Jaffe-Campanacci Syndrome

A Case Report and Review of Literature

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Jaffe-Campanacci syndrome is characterized by multiple nonossifying fibromas and multiple cutaneous café-au-lait spots without accompanying neurofibromas. To the best of our knowledge, the syndrome was first reported in 1958 by Jaffe[1]; in 1983, Campanacci et al.[2] described ten cases. Because of the presence of café-au-lait spots, several authors[1-4] have studied the relationship between Jaffe-Campanacci syndrome and neurofibromatosis. The occurrence of multiple nonossifying fibromas, a prominent feature of Jaffe-Campanacci syndrome, suggests that the syndrome should be considered distinctive from neurofibromatosis. To the best of our knowledge, only eighteen cases of Jaffe-Campanacci syndrome have been reported in the English-language literature[1-3,5-9]. We present a case in a young girl with lateralized skin and bone lesions. The patient and her parents were informed that data concerning her case would be submitted for publication, and they provided consent.

Case Report

Because of the presence of multiple café-au-lait spots, a two-year-old Chinese girl had been diagnosed with type-1 neurofibromatosis. At the age of six years, she had presented with pain over the right knee and right shoulder. Radiographs showed lytic changes in the proximal part of the right humerus, the distal part of the right radius, and the distal part of the right femur. Curettage and bone-grafting had been performed in the right humerus and right femur in a local hospital. The histological findings at that time are not known.

At the age of ten years, the patient was seen at our institution with symptoms of right shoulder and right thigh pain after a minor injury. A nondisplaced fracture of the proximal part of the humerus was suspected on the radiographs. A radiolucent lesion with an impending fracture was noted in the diaphysis of the right femur. The birth and developmental

Fig. 1

Multiple large café-au-lait spots over the right side of the face and neck, right arm, and right torso. Notably, only the right side is involved and the spots do not cross the midline.

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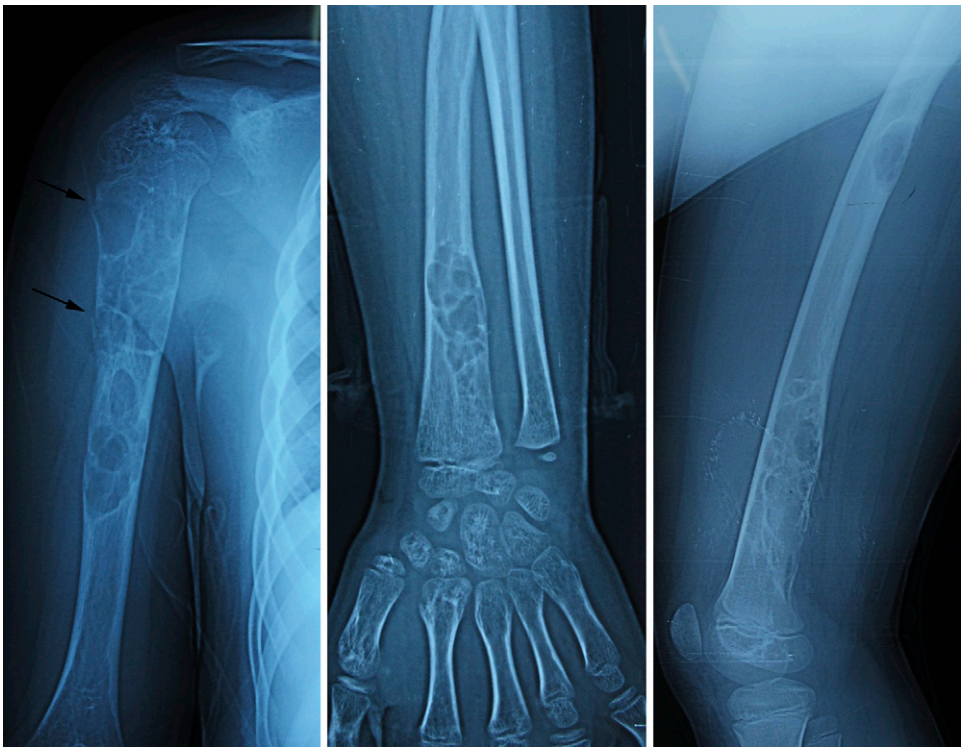

Fig. 2

Radiographs showing demarcated osteolytic lesions located in the metadiaphyseal regions of the proximal part of the right humerus (left panel), the distal part of the right radius (center panel), and the distal part of the right femur and femoral shaft (right panel). A pathologic fracture in the lateral aspect of the proximal part of the right humerus (black arrows) and an impending fracture in the right femoral shaft are present.

histories were normal; the family history was negative for features of type-1 neurofibromatosis and bone lesions. On physical examination, height and weight were appropriate for the patient's age. There was a 5 cm × 6-cm area of alopecia on the right side of the scalp. Corneal leukoma was found in the right eye; no Lisch nodules were present. There were no signs of neurofibromas, optic glioma, hypogonadism, or cardiovascular malformations. Multiple large café-au-lait spots were present over the right side of the face and neck, right limbs, and right torso without axillary freckles. Only the right side was involved, and the spots did not cross the midline. The café-au-lait spots were "coast of Maine" type with ragged and irregular outlines. Hyperkeratosis was noted on some of the spots (Fig. 1). There was no evidence of scoliosis. Neurological examination was normal. The patient did not have mental retardation.

Radiographs showed demarcated osteolytic lesions in the metadiaphyseal regions of the proximal part of the right humerus, the distal part of the right radius, and the distal part of the right femur and femoral shaft (Fig. 2). Magnetic resonance imaging (MRI) of the brain and spine showed no abnormal findings. The right arm was placed in a sling, and the pain was relieved after two weeks. Biopsy and bone-grafting were performed for the right femoral diaphyseal lesion to prevent fracture, and full weight-bearing was permitted two months after surgery (Fig. 3). Histological examination of tissue from the right femur showed spindle-shaped, fibroblastic cells with scattered osteoclasts, characteristic of a nonossifying fibroma (Fig. 4).

Three months after surgery, the patient reported relief of pain and normal function in both the upper and lower right limbs.

Discussion

Multiple café-au-lait spots and multiple nonossifying fibromas are two prominent features of the Jaffe-Campanacci syndrome. In our case, both skin and bone lesions were limited to the right side of the body. No café-au-lait spots crossed the midline. Kotzot et al.[7] also reported a case of both skin and bone lesions restricted to only one side of the body. However, other reported cases had bilateral involvement. It was suggested that somatic mosaicism in early embryonic development was possibly responsible for the lateralization of the lesions[7].

To identify any possible nonossifying lesions, Colby and Saul[4] suggested radiographs of the knee for patients suspected to have Jaffe-Campanacci syndrome. Nonossifying fibromas are likely to be located at the distal part of the femur or the proximal part of the tibia, but they can also be found in the upper limbs (as in our patient and some of the cases reported in the series by Campanacci et al.[2]). Therefore, a skeletal survey is likely indicated to localize all of the lesions. Moreover, the lesions are not limited to the metaphysis of the tubular bones: they usually extend extensively into the diaphysis in Jaffe-Campanacci cases. We also found a skip lesion with an impending fracture of the femoral diaphysis in our patient. Hau et al.[8] reported the case of a fifteen-year-old patient with Jaffe-Campanacci syndrome. In this patient, both of the lesions in the distal part of the femur were treated with intralesional

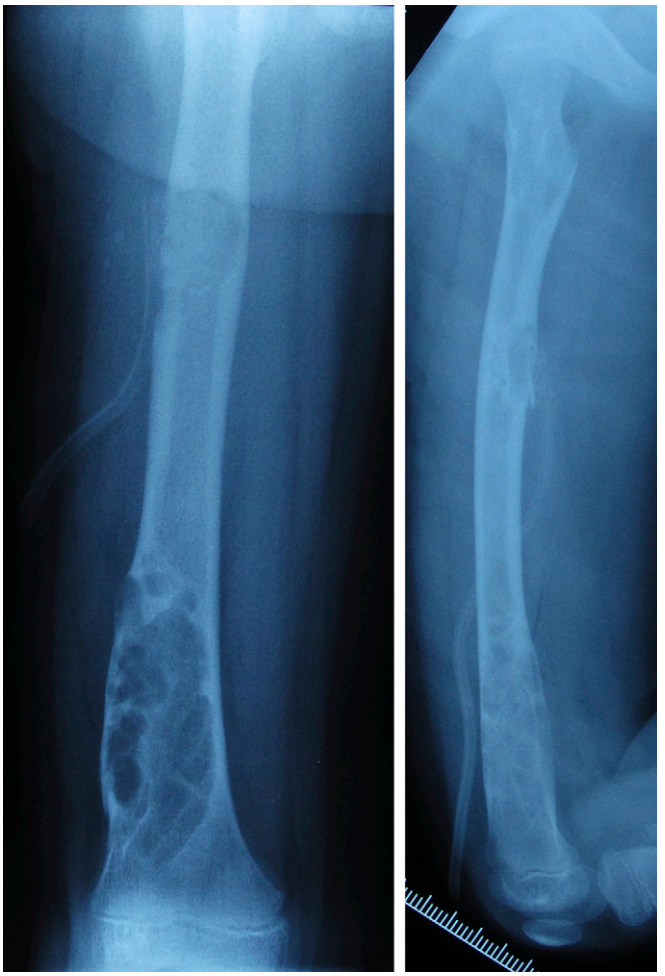

Fig. 3

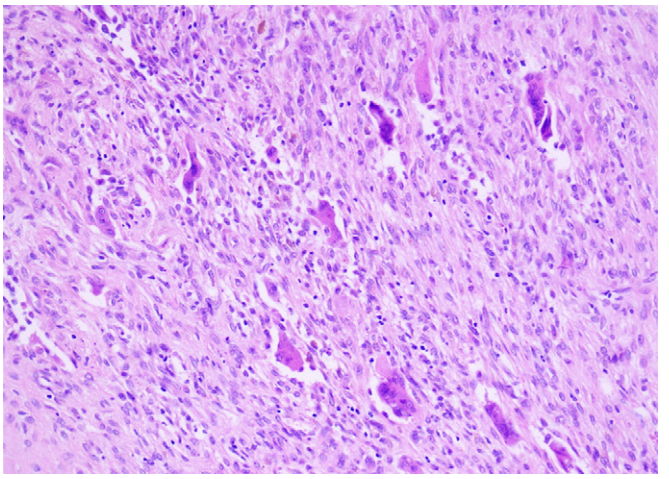

Fig. 4

Fig. 3 Anteroposterior (left panel) and lateral (right panel) radiographs of the right femoral shaft showing the diaphyseal lesion that was treated with curettage and bone allograft. **Fig. 4** Photomicrograph of tumor tissue demonstrating spindle-shaped fibroblastic cells (arrayed) in whirlpool fashion, which is characteristic of a nonossifying fibroma, with a diffuse distribution of osteoclasts (hematoxylin and eosin stain ×20).

excision, curettage, allograft strut-grafting, and plate fixation; healing was achieved six months after the operation. We performed a similar intralesional excision and allograft grafting, but, instead of rigid fixation, we suggested restricted weight-bearing for three weeks. Bone union was achieved after six months. These findings suggest that healing at the sites of the bone lesions was not impaired.

In conclusion, although Jaffe-Campanacci syndrome has been most commonly reported in whites[1-3,5-9], our case shows that it can also occur in the Asian population. ■

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