International Medical Case Reports Journal Dovepress

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CASE REPORT

Juvenile Xanthogranuloma Involving the Left Femur: A Case Report and Literature Review

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Background: Juvenile xanthogranuloma is a rare condition, and femoral involvement is even rarer. We report a case of juvenile xanthogranuloma affecting the femur. To the best of our knowledge, this is the first reported case of femoral juvenile xanthogranuloma in China.

Case Presentation: A 19-year-old boy began experiencing swelling in various parts of his body at the age of 1 year, followed by pain in his left hip. Preoperative pathology and imaging indicated juvenile xanthogranuloma of the left femur, leading to surgical intervention. Postoperatively, the patient experienced significant relief from left hip discomfort.

Conclusion: Surgery can be an effective treatment for juvenile xanthogranuloma involving the femur.

Keywords: juvenile xanthogranuloma, femur, rare case

Introduction

Juvenile xanthogranuloma is a benign non-Langerhans cell histiocytosis,[1] with an unknown etiology. Previous studies have suggested a potential link to abnormal lipid deposition in tissues and organs outside of blood vessels.[2] The prevalence of the disease has been associated with age, gender, and race. Histologically, the disease is characterized by mononuclear macrophages, foam-like cells, multinucleated giant cells, and occasional spindle-shaped cells.[3] Diagnosis relies mainly on histologic examination, as clinical and imaging findings are non-specific.

Juvenile xanthogranuloma is most commonly found in the superficial soft tissues of the body, including the skin, subcutaneous fat, and muscle. The incidence of juvenile xanthogranuloma is extremely low, with only 50 cases reported worldwide. Involvement of bone, particularly accessory bones such as the tibia and other long bones, is more common, though central axial bones, including the spine, can also be affected. Cases have also been reported in the skull and oral maxilla.[4,5] Patients with hyperlipidemia are at a higher risk of developing juvenile xanthogranuloma compared with those with normal lipid levels, and many have a family history of the disease.[6] We report the case of a 19-year-old patient from China diagnosed with juvenile xanthogranuloma in the proximal bone of the left femur. The patient underwent tumor shaving of the proximal left femur, followed by bone grafting and internal fixation device implantation to alleviate left hip pain.

Case Report

A 19-year-old male patient presented to our hospital with subcutaneous swelling that had been present for 18 years. According to his mother, a swelling was first noticed under the skin on the inner side of his left thigh when he was 1 year old. The swelling was round, about 1 cm in diameter, and showed no signs of redness or surface inflammation. At the time, the patient's parents did not seek medical attention, as they were unconcerned about the swelling. As the child grew older, swelling began to appear in several

Received: 4 July 2024
Accepted: 22 November 2024
Published: 7 December 2024

areas of his body, starting in his hips, then around his knees and elbows, and eventually in his hands. The lumps gradually increased in size. At the age of 14, the patient began experiencing pain in multiple joints, although it was tolerable at the time. In 2017, he underwent excision of subcutaneous masses from both hands at a local hospital, but no postoperative pathology was performed. By 2019, the masses had reappeared on both hands, and the pain in several joints had significantly worsened, requiring oral pain medication for relief, with the pain in his left hip being particularly severe. In April 2022, the patient presented to our hospital for evaluation. X-ray examination revealed structural abnormalities in the upper end of the left femur, characterized by mixed-density shadows, cortical thinning, irregular margins, and a lesion measuring 8.7 cm in length. Computed tomography showed a low-density lesion in the medullary cavity of the upper left femur, with dimensions of 3.1×3.1 cm and an intact bone cortex. Magnetic resonance imaging displayed an irregular abnormal signal in the medullary cavity of the upper left femur. Both T1- and T2-weighted images showed mixed high and low signals, with the lesion measuring 8.2 cm in length (As shown in Figure 1). Lipid examination results revealed elevated total cholesterol at 274.17 mg/dL and low-density lipoprotein at 238.21 mg/dL, both above the normal range, while high-density lipoprotein was reduced at 26.29 mg/dL. Following these investigations, a biopsy of the femoral lesion was performed, which showed hemorrhage, areas of necrotic bone, and inflammatory exudative tissue with a multinucleated giant cell reaction. Immunohistochemistry confirmed the diagnosis of juvenile xanthogranuloma in the proximal left femur. The lesion had a Mirels score of 10, indicating the need for surgical intervention to prevent a pathological fracture.[3] After a thorough evaluation, the patient underwent surgical treatment. Postoperative pathology revealed fibrous tissue

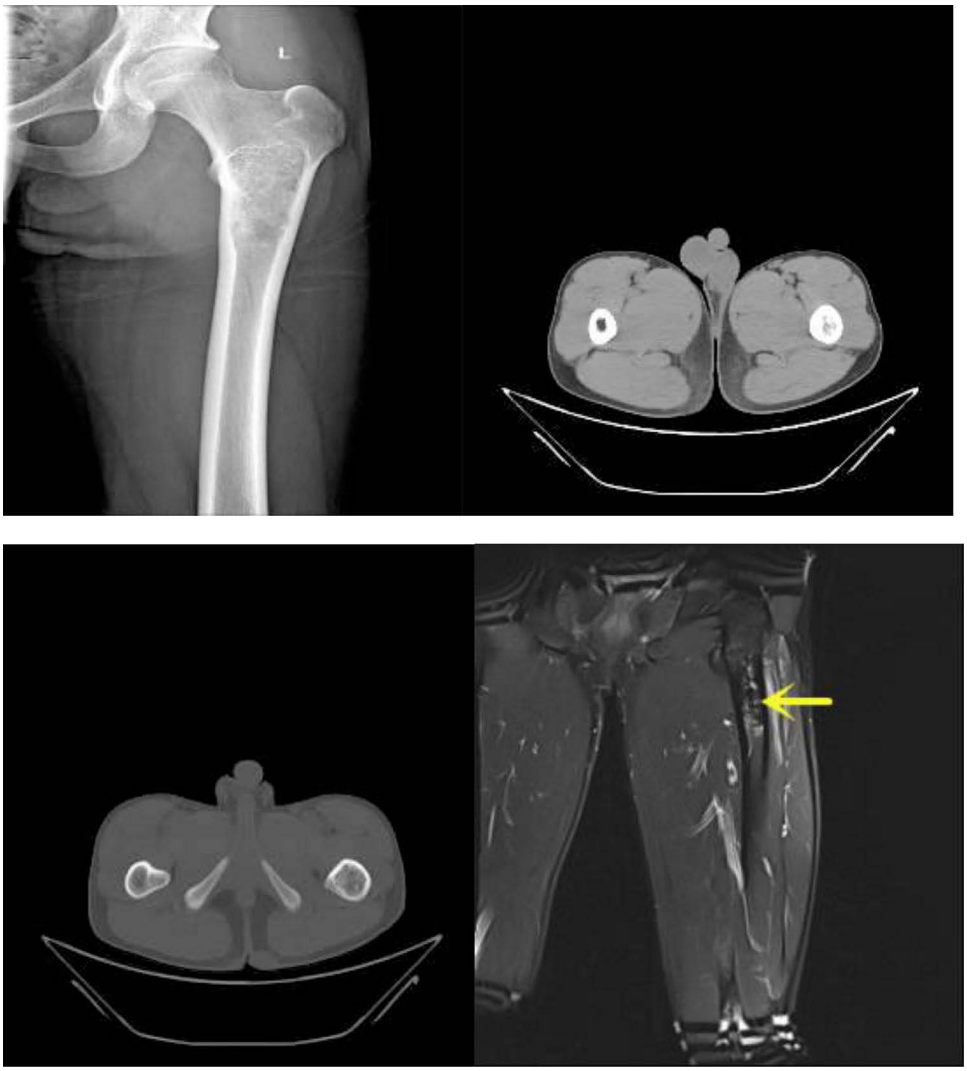

Figure 1 Preoperative imaging: X-ray and CT examination indicate a mixed-density lesion at the proximal end of the left femur, with both high and low attenuation areas.

proliferation and cholesterol crystal formation, with immunohistochemical findings consistent with juvenile xanthogranuloma. Following surgery, the patient experienced significant pain relief. During regular follow-up, the patient remained pain-free in the left femur by 6 months postoperatively.

Discussion

To the best of our knowledge, this is the first reported case of juvenile xanthogranuloma involving the femur in China. Juvenile xanthogranuloma is a rare benign condition,[3] with approximately 50 cases documented worldwide. The exact etiology of the disease remains unclear. Some researchers attribute it to abnormal cholesterol deposition in tissues outside of blood vessels, while others suggest it may result from reactive proliferation of reticular histiocytes.[1]

A previous study indicated that juvenile xanthogranuloma is often seen in patients with hyperlipidemia, particularly those with familial hyperlipidemia.[7] Our patient's preoperative examination confirmed hyperlipidemia, supporting these findings. The onset of juvenile xanthogranuloma varies widely, occurring between the ages of 1 and 70 years, with two-thirds to three-quarters of patients developing the condition before age 25.[1] Our patient presented with the disease around 1 year of age and experienced significant symptom worsening around 14 years of age, which aligns with this pattern and supports the diagnosis of juvenile xanthogranuloma. When bone involvement occurs, it mainly affects appendicular bones such as the tibia and other long bones, although axial bones like the spine can occasionally be involved. The condition has also been reported in the skull and maxilla. Literature on juvenile xanthogranuloma of the bone is scarce, and the clinical presentation and imaging findings are not specific, complicating diagnosis. As a result, the diagnosis relies largely on histologic biopsy. Typical histologic features of juvenile xanthogranuloma include numerous foamy cells, nonfoamy mononuclear or multinucleated giant cells, occasional Touton-type giant cells, and several cholesterol crystals within the lesion.[1,3] Histocyte immunostaining showed positivity for CD68 and CD163, and negativity for CD1a and S100.[3,4] The patient exhibited fibrous tissue hyperplasia with cholesterol crystal formation (CD68+), consistent with the findings reported in the literature (As shown in Figure 2).

Treatment for juvenile xanthogranuloma varies depending on the site of involvement. For hip joint involvement, total hip replacement may be performed to alleviate pain.[1] In cases where the proximal humerus is affected and there is a pathological fracture, proximal humeral resection and prosthetic replacement may be necessary.[5] For involvement of the proximal femur, the standard approach typically includes tumor bone curettage, bone grafting, and internal fixation device implantation.[7] Prophylactic internal fixation is determined based on the lesion location and the extent of involvement. Prophylactic internal fixation is often necessary because the pelvis, proximal femur, and tibia are particularly vulnerable to pathologic fractures. In our patient, an osteolytic lesion measuring 8.2 cm in length, with a Mirels score of 10 in the left proximal femur, required surgical intervention to prevent a pathologic fracture. Consequently, we performed tumor debridement, bone grafting, and placement of an internal fixation device to address the potential instability of the femoral lesion. Postoperative radiographs confirmed that the internal fixation device was properly positioned, and the patient's pain symptoms were significantly relieved (As shown in Figure 3). After a prolonged follow-up period, the patient was completely pain-free at the 6-month postoperative mark.

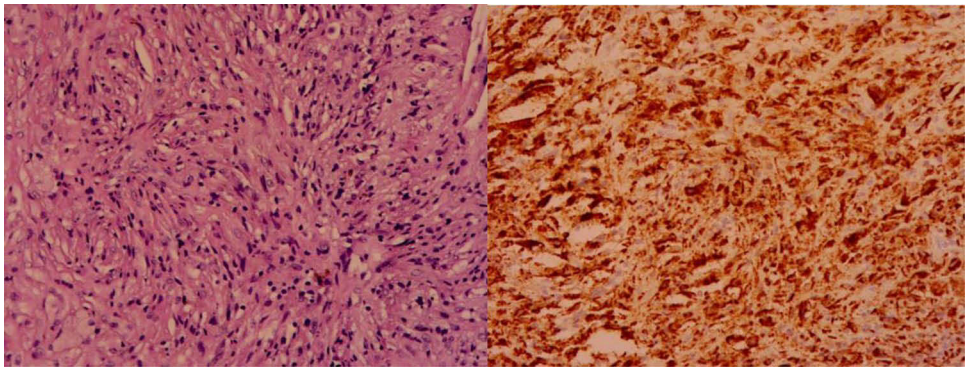

Figure 2 The left image shows pathological histology with cholesterol crystals and calcified deposits, while the right image displays CD68 positivity with a diffuse distribution of foamy, tissue-like cells.

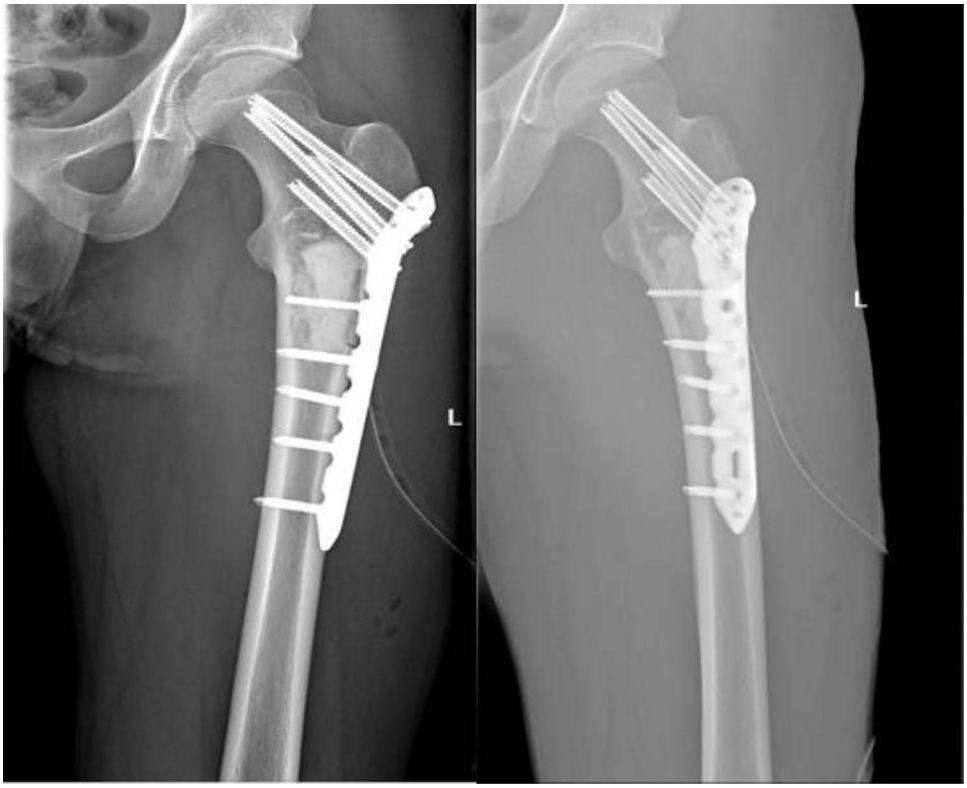

Figure 3 Postoperative anteroposterior and lateral radiographs confirmed that the internal fixation device was properly positioned.

The patient had hyperlipidemia preoperatively, which can predispose individuals to cardiovascular diseases. Therefore, it is essential for him to monitor and manage his blood lipid levels, as recommended. Currently, the patient's pain symptoms have completely resolved. We will continue to follow up with him to assess the long-term effects of the treatment.

Conclusion

In conclusion, we present a rare case of juvenile xanthogranuloma of the proximal femur associated with hyperlipidemia. The variability in clinical presentation and the low specificity of imaging findings complicate the diagnosis of this condition. Currently, pathology remains the primary diagnostic tool. Surgical intervention proves to be an effective treatment approach when the proximal femur is involved.

Ethics Statement

Written informed consent was obtained from the individual(s) for the publication of any potentially identifiable images or data included in this article.

Ethics Approval and Consent to Participate

Our institution does not require ethical approval for reporting identity-nullified individual cases or case series.

Author Contributions

All authors made a significant contribution to the work reported, whether that is in the conception, study design, execution, acquisition of data, analysis and interpretation, or in all these areas; took part in drafting, revising or critically reviewing the article; gave final approval of the version to be published; have agreed on the journal to which the article has been submitted; and agree to be accountable for all aspects of the work.

Disclosure

The authors declare that they have no competing interests.

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