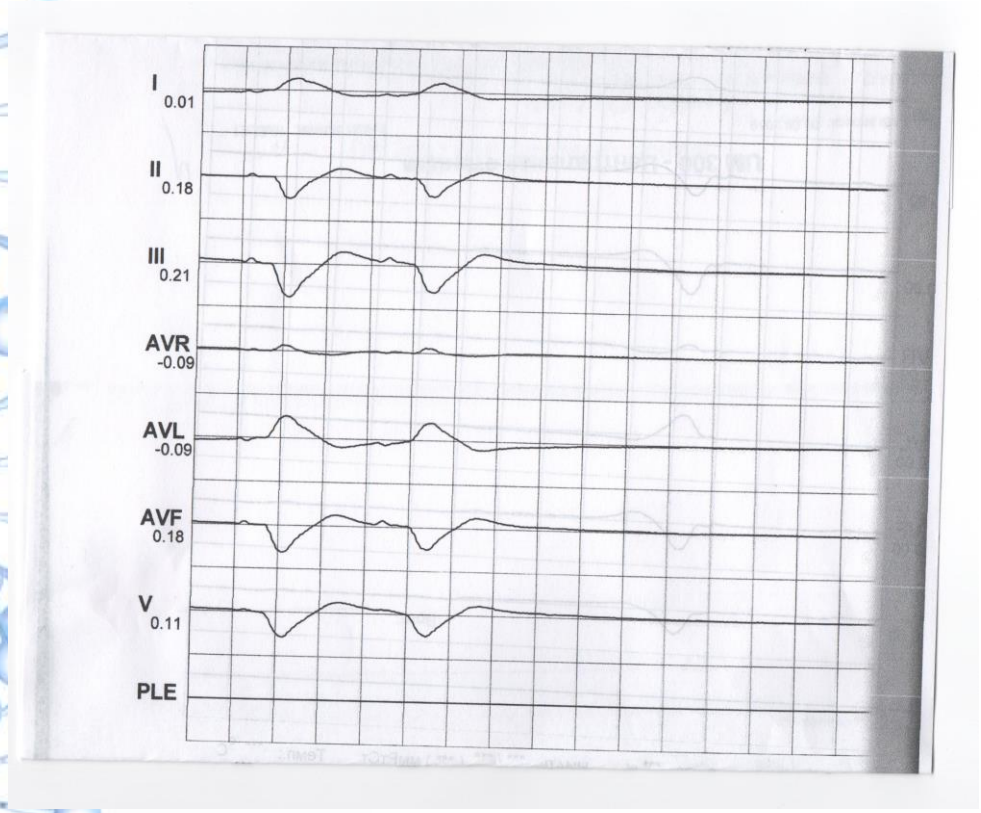
Pulmonary system - spontaneous pneumothorax
Skin and integument - striae atrophicae (stretch marks) without marked weight gain, pregnancy, or repetitive stress

Clinical case

08/06/2019

42 years old female patient was admitted to the emergency department in a grave condition.

She was urgently hospitalized in the cardiopulmonary resuscitation department with complaints of pain behind the sternum and lower abdomen, shortness of breath, weakness, dizziness, interruptions in the work of the heart.



Despite the therapy, the patient's condition progressively worsened, Consciousness, reflexes, attempts of spontaneous breathing were absent.
Blood pressure, pulsation of large vessels were not determined.
According to ECG monitoring - persistent asystole. Biological death was ascertained.

Atrioventricular rhythm passing into asystole

ANAMNESIS MORBI

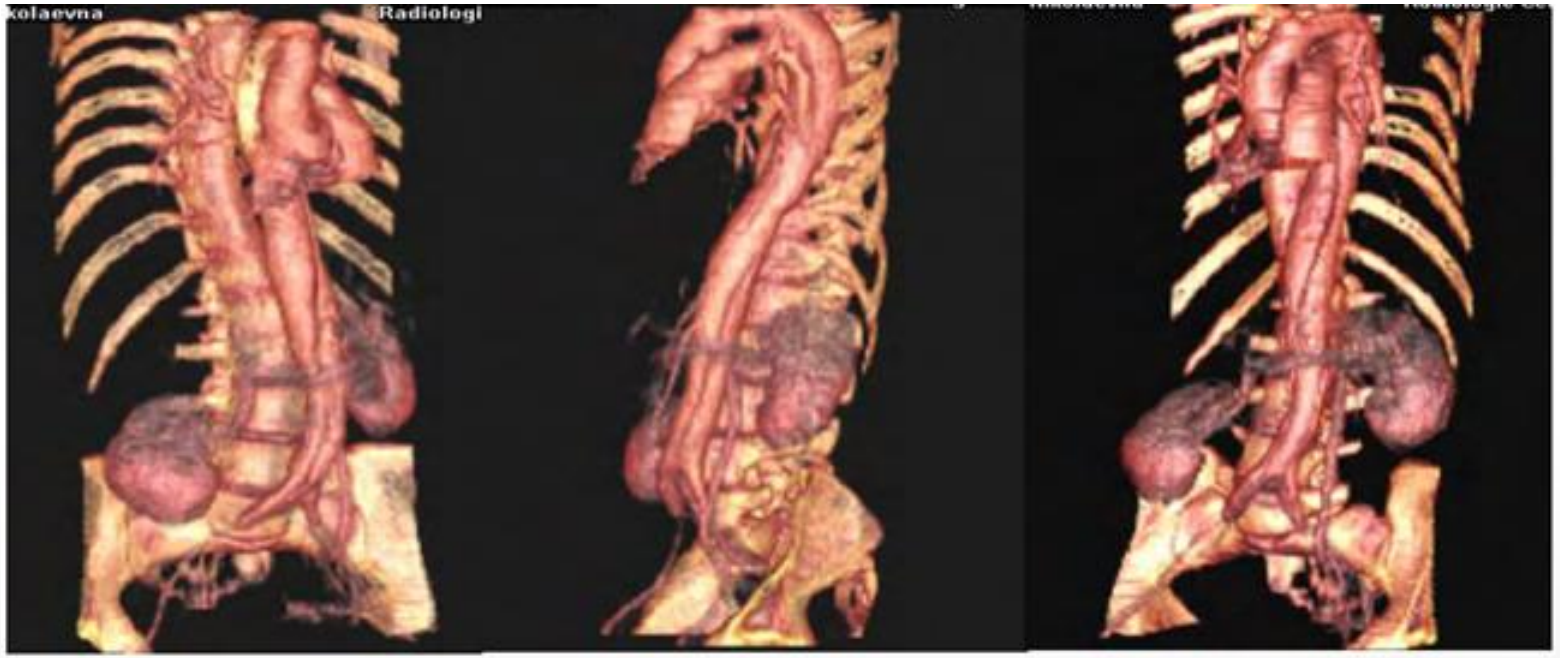
The patient’s first visit to the hospital was at age 33 (in 2011)
She presented complaints of a feeling of discomfort in the chest, headache, a feeling of heart palpitations up to 120 beats per minute, and an increase in blood pressure.

Objective examination revealed asthenic constitutional type
(weight 65 kg, hight 180 cm, BMI=20,1 kg/м2),
funnel chest deformity,
signs of scoliosis, BP 140|100 mm Hg

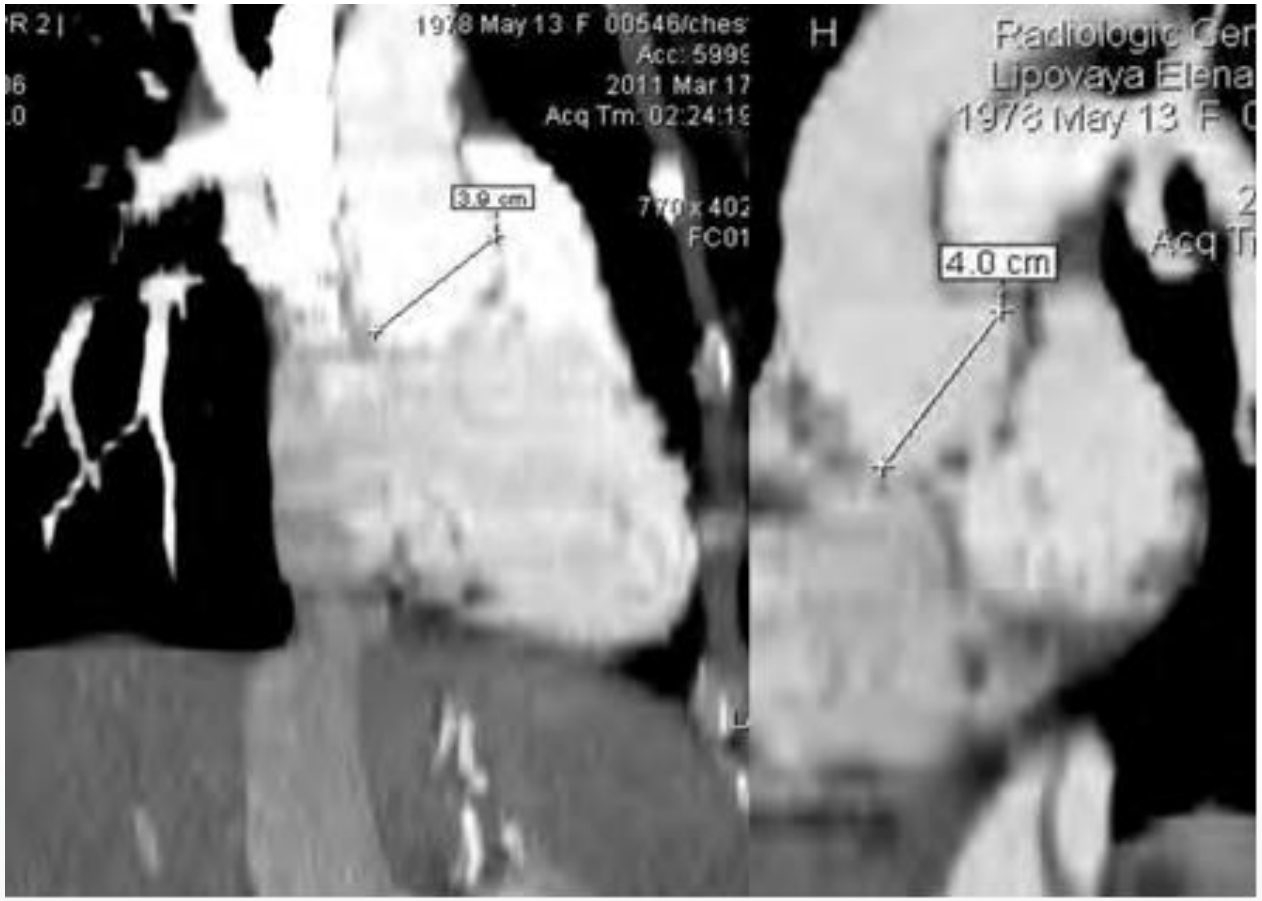
The **patient’s father** died at the age of 33 because rapture of aortic aneurysm.

CT of the case

- Aortic aneurysm with dissection (DeBakey type III, Stanford type B) from the origin of the left subclavian artery, along the entire thoracic, abdominal aorta, common iliac arteries on both sides.



- **MRI of the case**
- Frontal and sagittal planar reformation of ascending aorta.



Diagnosis

According to data of instrumental examination (CT: exfoliating aortic aneurysm with dissection (DeBakey type III, Stanford type B) from the origin of the left subclavian artery, along the entire thoracic, abdominal aorta, common iliac arteries on both sides) was established diagnosis:

- Marfan syndrome
- Aortic aneurysm, DeBakey class III. Stanford type B
- Mitral valve prolapse III grade
- Arterial hypertension II stage 2 grade.

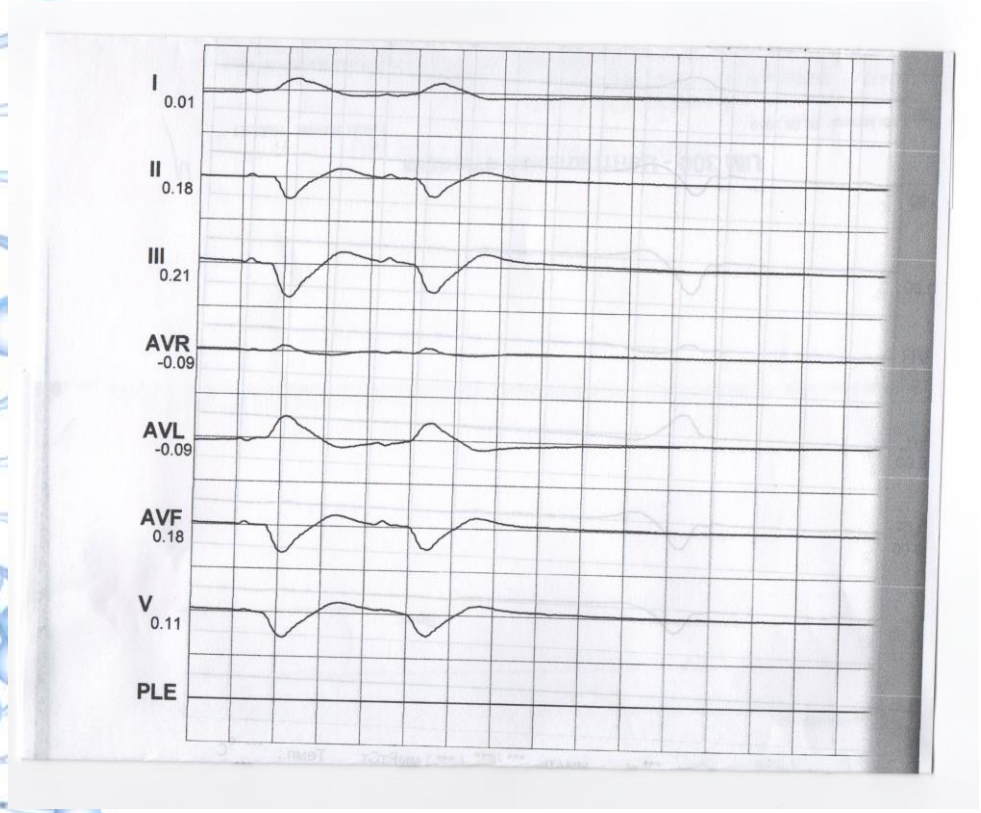
Patient was consulted by cardiosurgerist: surgical treatment **has not been indicated in relation to the magnitude of dissection.**

Patient progression

Next 8 years the patient was regularly hospitalized because of high blood pressure, got antihypertensive treatment. Annually repeated CT and Echo CG did not show any changes in aortic dissection length.
On July 2019 she was hospitalized because of high BP (170/100 mmHg) and arrhythmia.
After proper treatment the patient was discharged from the hospital in stable condition with BP 120|80 mmHg.

Patient progression

In 1 month after discharging from the hospital the patient felt worsening of her condition: pain in the abdomen, chest, sudden weakness and dizziness
At an extremely grave condition she was admitted to the hospital with BP 70/40 mmHg, HR 100 bpm



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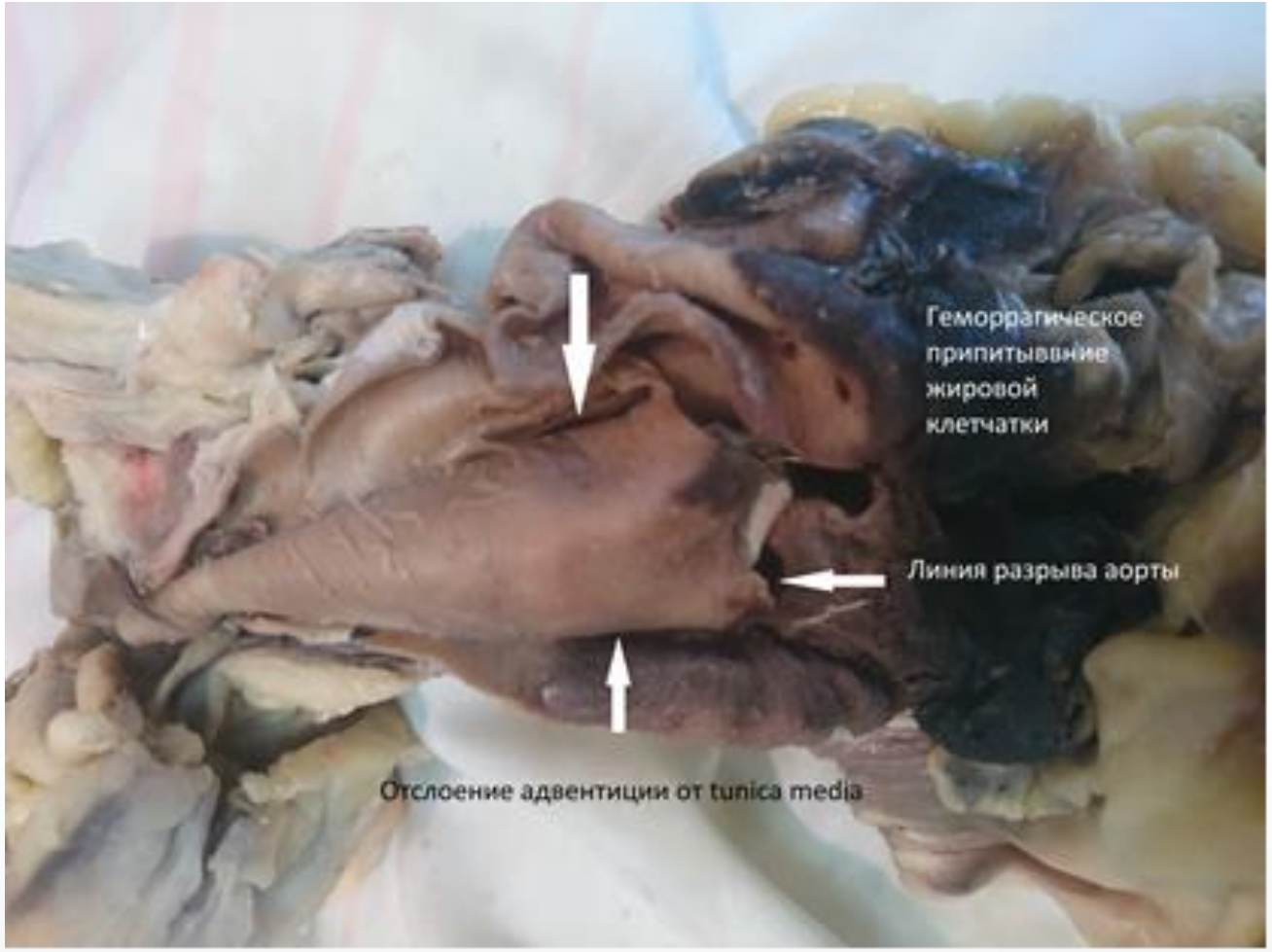
Atrioventricular rhythm passing into asystole

- Pathological preparation of aorta with left ventricular endocardial fragments.

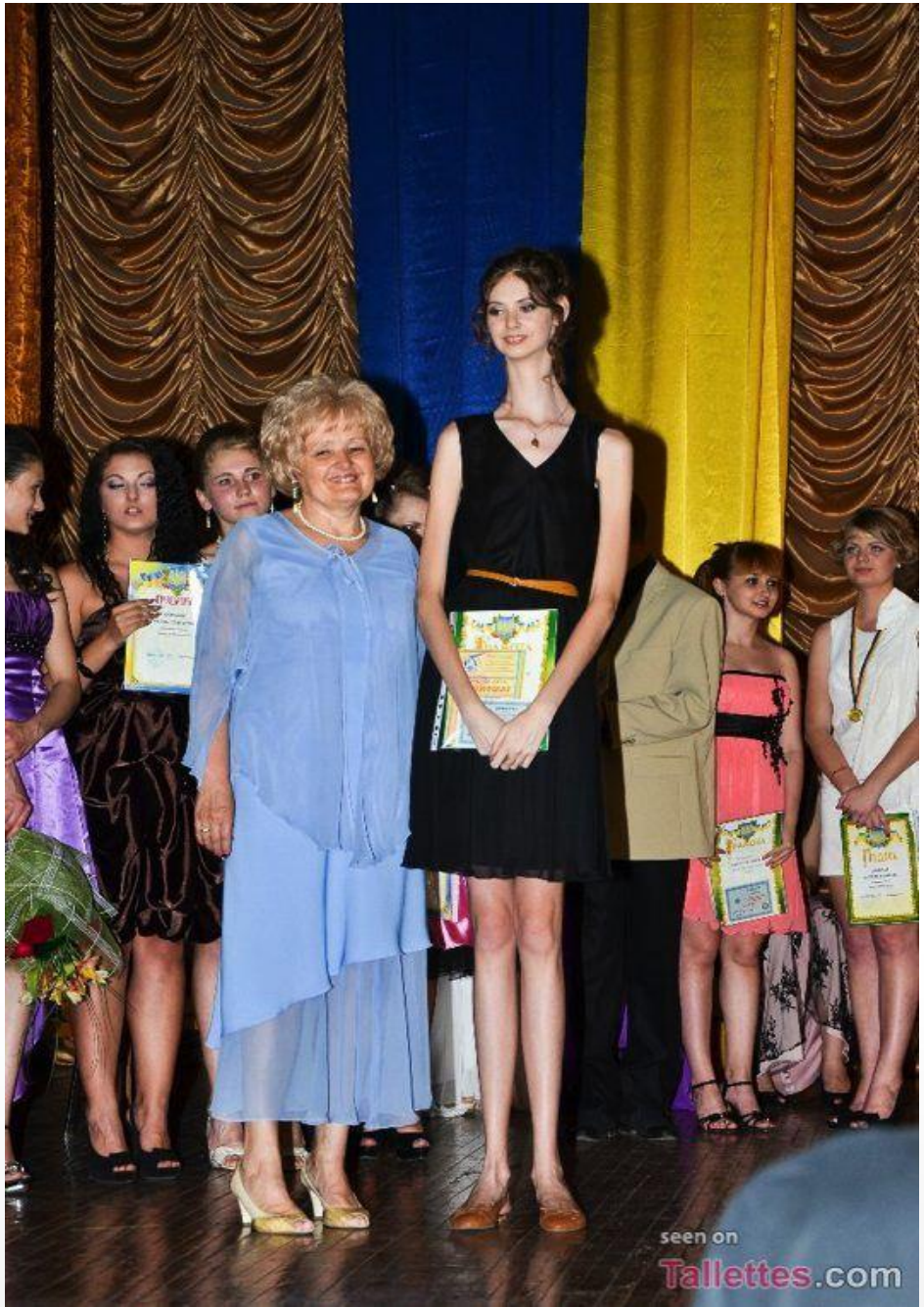


Conclusion

The presence of connective tissue disorders predisposes patients to early dissection of the aorta. Identification of such patients at the earliest stages will provide focused therapy and early intervention for prevention of ruptures and following mortality.

Remember how do they look and may be you will save somebody's life!

(from the Marfan Foundation)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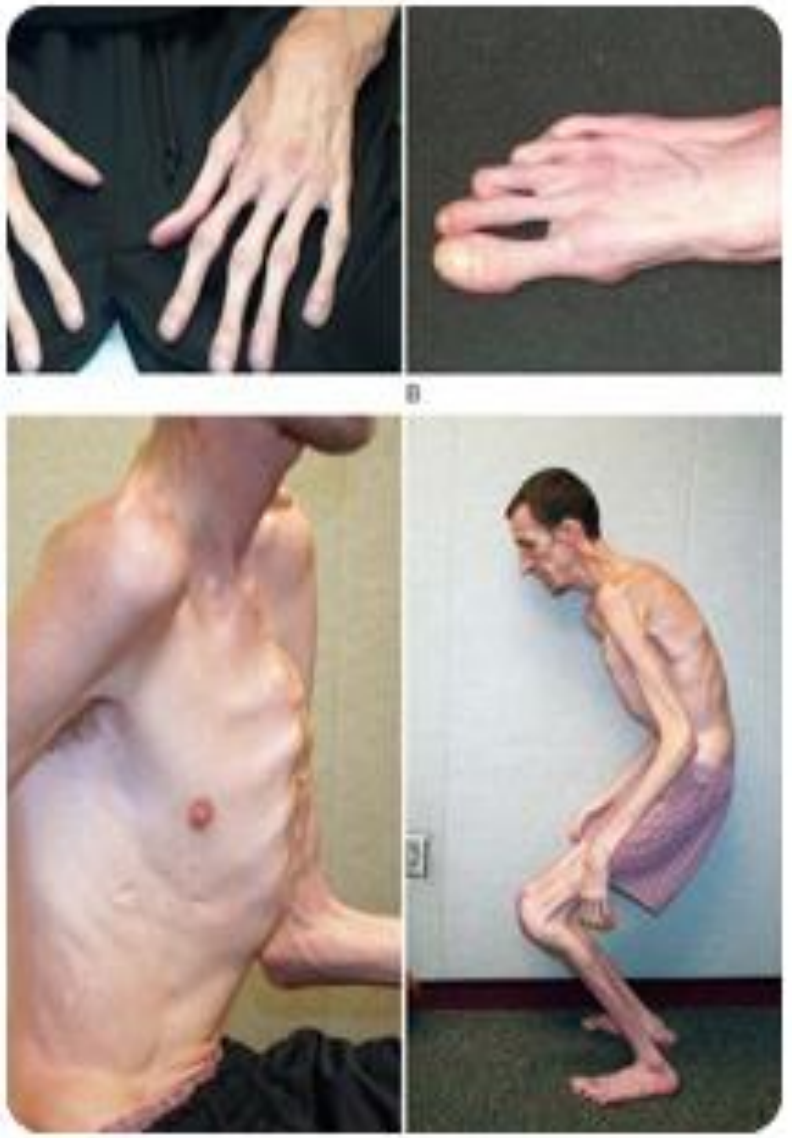


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THANK YOU
FOR YOUR
ATTENTION