Rare disease

Pathological fractures on both lower limbs with Jaffe-Campanacci's syndrome

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Summary

Jaffe-Campanacci's syndrome is a very rare condition and was first described by Jaffe in 1958. It is presented by non-ossifying fibromas, café-au-lait spots and axillar freckling. Non-ossifying fibromas are usually found after minor traumas or stress fractures. Differential diagnosis to neurofibromatosis include the absence of Lisch granules, neurofibromatous skin lesions and schwannomatous soft tissue masses. In this case, we report a 13-year-old white boy with Jaffe-Campanacci's syndrome, and bilateral pathological lower limbs fracture.

BACKGROUND

Jaffe-Campanacci's syndrome is a very rare condition and was first described by Jaffe in 1958.¹ This syndrome can be misdiagnosed as cases of neurofibromatosis because orthopaedic surgeons generally focus in treating patient.² We would like to call attention to such an interesting, rare and important clinical condition.

CASE PRESENTATION

A 13-year-old white boy was referred to orthopaedic and traumatology clinic with a pathological fracture of the distal part of the left tibia. The patient was born with multiple, variously sized café-au-lait spots and axillar freckling (figure 1). He was operated on for cryptorchidism in his first year but had not been previously diagnosed as having neurofibromatosis. He remained symptomless until a complaint of a tibial fracture. He was receiving education at a special rehabilitation school for children with learning disorders.

INVESTIGATIONS

The patient's blood pressure was within normal limits. He had no abnormal ECG findings. Both pupil inspections were normal and no lish granules were seen. The patient was mildly retarded (IQ 69). No visceral, central nervous system or other system anomalies were detected. None of the family members had skin lesions, such as café-au-lait spots, axillar freckling and long bone fracture history. Familial research showed no genetic errors. All laboratory studies were within normal limits. Plain radiographs showed a fracture on the distal part of the left tibia with mildly expanded cystic lesions (figure 2). Similar lesions were found in the contralateral distal part of the femur.

DIFFERENTIAL DIAGNOSIS

Non-ossifying fibroma, polyostotic fibrous dysplasia, neurofibromatosis, von Recklinghausen, and malignant tumours should be kept in mind for differential diagnosis.

Non-ossifying fibroma of bone (NOF) is a common entity, more frequently found in male children and consisting of a solitary eccentric, lytic expanded lesion in the metaphysis of a long bone. The disorder is benign and most often asymptomatic but may result in a fracture, requiring therapy. Of some importance is to distinguish NOF from another very similar but smaller lesion, fibrous cortical

Figure 1 Clinical view of patient with axillary freckling.

Figure 2 Anterior – posterior and lateral radiographs of fracture of distal part of left tibia.

defect, which is almost always asymptomatic and eccentrically located. Even more striking is a very rarely encountered lesion known as Jaffe-Campanacci's syndrome, which also occurs in children who present with typical non-ossifying fibromatous tumours but in multiple sites. In addition, these patients have some systemic and dermal findings resembling those seen in patients with type 1 neurofibromatosis.³

TREATMENT

The patient underwent an operation for fracture of the distal part of the left tibia. Curettage, grafting with allograft, and fixation with conventional plate were performed (figure 3). Four weeks later, the patient was admitted to hospital because of pain around the right knee. Plain radiographs showed a fracture on the distal part of the right femur (figure 4). The same procedure was performed for fracture treatment (figure 5).

OUTCOME AND FOLLOW-UP

Histological findings showed highly cellular colloganeous tissue with storiform pattern in both tibial and femoral curettage materials. These findings were consistent with the characteristic of the non-ossifying fibroma (figure 6). Both tibial and femoral fractures healed on time. No signs were found in favour of recurrence.

DISCUSSION

Jaffe-Campanacci's syndrome is a very rare condition which can be overlooked by the clinician. It is mostly

Figure 3 Anterior – posterior and lateral radiographs of left tibia postoperatively.

diagnosed at 10–15 years of age; males and females are equally affected.⁴ Most studies are case reports and usually present axillary freckling,⁵ ocular anomalies (Lisch granules), cardiovascular anomalies (aortic stenosis),

Figure 4 Anterior – posterior and lateral radiographs of fracture of distal part of right femur.

Figure 5 Anterior – posterior and lateral radiographs of right femur postoperatively.

genitourinary anomalies (hypogonadism) and chylothorax.⁶ Kourelis *et al* reported pleuritic chest pain and small pulmonary embolus after trauma in their case.⁷ Kotzot *et al* also reported chylothorax and chylopericardium in their case with Jaffe-Campanacci's syndrome.⁸ Our case had no thorax abnormality. Mental retardation and hereditary genetical error have been also reported.⁹ Reciprocal chromosomal translocation involving bands 1–31 and 4q34 (t(1;4)(p31;q34)) for non-ossifying fibroma was reported by Nelson *et al*,¹⁰ but no genetic errors were found in our case. Also, it has been found chromosomal abnormality,^{5,7} but our patient's karyotype analyses were normal.

Histologically bone lesions consist of a fibroblastic, highly cellular, collagenous stromal tissue with a storiform pattern.¹¹ Fibroblasts and fibrocytes with giant cells can be seen, but Schwann cells or verocay bodies must

Figure 6 A histopathological image of tibial curettage material (H&E ×200).

not be present in tissues.¹² Patients are mostly referred to hospital with a pathological fracture in the lower limbs as in our case. Although the patient had surgical history for cryptorchidism, he had not been diagnosed with Jaffe-Campanacci's syndrome previously. It should be differentiated from polyostotic fibrous dysplasia, and neurofibromatosis, because they are very similar presentation. Polyostotic fibrous dysplasia generally tends to involve the skull and only one side of the body.¹³ Fibrous cortical defects are mostly asymptomatic, and seen by a radiograph performed for different problems, but non-ossifying fibromas are more often symptomatic. Neurofibromatosis type 1 can be differentiated through the presence of Lisch nodules, cutaneous neurofibromas, and optic gliomas.³ These findings were not seen on our patient. Non-ossifying fibroma lesions are generally located in the metaphyses of long bones, usually in the lower limbs, especially the distal third of the femur and tibia,¹⁴ and most patients with non-ossifying fibromas are asymptomatic. In non-ossifying fibromas, cortices can be expanded, stress-type fractures and irregular bone formation can also be seen on radiographs, but non-ossifying fibromas have no periosteal reaction or Codman triangle and associated soft tissue mass.¹⁵ The differentiation of non-ossifying fibromas can sometimes be difficult with malignant tumours in pathological fractures, as soft tissue penetration and cortical destruction cannot be excluded. We recommend a two-stage operation in those situations. Biopsy can be performed before definitive surgery under certain conditions. Performing prophylactic surgery is contradictory and most authors recommend that no surgery should be done in asymptomatic patients. Colby and Saul reported that appropriate intervention (exercise restriction, bracing and/or surgery) might decrease the long-term disability associated with Jaffe-Campanacci's syndrome.¹⁶ As Hau *et al*,⁵ we also believe that further genetic studies should be performed in patients with this syndrome. Our case, unusual as the one published by Campanacci in 1983, carried cryptorchidism and multiple non-ossifying fibromata.⁹

Learning points

- ▶ Jaffe-Campanacci's syndrome is a very rare syndrome.
- ▶ Pathological fractures can occur in many conditions.
- ▶ Clinical manifestations of Jaffe-Campanacci's syndrome can lead to severe functional disturbance. It should be considered in these very unique circumstances with multiple non-ossifying fibromas.

Competing interests None.

Patient consent Obtained.

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