PORTUGUESE JOURNAL OF PEDIATRICS

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Hereditary multiple osteochondromas

Osteocondromatose múltipla hereditária

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Keypoints

What is known

- Hereditary multiple osteochondromas is characterized by osteocartilaginous formations in the growth plates of long or flat bones, which stabilize at puberty.
- Complications include tendon/nerve/vascular compression, short stature, pathological fractures and malignancy. Surgical intervention is rarely needed.

What is added

- Awareness about hereditary multiple osteochondromas is important in the differential diagnosis of bone tumors.

A three-year-old boy's mother reports the appearance of painless swelling at the right tibia's proximal end, with one year of evolution, without previous trauma or local/systemic inflammatory signs, namely fever or lymphadenopathy. There was no family history of similar malformations. An X-ray (Fig. 1) revealed "exostotic formations on the distal end of the external surface of the peroneal diaphysis, external femoral supracondylar region and proximal metaphyseal-diaphyseal region of the tibia". An ultrasound highlighted a "procidence of the tibial anterior tuberosity of the proximal end, without cortical discontinuity". Further studies included a full-length X-ray of the lower limbs (Fig. 2) that showed multiple exostotic formations in both femurs, tibias and fibulas. For better characterization, an MRI was performed (Fig. 3), which identified several other metaphyses' osteochondromas along the lower limbs and signs of malignancy were excluded.

The most likely hypothesis –hereditary multiple osteochondromas– is characterized by osteocartilaginous formations in the growth plates (metaphyses) of long or flat bones, due to a deleterious variant in *EXT1* or *EXT2* tumor suppressor genes[1]. It results from autosomal dominant transmission and its prevalence is 1:50.000[2]. There is no gender difference regarding prevalence, but male subjects can have a more pronounced disease. The formations appear and increase in size and number during the first decade, stabilizing at puberty with the closure of growth plates. However, the number, size and location of osteochondromas vary[3]. Generally asymptomatic and symmetric, the predominant locations are the femur (30%), tibia (20%), radius/ulna (13%) and fibula (13%); hands are also commonly affected[4]. The diagnosis is based on radiological findings of at least two osteochondromas of the juxta-epiphyseal region of long bones and, in the majority of patients, a positive family history or a documented mutation in one of the *EXT* genes, if available. Almost 10% of affected individuals have no family history. Clinical and radiological findings can be supplemented with histological

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Received: 11-11-2023
Accepted: 13-11-2023
https://pjp.spp.pt

Available online: 07-02-2024
Port J Pediatr. 2024;55(1):69-71
DOI: 10.24875/PJP.M23000127

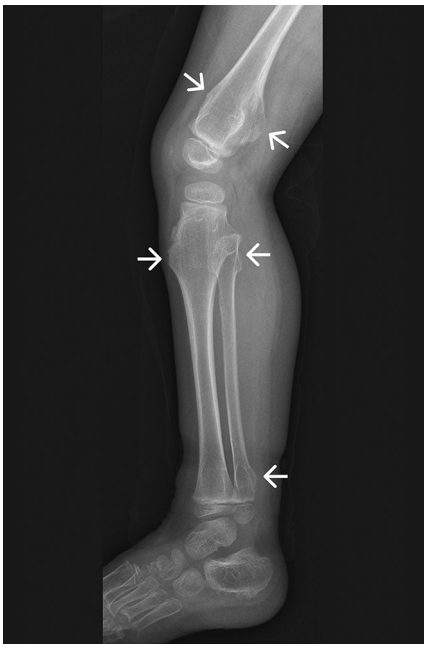

Figure 1. Right lower limb X-ray showing exostotic formations on the distal femur, proximal tibia and distal fibula (white arrows). Note the typical sessile lesions on the surface of the bone which point away from the joint. There is an associated widening of the diametaphysis resulting in an erlenmeyer flask deformity.

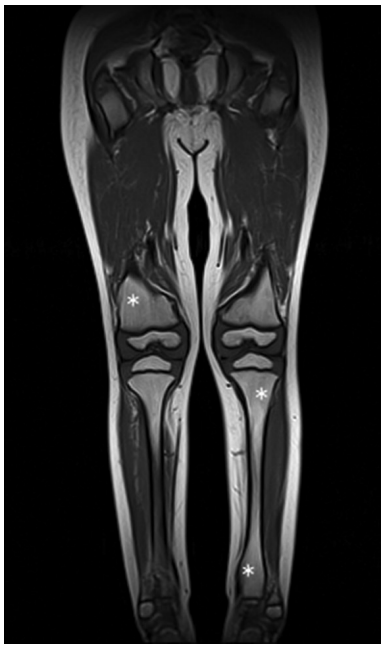

Figure 3. MRI coronal plane of the lower limbs showing multiple exostotic formations (white asterisks), highlighting the shape of the osteochondromas and its continuity from the cortical bone. This allows a direct study of the cartilage cap and the soft tissues surrounding the lesions

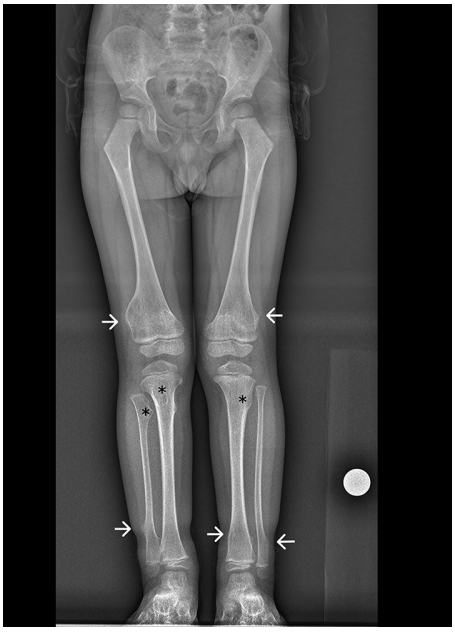

Figure 2. Lower limbs full-length X-ray showing multiple exostotic formations in both femurs, tibias and fibulas (white arrows and black asterisks). Note the characteristic metaphysis widening.

evaluation of the osteochondromas, although not mandatory for diagnosis[4,5]. Complications include tendon/nerve/vascular compression, functional impairment, short stature, fractures and malignancy (1-5%). Surgery may be necessary if any complications arise[1-3]. Differential diagnoses such as simple or aneurysmal cyst, metachondromatosis (most frequently small osteochondromas in the hands and feet, predominantly in digits and toes, that point towards the adjacent growth plate and tend to spontaneously decrease in size or regress), enchondromatosis (predominantly unilateral cartilaginous medullary tumors of short tubular bones), periosteal chondroma (typically includes bone cortex erosion), osteoid osteoma, osteoblastoma and chondroblastoma (usually later and symptomatic onset) must be excluded[2,5].

Funding

None.

Conflicts of interest

None.

Ethical disclosures

Protection of human and animal subjects. The authors declare that no experiments were performed on humans or animals for this study.

Confidentiality of data. The authors declare that they have followed the protocols of their work center on the publication of patient data.

Right to privacy and informed consent. The authors have obtained the written informed consent of the patients or subjects mentioned in the article. The corresponding author is in possession of this document.

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