

Hereditary multiple osteochondromatosis in children

Osteocondromatosis múltiple hereditaria en niños

Geiser Reinaldo Hernández Villard¹, Margionnis Torres Silot², Edwin Paulino³

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ABSTRACT

Introduction: Hereditary multiple osteochondromatosis is a rare entity characterized by the growth of bony masses called osteochondromas, which constitute the most frequent benign cartilaginous tumors in children, accounting for 10%-15% of all bone tumors. **Objective:** To present a clinical case with a presumptive diagnosis of hereditary multiple osteochondromatosis.

Case presentation: We report the case of a 12-year-old male patient who presented with slight pain in the right knee, more accentuated while walking. On physical examination, a painless enlargement of the right knee towards the external aspect was noticed. On palpation, a hard, firm mass was palpable. Similar tumors were also found at the level of the left knee, both wrists and ankles. X-rays of both knees, ankles and wrists were taken, showing bone lesions in the metaphysis, predominantly in long bones, demonstrating cortical and medullary continuity, pointing to a benign lesion. Surgical excision of the tumors located at the distal end of the ulna and the distal end of the left tibia and fibula was performed and the biopsy confirmed the diagnosis.

Conclusions: The knowledge of the spectrum of radiological findings of this lesion is essential for the multidisciplinary team, especially to make the differential diagnosis (osteochondroma vs chondrosarcoma) and to implement the proper management.

Keywords: osteochondromatosis; cartilaginous tumors; radiological findings.

RESUMEN

Introducción: La osteocondromatosis múltiple hereditaria es una entidad poco frecuente que se caracteriza por el crecimiento de masas óseas denominadas osteochondromas, los cuales constituyen los tumores benignos cartilaginosos más frecuentes en niños, suponiendo el 10 %-15 % de todos los tumores óseos. **Objetivo:** Presentar un caso clínico con diagnóstico presuntivo de Osteocondromatosis Múltiple Hereditaria. **Presentación del caso:** Se reporta el caso de un paciente masculino de 12 años que acude a consulta por presentar ligero dolor en la rodilla derecha, más acentuado con la marcha. Al examen físico se identifica un aumento de volumen de la rodilla derecha hacia cara externa, sin dolor. A la palpación se palpa masa dura, no móvil. También se constatan tumoraciones similares a nivel de rodilla izquierda, ambas muñecas y tobillos. Se realizan radiografías de ambas rodillas, tobillos y muñecas, observándose lesiones óseas en la metáfisis, a predominio de huesos largos, con continuidad cortical y medular, signo radiológico que habla a favor de una lesión benigna. Fue posible realizar el tratamiento quirúrgico, escisión tumoral del extremo distal del cúbito y del extremo distal de la tibia y fibula izquierdas. **Conclusiones:** El conocimiento del espectro de hallazgos radiológicos de esta lesión constituye un reto para el equipo multidisciplinario, pero al mismo tiempo brinda la cultura radiológica a los médicos generales para poder realizar un correcto diagnóstico diferencial (osteochondroma vs condrosarcoma) y a los imagenólogos a contribuir al mismo, y de esta forma ofrecer al paciente una terapéutica correcta.

Palabras clave: osteocondromatosis; tumores cartilaginosos; hallazgos radiológicos.

¹Bachelor in Imaging and Radiophysics. Punta Gorda Community Hospital. Punta Gorda. Belize. ORCID <https://orcid.org/0000-0002-7720-2845>
Correspondence: geisereinaldo@gmail.com

²Bachelor in Imaging. Punta Gorda Community Hospital. Punta Gorda. Belize

³Bachelor in Imaging. Technical Chief of Staff. Radiology Service. Punta Gorda Community Hospital. Punta Gorda. Belize

INTRODUCTION

The encondromatoses comprise a heterogeneous group of syndromes, characterized by the presence of multiple encondromas.¹ The first description of a patient with multiple osteochondromatoses is attributed to Hunter in 1786. Boyer, in 1814, published the first description of a family with hereditary multiple osteochondromatoses, followed by Guy's description of a second family in 1825.²

Multiple Hereditary Osteochondromatoses (MHO) also called multiple cartilaginous exostosis, multiple hereditary exostosis or congenital osteochondromatoses, is an autosomal dominant entity characterized by the growth of multiple benign tumors called osteochondromas and it is usually diagnosed in the first decade of life. Its frequency rates from 1 x 50 000 to 100 000 population and it affects more males than females in a 2 to 1.5 ratio.^{3,4}

MHO is known to have a genetic basis. EXT1 and EXT2 genes have been located on chromosomes 8 and 11, respectively; while EXT3 has been found on the short arm of chromosome 19. Some studies have shown that mutations in EXT1 and EXT2 are responsible for MHO.^{5,6}

The imaging gold standard to evaluate the patient with osteochondromas is plain radiography, except in cases without mineralization, in which computed tomography (CT) should be used. In most cases of osteochondromas with a small and asymptomatic lesion, conservative treatment is recommended. If the lesion is extensive and symptomatic, surgical treatment is recommended.⁶⁻⁸

The occasional presentation of this condition, in addition to the few existing case reports in the Belizean literature, and the motivation to direct the management of the patient towards a correct therapy, justify this presentation; which aims to present a clinical case with presumptive diagnosis of MHO.

CASE PRESENTATION

We report the case of a 12-year-old male patient, date of birth August 2, 2011, who is consulted for presenting slight pain in the right knee, more accentuated with walking, which does not radiate and barely relieves with the use of non steroidal anti inflammatory drugs (NSADs). This pain has been present for two months and is associated with an increase in volume near the right knee.

In her prenatal history (G₁ P₀ A₀), it is noted that her mother suffered from urinary tract infection in the third trimester of pregnancy and no other conditions were reported. She was a product of delivery at 38.2 weeks of gestational age with a weight of 3,090 grams, adequate for her gestational age, with normal Apgar score at birth. No postnatal pathological history is reported and a complete vaccination schedule is on record. There is no family history of pathology.

Physical examination

Nutritional assessment: weight of 46 kg and a height of 136.3 cm, for a body mass index of 24.76. Increase in volume of the right knee towards the external aspect. On palpation, a hard, not tender, firm mass adhered to deep plane, was found; similar findings were located in the left knee, both wrists and ankles (Figure 1 and 2).

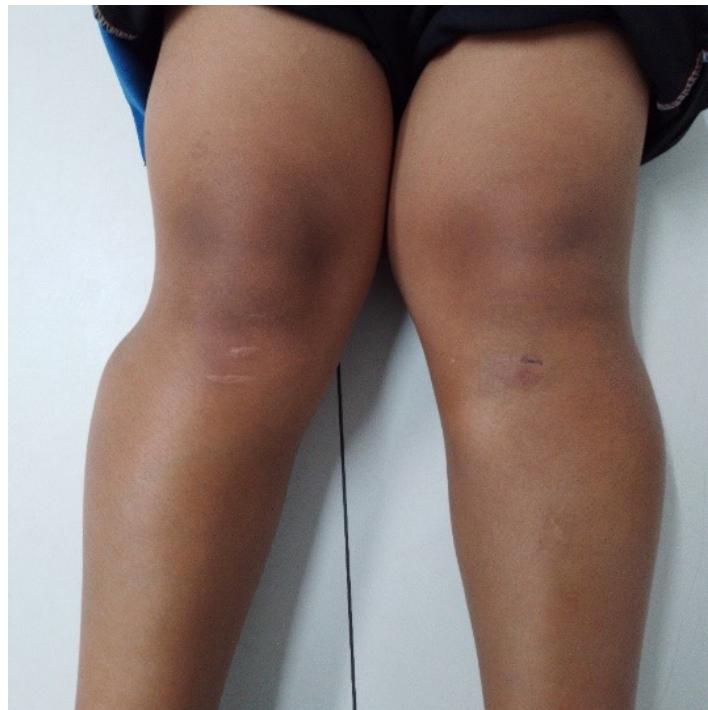


Fig. 1. Bilateral volume increase and deformities at the level of the proximal end of both tibias, close to the knees, more accentuated on the right side.



Fig. 2. Increase in volume and deformity at the level of the distal end, towards the medial aspect of the right forearm.

X-rays were taken of both knees (Figure 3), ankles (Figure 4) and right forearm (Figure 5), visualizing bone lesions in the metaphysis, predominantly in long bones, showing cortical and medullary continuity, a radiological signs suggestive of a possible bone tumor.



Fig. 3. Simple X-ray of both knees showing metaphyseal bone lesions, with cortico-medullary continuity, well delimited, no calcifications, no soft tissue mass at the level of the distal ends of the right and left femur, in addition to the proximal ends of both tibia and fibula.



Fig. 5. Simple X-ray of the right forearm, showing prominent bone lesions.

MHO is suggested as a possible diagnosis. Surgical excision of the tumors located at the distal end of the ulna and the distal end of the left tibia and fibula was performed. The biopsy confirmed the diagnosis of MHO.

MHO is proposed as a possible clinical-radiological diagnosis. It was possible to perform surgical treatment, using this therapeutic variant for those affected who present symptoms and/or deformities caused by osteochondromas. Furthermore, this therapeutic option is ideal when possible compression injuries of neurovascular structures are suspected and when extracting a sample for evaluate the histology of the tumor. All of these previously stated criteria justified the surgery performed on this patient.



Fig. 4. Simple X-ray of both ankles showing bone lesions.

DISCUSSION

The interest of this case lies in the necessary differential diagnosis of bone lesions, whether or not they are of tumor origin, regardless of whether they are discovered by chance or as a result of a symptom.

Sáez Moreno MA, et al⁹ sustain the idea that benign bone tumors can appear in any location and can be dangerous due to compression of healthy tissue. Their incidence depends on the type of tumor and they most frequently appear between 10 and 30 years of age, being osteochondroma the most common. On the other hand, Santos-Guzmán J, et al⁶ reported that about two thirds of patients have a family history of the disease and that MHO affects men 1.5 times more often than women. However, other authors such as Guerrero Tamayo P, et al² state that it seems to affect both sexes in a similar way.

In our particular case, we agree with Sáez Moreno MA, et al,⁹ because this type of lesions can appear at any anatomical point, generally at the distal ends of the long bones. On the other hand, the possible neurovascular complications associated with this entity could be clinically confirmed due to the palpated bone deformities. Furthermore, it is proposed that the appearance of this disease is very common in the age range from 10 to 30 years, corresponding to this patient.¹⁰

Penetrance is 96% in women and 100% in men. The affected person has a 50% probability of transmitting the trait to his/her offspring. The average age of presentation of MHO is 3 years and 96% of cases have been diagnosed before 12 years of age.^{2,6}

In our case we include the family history of the father, because he presented similar bone lesions, without specifying the name of the disease, which began to appear at an early age. The condition is characterized by the development of two or more osteochondromas. The most common location of osteochondromas is in long bones, mainly in the femur, distal end, tibia and fibula, proximal ends; however, bones of the spine, pelvis, ribs and scapulae can also be affected.^{6,11}

They are particularly frequent around the knee, where they are more likely to suffer malignant transformation. For some specialists, the probability of affecting the knee is described as 94%. In this case there is considerable involvement of the knee, presenting evident clinical and radiologically demonstrated deformities, which make it difficult for the patient to walk correctly.¹¹⁻¹³

The location of the tumors and the signs and symptoms presented by the diagnosed patient, correspond to those described in the literature on hereditary multiple exostoses. The diagnosis is established clinically and radiologically. In case of diagnostic doubt, in order to provide more elements, from the imaging point of view, it is necessary to perform high-

tech studies, CT and Magnetic Resonance Imaging (MRI), the latter provides an excellent demonstration of arterial and soft tissue affection, if any.^{5,7}

Ultimately, excisional biopsy of the injured area is suggested. Deformities of the forearm and ulnar shortening occur in 30% to 70% of patients with MHO, producing angular deformities and loss of mobility.⁹ The treatment that currently has demonstrated effectiveness and better results is surgery. For this particular case, a corrective osteotomy was performed. This is reserved for those affected who present symptoms and/or deformities caused by osteochondromas.² Typically, surgical procedures performed include tumor excision, corrective osteotomies, procedures to align or enlarge bones, epiphysiodesis, and hemiepiphysiodesis.^{9,10}

Palomo H, et al¹⁴ stated that given the complexity of the deformity at the level of the forearm, in some cases it is necessary to combine the osteotomy with other surgical procedures (ulnar lengthening plus radial hemiepiphysiodesis, radial osteotomies) as a guideline to follow in the correction of deformities due to MHO of the forearm. In our case, excision of the ulnar deformity tumor was performed, due to the complexity of the lesion and deformity present in the patient's right forearm.

The patient is currently recovering from surgery, pending post-surgical radiological follow up investigations and waiting for further surgical treatment of the lower limbs.

CONCLUSIONS

MHO is an infrequent condition associated with skeletal deformities. Due to its possible bone complications, associated neurological and vascular disorders, as well as the risk of malignancy, its early diagnosis and the timely correction of the bone deformities it causes are of importance. Its diagnosis is basically clinical and radiological.

The knowledge of the spectrum of radiological findings of this lesion is essential for the multidisciplinary team, especially to make the differential diagnosis (osteochondroma vs chondrosarcoma) and to implement the proper management.

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Authorship

Geiser Reinaldo Hernández Villard (Conceptualization. Visualization. Formal analysis. Research. Methodology. Supervision Original drafting-drafting. Drafting, revision and editing).

Margionnis Torres Silot (Conceptualization. Research. Methodology. Supervision Original drafting-drafting. Writing, proofreading and editing).

Edwin Paulino (Research. Methodology. Writing, revising and editing).

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