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Olgu Sunumu / Case Report

Gluteal Congenital Fibrous Hamartoma of Infancy: A Case Report

Konjenital Infantil Fibröz Hamartom: Bir Olgu Sunumu

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ABSTRACT

Fibrous hamartoma of infancy (FHI) is a rare benign subcutaneous tumor which is commonly seen in the first year of life. Patients usually present with a single, painless, rapidly growing mass located mostly on the upper extremities. It can be difficult to distinguish this lesion from the malignant soft tissue sarcomas because of the rapid growth and radiological characteristics. Here we report a 5-month-old boy with a large gluteal mass and cutaneous findings which was initially thought to be an infiltrative malignant tumor and finally diagnosed as FHI.

Key Words: Fibrous hamartoma of infancy, congenital, gluteal

ÖZET

İnfantil fibröz hamartom sıklıkla hayatın ilk yılında görülen, nadir ve benign bir subkutan tümördür. Hastalar genellikle üst ekstremite yerleşimli ağrısız ve hızlı büyüyen bir kitle nedeniyle başvurmaktadır. Kitlenin radyolojik özellikleri ve hızlı büyüme özelliği nedeniyle yumuşak doku sarkomlarından ayırt edilmesi zor olabilir. Burada büyük bir gluteal kitle ile başvuran 5 aylık bir erkek çocukta infiltratif malign tümör özellikleri gösteren fibröz hamartom olgusu sunulmaktadır.

Anahtar Kelimeler: İnfantil fibröz hamartom, konjenital, gluteal

INTRODUCTION

Differential diagnosis of the soft tissue masses located around the pelvis can be difficult to make due to indistinguishable physical examination and radiological findings. Teratoma, yolk sac tumor, rhabdomyosarcoma and other soft tissue sarcomas, vascular lesions (hemangioma, vascular malformation), adipose tissue tumors, fibroblastic or fibrohistiocytic tumors are among the most common lesions of this area[1,2]. Most of these lesions may initially resemble a dermatologic disorder beacause of various cutaneous findings such as papule or nodule formation, color changes or increased hair growth around the lesions[3,4]. Fibrous hamartoma of infancy (FHI) is an uncommon benign tumor of hamartomatous origin which is usually diagnosed in the first year of life. Males are more commonly affected than girls and approximately 25% of cases are congenital. Patients usually present with a rapidly growing solitary mass commonly located

at upper extremities and upper trunk[5,6]. Here we report a congenital case of gluteal FHI with increased hair growth on the overlying skin.

CASE REPORT

An otherwise healthy 5-month-old boy presented with a rapidly growing gluteal mass which was noticed at birth. On physical examination there was a 10x7 cm non-tender right gluteal mass with multiple coarse hairs on the overhanging skin. Magnetic resonance imaging (MRI) showed a solid mass located at the presacral region infiltrating the subcutaneous tissue (Figure 1A and B). Serum alpha-fetoprotein was within normal values. Initial diagnosis was a malignant solid tumor because of the infiltrative radiological features. Surgical excision was performed. The gluteal part of the mass was totally excised with the surrounding adipose tissue. Since, total excision was not possible for the presacral part of the mass, it was excised in pieces and a small amount of residual tumor was left. Histopathological examination revealed a tumor consisting of various amounts of fibrous tissue, mature adipose tissue and immature mesenchymal cells with low mitotic activity (Figure 2). Local infiltration to the adjacent muscles was observed. Immunohistochemical staining showed diffuse vimentin, focal desmin and smooth muscle actin positivity. These findings were compatible with FHI. The patient is under routine follow-up with a residual perirectal mass. Radiological and clinical follow up showed no growing of the residual mass two years after the surgery.

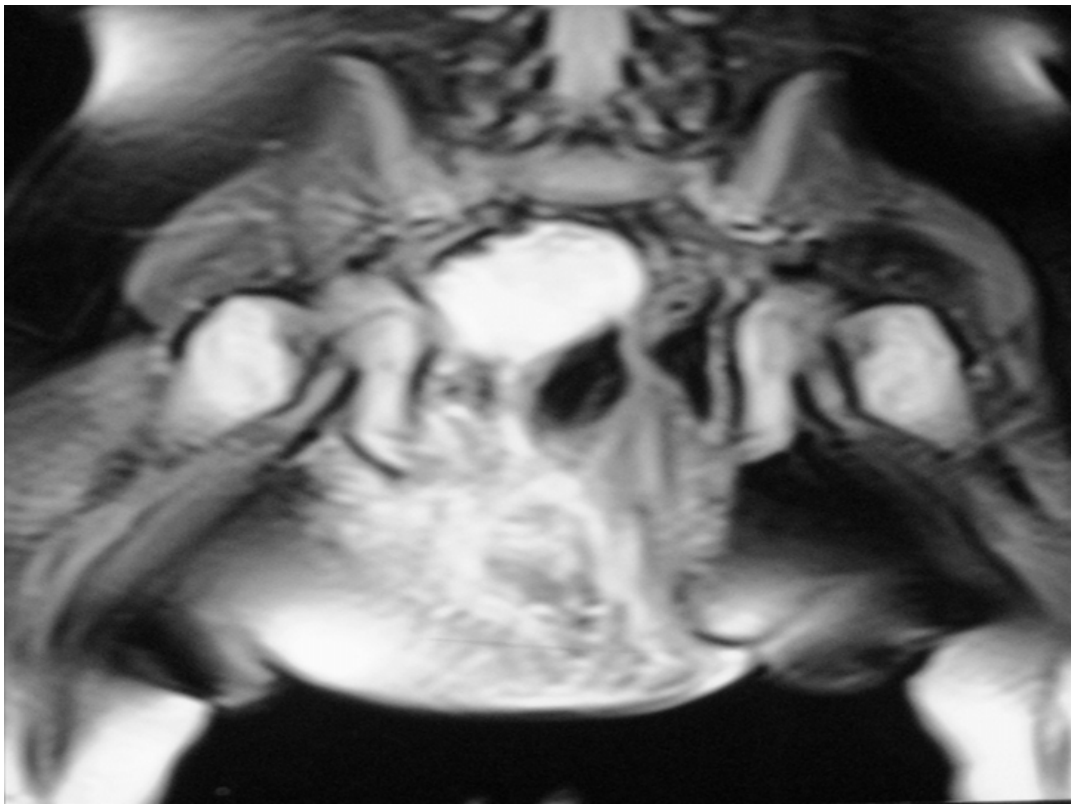

Figure 1A. Sagittal T1-weighted contrast enhanced MR image demonstrates enhancing presacral mass and subcutaneous tissue.

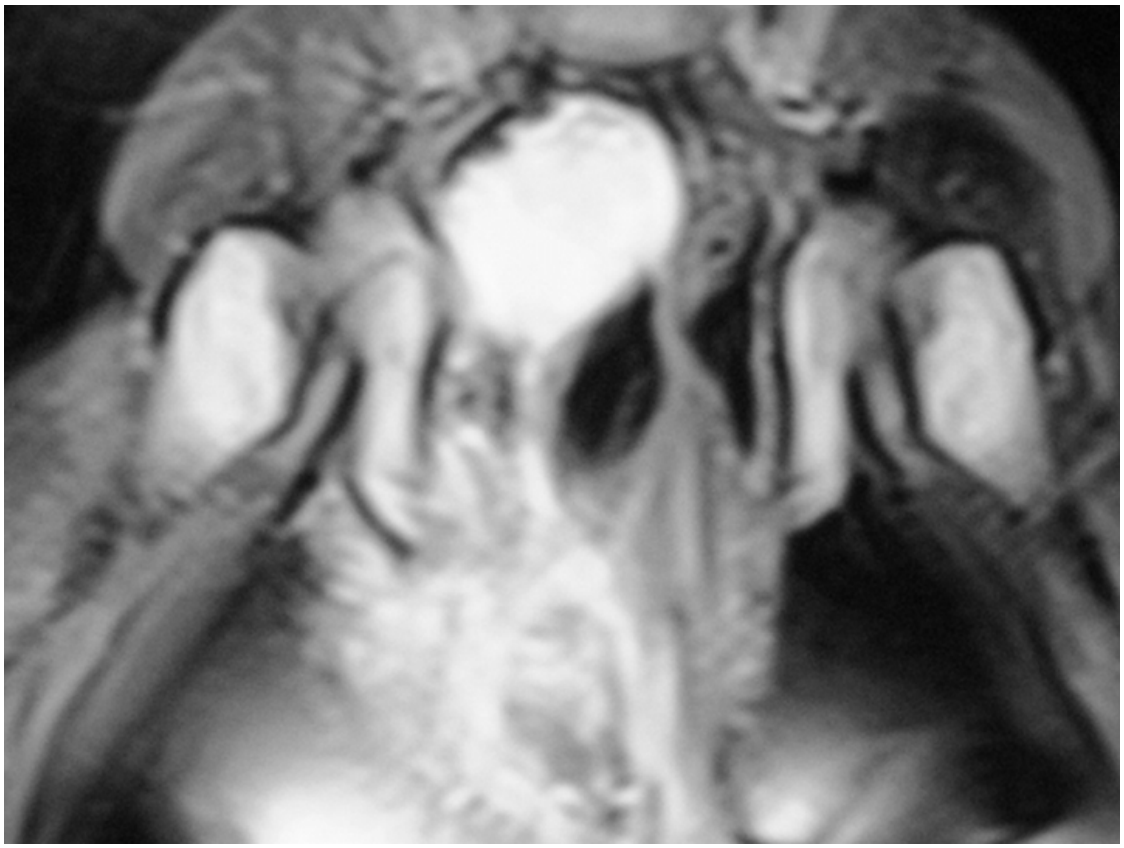

Figure 1B. Coronal T2-weighted MR image shows pelvic origin of the solid tumor with high signal intensity. Note is infiltrating subcutaneous tissue.

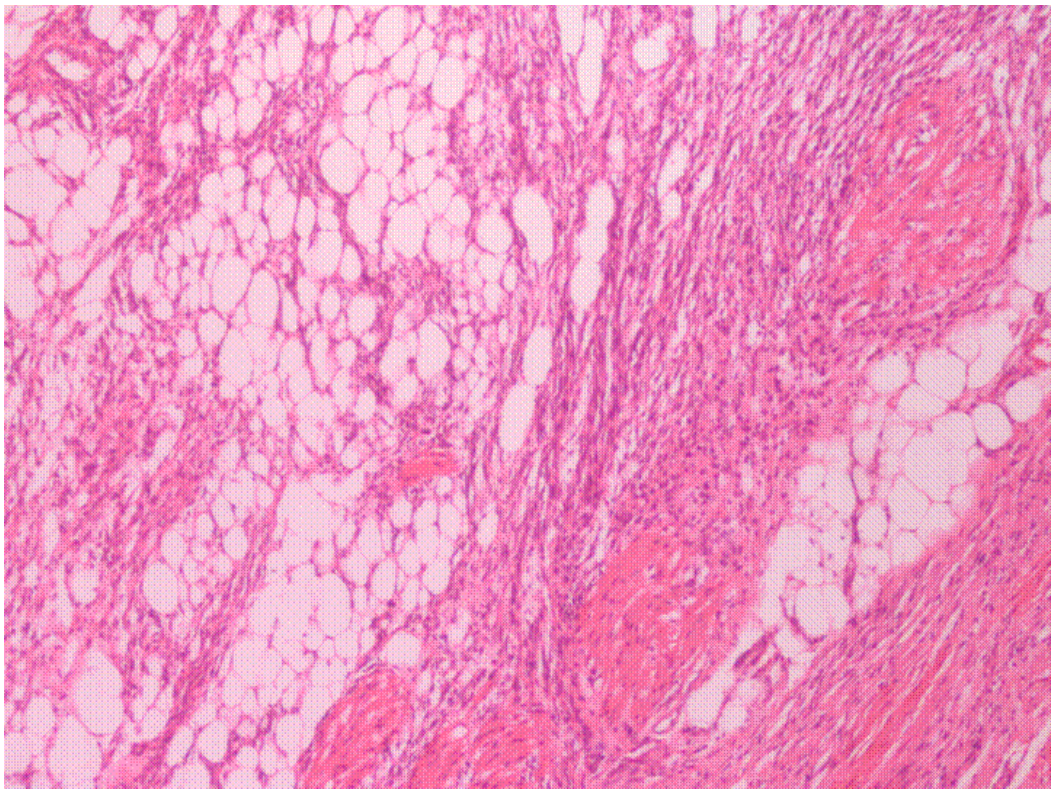

Figure 2. Tumor consisting fibrous tissue, mature adipose tissue and immature mesenchymal cells. HE. Original magnificationX100.

DISCUSSION

Fibrous hamartoma of infancy is a rare benign tumor which is located within the subcutis rarely involving the lower dermis. Despite the rapid growth at presentation, the lesion has benign histopathological features and the growth pattern usually slows down as the patient gets older[5]. Because of the rapid growth and the infiltrative radiological features on MRI scan, initially a malignant tumor was considered in this patient.

Most lesions are detected in the first year of life with a male/female ratio reaching up to 3[5,6]. Approximately a quarter of cases are congenital[5]. Overlying skin changes such as increased hair growth[7,8], pigmentation changes[7], eccrine gland hyperplasia, eccrine duct ectasia, syringometaplasia, and adnexal hyperplasia with differentiation[9] were reported. Overlying hypertrichosis could have been a clue to the diagnosis in our case. However, such a finding could also be seen in benign lesions like occult spinal dysraphism[10], teratoma[4], or tufted angioma[11].

Although the shoulder girdle area and upper extremities are the most common sites of involvement, the lesion can be located at anywhere throughout the body. Gluteal or sacral regions are relatively uncommon locations and less than twenty cases were reported at these sites[5,6,8,12]. Especially the sacrococygeal lesions in this age group should be distinguished from other benign or malignant tumors such as sacrococygeal teratomas or yolk sac tumor. Alfa-fetoprotein can be useful in the differential

diagnosis. The lesions tend to be solitary and less than 5 cm in diameter. However, tumors reaching up to 20 cm[13] and separate multiple synchronous[14] and multinodular lesions[8] were also reported.

The treatment of choice is wide local excision with clear surgical margins[5,6]. Although the malignant degeneration has not been reported, the main reason of the surgery is the need for histopathological diagnosis. Furthermore, as spontaneous regression is not expected, observation without excision seems to be a time consuming approach. Local recurrence rate is approximately 15% and is usually seen when radical excision can not be done. Patients should be followed for local relapses especially in case of subtotal excision as in this patient.

In conclusion, malignancy should not be regarded as a rule in case of rapidly growing mass in infants. Overlying cutaneous findings can be a clue to the diagnosis of FHI.

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