

Aneurysmal Bone Cyst presenting Incidentally as a Traumatic Foot Injury

Case Report

Abstract:

Aneurysmal bone cyst is a rare, benign neoplastic lesion affecting metaphysis of long bones and vertebrae, characterized by blood filled spaces separated by fibrous septae.[1] Diagnosis depends on typical features on magnetic resonance imaging and confirmation by biopsy is essential before surgery is considered. We present an aneurysmal bone cyst in a 7-year-old child with characteristic X ray findings. Diagnosis of ABC should be suspected in young patients presenting with limb pain or swelling and pain disproportionate to the mechanism of injury could indicate pathologic fracture. Early detection of ABC is possible as an incidental finding by closely examining routine X rays so that appropriate treatment can be instituted early and complications like pathologic fractures can be avoided. This case will help to increase its awareness as early diagnosis and treatment can limit disability.

Key words:

Aneurysm, cyst, metaphysis, vascular lesion, USP6 gene

Introduction:

Aneurysmal bone cyst (ABC) is a benign, highly vascular lesion of the bone affecting mainly in the first two decades of life. This is characterized by expansile lesions with multiseptated blood filled spaces in the metaphyseal areas of long bones and vertebrae.[2]

Epidemiology

The annual prevalence is 0.14-0.32 per 100,000 individuals (range, 0-1.238) for aneurysmal cysts, with a 1.8:1 male to female ratio and a median age of the patients of 11.1 years (range, 1-19.7 years).[3,4] About 30% of ABCs are secondary, without translocation; they occur in reaction to another, usually benign, bone lesion.[5] Very rare malignant transformation has been reported in some cases.[6]

Diagnosis

Both radiographic and Magnetic Resonance Imaging appearances help in the diagnosis although biopsy may help to differentiate other sinister causes. The differential between aneurysmal bone cysts and unicameral bone cysts usually is clear

Thomas Mathew, Tom Jose,
Ayotunde Oguntade, Rajinder
Thripathy & Awais Iqbal

Go-To-Doc Health Care, Royal
Preston Hospital, Preston, UK

Corresponding author:
tommjoe35@yahoo.com

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clinically and radiographically.

Occasionally there are cases in which the diagnosis is not clear. The presence of a double density fluid level and the presence of septations within the lesion and signal characteristics of low intensity on T1 images and high intensity on T2 images, was compatible with an aneurysmal bone cyst. [7]

Because natural history and treatment are different, the ability to distinguish between unicameral and ABCs before surgery is important. ABCs are potentially more aggressive, with a risk of bone destruction. Diagnosis must systematically be confirmed by biopsy, identifying soft-tissue parts, as telangiectatic sarcoma can mimic ABC.

Genetic basis

About two-third of primary ABCs have a rearrangement of the USP6 gene, which is not present in the ABC-like changes that occur secondary to other primary bone tumours (i.e., secondary ABC).[8] A neoplastic basis in primary ABC is evidenced by demonstration of clonal chromosome band 17p13 translocations that place the USP6 (TRE2 or TRE17) oncogene under the regulatory influence of the highly active CDH11 promoter. It is believed that primary ABCs are mesenchymal neoplasms exhibiting USP6 and/or CDH11 oncogenic rearrangements [9]

Treatment

The therapeutic options of ABC range from

curettage with or without adjuncts such as phenol, liquid nitrogen, argon laser and bone grafting or bone substitutes to more recently employed alternatives such as image-guided sclerotherapy with various sclerosing agents and monoclonal antibodies (e.g., Denosumab). [10] Intra-lesional sclerotherapy with alcohol is an effective treatment. In spinal ABC and in aggressive lesions with a risk of fracture, surgical treatment should be preferred, possibly after preoperative embolization. The risk of malignant transformation is very low, except in case of radiation therapy.[5]

Case report:

A 7-year-old boy was brought to the Urgent Care Centre after sustaining injury to his right foot whilst playing football. The foot x ray revealed a well-defined expansile lucent lesion within the 2nd metatarsal. There was an associated modelling deformity to the 3rd metatarsal shaft suggestive of a long-standing benign process.

The differential diagnoses were a simple bone cyst, ABC or an enchondroma with no acute fracture. Subsequent MRI scan confirmed the cystic expansile lesion with cortical thinning but no cortical destruction, suggestive of ABC.

The patient was referred for biopsy confirmation and surgical treatment to the local Paediatric surgical centre.



Discussion:

ABCs commonly occur in the metaphysis of long bones and vertebral column with high vascularity and marrow content especially in weight bearing ones.[5] The clinical presentation is variable, may present as an asymptomatic lesion or as a rapidly growing, expansile, destructive lesion causing pain, swelling, deformity, neurologic symptoms and pathologic fracture.[11]

Plain radiograph shows lytic, expansile lesion. MRI is the radiological investigation of choice. The gold standard investigation is a biopsy. Curettage with bone grafting is the treatment of choice.[12]

Conclusion:

Diagnosis of ABC should be suspected in young patients presenting with limb pain or swelling and pain disproportionate to the mechanism of injury could indicate pathologic fracture. Early detection of ABC is possible as an incidental finding by closely examining routine X rays so that appropriate treatment can be instituted early and complications like pathologic fractures can be avoided.

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Conflicts of interest:

There are no conflicts of interest.

Author's contributions:

All authors have critically reviewed and approved the final draft and are responsible for the content of the article.

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