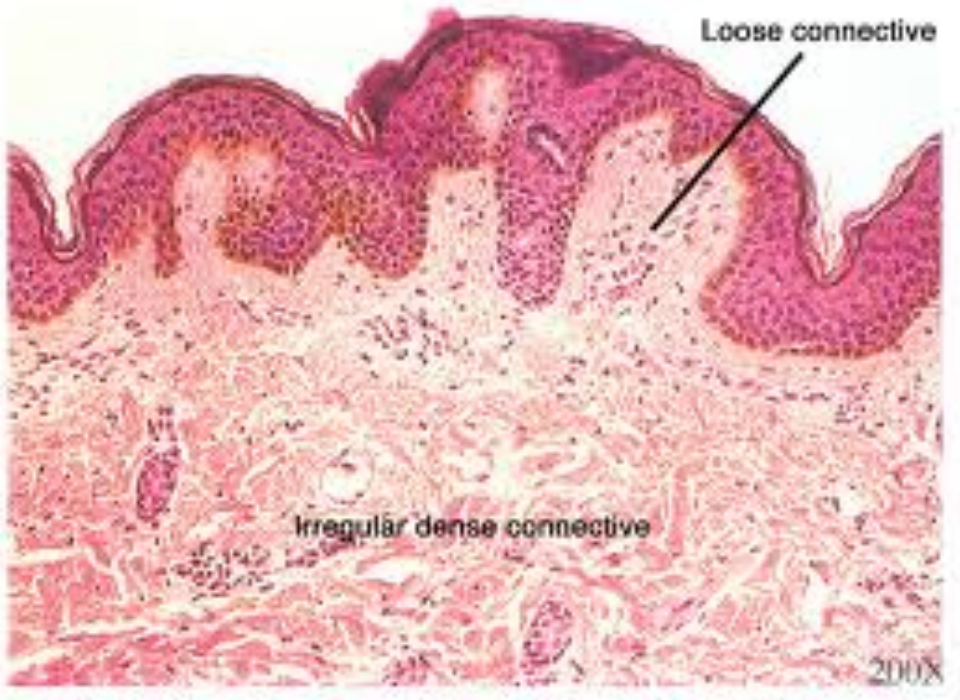
Marfan Syndrome

By Taylor Aulson

What Is It

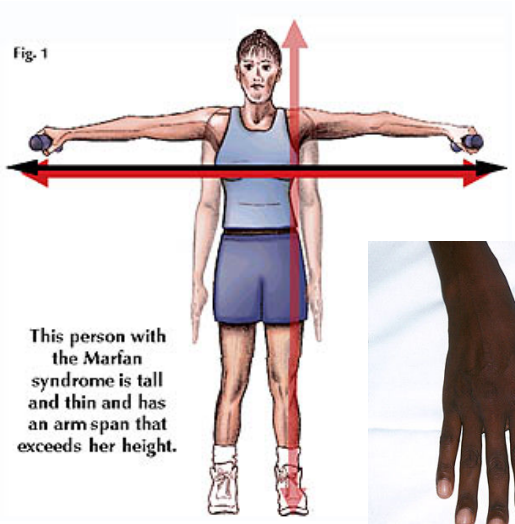
- A disorder of the connective tissue
- It can affect many body systems

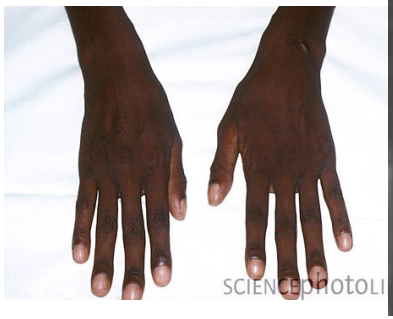


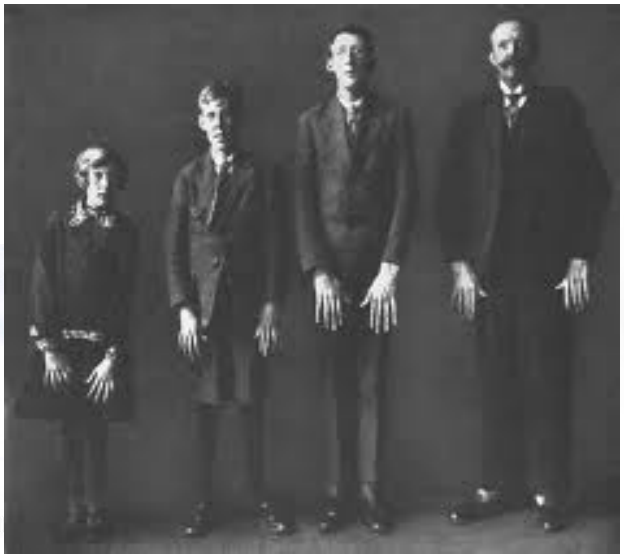
OTHER NAMES:

- MLS
- Marfan's Syndrome

- Affects men, women and children
- All races and ethnic backgrounds
- At least 1 in every 5,000 people in the United States have the disorder







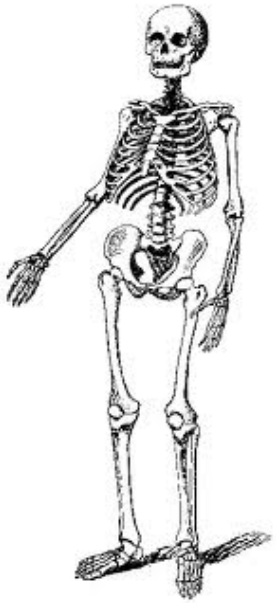
Who it Affects

- Skeleton
- Eyes
- Cardiovascular System
- Nervous System
- Skin
- Lungs

WHAT IT AFFECTS

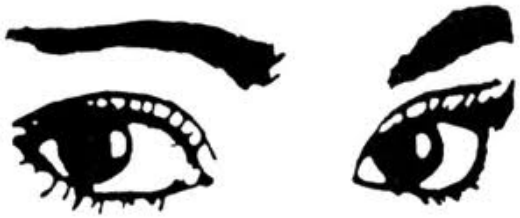
SKELETON:

- Tall and slender bodies
- Long fingers and toes
- Arm spans longer than their body height
- Unusually flexible joints
- Narrow face
- High arched roof of mouth
- Crowded teeth
- Scoliosis
- Sunken or protruding chests



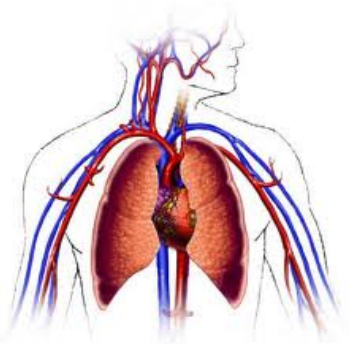
EYES:

- Dislocation of one or both eye lenses
- Retinal detachment
- Nearsightedness
- Glaucoma
- Cataracts



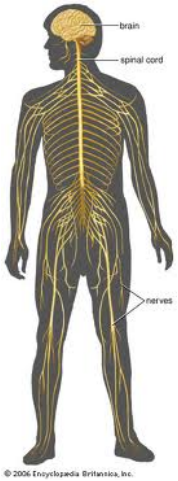
Signs and Symptoms

CARDIOVASCULAR SYSTEM:



- Aortic dilatation
  - The wall of the aorta may be weakened and stretched
- Aortic dissection
  - When the aorta tears
- Sudden death
- Heart murmurs
  - Doctors hear through stethoscopes
  - Large murmurs can result in shortness of breath, fatigue, and palpitations

NERVOUS SYSTEM:

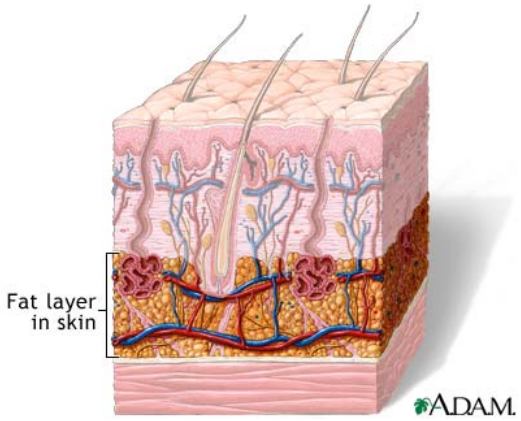


- Dural ectasia
  - Weak and stretched duras begin to weigh on the vertebrae in the lower spine and wear away the bone surrounding the spinal cord
  - Can lead to mild discomfort or to radiated pain in the abdomen, pain, numbness, or weakness in the legs.

Signs and Symptoms

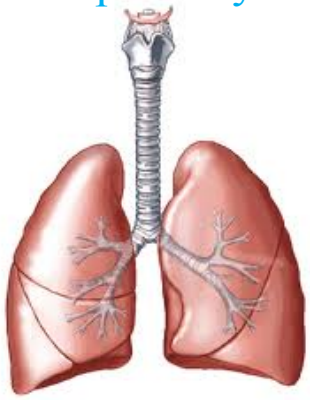
SKIN:

- Stretch marks
  - Even without weight change
  - Can occur at any age and involve no health risks
- Abdominal or inguinal hernia
  - A bulge containing parts of the intestines



LUNGS:

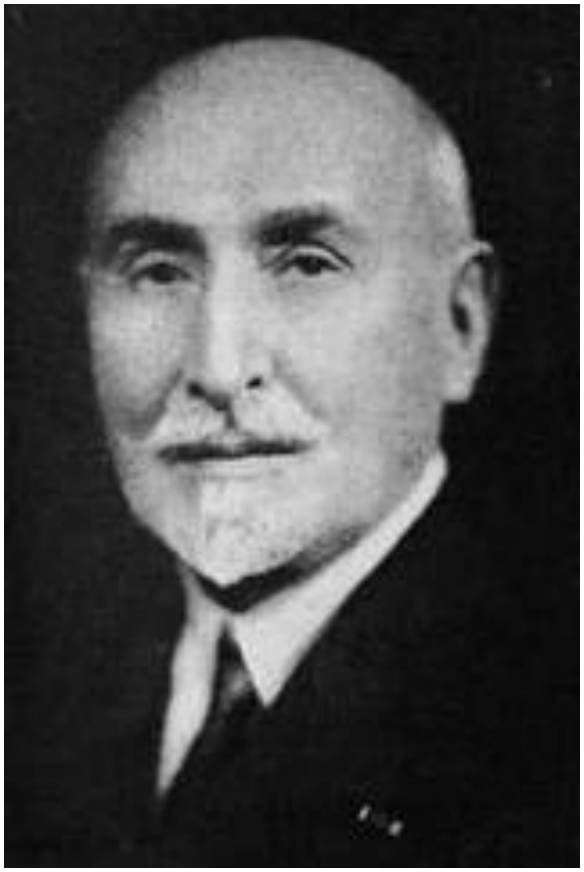
- No noticeable problems
- If tiny air sacs become stretched or swollen, the risk of lung collapse may increase
- Sleep-related breathing disorders
  - Snoring
  - Sleep apnea
    - When breathing briefly stops



Signs and Symptoms

History of the Disorder

Marfan Syndrome was named after a French man, Bernard Marfan. He became Assistant Professor of Pediatrics in the Paris faculty in 1892. Using a 5 year old girl, Gabrielle, who seemed to have long limbs that were disproportioned, he shared his findings of the symptom with others. Other symptoms like arachnodactyly (long digits), cardiovascular abnormalities, and ocular lens dislocation were found in later studies. Marfan gained a world-wide reputation and was recognized as a pioneer of pediatric medicine in France. By the time 1934 had arrived, Bernard received an honorary fellowship of the Royal Society of Medicine in Britain. He later died in 1942, but his findings are still being investigated today.



BERNARD MARFAN

There isn't any specific laboratory tests to diagnose Marfan Syndrome. Doctors rely on observations and medical history like…

- Information about family members who may have the disorder or who had an early, unexplained, heart-related death
- A thorough physical examination
- An examination of the eyes
- Heart tests

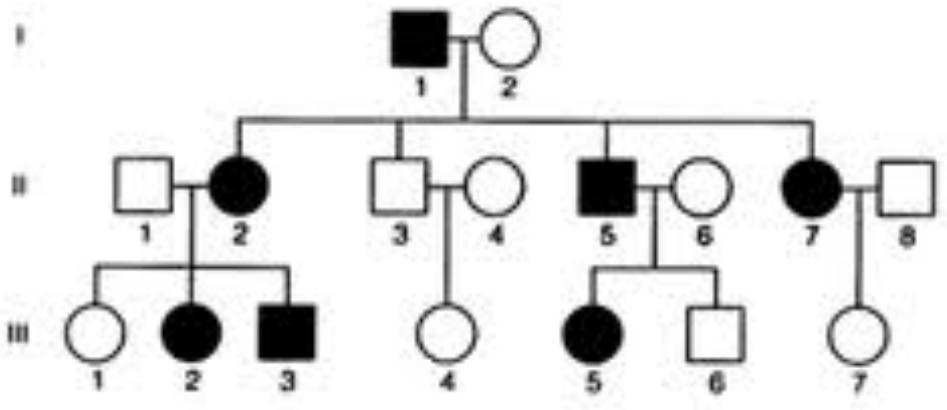
If the family has a history of the disease, a doctor may diagnose it. For someone with no family history of the disease, at least three body systems must be affected before a diagnosis is made. Genetic analyses may be helpful when creating a diagnosis of Marfan syndrome, but these analyses are time consuming and might not provide any additional information that is helpful for the doctor and the patient. Family members of a diagnosed person shouldn't assume they aren't affected if there isn't any knowledge that the disorder existed in previous generations.

How It Is Diagnosed

Genetics of the Disorder

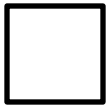
Mutations in the FBN1 gene cause Marfan syndrome. A protein called fibrillin-1 is made by following instructions that the FBN1 gene provides. The fibrillin-1 binds to itself and other proteins and molecules and creates filaments called microfibrils. Microfibrils become part of the fibers that give connective tissue it's strength and flexibility. A mutation in the FBN1 gene can reduce the amount and/or quality of the fibrillin-1 that's available to form microfibrils. Resulting with the growth factors being released inappropriately, causing the features of Marfan syndrome.

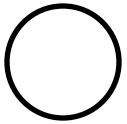
It is inherited by an autosomal dominant pattern, which means one copy of the carrier gene in each cell is needed to cause the disorder. Meaning that if one of the parent's have MFS, then the offspring will obtain it.




Affected Male


Affected Female


Unaffected Male


Unaffected Female

KINDS OF DOCTORS TO SEE:

- A general practitioner or pediatrician
  - Oversees routine health care and refer the patient to specialists
- Cardiologist
  - Specializes in heart disorders
- Orthopaedist
  - Specializes in bones
- Ophthalmologist
  - Specializes in eye disorders
- Geneticist
  - Specializes in genetics

There is no official cure for Marfan Syndrome. Scientists are working on identifying and changing the specific gene that is responsible for the disorder before birth. A range of treatment can help minimize and sometimes prevent complications. The appropriate specialists will develop an individualized treatment program. Approaches the doctors use depends on which systems have been affected in the body. Doctors and scientists want to better understand what happens once the genetic defect or mutation occurs. They are studying the genes themselves and by studying large family groups affected by the disease. Other doctors are experimenting different medicines to help cure Marfan Syndrome.

Treatments

General Knowledge Sites:

- http://www.medicinenet.com/marfan_syndrome/article.htm
- http://emedicine.medscape.com/article/1258926-overview#showall

Medical Sources:

National Marfan Foundation
22 Manhasset Avenue
Port Washington, NY 11050-2023
Phone: 800-8-MARFAN (862-7326) (free of charge)
Fax: 516-883-8040
E-mail: staff@marfan.org
www.marfan.org

Suggested Readings/Sources

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The End