

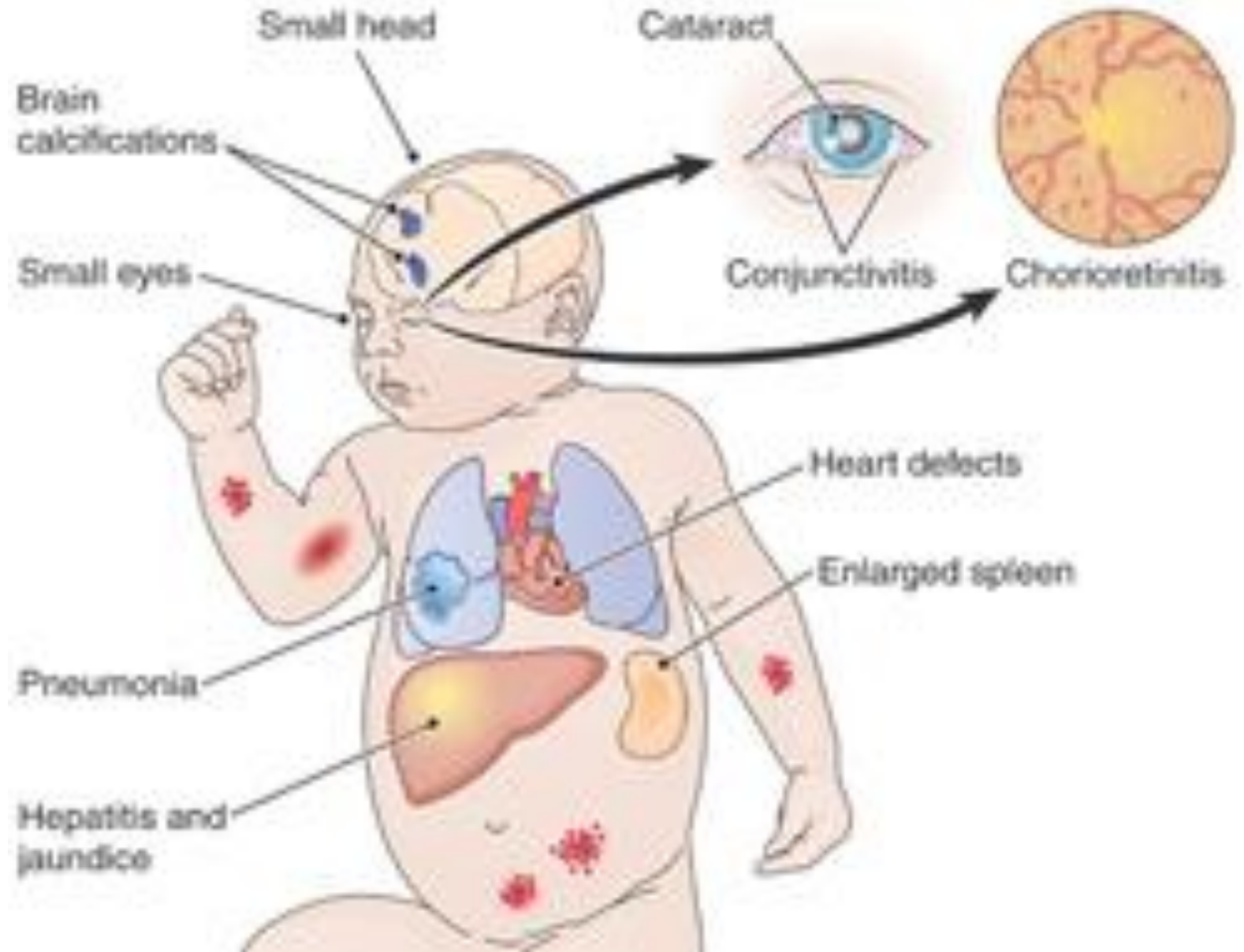


GENETIC DISEASES

DOWN SYNDROME



■ Down Syndrome



DOWN SYNDROME

How To Identify a Down Syndrome Baby



Flat nasal bridge



macroglossia
and protruding
tongue



Irregularly shaped
mouth



An almond shape to the
eyes caused by an epicanthic
fold of the eyelid



microgenia



upslanting
palpebral fissures



single palmer
crease, short
fifth finger that
curves inward



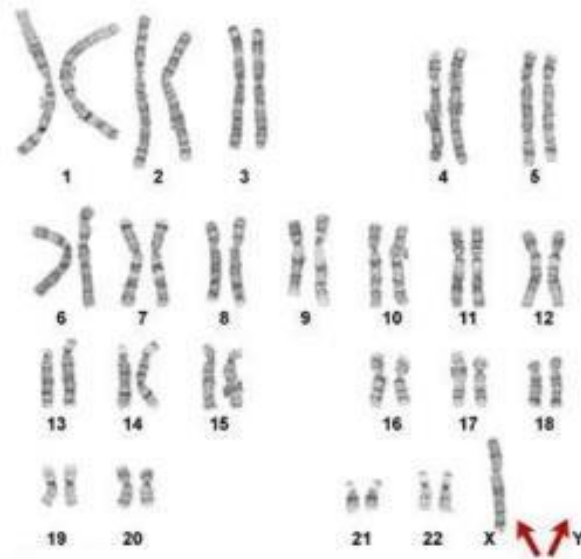
widely separated
first and second
toes and increased
skin creases



TURNER'S SYNDROME

TURNER'S SYNDROME

- Missing a X chromosome on 23rd chromosome
- Causes underdeveloped ovaries, short stature, webbed, and only found in women.
- Bull neck and broad chest. Individuals are sterile and lack expected secondary sexual characteristics.
- Mentally handicapped typically not evident.



TURNER'S SYNDROME

Turner Syndrome Phenotype: Example

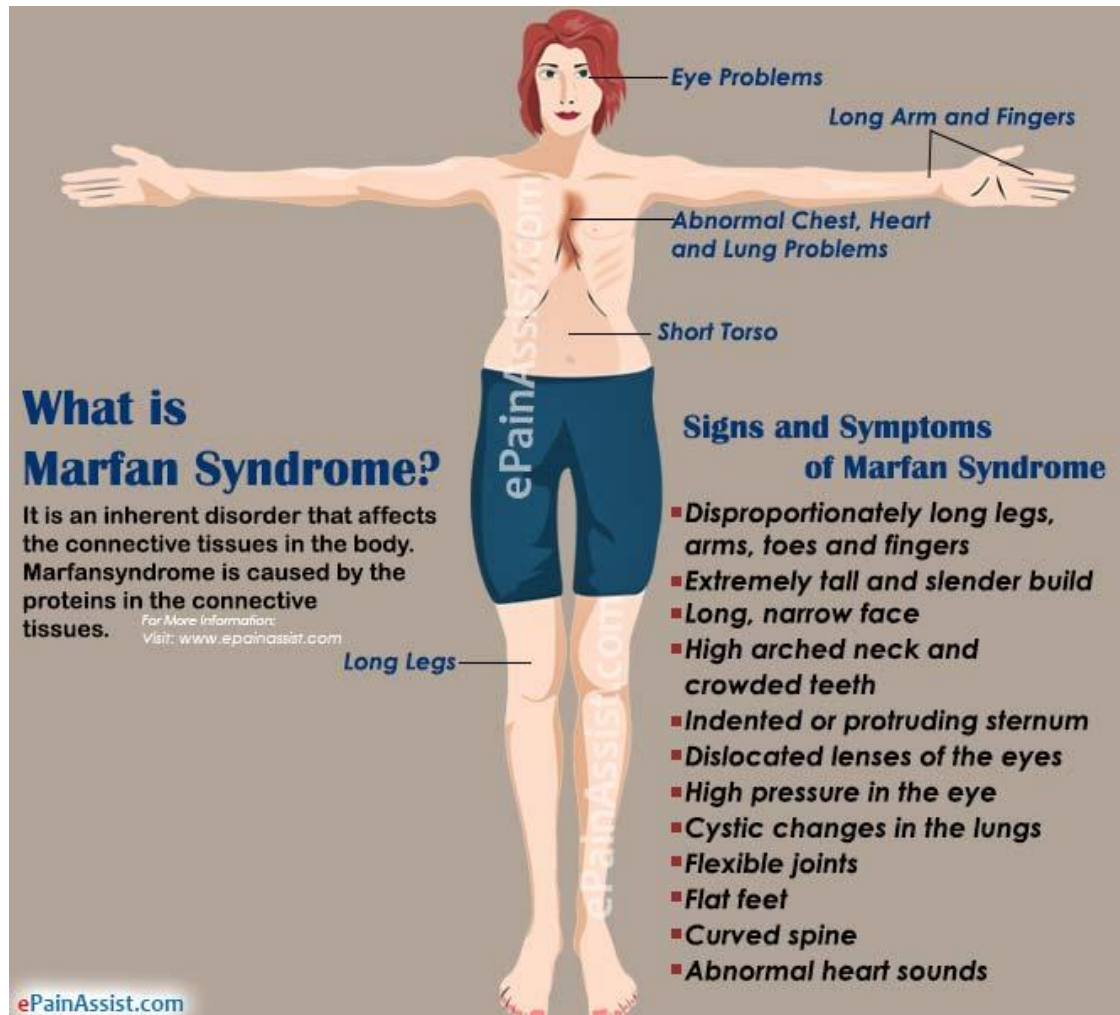


- Short stature
- Webbed neck
- Shield-like chest
- Edema of wrists and ankles

MARFAN SYNDROME



MARFAN SYNDROME



What is Marfan Syndrome?

It is an inherent disorder that affects the connective tissues in the body. Marfan syndrome is caused by the proteins in the connective tissues.

For More Information:
Visit: www.epainassist.com

Signs and Symptoms of Marfan Syndrome

- Disproportionately long legs, arms, toes and fingers
- Extremely tall and slender build
- Long, narrow face
- High arched neck and crowded teeth
- Indented or protruding sternum
- Dislocated lenses of the eyes
- High pressure in the eye
- Cystic changes in the lungs
- Flexible joints
- Flat feet
- Curved spine
- Abnormal heart sounds

ePainAssist.com

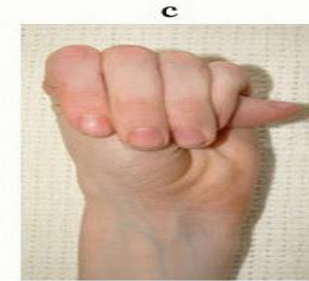
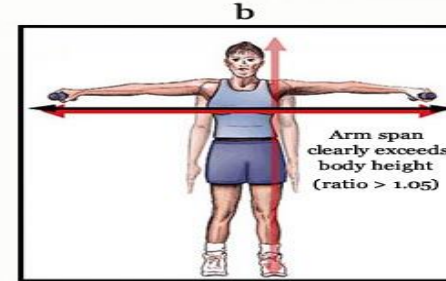
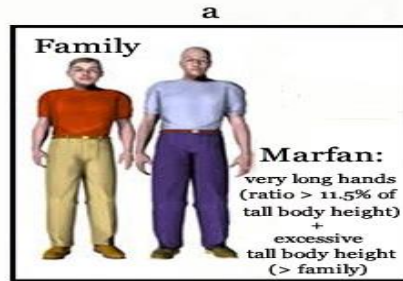
MARFAN SYNDROME

- Marfan Syndrome Hand Test -

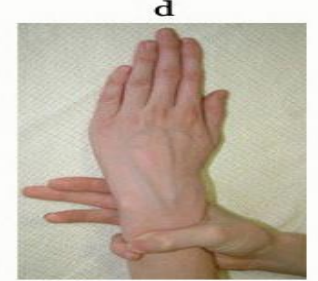
(Adults: 17 points or more required for positive testing; children: 18 points or more)

PICTURES

HAND SHAPE:



4 points

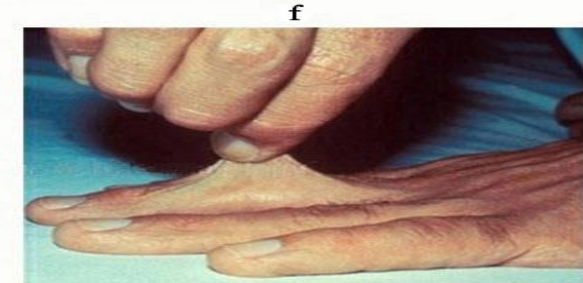


4 points

HAND SKIN QUALITY:



2 points

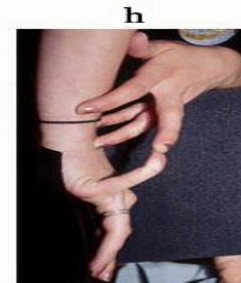


2 points

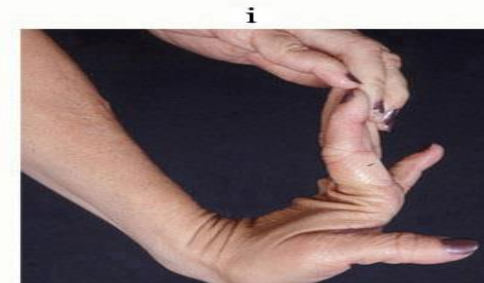
HAND MOTORICS:



1 point



1 point



1 point



1 point