Congenital multiple exostoses

Multiple osteocartilaginous exostoses syndrome is the most common bone dysplasia with a frequency of 1:50,000, autosomal dominantly inherited, characterized by the growth of *exostoses in the metaphyseal region of* long bones, pelvis, scapula and ribs.

Patogenesis

Multiple congenital exostoses are caused by mutation of genes encoding the signaling molecule **Ihh** (*Indian hedgehog*, controlling the development of chondrocytes in the growth plate, see Congenital limb defects). Subsequently, there is an excess or lack of chondrocytes. When there is an excess of chondrocytes, osteochondroma develops, on the contrary, when they are lacking, bone growth slows down in length, the so-called angulation of the bone.

Template:Infobox - Genetic Disease

Clinical Aspect

Already in preschool age, multiple congenital exostoses manifest themselves in the form of osteochronomas, which are painful palpable formations. They can also be manifested by unequal length of the limbs or angular deformity of the limbs, the so-called pseudomadelung deformity. With this deformity, the ulna is shortened, the radium bends and ulnar deviation of the wrist occurs (Madelung's deformity), as well as lower leg deformity (genu valgum). As a result of the pressure of the osteochondroma, a dislocation occurs, which is called a subluxation. In this dislocation, the momentum is broken. In some cases, a malignant reversal in chondrosarcoma is possible.

X-Ray Image

From the radiograph, it is possible to see that the exostoses grow from the bone in a pedunculated or sessile manner. The active part here is the cartilaginous cap of the exostosis.

Therapy

In case of soft tissue irritation, surgical ablation is performed. This involves the removal of the entire cartilaginous cap with the adjacent periosteum, but the epiphyseal growth plate must not be broken. On the forearm (Chomiak et al.), timely removal of exostoses is necessary, which enables correction of the forearm deformity by fusion. At the same time, lengthening (calotaxy) and centering the ulna into the distal radioulnar joint is required. This treatment improves wrist function and cosmetic appearance.

Links

Related articles

- Achondroplasia
- Diastrophic dwarfism
- Tanatoforic dwarfism
- Pes equinovarus congenitus
- Enchondromatosis

Refrences

- DUNGL, P., et al. *Ortopedie.* 1. edition. Praha. 2005. ISBN 80-247-0550-8.