Marfan Syndrome

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Description

- A hereditary disorder of the connective tissue that maintains the structure of the body and support internal organs.It affects many parts of the body including heart,blood vessels,bones,joints,eyes.Typical characteristics;
- Being tall
- Abnormally long and slender limbs,finger and toes
- Heart defects
- Lens dislocation
- Loose joints
- Crowded teeth
- Scoliosis or kyphosis

Inheritance

- Children usually inherit from one of the parents(in 75% of the cases).It is an autosomal dominant disease which means one copy of altered gene in each cell is sufficient to cause the disorder . Mutaion in FBN1 gene which lead to defective fibrilin 1 protein.Reduce amount of funtional fibrilin 1 to form microfibrils(thread like filaments).
- At least 25% of the cases result from new mutation in FBN1 gene.
- Frequency;1 in 5000 worldwide

Diagnosing

- It can be difficult as the symtopms vary from person to person.Symptoms do not always develop during childhood.Genetic testing is used to confirm the test,but it is an expensive process. Physical examination and detailed patients history and family history usually done.
- Genetic testing:
- Molecular:study single genes
- Chromosomal:analyze whole chromosome
- Biochemical:study amount or activity level of proteins

Treatment

- There is no cure for the disease. Symptomatic treatment and reducing risk of complication can be done,but it varies depending on the severity.A serious comlication is when heart and aorta are affected but other disorders caused by the disease should be treated as well to prevent further comlications. A regular check ups with specialists are essential.Physiotheray and psychotherapy are important to acheive good results.

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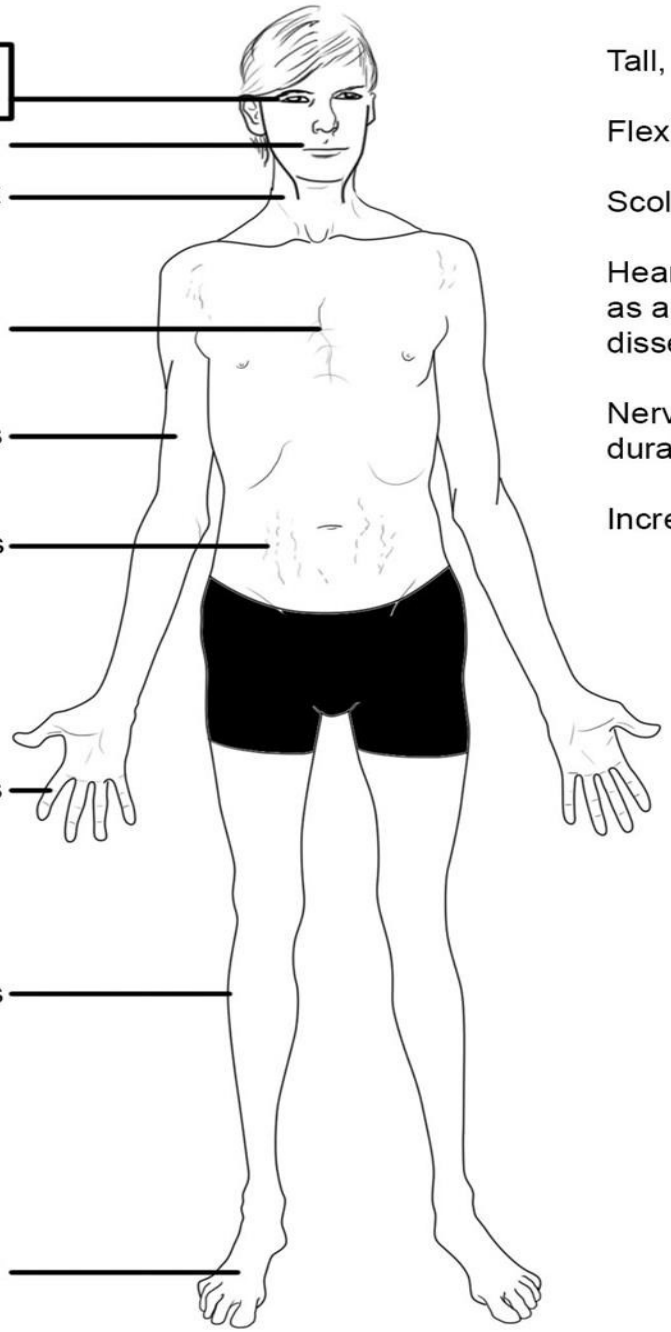

Intraocular lens dislocation
Ocular hypertension
Crowded teeth
High, arched neck
Pectus carinatum or excavatum
Disproportionately long arms
Stretch marks
Long fingers
Disproportionately long legs
Flat feet

Tall, thin build

Flexible joints

Scoliosis

Heart and blood vessel complications such as aortic dilation, aortic aneurysm,aortic dissection or mitral valve prolapse

Nervous system complications such as dural ectasia

Increased risk of pneumothorax