

Fibrous Hamartoma of Infancy, Report of Two Cases

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Abstract

Objective: Fibrous hamartoma of infancy (FHI) is a rare, benign soft tissue tumor that typically occurs within the first two years of life. It has a specific histologic appearance comprising of three different mesenchymal tissues with variable proportions in an organoid fashion. The clinical course is typically benign with excellent prognosis. We report two cases of this rare lesion and review its clinicopathologic characteristics.

Case(s) Presentation: The first case was a 15-month-old girl who had a subcutaneous mass in the right axillary region and the other one was an 18-month-old boy with a mass on the medial surface of his right ankle. The masses were successfully excised. After 6 and 30 months follow up no recurrence occurred.

Conclusion: Although the clinical and imaging findings of FHI are quite similar to those of malignant soft tissue tumor, histologic characteristics of this tumor will guide to the definite diagnosis that will prevent aggressive and mutilating procedures.

Key Words: Fibrous hamartoma of infancy; Tumors of infancy; Soft tissue tumor; Hamartoma; Infancy

Introduction

Fibrous hamartoma of infancy (FHI) is a rare, benign soft tissue tumor that usually occurs within the first two years of life. It was first described by Reye in 1954 as subdermal fibromatous tumor of infancy and later its current name was coined by Enzinger^[1]. The lesion is characterized by rapid initial growth, but generally it does not cause any symptoms.

The male-to-female ratio is 2.4, and there is no apparent familial or syndromic association.

FHI is most commonly found in the axilla, shoulder, upper arm, upper trunk and external genital areas^[2]. Histologic evaluation demonstrates a typical organoid pattern, with a mixture of fat, mature myofibroblasts arranged in fascicles and immature mesenchymal cells. Although this lesion is benign, it is very important to differentiate it

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from other soft tissue masses. As far as we know, these are the first cases of FHI reported in Iran.

Case(s) Presentation

Case 1: A 15-month-old girl presented with a soft, mobile subcutaneous mass without adhesions in the right axillary region (Fig 1). Her mother had noticed this lesion a week before admission. Excisional biopsy was performed. Macroscopic examination revealed a lobulated and poorly circumscribed yellowish and fragile tissue resembling fat with scattered gray-white fibrotic foci measuring 3.7×1.9×0.7cm (Fig 2).



Fig 1- subcutaneous mass in right axillary region



Fig 2- ill-defined fatty mass with bands of white fibrous tissue

Histologically, it was composed of three types of tissue, interlacing fibrous trabeculae, islands of loosely arranged spindle-shaped cells, and mature adipose tissue (Fig 3, 4). Immunohistochemical staining revealed a positive reaction for vimentin in primitive mesenchymal cells. Smooth muscle actin, desmin and S-100 stains were negative. After 6 months following surgery, the patient shows no sign of recurrence.

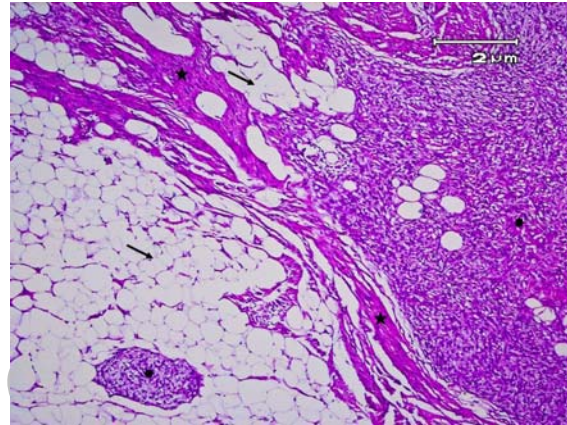


Fig 3- FHI showing a characteristic organoid pattern composed of interlacing fibrous trabeculae (★), islands of loosely arranged spindle-shaped cells (●), and mature adipose tissue (→) (H&E; ×100)

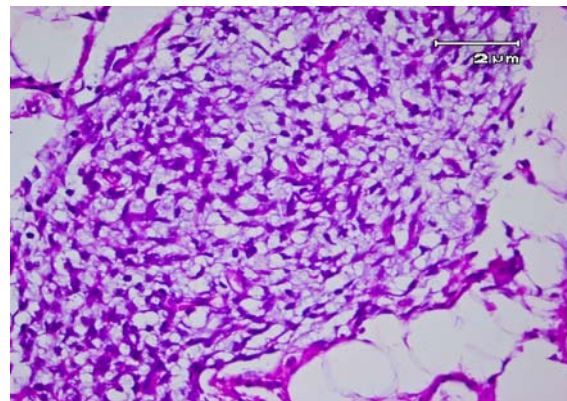


Fig 4- High power view of cytologically bland spindle-shaped cells deposited in myxoid stroma in FHI (H&E; ×400)

Case 2: An 18-month-old boy presented with a history of slowly growing mass on the medial

surface of his right ankle, noticed shortly after birth. On physical examination patient appeared healthy. A fixed and soft 3×2 cm mass was palpable on the medial surface of the right ankle. The mass appeared extra-articular and the range of motion was not decreased. Radiologically, there was no evidence of bone involvement. Complete excision of the lesion was performed. Macroscopic examination revealed 5 fragments of fibrofatty tissue measuring 3×1.2×0.5 cm to 0.3×0.2×0.1 cm. Microscopic examination was indicative of FHI. After 30 months follow-up no recurrence occurred.

Discussion

Some varieties of soft tissue lesions are noted among children, which have no clinical or morphologic counterpart in adults. FHI is one of these lesions with unique clinicopathologic features. It is an uncommon benign fibroblastic and myofibroblastic proliferation, that typically occurs within the first two years of life and nearly 25% occur congenitally^[2]. Most cases presented as solitary masses, but multiple separate synchronous lesions have been reported rarely^[3]. The size ranges from 0.5 cm to very large masses over 20 cm^[2].

Preoperative diagnosis by fine-needle aspiration (FNA) has been established only in a few cases^[4]. Thus for definitive diagnosis, we need to have a complete excision biopsy.

Grossly, it is poorly circumscribed with firm gray-white tissue admixed with fat. FHI has a specific histologic appearance comprising of three different mesenchymal tissues with variable proportions in an organoid fashion: fibrous tissue with amounts of collagen, mature adipose tissue, and loosely textured areas consisting of immature small rounded to stellate cells embedded in a myxoid matrix. The spindle cells react primarily with vimentin and to a lesser extent with smooth muscle actin. Ultrastructural examination reveals an admixture of fibroblastic and myofibroblastic cells^[1].

Two cytogenetic analyses have been reported to date in FHI^[5,6]. First one revealed a reciprocal translocation, t(2; 3) (q31; q2). A possible gene on chromosome 2q31 may be related to the development of FHI. The vitronectin receptor- α subunit gene belongs to the integrin family. Integrins serve as the major receptors for extracellular matrix-mediated cell adhesion and cellular proliferation, apoptosis, and differentiation^[5]. Recently Rougemont et al, showed a complex translocation(6; 12; 8) (q25; q24.3; q13) in FHI^[6]. This finding confirms that this lesion represents a true neoplasm with a monoclonal origin. The differential diagnoses to be considered are lipofibromatosis, infantile digital fibroma, myofibroma, fibromatosis, infantile fibrosarcoma and also embryonal rhabdomyosarcoma especially in the genital region^[1,5].

Pediatric soft tissue tumors composed of adipose tissue and fibroblastic elements have been recently classified as lipofibromatosis by Fetsch et al^[7]. Histologically they are composed of abundant adipose tissue traversed by bundles of fibroblast-like cells but there are no primitive mesenchymal cells as FHI. For differentiating FHI from infantile digital fibroma sites of involvement and presence of intracytoplasmic perinuclear inclusions can be useful. Myofibroma has a characteristic hemangiopericytoma-like pattern and calcifying aponeurotic fibroma has a zonation pattern which can be helpful in differentiating these lesions from FHI^[1,5].

Because some FHI occur in the scrotal region, the spindle cell form of embryonal rhabdomyosarcoma enters in the differential diagnosis but this lesion generally occurs in older children and is composed of cells with more cytologic atypia. Awareness of characteristic organoid pattern also facilitates distinction from infantile fibromatosis and infantile fibrosarcoma^[1].

Carretto et al evaluated 18 children with FHI whose data were collected from 6 centers of pediatric surgery and pediatric oncology. Their results show that complete excision is the treatment of choice for FHI; even with

incomplete excision it has a low recurrence rate between 10-15%. There is no evidence of malignant transformation or spontaneous regression. Long-term clinical follow-up indicates a benign biological behavior^[8].

Conclusion

FHI is a rare and benign soft tissue tumor that typically occurs within the first two years of life. Although its clinical and imaging findings are quite similar to those of malignant soft tissue tumor, the histologic characteristics of this tumor will guide to the definite diagnosis that will prevent aggressive and mutilating procedures.

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