Marfan syndrome

Marfan's syndrome (*dolichostenomelia*) includes a wide range of symptoms; when fully expresses, the bone, ocucal and cardiovascular systems are affected. The basic criteria are:

1. ***tall figure***
2. ***long slim limbs***
3. ***long thin fingers*** (***arachnodaktyly***)
4. ***dislocation of the lens*** (***ectopia lentis***)
5. ***cardiovascular anomalies*** (mitral valve prolapse, aortic aneurism affecting Valsalva`s sinuses or aortic regurgitation, pulmonary dilatation)

It is an inherited AD disease but spontaneous mutation are common, with a frequency of approximately 1:10 000. The violinist Niccolò Paganini, for example, suffered from this syndrome.

A common causes of death at an early age are aneurysm ruptura, aortic dissection or heart failure due to aortic regurgitation (possibly mitral regurgitation).

Etiopathogenesis

- Hereditary disorder of the mesenchyme (mutations in the gene for **fibrilin 1** (FBN1; 15q21.1) – closely related to elastin, mutations in the procollagen I gene have also been found)
- Disorders of mucopolysaccharide metabolism are characteristic

The clinical picture

- **Graceful skeleton** with a shorter torso (ratio of upper and lower body segment <0.85) and noticeably long and **elongated fingers and toes**, **dolichocephaly**
- **„Thumb flag“** (thumb extending beyond the ulnar margin of the hand with the fingers clenched into a fist)
- **„Wrist symptom“** (when the wrist is freely embraced, the thumb exceeds the little finger)
- Hypotonic muscles, soft fibrous apparatus (**ligament laxity**) → scoliosis, kyphosis, chest deformities (*pectus excavatum / carinatum*)
- **Hypermobile joints**, *scapulae alatae, genua valga, genua recurvata, pedes planovalgi*
- Habitual subluxation, joint **dislocation**
- PIP flexion **contractures**, sometimes of the elbow and knee joints
- **Eye defects** (ectopia lentis, flattened cornea)
- **Aortic root dilatation**
- **Aneurysm of the ascending aorta** (Erdheim cystic medionecrosis)
- **Dental** anomalies, gothic floor
- Risk of spontaneous pneumotorax, apical lung bubble
- Striae atrophicae on the skin and recurrent hernia
- Hydroxyproline in urine

X-ray image

- Dilation of the medullary cavity at the expense of compact bone
- Metacarps, metatarsis and basic phalanges elnogated x middle and distal phalanges tend to be shorter

Therapy

- Prevention of contractures by exercising
- Unusually symptomatic surgical treatment (treatment of scoliosis and spondylolisthesis, reverse pelvic osteotomy with acetabular protrusion)
- Monitoring of cardiovascular and pulmonary disability, ev. intervention (life-threatening aortic aneurysm, spontaneous pneumothorax)

English video, definition, pathogenesis, symptoms, complications, treatment.

Prognosis

- Unfavorable (especially due to damage to the cardiovascular system):
  - Aneurysm rupture
  - Aortic dissection

    - Heart failure due to aortic regurgitation
    - Arrhytmias in mitral prolapse

- It is considered as a contraindication to pregnancy (or indication for an abortion)
- During continued pregnancy: regular check-ups and no greater physical activity, β-blockers, delivery usually by a caesarean section

Differential Diagnosis

Homocystinuria: An AR disorder presenting with Marfanoid-Habitus. Homocystinuria may have both Healthy Parents (but both of them carriers) on the Family Pedigree. Marfan Syndrome is AD so you can see at least one parent manifesting the disease.

References

Related articles

- Achondroplasia
- Tanatophoric dwarfism
- Diastrophic dypplasia
- Spondyloepiphyseal dysplasia
- Larsen syndrome
- Congenital multiple exostoses
- Fibrous bone dysplasia
- Cleidorcanial dystosis
- Morbus Albers-Schönberg
- Osteopoikillosis
- Arthrogryposis multiplex congenita
- Mucopolysaccharidosis
- Hereditary osteoonychodysplasia (nail-patella syndrom)

External links

- eMedicine: Marfan syndrome (https://emedicine.medscape.com/article/946315-overview)
- Marfanův syndrom – video (https://www.youtube.com/watch?v=p4Ev9KEyw78)

References

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