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Case Report

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A Case Report of Hereditary Multiple Exostoses

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ABSTRACT

Hereditary multiple exostoses (HME), also known as hereditary multiple osteochondromas, is a rare autosomal dominant genetic disorder featuring multiple exostoses (osteochondromas) mostly diagnosed in childhood, they arise near the growth plates of bones such as ribs, pelvis, vertebrae and predominantly long bones. CT and MRI imaging are required to examine anatomically difficult regions such as the pelvis and thorax, and X-rays are additionally helpful in finding associated deformities. In this article and through a case report, we mainly outline the input of imaging in the diagnosis, prognosis and therapeutic management of such condition.

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Introduction

Hereditary multiple exostoses (HME), also referred to as hereditary multiple osteochondromas, is a rare autosomal dominant genetic disorder characterized by multiple exostoses (osteochondromas) most commonly diagnosed in childhood [1], it develops near the growth plates of bones such as the ribs, pelvis, vertebrae and especially the long bones.

The disease has a variety of clinical features, including chronic pain syndromes, limited range of motion, limb deformity, short stature, scoliosis and neurovascular impairment [2]. Malignant transformation of exostosis is rarely seen.

Axial imaging by computed tomography (CT) or magnetic resonance imaging (MRI) is usually mandatory to study difficult anatomical regions such as the pelvis and thorax, and X-rays are also useful to detect and study the HME associated deformities. Although no medical treatment is currently available, surgery is only recommended in symptomatic exostoses or in cases where a malignant transformation is suspected.

Case Report

We report a case of a nine-year-old boy with a progressive, painless left thoracic palpable mass evolving since the age of 7 years without any particular irradiation (figure 1). No similar cases were found in the patient's family history. Radiological examination found multiple osteochondromas, the largest tumor was located in the 7th left rib, A thoracic CT scan was then performed and showed two growths of bone extending outwards and originating from the anterior arches of the 7th both left and right rib measuring respectively 3.2 x 3.3 cm and 1. 6x 0.5 cm (figure 2 and 3), they are covered by a dense and irregular cartilaginous cap measuring 3 mm on the left and 1.5 mm on the right (figure 4).

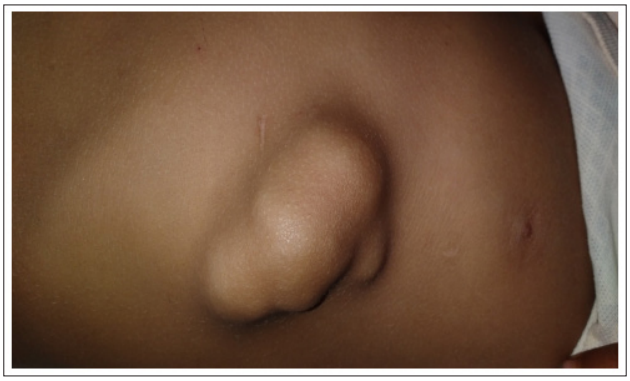

Figure 1: An Image Showing Left Thoracic Palpable Mass

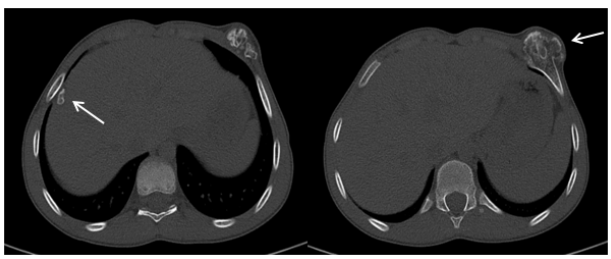

Figure 2: Thoracic CT scan (Axial) Showing Two Growths of Bone Extending Outwards and Originating from the Anterior Arches of the 7th both Left and Right Rib.

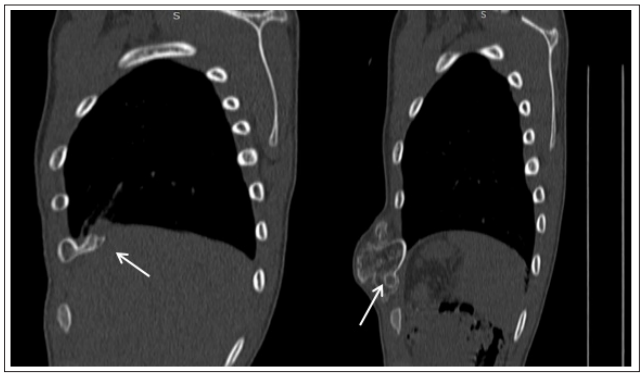

Figure 3: Thoracic CT scan (sagittal) Showing two Growths of Bone Extending Outwards and Originating from the Anterior Arches of the 7th both Left and Right Rib

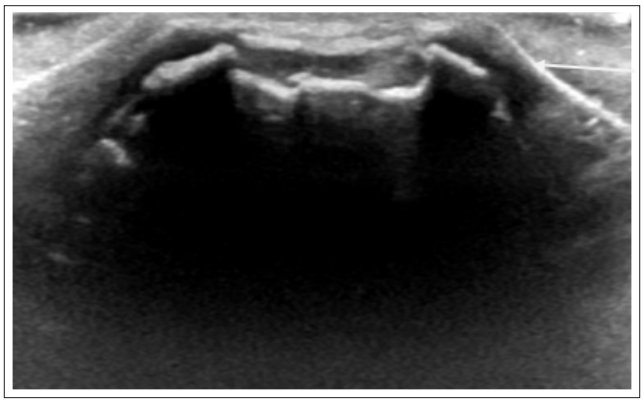

Figure 4: Ultrasound Examination of the Thoracic Mass Showing the Cartilaginous Cap (Arrow)

The patient was referred to the pediatric surgery department for further follow-up.

Discussion

Hereditary multiple exostoses (HME) is a rare genetic condition in which several benign cartilage tumors arise from the perichondrium and frame the cartilage growth, it is carried down in families as an autosomal dominant genetic trait. Estimates of its prevalence have ranged from between 0.9 and 1.4 in 100 000 in two european populations, to 100 in 100 000 in a small closed population of the Chamorros of Guam, some reports have documented a male predominance in the HME [1, 3].

Patients are commonly diagnosed before the age of 12. Usually, they come to the physician's attention following the appearance of a palpable mass near the joints. Shoulders, knees, wrists and ankles are the most implicated joints in the diagnosis, however, a variety of clinical manifestations including chronic pain syndromes, restricted range of motion, limb deformity, short stature, scoliosis and neurovascular alteration can be present.[4, 5]

The majority of patients are asymptomatic at birth, so the diagnosis can only be pursued by genetic screening of newborns from affected families, the median age of diagnosis is 3 years.; fifty percent of the patients have a noticeable tumor and are diagnosed by the age of 5 and 80% by the age of 10. In most cases, patients are diagnosed by the age of 10 to 12.

Computed tomography (CT) scan or magnetic resonance imaging (MRI) is essentially required to investigate difficult anatomical areas such as the pelvis and the thorax, the spine should also be examined in HME patients with an MRI of the entire spine and the screening must be performed once every two years during the follow-up period.

Performing an MRI is important for planning the surgical removal of exostosis; magnetic resonance imaging can emphasize the shape of the exostosis and its continuity from the cortical bone; but above all, it provides a detailed and precise analysis of the cartilaginous cap and soft tissues around the lesions. Cartilage cap can also be evaluated using ultrasonography and CT scan images but MRI provides a much better resolution, measurement of the cartilage cup is a reliable method to forecast malignant transformation of osteochondromas. The measurement should be done from the osseous interface of the exostosis stalk to the edge of the cartilage cap in its largest portion.

In childhood and adolescence, the cap thickness can be more than 3 cm. On the other hand, in adults, malignancy should be suspected when the thickness is more than 1–2 cm. Various authors have set the limit at 1 or 2 cm; Kok stated that a limit value of 2 cm is sufficient to diagnose malignancy with 95% specificity and sensitivity.[5, 6]

Additional radiological signs of malignant transformation are an irregular surface of the cap, a lytic foci inside the exostosis or near the bone, and the presence of soft tissue mass surrounding the exostosis, particularly if it contains scattered or irregular calcifications.

To the best of our knowledge, there is still no available medical treatment for HME, Surgical excision was carried out by removing the exostosis at the base of the bone, with subsequent removal of the cartilage cap, usually when the cartilage cap is entirely removed, there are no recurrences, however, complete removal in some regions could lead to the necessity of reconstruction techniques like bone grafting and internal fixations. [7, 8]

Conclusion

Hereditary multiple osteochondromas is a genetic condition for which the orthopedist, geneticist, radiologist, and pediatrician must all study and treat the disease.

The complexities and unresolved issues associated with HME are a challenging task for both researchers and clinicians, despite the significant progress that has been made in recent years.
Precise monitoring and proper surgical management are today imperative to guarantee the best possible prognosis for patients with HME, which usually have a low quality of life and several disabilities caused by their pathology.

Disclosure of Interest

The authors declare that they have no conflicts of interest concerning this article.

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