JOURNAL OF CLINICAL AND HEALTH SCIENCES

CASE REPORT

Skeletal Deformity in a Preschool Child: Recognizing Enchondromatosis (Ollier Disease) in a Primary Care Setting

Ahmad Muslim Ahmad Roslan[1], Farnaza Ariffin[2], Mohd Farhan Hamdan[3]

1 Mahmoodiah Health Clinic, Johor Bahru, Johor, Malaysia
2 Department of Primary Care Medicine, Faculty of Medicine, Universiti Teknologi MARA (UiTM), Sungai Buloh, Selangor, Malaysia
3 KPJ Tawakkal KL Specialist Hospital, Wilayah Persekutuan, Kuala Lumpur, Malaysia

Received
3rd December 2021
Accepted
17th May 2022
Received in final revised form
2nd July 2024
Published
1st September 2024

Corresponding author:
Dr Ahmad Muslim Ahmad Roslan,
Family Medicine Specialist,
Mahmoodiah Health Clinic,
Jalan Mahmoodiah,
80000 Johor Bahru, Johor, Malaysia.
Tel: 019-2889685
Email: muslim.best@gmail.com

ABSTRACT

Enchondromatosis is a rare disease which has a risk to transform into secondary chondrosarcoma. This is a 4-year-old girl presented with a one-year progressive history of her right ankle joint deformity. Plain radiograph showed bone lesion over right distal fibula. Magnetic resonance imaging suggested features of enchondroma with multiplicity lesions. Skeletal survey showed multiple enchondromas over right distal ulnar, proximal femur, pelvis and both proximal fibulas and feet. She was planned for corrective osteotomy to prevent further progression of the deformity. She has regular follow-up under paediatric orthopaedic team and primary care physician for long term monitoring and overall health care. This case demonstrates the importance of recognizing the features of non-malignant bone tumour clinically and radiologically for early detection and referral. Long term follow-up and monitoring is necessary due to the risk of malignant transformation in enchondromatosis.

KEYWORDS: Enchondromatosis, Ollier Disease, Chondrosarcoma, Preschool Child, Primary Care

INTRODUCTION

Enchondromatosis, commonly presented as Ollier disease (enchondromatosis Spranger type I) is a rare disease with estimated prevalence of 1 in 100,000 [1-3]. It is characterised by cartilage lesions with unilateral predominance and the features can be extremely variable in different cases [1]. The condition is often discovered incidentally at first decade of life as the patient is usually asymptomatic [1, 4]. Patients can present with skeletal deformities, limb length discrepancy and pain. Ollier disease has a risk of malignant transformation into chondrosarcoma ranging from 5-50% [1, 3]. Its prognosis is variable depending on its extent and severity of the disease [2]. This case discusses a child with right ankle joint deformity presenting at a primary care clinic and subsequently was diagnosed with enchondromatosis. It highlights the importance of recognizing the radiological features of benign tumour bone, the more serious differential diagnoses, natural course of the disease and management in a primary care setting.

CASE PRESENTATION

A 4-year-old girl was brought by her mother to a primary care clinic with a complaint of deformity over her right ankle joint. Her mother noticed this deformity since the child was three years old but did not seek any medical attention at the time as the child was, otherwise, asymptomatic and healthy. For the past few months, the deformity looked more obvious. The mother noticed a swelling over the medial aspect of the right ankle joint with external angulation. The child did not complain of pain and there was no difficulty in walking, running or

Vol 9(2) (2024) 65-70 | jchs-medicine.uitm.edu.my | eISSN 0127-984X
https://doi.org/10.24191/jchs.v9i2.17265

jumping. There was also no episode of fall or trauma. She had no fever or constitutional symptoms. The girl was the second of twins, born at 36 weeks of gestation through caesarean section due to precious pregnancy. There was no complication during antenatal and postnatal period. Her developmental milestone was comparable with her twin brother which was according to age. Her twin brother did not have similar problems and there was no family history of musculoskeletal disease.

Clinically, she was an active girl with no syndromic facies. Her weight (10.6 kg) and height (90 cm) were below 3rd centile while her BMI (13 kg/m^2) was just above 3rd centile for her age. She has normal gait. On examination of her right ankle, there was a bony swelling over medial malleolus. The swelling was fixed to the bone, non-tender, normal temperature and no skin changes overlying it. There was also valgus deformity of the right ankle joint involving hind foot with concomitant plano valgus. The right ankle joint had restricted dorsiflexion but normal plantarflexion movement. Examination of her left ankle joint showed mild valgus deformity with short and small third toe. No restriction in the dorsiflexion and extension of the left ankle joint. Sensory and power of both lower limbs are normal, and no obvious similar deformity was noted over other joints, toes or fingers.

Plain radiographs of both ankles showed multiple radiolucent lesions with oval shape involving the metaphysis of the right distal fibula. No similar lesion found over left ankle (Figure 1 and Figure 2). Skeletal survey revealed similar lesions with variable degree at right distal ulnar, proximal femur and pelvis besides both proximal fibulas and feet (Figure 3). Following the radiograph findings, a Magnetic Resonance Imaging (MRI) of the right ankle joint was done and showing an expansile intramedullary lobulated lesion at the distal fibula and lateral malleolus with involvement of distal diaphysis, metaphysis and epiphysis. It extends through the bony cortex and causing bone expansion (Figure 4). These radiological findings were discussed with the radiologist and the oncology team, and they agreed on the diagnosis of enchondromatosis due to its typical radiological presentation. The child was referred to paediatric orthopaedic team for further management and follow up. Due to the significant malalignment of the right ankle joint, a corrective supramalleolar osteotomy was planned to reduce the progression of the deformity and complications.

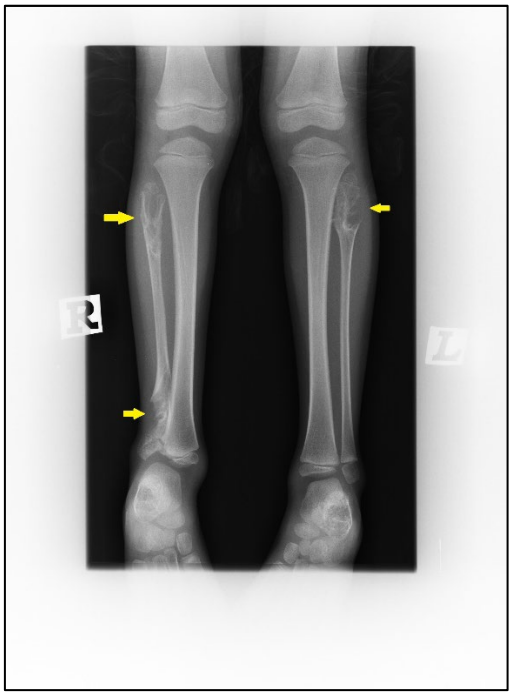

Figure 1 AP bilateral tibia/fibula radiograph. Expansile intramedullary lucent lesion in both proximal fibular and right distal fibular metadiaphysis with associated endosteal scalloping (yellow arrow)

Vol 9(2) (2024) 65-70 | jchs-medicine.uitm.edu.my | eISSN 0127-984X
https://doi.org/10.24191/jchs.v9i2.17265

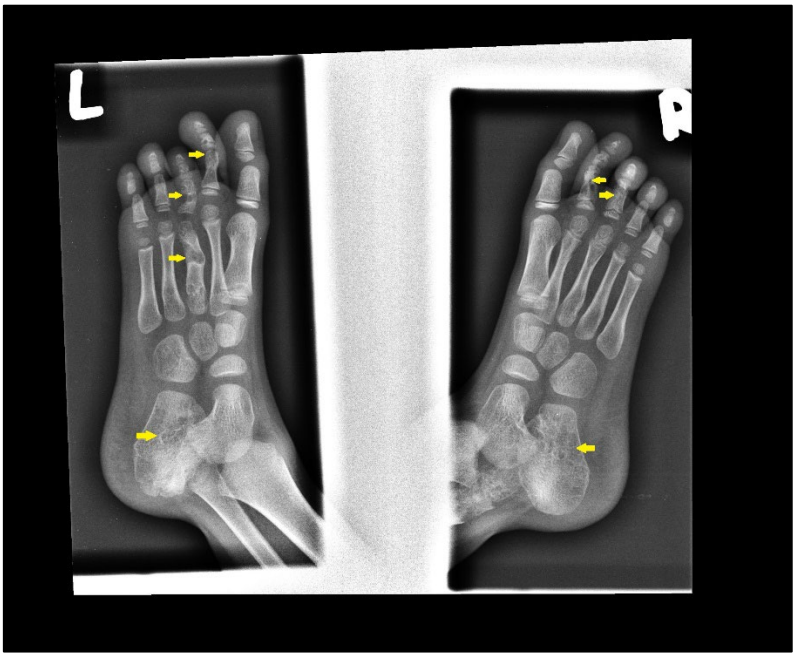

Figure 2 Oblique bilateral feet radiograph. Multiple small intramedullary lucent lesions of varying sizes in the 3rd left metatarsal, 2nd and 3rd phalanges bilaterally and both calcaneus (yellow arrow)

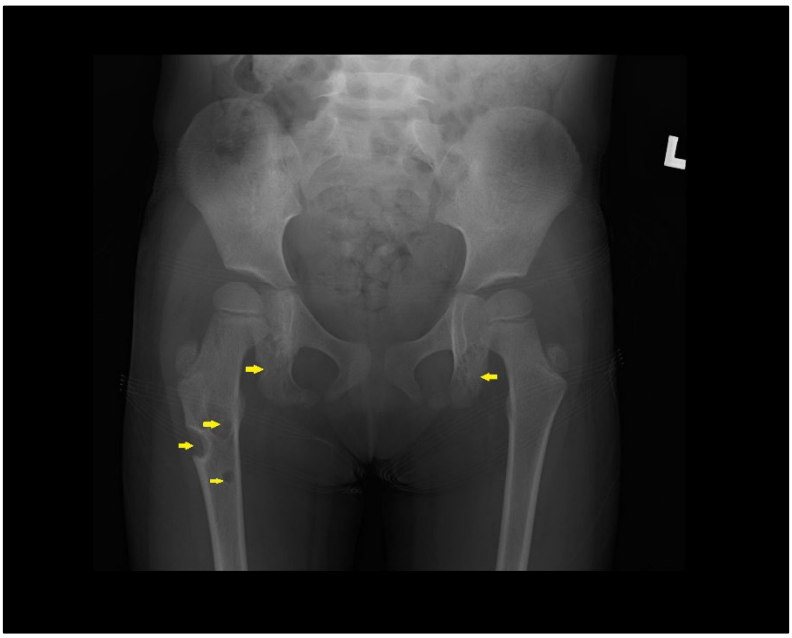

Figure 3 AP pelvis radiograph. Several lucent lesions in the proximal right femur diaphysis with slight expansion of the femur. Multiple small lucent lesions in both inferior pubic rami are also present (yellow arrow)

Vol 9(2) (2024) 65-70 | jchs-medicine.uitm.edu.my | eISSN 0127-984X
https://doi.org/10.24191/jchs.v9i2.17265

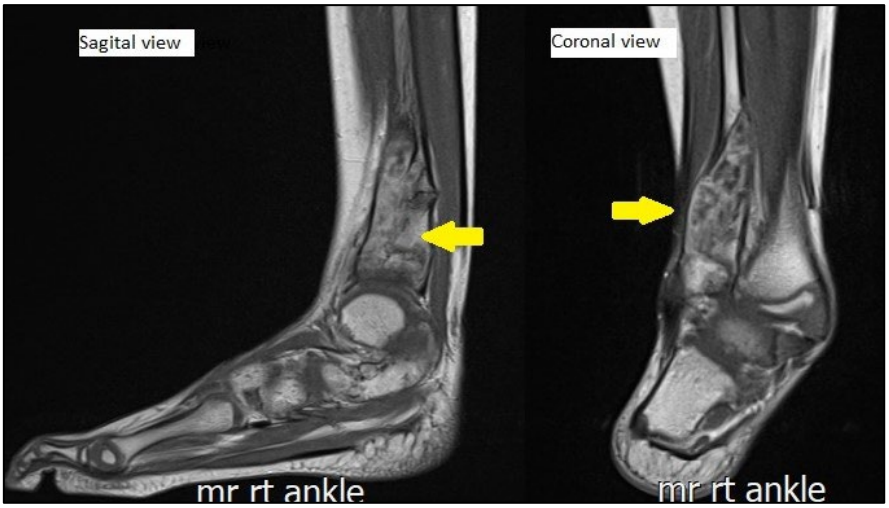

Figure 4 Coronal/sagittal right ankle MRI post gadolinium administration. Expansile intramedullary lobulated lesion at the distal fibula and lateral malleolus with involvement of distal diaphysis, metaphysis and epiphysis (yellow arrow).

DISCUSSION

Enchondroma is a common non-malignant bone tumour in children which accounts for about 15.6% of benign tumour bone [4]. It is included as cartilage-forming tumour which develops in the intramedullary region, mostly at the metaphysis and diaphysis of the long and short bones [4]. It typically affects the phalanges and metacarpals, followed by tibia and femur, then pelvis, fibula and humerus [2]. Enchondroma usually occurs as a solitary lesion [4]. When there are three or more bones affected with enchondroma, it is called multiple enchondromas or enchondromatosis [2, 4].

According to modified spranger classification system, there are eight subtypes of enchondromatosis [2]. Ollier disease (enchondromatosis with asymmetrical distribution) and Mafucci syndrome (enchondromatosis with hemangiomas) are the most common subtypes of enchondromatosis [4]. While Ollier disease and Mafucci syndrome are non-hereditary diseases, other subtypes are either autosomal dominant or recessive [3, 5]. This case is suggestive of Ollier disease because the patient has multiple enchondromas distributed asymmetrically with right sided predominance and no soft tissue hemangioma or other subtype's features noted. Ollier disease is often diagnosed late because it is asymptomatic. In this case, the patient was diagnosed at the age of four years old due to the presence of right ankle deformity. The reason for early identification in this patient is the presence of palpable bony mass over her right ankle joint with prominent external angulation. Ollier disease can appear during the first decade of life with palpable bony mass over fingers or toes, asymmetric shortening of lower limbs or osseous deformities with or without pathological fracture [1]. The exact cause of Ollier disease is unknown, but it is suggested to be sporadic with random spontaneous mutation during early development [4, 5]. In this patient, there was no similar condition or deformity in her twin as well as her other family members.

Radiological imaging is important to diagnose Ollier disease. The initial imaging method conducted for this case was two plane conventional radiographs of both ankle joints. However, the lesion over the right ankle was not recognized at first impression. Hence, proceeded with MRI evaluation which was consistent with the features of enchondroma of right distal fibula with possible enchondromatosis due to detection of multiple small similar lesions scattered over calcaneum, second and third metatarsals and proximal phalanges. Skeletal survey over several other joints confirmed the multiplicity of the lesion with no or minimal joint deformity. It is sufficient to diagnose enchondromatosis with conventional radiograph with its clinical features [1]. The radiographic features may include multiple expansile lytic lesions, occasionally with punctate calcification [2]. It frequently assembles in clusters, thus resulting in metaphysis widening and endosteal scalloping of the cortex [1]. The MRI and Computed Tomography (CT) scan should be used when the

Vol 9(2) (2024) 65-70 | jchs-medicine.uitm.edu.my | eISSN 0127-984X
https://doi.org/10.24191/jchs.v9i2.17265

diagnosis is difficult as they have better visualization of the periosteal bone formation, calcification, cortical destruction and soft tissue involvement [4]. Among differential diagnoses that need to be considered are hereditary multiple exostosis and low-grade chondrosarcoma [1, 3, 4].

Ollier disease has a risk to transform to secondary chondrosarcoma for up to 50% higher than enchondroma which has a risk of up to 4% [1, 3, 5]. Therefore, radiographic risk factors of secondary chondrosarcoma should be observed in any imaging modalities of affected bone. Malignancy should be suspected if there are radiographic features like cortical destruction, moth-eaten osteolysis, periosteal reaction, oedema surrounding the tumour, and soft tissue mass [4]. The patient's imaging did not show any of these features. However, since the potential of progression into secondary chondrosarcoma is high in Ollier disease, the need for clinical and radiological follow up is necessary.

There are several recommendations on follow up and surveillance of the disease. A study recommends staging for enchondromatosis with technetium scan and x-rays of each enchondroma for baseline [4]. Recent review recommends plain radiographs for standard initial diagnostic approach, supported by MRI or CT scan over anatomically difficult regions like pelvis and scapula [4]. Bone scintigraphy or whole-body MRI can be used to screen underlying enchondromatosis [4]. If there is suspicion of malignancy transformation during initial diagnosis or follow up, case should be discussed in a multidisciplinary team involving orthopaedic and oncology teams and biopsy maybe required. Subsequent follow up will depend on histology finding. If the tumour is located at the high-risk region like in the pelvis, femur and scapula and/or size is more than 5 to 6cm, a yearly clinical and MRI follow up should be done [4]. Other than that, clinical survey should continue annually with radiographic control every two to three years [2, 4]. This follow up may be benefitted lifelong [4].

Our patient is planned for corrective osteotomy in view of her right ankle joint deformity. She may need multiple corrective surgery as the recurrence is high following surgery in Ollier disease [2]. However, without corrective surgery the deformity may worsen over time and lead to other complications such as limb length discrepancy and pathological fracture. There is no medical treatment for Ollier disease. Surgical treatment is indicated only for complications above and malignancy transformation [2]. In general, prognosis of Ollier disease is variable, depends on the location, size, extension of the lesions, the severity of deformity, the age onset and the risk factors for secondary chondrosarcoma transformation [1, 2, 5]. On the other hand, the secondary chondrosarcoma has relatively good prognosis with low occurrence of metastasis and 90% overall survival rate at five years [4]. Although it is still early in the disease with mild-moderate skeletal deformity and no symptom for our patient, it is important to properly monitor the disease and correct the deformity. The skeletal deformity if not manage early will subsequently impair the growth of the child in term of his/her gross motor as well as mental development.

CONCLUSION

In conclusion, the learning point from this case is the importance of radiological imaging in identifying enchondromatosis in a young child. Children often present to the primary care clinic with musculoskeletal complaints. It is pertinent to consider rare conditions to not miss serious causes. This case demonstrates that plain radiographs in two planes is the best initial imaging to evaluate primary bone lesion in children. This should be followed by a skeletal survey once a solitary lesion is detected to screen for multiplicity of the lesions. The clinical and radiological features help in early diagnosis and early referral to multidisciplinary team for further management. Understanding the possible serious complications of enchondromatosis for malignant conversion will ensure that the child is assessed and followed up appropriately in the long-term management.

Conflict of Interest

Authors declare none.

Acknowledgements

We would like to thank the patient and her parents who provided clinical information for this case and permission to publish this case report.

Authors' contribution

Literature review and manuscript drafting were prepared by AMAR. Clinical input and radiological interpretation were done by FA and MFM. The final manuscript was revised and edited by AMAR and FA. All authors approved the final version of the manuscript submitted for publication.

REFERENCES

1. Jurik AG. Multiple hereditary exostoses and enchondromatosis. Best Pract Res Clin Rheumatol. 2020; 34(3): 101505.
2. Kumar A, Jain VK, Bharadwaj M, Arya RK. Ollier disease: Pathogenesis, diagnosis, and management. Orthopedics. 2015; 38(6): e497–506.
3. Pansuriya TC, Kroon HM, Bovée JVMG. Enchondromatosis: Insights on the different subtypes. Int J Clin Exp Pathol. 2010; 3(6): 557–69.
4. Herget GW, Strohm P, Rottenburger C, Kontny U, Krauss T, Bohn J, et al. Insights in Enchondroma, Enchondromatosis and the risk of secondary Chondrosarcoma. Review of the literature with an emphasis on the clinical behaviour, radiology, malignant transformation and the follow up. Neoplasma. 2014; 61(4): 365–78.
5. Pansuriya T, Bovée J. Enchondromatosis. Atlas Genet Cytogenet Oncol Haematol. 2011;13(7):524–6.

Vol 9(2) (2024) 65-70 | jchs-medicine.uitm.edu.my | eISSN 0127-984X
https://doi.org/10.24191/jchs.v9i2.17265