Osteoid Osteoma as a Cause of Macrodactyly

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Abstract: We report a case of macrodactyly secondary to an osteoid osteoma in the distal phalanx of a toe, and describe the clinical presentation, radiological findings and successful outcome after surgical excision of the lesion.

A review of the current literature is also provided.

Keywords: Osteoid osteoma, macrodactyly, orthopaedics.

INTRODUCTION

Osteoid osteoma is a benign bone tumour that usually affects children and young adults [1]. It was first identified as a clinical entity by Heine in 1927 [2], Bergstrand in 1930 [3], and Jaffe in 1935 [4]. Well-localised pain is often the only symptom of the disease and is typically described as mild and intermittent at first, becoming more constant and severe especially at night [5]. The pain is dramatically relieved by small doses of aspirin (or other prostaglandin inhibitors). The tumour is a very vascular one and thus anything that causes vasodilation (including alcohol) may exacerbate or induce severe pain. Tumour regression sometimes occurs after some time. Whilst the precise mechanism of this regression is not known, tumour infarction is the most likely explanation [6].

Macrodactyly is defined as a hamartomatous enlargement of the soft tissues and underlying bone. This growth can be static, commensurate with the rest of the limb or progressive. It typically involves the hands rather than the feet. Causes can be broadly divided into congenital or acquired. Congenital causes include Klippel Trenaunay Weber syndrome, macrodystrophia lipomatosa, neurofibromatosis and lipoatrophic diabetes. Acquired causes include trauma and an acute inflammatory response, tumours such as lipofibromatous hamartomata, arteriovenous malformations and amyloidosis. These will be discussed further later on.

We report a case of macrodactyly secondary to an osteoid osteoma in the distal phalanx of a toe, and describe the clinical presentation, radiological findings and successful outcome after surgical excision of the lesion.

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CASE PRESENTATION

A fourteen-year-old girl presented to the outpatients' department with a one year history of a gradually enlarging left second toe. The increase in size was gradual in onset, with only occasional episodes of pain localised to the distal phalanx. There was no variation of the pain in relation to the time of day. There was no history of injury to the toe and no similar swellings in any other digits.

By the time she presented, the affected toe was hypertrophied compared to the contralateral toe. The hypertrophy affected mainly the distal phalanx and overlying nail (Figure **1**). The toe was slightly tender to palpation. There were no local signs of infection or recent trauma.

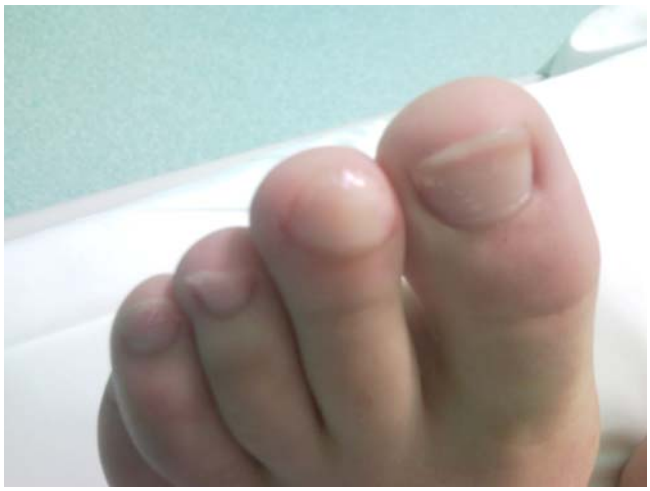

Figure 1: Photo of the affected left second toe.

A plain radiograph of the foot (Figures **2A** and **2B**) revealed a radiolucent lesion measuring about 5mm in diameter in the tuft of the distal phalanx. The overlying soft tissue shadow is larger than that of the other toes.

Surgical curettage of the lesion was performed and subsequent histological analysis of the specimen confirmed the diagnosis of osteoid osteoma. There was

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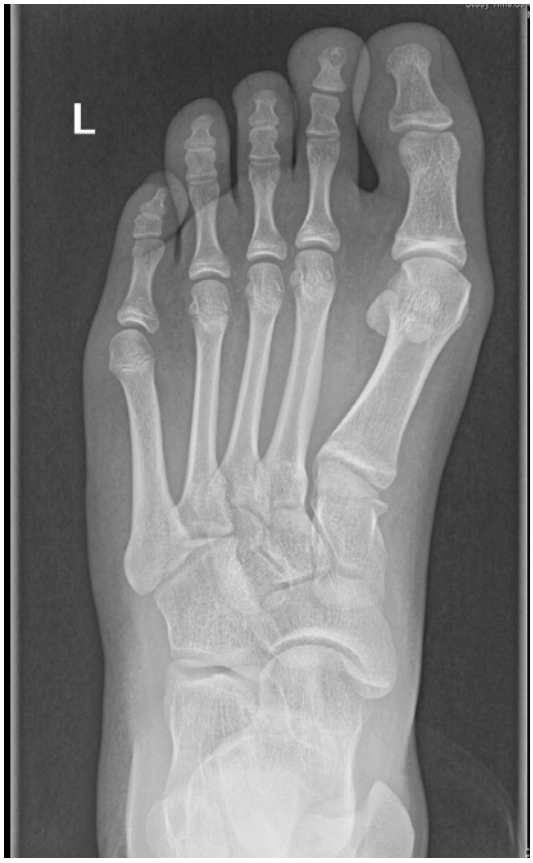

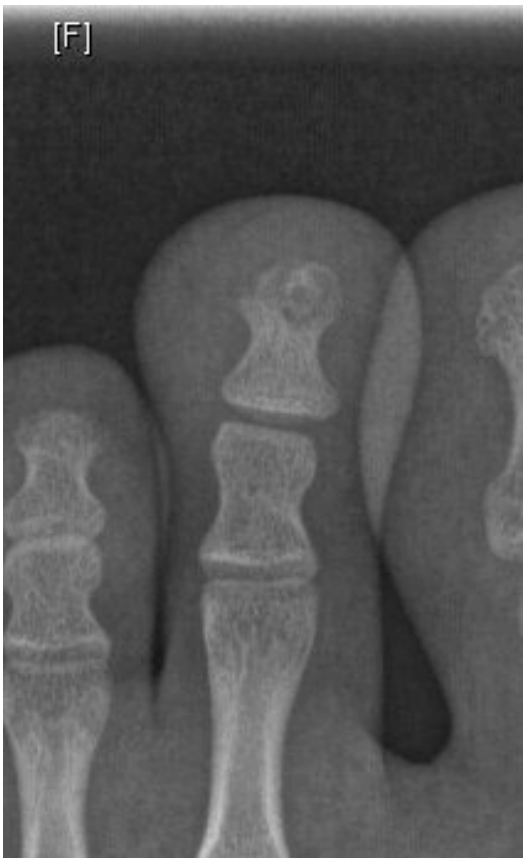

A **B**

Figures 2: A and **B**. Plain radiograph of the left foot with a close up of the left second toe distal phalanx showing a radiolucent lesion in the distal half of the left second distal phalanx and the expansion of the soft tissue shadow.

no cartilage in the tissue submitted for pathological analysis and no signs of malignancy, thereby excluding an endochondroma or osteoblastoma.

One year following surgical treatment of this lesion, the patient is completely free of symptoms and the toe looks completely normal (Figure **3**).

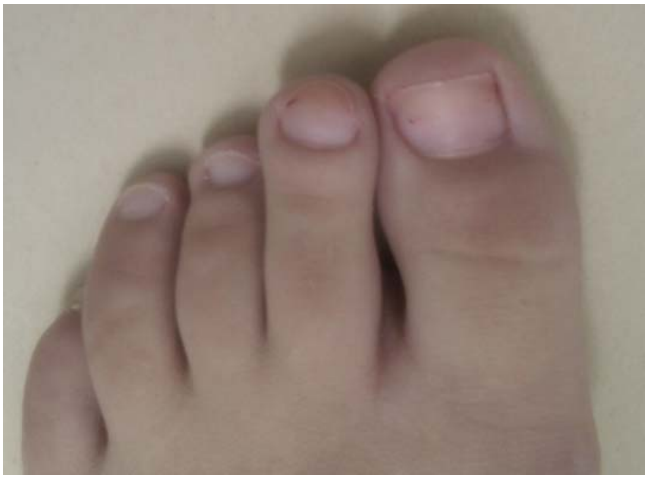

Figure 3: Photo of the affected toe (on the left) one year post-operatively.

DISCUSSION

Osteoid osteoma is typically a small, painful, benign bone tumour of the growing skeleton representing approximately 10% of all benign bone neoplasms. It usually affects children and young adults, with a preponderance of male patients reported in most cases [18]. Normally the tumour does not exceed 1 cm in diameter. Osteoid osteoma has a predilection for the lower extremity, with half or more of the lesions occurring in the femur and tibia. The remaining lesions are distributed approximately equally among the spine, hand and foot [8]. Rarely, this tumour can be almost painless in children [7-9], as was the case with our patient.

Macrodactyly, or enlargement of a digit, is rare and is usually congenital. The vast majority of cases are unilateral involve more than one digit. The digits involved typically correspond to the territory of one of the peripheral nerves. The macrodactyly can be static, growing commensurately with the growth of the limb, or progressive, growing faster than the rest of the limb. Involvement in the hand is more common than

involvement in the foot. Three types of macrodactyly are described—nerve territory oriented macrodactyly where there is lipofibromatous harmatoma growth of a peripheral nerve [16], macrodactlyly associated with neurofibromatosis and the rare hyperostotic macrodactyly where osteocartilagenous protuberances arise around the digital joints and limit motion.

There are very few reports in the current literature that consider osteoid osteoma as part of the differential diagnosis in a child with macrodactyly, especially macrodactyly of the toe. Indeed there is literature stating that in the hands and feet, presentation is rare in the terminal phalanx and exceptional in the middle phalanx [19-22]. The differential diagnosis of macrodactyly [17] includes both congenital or acquired conditions. Congenital causes include Klippel Trenaunay Weber (KTS) syndrome (syndromal condition associated with port-wine stains, venous and lymphatic malformations and soft-tissue hypertrophy of the affected limb), macrodystrophia lipomatosa (a rare congenital disorder with progressive overgrowth of all the mesenchymal elements in a digit, with a disproportionate increase in the fibroadipose tissue; can be associated with type I neurofibromatosis or KTS), neurofibromatosis and lipoatrophic diabetes. Acquired causes include trauma and an acute inflammatory response, infection, tumours such as lipofibromatous hamartoma and lymphangioma, arteriovenous malformations and amyloidosis.

Osteoid osteoma causes an intense and chronic inflammatory response in the surrounding tissues which include a periosteal reaction, sclerosis of bone and synovitis [10, 11] because of the production of prostaglandins by the tumour [12, 13]. This response is most probably responsible for the increase in size of the toe in our case. This reaction often regresses spontaneously after removal of the nidus. Increased blood flow through the tumour [14] may explain the pain. This increased blood flow may causes high pressure which directly stimulates the nerve endings within the tumour. Furthermore, prostaglandins may directly stimulate free nerve endings inside or close to the tumour by lowering the nociceptive threshold [15].

An early diagnosis of osteoid osteoma based on clinical and radiographic findings can usually easily be made when long bones are affected. However, when the bones of the hands and feet are involved, both the atypical presentation and the subtle radiologic changes make diagnosis difficult. Because this difficulty can result in a delay in definitive treatment, we would recommend early investigation with CT or MRI if clinically warranted and if initially radiographs do not provide a definitive diagnosis.

CONCLUSION

Although the presentation of an osteoid osteoma is often typical, and a provisional diagnosis can be made with a good history and plain imaging of the affected area, the exceptions to the rule can present a diagnostic challenge. As with this atypical case, it is important to think of osteoid osteoma when considering the differential diagnosis of macrodactyly.

With regards to treatment of osteoid osteomata, surgical excision is the treatment of choice because it abolishes the pain and reverts the chronic inflammatory response characteristic of an osteoid osteoma.

CONFLICTS OF INTEREST

None.

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