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CASE REPORT

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Congenital fibrous hamartoma of the knee

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Abstract A full-term male infant presented at birth with a hard swelling of the left knee. The lemon-sized lesion was fixed to the underlying knee muscles, while the overlying skin was stretched and shiny; there was no bruit. Radiography, sonography and MRI suggested a soft-tissue tumour. After surgical excision, histology showed the presence of fibrous and mesenchymal tissue, with mature adipose tissue. Fibrous hamartoma of infancy was diagnosed. Among soft-tissue tumours, fibrous hamartoma of infancy is a rare and benign lesion, occurring in the first 2 years of life. The tumour mainly affects the trunk, axilla, and upper extremities. This infant had unique involvement of the knee. The treatment of choice is local excision.

Keywords Knee · Tumour · Fibrous hamartoma of infancy · Radiography · Ultrasound · MRI

Introduction

Fibrous hamartoma of infancy (FHI) is a rare, benign, but persistent subcutaneous fibrous tumour [1, 2]. The vast majority of these cases occur in the first 2 years of life [3]. The tumour occurs mainly on the trunk, especially the axilla, and upper extremities [1–3]. Histology consists of an 'organoid' mixture of three components: fibrous tissue, adipose tissue, and nests of immature mesenchyme [3]. It is benign, and the treatment of choice is local excision [1–3]. Local recurrences have been reported [3]. We describe a unique case with involvement of the knee.

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Case report

A full-term male infant (weight 3,140 g, length 49 cm) was born vaginally after an uneventful pregnancy. Antenatal sonography at 33 weeks had not revealed any abnormality. Examination at birth showed a voluminous 6×7.5×4-cm swelling of the left knee (Fig. 1). The lesion was fixed to the underlying knee muscles and the overlying skin was stretched and shiny; there was no bruit. There was no family history of cancer or chronic illness.

Radiography of the left knee showed a soft-tissue mass without any bony abnormality or intralesional calcifications (Fig. 2). Sonography demonstrated a solid mass that was homogeneously hyperechoic in its deepest part and localized within the soft tissues (Fig. 2). The lesion did not involve the capsule of the knee joint.

On MRI the mass was seen to have a lobular structure with septa. Fatty tissue (fat-equivalent signal) was evident in the deepest and in the paraseptal parts of the lobules. The signal intensity of the remaining tissue was low, but slightly higher than the signal intensity in the muscles on T1-weighted (T1-W) sequences. The tumour was hyperintense on the STIR sequence and after intravenous (IV) administration of Gd-DOTA (Fig. 3).

The mass was surgically excised. During excision, the tumour was found to be separate from the muscular fascia. Grossly, the surgical sample measured 9×7×5.4 cm and was partially covered by normal, intact skin. Sections revealed noncircumscribed, glistening, grey–white fibrotic tissue alternating with yellow fat. Microscopically, the tumour revealed three distinct components: (1) well-defined interlacing trabeculae of fibrous tissue composed of bundles of spindle-shaped cells separated by bundles of collagen; (2) primitive, undifferentiated, loosely arranged round cells embedded in a myxoid stroma; and (3) mature adipose tissue admixed with the other components. A

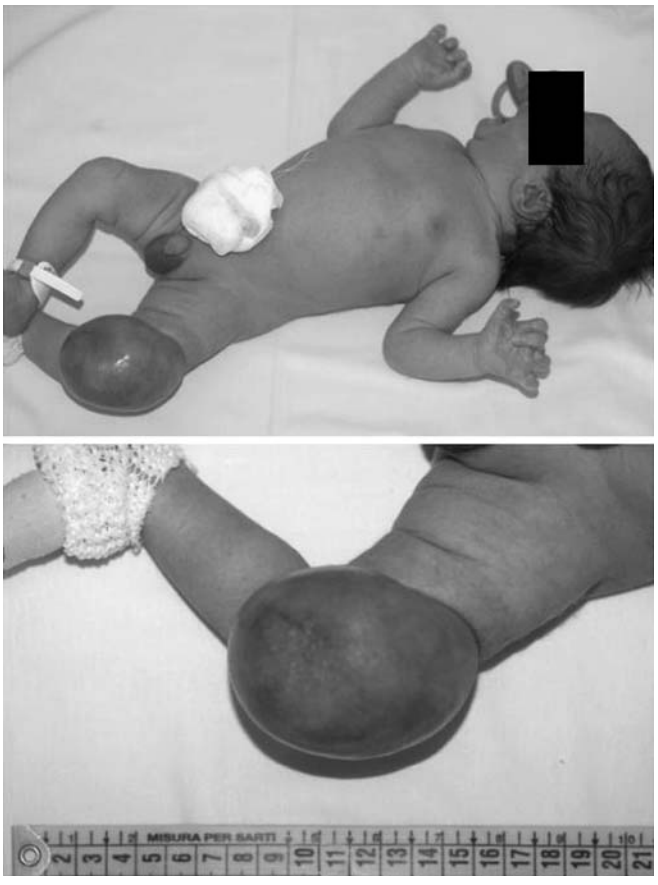

Fig. 1 The patient at the age of 3 days. Note the voluminous mass at the left knee

variably rich eosinophilic infiltrate was associated with the tumour. The epidermis overlying the lesion was slightly hyperplastic and punctuated by small foci of eosinophilic spongiosa. Immunohistochemistry of the primitive mesenchymal cells revealed positive staining with vimentin. The myofibroblastic cells of the fibrous tissue were positive for smooth muscle actin and focally for muscle-specific actin. S100 protein was positive in the adipose tissue. The final diagnosis was FHI.

Discussion

FHI is a rare, benign, but persistent subcutaneous fibrous tumour that is predominantly observed in males [1, 2]. The majority of these cases (about 90%) occur within the first 2 years of life and 23% of these are congenital [3].

The tumour predominantly occurs on the trunk, especially the axilla, and on the upper extremities, but in rare cases it has also been reported on the distal extremities, head, neck, and scrotum [1–3]. Only 1% arise in the legs [4]. To the best of our knowledge, this is the first report describing a fibrous hamartoma of the knee. Except for the sole report by Sotelo-Avila and Bale [4] regarding two patients who presented with simultaneous occurrence of a double lesion, FHI has always been reported as a single lesion, usually spanning 3–5 cm in diameter [2]. The tumour generally grows slowly. It presents as a painless mass that is usually freely movable, solitary, firm, and showing no involvement of the overlying skin [3]. There is

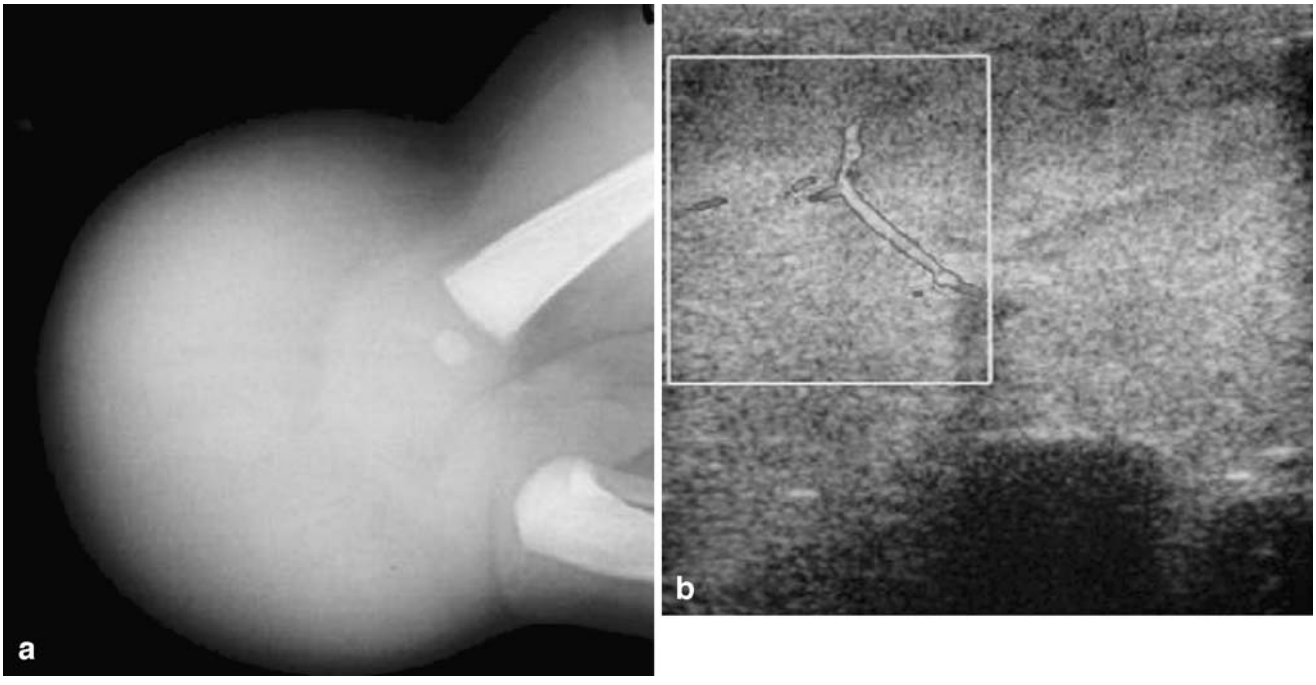

Fig. 2 Radiography (**a**) and sonography (**b**). **a** There is a large soft-tissue mass at the left knee. There is no bony abnormality and no calcification. **b** Colour Doppler US in the transverse plane. The peripheral tissue is homogeneously hypoechoic compared with the deepest homogeneously hyperechoic tissue. A large vessel coursing through the lesion is evident. The articular capsule was spared

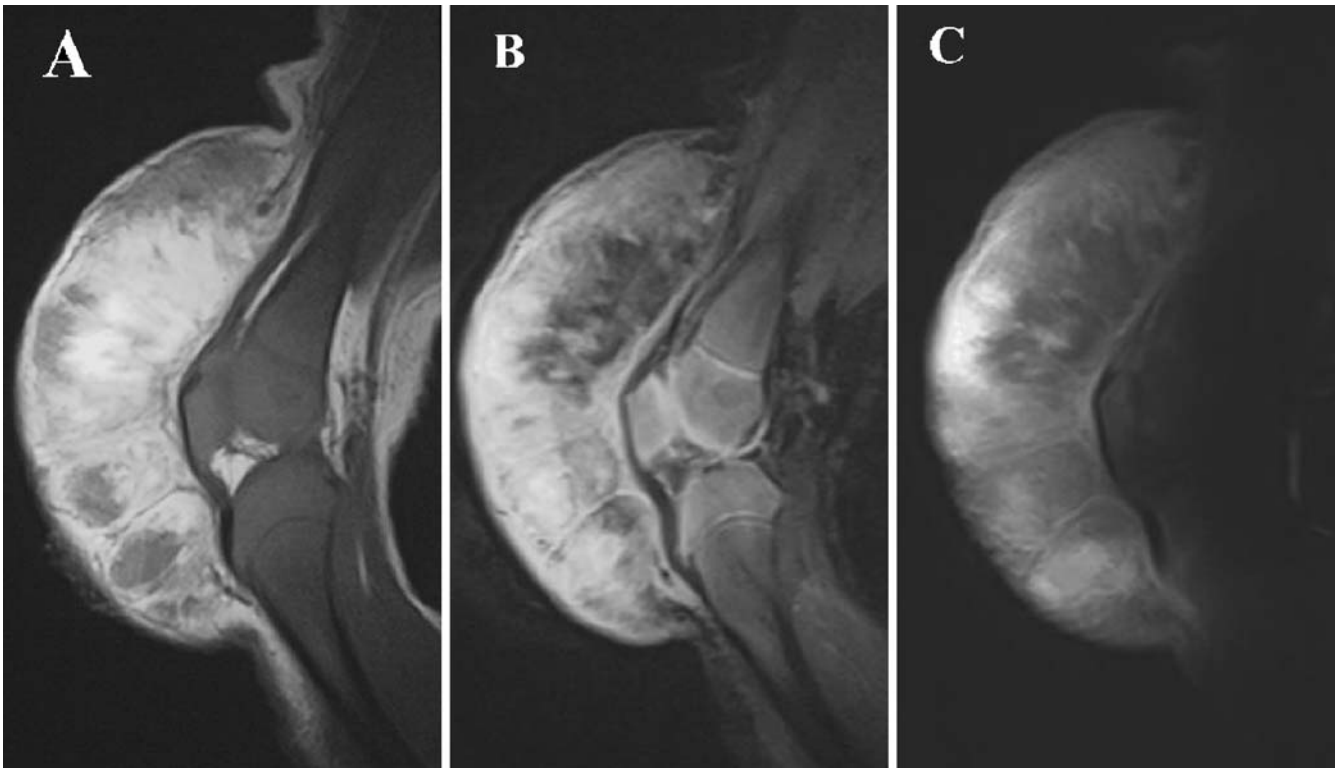

Fig. 3 MRI. Sagittal T1-W spin-echo image (**A**), sagittal STIR image (**B**), and sagittal T1-W spin-echo image with fat saturation after IV administration of contrast medium (**C**). The mass has a lobular structure and septa that are hypointense on the T1-W image and hyperintense on the STIR image. On the T1-W image (hyperintense) and STIR image (hypointense) the deepest and most peripheral parts of the lobules have the same fatty intensity as Hoffa's fat pad. The other tissue is slightly higher signal than normal muscle on the T1-W image and hyperintense on the STIR sequence. The mass shows contrast enhancement after IV injection of Gd-DOTA. The superficial part of the mass has high signal on all sequences

no evidence of regression, transformation, or malignant degeneration in the natural history [3]. The treatment of choice is local excision [1–3]. Local recurrences have been reported in up to 16% of cases due to inadequate primary excision of the lesion [3].

In our case, radiography and sonography revealed a non-specific soft-tissue mass. The sonographic pattern was probably due to the large number of interfaces that caused increased echogenicity and impaired assessment of its anatomical structure. We were able to detect the three distinct components by MRI. The trabeculae were hypointense on T1-W spin-echo sequences. Furthermore, they were hyperintense both on STIR sequence and after IV administration of contrast medium. The myxoid stroma appeared slightly hypointense on T1-W sequences and hyperintense on the STIR sequence and after the IV injection of Gd DOTA. Only fat tissue was characterized.

In the newborn, the differential diagnosis includes soft-tissue tumours and tumour-like lesions, i.e. vascular, fibrous and fatty lesions. The clinical features and fat-equivalent signal on MRI in our patient excluded haemangioma and vascular malformations. There were no vascular structures on US to suggest a haemangioma. Venous vascular malformations are usually soft, easily compressible, cold, bluish, and have normal overlying skin. They increase in size with various postures, evoke crying on compression, and have cystic lymphatic structures. None of these findings was detectable in our patient. The high degree of fat-equivalent signal on MRI allowed us to exclude infantile myofibromatosis, infantile fibrosarcoma and congenital embryonal rhabdomyosarcoma. However, the MRI appearance did not allow the exclusion of a lipoblastoma that typically reflects the varying contribution of mature lipoblasts vs. myxoid matrix and fibrous network.

The histological features we observed in our case pointed to a diagnosis of FHI. An interesting, and hitherto undescribed feature was the eosinophilic infiltration within the tumour and its association with foci of eosinophilic spongiosa. Furthermore, the present case was characterized by the unusually large size of the tumour.

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