

Fibrous hamartoma of infancy. Case report

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Abstract

Background: Fibrous hamartoma of infancy (FHI) is an infrequent benign proliferation of the soft tissues. Ninety one percent of cases occur during the first year of life. FHI is characterized as a subcutaneous lesion with characteristic morphology with a triphasic organoid growth pattern.

Clinical case: We present the case of a 6-month-old male infant with a tumor in the medial plantar region of the left foot, which was completely withered. Histopathological study reported a fibrous hamartoma of infancy.

Conclusions: Fibrous hamartoma of infancy most frequently affects males. It may be localized at any anatomic site, although 5-10% of cases affect the lower limbs. Few cases are reported in the foot. This is a lesion with a characteristic morphological pattern; however, in small biopsies, its differential diagnosis is important with other fibroadipose lesions for appropriate treatment. Ample surgical resection with lesion-free borders confers a good prognosis for these patients.

Key words: fibrous hamartoma of infancy, pediatric soft tissue tumors, lipofibromatosis, hamartoma, fibrous tumors.

Introduction

Fibrous hamartoma of infancy (FHI) is an infrequent, benign soft tissue tumor. It was originally described by Reye in 1956 as “subdermal fibrous tumor of childhood”¹⁻⁴ and was later reported by Enzinger and coined as “fibrous hamartoma of infancy” in 1965.⁵ It is usually diagnosed during the first 2 years of life. It is observed in 91% of cases within the first year of age; 15-20% of these tumors are present at birth and they are seen more often in males.² Its anatomical location is variable and may be located anywhere in the body. However, it is rarely found in the foot region where it has only been reported in a few cases.^{1,6,7} From a clinical and radiological standpoint it is not a distinct lesion and is characterized by rapid growth. It is usually asymptomatic, similar to malignant

lesions of the soft tissues. Definitive diagnosis is made after histological evaluation. It presents a characteristic three-phase growth pattern that differentiates it from other fibrous lesions, which is important for proper surgical treatment.

Case Report

We present the case of a 6-month-old male who was the product of a normal pregnancy with no medical history pertaining to the present illness. His clinical picture began at 6 months characterized by the presence of a soft-tissue tumor of the medial plantar region of his foot, which progressively increased, becoming more evident after he began to wear shoes.

During physical examination, a subcutaneous tumor was identified with an axis >3 cm in the medial plantar region of the left foot. The tumor was well delimited, nonpainful, movable and not fixed to deep structures. The skin did not show trophic changes. Increase of the radiopacity of the soft tissues in the medial plantar region of the left foot and normal bone structure were noted on radiographic study (Figure 1). A hypoechoic tumor was identified on ultrasound that was homogeneous, oval, and with regular and well-defined borders measuring 35 × 9 × 13 mm (Figure 2).

An incisional biopsy was initially performed with the diagnosis of “lipoblastoma” and surgical treatment was decided upon. Subsequently, wide and complete resection of the lesion was performed (after regional anesthesia and sedation) through an incision of the skin on the medial plantar region and dissection through identified planes of the lesion. It was resected with wide

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Figure 1. Dorsoplantar and lateral x-ray of the left foot with soft tissue technique. Increase of the soft tissue in the region of the medial half and left hind. Bony structures are normal.

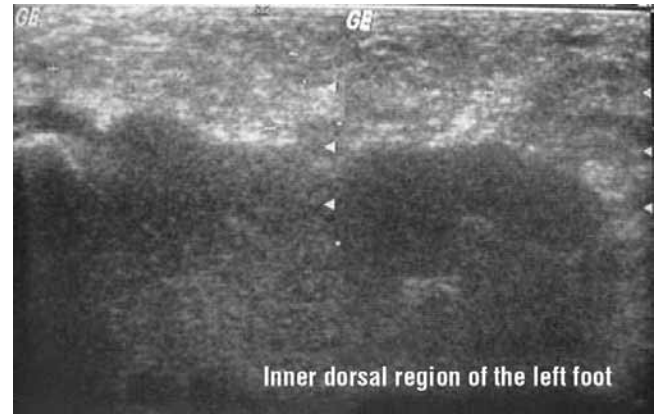


Figure 2. Ultrasound of the soft tissue of the medial plantar region of the left foot. Hypoechoic, homogeneous, oval tumor with regular and well-defined edges.

margins of healthy tissue. Postoperative evolution was without complications.

In the anatomico-pathological study, the macroscopic sample corresponded to a surgical specimen that measured $5 \times 3 \times 1$ cm and was composed of a spindle of skin 1.7×4 cm with light-brown epidermis and subcutaneous injury. The specimen was unencapsulated with poorly defined borders and was grayish-white and light yellow in color. It was lobulated with a fibrofatty appearance. Dissection was rubbery with fibrosis areas (Figure 3).

In the microscopic sections we identified a subcutaneous lesion (Figure 4) consisting of a homogeneous mixture of mature adipose tissue intercepted by fibrous tissue septa. We identified areas with immature mesenchymal tissue (Figure 5) composed of small oval cells with spindle-shaped nucleus and starry aspect embedded in a myxoid matrix rich in mucopolysaccharides evidenced with alcian blue stain (Figure 6). Fibroblast-like cells of the mesenchymal tissue were immunopositive for vimentin in the immunohisto-

chemical study (dilution 1:75 μ l, Monoclonal Mouse Anti-Vimentin, Dako Cytomation, Carpinteria, CA). Adipose tissue was immunopositive for S-100 protein (dilution 1:200 μ l, Polyclonal Rabbit Anti-S100 Dako Cytomation). Fibroblastic areas were immunopositive for actin (dilution 1:100 μ l, Monoclonal Mouse Anti-Human Muscle Actin, Dako Cytomation) and vimentin (dilution 1:75 μ l, Monoclonal Mouse Anti-Vimentin, Dako Cytomation). Histopathological diagnosis was FHI.

Discussion

FHI is a benign soft tissue tumor of infrequent presentation. It is a mesenchymal lesion of uncertain histogenesis with a variable



Figure 3. Surgical specimen: subcutaneous ill-defined tumor margins and fibrofatty appearance.

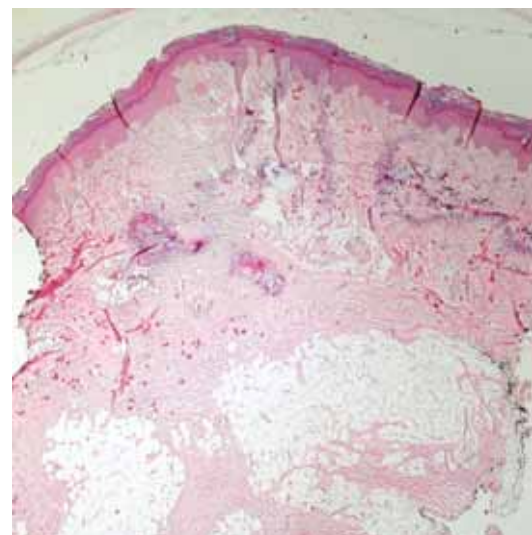


Figure 4. Histological cut, panoramic view. Subepidermal lesion with characteristic organoid arrangement of its components.

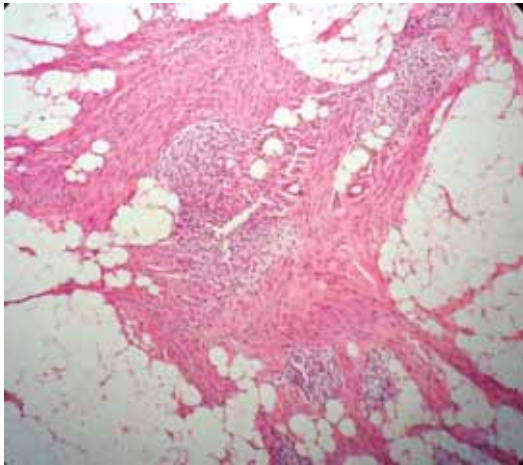


Figure 5. FHI composed of a mixture of mature adipose tissue, septa of fibrous tissue and immature mesenchymal tissue.

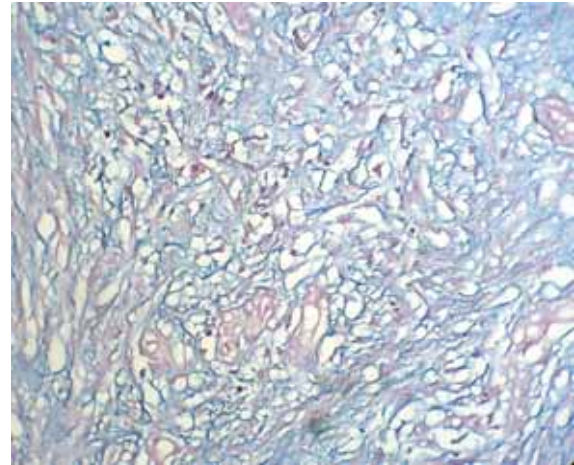


Figure 6. Component mesenchymal cells of stellate appearance embedded in myxoid matrix rich in mucopolysaccharides with alcian blue stain.

but distinctive histological appearance. It was initially described by Reye in 1956 as a tumor derived from myofibroblasts and referred to as a “subdermal fibrous tumor of childhood.”¹⁻⁴ In 1965 Enzinger referred to it as a “fibrous hamartoma of infancy” and, in turn, divided fibrous proliferations of children into two groups:⁵

1. Injuries that due to their location, morphology and behavior are similar to those found in adults, such as facial fibromatosis and desmoid tumor
2. Lesions without clinical or morphological counterpart in adult life

WHO refers to it as “a pediatric tumor, benign, poorly delimited, superficial to soft tissues, characterized by three components mixed in an organoid pattern: trabeculae of fibrocollagenous tissue, well-defined, loose areas of mesenchymal cells with an immature appearance, and small, rounded and mature adipose tissue.”⁸

In recent years, Fetsch et al. referred to FHI as part of a group of lesions whose main component is a mixture of adipose tissue and fibroblastic elements, designating them in a new classification as “lipofibromatosis” including fibromatosis (congenital and infantile and juvenile), calcifying aponeurotic fibroma, lipoblastoma and FHI in the differential diagnosis.⁹

Although Reye described it as a reparative process, there are no histological characteristics that suggest this as a local response to injury, rather it seems consistent with a hamartomatous natural injury, but we cannot exclude the possibility that this is a benign neoplasm.⁴ In a recent study, Lakshminarayanan reported a case of FHI with t(2;3)(q31;q21), which may confirm the neoplastic origin.¹⁰ History of trauma has occasionally been reported at the time of presentation of FHI but apparently is not related to its histopathogenesis.^{1,4}

FHI generally is diagnosed during the first 2 years of life, and ~15-20% of cases occur in newborns^{1,2,6,11} with ~25% being

congenital. In larger series studied, it has been reported that up to 91% of the cases occur during the first year of life⁶ with a higher prevalence in males (2.4:1).^{3,6,12} Female African patients have reported a higher prevalence, with a maximum age of presentation at 10 years of age.¹¹

FHI is a solitary lesion that can occur in any anatomic site. It generally occurs in the upper extremities but may be seen in axilla, sacral region, perineum (scrotum), buttocks, and head and neck area,^{2,6} with a similar distribution in males and females. Although most cases involve a subcutaneous nodule, there have been some reports of multiple nodules.^{6,13,14} Occurrence of these in the lower extremities has been reported in 5-10% of the cases, but its presentation in the foot is very uncommon.^{1,2,6} In the case reported, it was located in the medial plantar region of the foot. With respect to this location in the literature reviewed, we identified only four cases previously reported in this anatomic site, which occurred in male patients <2 years of age.^{1,6,7}

Clinically, this lesion is characterized by a progressive and asymptomatic growth pattern. On physical examination, these lesions are solid, nonpainful subcutaneous nodules, which may or may not be fixed to deep structures,^{2,4-6} similar to some malignant lesions, an important reason to arrive at a differential diagnosis.^{3,12,15} Cutaneous and trophic changes on the skin are not apparent; however, in histological sections we have seen reports of hyperplasia and ductal eccrine ectasia, squamous syringometaplasia, intraluminal papillary formations, and follicular basal cell hyperplasia.^{6,16} On x-rays they may be similar to other fibrous soft tissue lesions; however, ultrasound and magnetic resonance imaging may support the diagnosis specifically.^{17,18}

These lesions are regularly delimited and can measure 0.5-10 cm in diameter;^{2,4-6,12,14} however, in those cases with a long clinical course the lesions present a well-defined capsule that can measure up to 20 cm in diameter.¹¹ They are lesions that are identified by

dissection due to their rubbery texture and grayish-brown and light-yellow coloring.^{2,4-6,11,12,16}

In histological dissections we identified a subcutaneous or reticular dermis lesion consisting of three principal components mixed together: dense bands of fibrous tissue that project and intercept the adipose tissue, tissue areas rich in primitive mesenchymal cells in mucopolysaccharides represented by immature round cells, and mature adipose tissue in varying amounts arranged in a characteristic organoid pattern.^{2,4-6,8,11,12,16} IHC studies identified vimentin + fibrous and mesenchymal component as well as positive actin in the fibrous component. S-100 protein is only positive in adipose tissue.^{4,6,11,19-21}

The treatment of choice for FHI is local excision^{6,11,12} with wide surgical limits and with deep extension including adjacent soft tissues.²² Recurrence after complete resection occurs in 10-16% of the cases and no malignant changes have been reported with this injury so far.^{2-6,12}

In conclusion, FHI is a lesion with typical morphological characteristics, however, its diagnosis of small size biopsies can be misleading when the typical organoid pattern and tri-phase of the lesion are not recognized by the examiner. These may be mistaken for sarcomas or other fibroadipose lesions that should be taken into account for its differential diagnosis, coupled with the clinical correlation. Radiographic studies are not specific to the diagnosis, but an ultrasound helps to characterize and determine the location of the lesion.

The recognition and differentiation of FHI is important in conjunction with other forms of fibromatosis because this is a benign lesion with good prognosis and whose treatment of choice is complete surgical excision with wide margins. Recurrence occurs in up to 16% of cases treated with re-excision. In the case presented here, the patient was monitored for 20 months with an asymptomatic course and without recurrence of the lesion. FHI is a lesion with a very rare presentation in the foot; however, it is very important to consider it as a possible diagnosis in order to provide proper treatment.

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