

## CASE REPORTS

# JUVENILE HYALINE FIBROMATOSIS: REPORT OF A CASE

## JUVENİL HYALEN FİBROMATOSİS : OLGU SUNUMU

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**SUMMARY:** This paper presents a case of Juvenile Hyaline Fibromatosis located on the back of a child. After total excision of the lesion, the microscopic appearance showed hyalinized lobules separated by spindle cells and Juvenile Hyaline Fibromatosis was the diagnosis. This is the second localized case reported from Turkey in the English literature.

**Key Words:** Fibromatosis, Juvenile Hyaline Fibromatosis.

### INTRODUCTION

Juvenile Hyaline Fibromatosis (JHF) was first described by Drescher in 1967 under the name "Fibromatosis Hyalinica Multiplex". Since then 40 cases have been reported in the literature and the details about its clinical presentation, pathogenesis and genetical aspects were described (1-3). In this paper, a case report of JHF is presented and its differential diagnosis discussed.

### CASE REPORT

The child was born to 33-year-old mother in July, 1991. He was born at term by normal delivery as a second child and 3800 g in weight. There were no significant perinatal and postnatal problems. He had a healthy motor and mental development. His parents were not relatives and his parents and brother were no significant health problems. On day 20, the lesion was first noticed on his back by his mother as a nodule about 1 cm

**ÖZET:** Bu çalışmada küçük bir çocuğun sırt bölgesinde yerleşen Juvenil Hyalen Fibromatosis igusu sunulmaktadır. lezyon tamamen çıkarılmış, iğsi hücrelerden oluşan hyalinize lobullerle karakterli lezyon saptanmıştır. bu olgu, ingilizce literatürde Türkiye "den bildirilen ikinci olgudur.

**Anahtar Kelimeler:** Fibromatosis, Juvenil Hyalen Fibromatosis.

in diameter. During the next 14 months the size of the lesion increased gradually.

When the patient was admitted to our hospital, a painless, firm and mobile nodule, 2x2 cm in length was palpated under his left scapula. Computerized tomographic (CT) scan revealed an encapsulated mass, 16x35 mm in diameter in the soft tissue of the chest wall and there were no connections with the bony structures. It also had a hypodense area, compatible with the soft tissue lesion called "Hypertrophied musculus infraspinalis/rhamboides". The lesion was excised totally.

On macroscopic examination, the excised lesion consists of a nodular, encapsulated mass, 35x25x15 mm in diameter. It has a gray-white cut surface with foci of hemorrhage. Microscopically, the tumor displays a nodular growth pattern characterized by uniform hyalinized lobules separated by spindle cells (Fig.

1). The centre of the nodule is more cellular than its periphery and associated with cleft-like, richly vascular areas. Some of the spindle cells are in lacunes. The nuclei are slender and serpentine-like (Fig. 2).



Fig. 1: Low-power picture shows nodular growth pattern characterized by uniform hyalinized lobules separated by spindle cells (H&E x40).

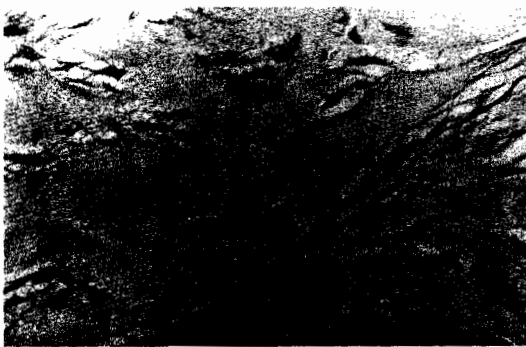


Fig. 2: The nuclei in lacunes are slender and serpentine-like (H&E x200).

Histochemically, Phosphotungstic acid-Hematoxylin (PTAH) preparation fails to demonstrate myofibrils and silver preparation demonstrates slight increase in reticulin fibers. The hyalinized matrix is not positive on staining with Periodic Acid-Schiff (PAS) and Congo-red methods. Immunohistochemically the cells are negative with S-100, Desmin and Actin.

## DISCUSSION

Juvenile Hyaline Fibromatosis (JHF) is an autosomal recessive disease characterized by slow growing multiple subcutaneous nodules, gingival enlargement and joint deformities which appear early in life (1-5). There were 40 cases reported in the literature and a Turkish child was recently published (3). In our case we noticed that the nodule was growing slowly but no gingival enlargement or joint deformities as mentioned in the literature were observed (6, 7). These were not seen in the case of Uğraş et al and De Rosa et al (3, 8). De Rosa et al also postulated the existence of two distinct forms of JHF localized which is characterized only by limited cutaneous involvement as seen in our case as well as in Uğraş et al and the diffuse form in which cutaneous involvement is wide, with large and rapidly growing tumors which can be associated with visceral involvement with small skin tumors and very slow growth.

There are several soft tissue tumors of the pediatric age group. In general, soft tissue tumors can be categorized according to their cell type of origin; fibroblastic-myofibroblastic, fibrohistiocytic, lipomatous, myogenous, neurogenous, pigmented and vascular (or endothelial) (9). History and physical examination, with a focus on the child's age at presentation, location of the tumor, and rate of growth are the initial steps in evaluation of a soft tissue mass. We did not suggest lipomatous, pigmented or vascular tumor due to the clinical setting and the CT scan. Tumors of fibrous origin comprise most of these tumors in children and are usually benign. Because the diversity of pediatric soft tissue tumors can complicate diagnosis, surgical management is essential. All of these lesions are characterized by fibroblasts and collagen fibers with varying degrees of cellularity. The entities within this group include fibroma, giant cell fibroblastoma, fibromatosis, proliferative fasciitis (PF) and myositis (PM), dermatofibroma, dermatofibrosarcoma protuberans (DFSP), fibrosarcoma (FS), neurofibroma (NF) and in the childhood period; Whinchester's syndrome, lipid proteinosis (hyalinosis cutis et mucosae) and nodular amyloidosis (1,3,10).

Fibroma is a benign tumor of fibroblasts that can develop anywhere on the body. It is usually

painles similar to our case but is more cellular.

Giant cell fibroblastoma is a childhood tumor of subcutaneous tissue found in the back. It presents in the first decade of life as well but histologic examination makes the difference clear: It contains numerous giant cells in contrast to JHF.

Proliferative fasciitis and myositis are lesions of adults. They grow rapidly within several weeks and in general, they are poorly circumscribed. On histologic examination, a gray zone between dermis and subcutis can be seen. PF and PM consist of spindle cells. These cells were seen in our case but the difference is that the former have myxoid appearance. Also there are small foci of inflammation and necrosis and numerous basophilic giant cells.

Dermatofibroma is a slow-growing nodular fibrocellular proliferation involving dermis and subcutis similar to our case but the margins of DF are not sharply defined and the tumor consists of short intersecting fascicles of fibroblastic cells. The fascicles usually form a storiform pattern and are never separated by collagenized matrix. Furthermore, histiocytic cells, xanthoma cells and multinucleated giant cells of the foreign body or Touton type, which contain phagocitized lipid and hemosiderin can be seen in DF.

Dermatofibrosarcoma protuberans typically presents as a nodular, cutaneous, slow growing mass but seldom occurs in children and the first manifestation is the development of a plaque like lesion of the skin, having a red to blue discoloration. Storiform pattern and pleomorphism are helpful in differential diagnosis from JHF but the distinction may be difficult when only the superficial portion of the DFSP is present in a biopsy specimen..

Well differentiated fibrosarcoma may resemble JHF clinically but subcutaneous localization of FS is rare, the lesion is more cellular and there is more mitosis than an JHF.

Neurofibroma typically presents at adult age but must be distinguished from JHF because of its slow-growing subcutaneous nature and the slender, serpentine-like nuclei of its cells. The matrix is fibrillary rather than hyaline and there are more stromal mucosubstances, mast cell, lymphocytes and xanthoma cells in the former.

Neurofibromatosis is diagnosed clinically on the basis of cafe-au-lait spots, multiple neurofibromas and bony lesions. Cafe-au-lait spots can be congenital or develop during the first year of life and neurofibromas often develop around puberty and increase in number and size with time (7). We did not notice cafe-au-lait spots or bony lesions. Despite the slow growing subcutaneous nature seen in JHF, the nuclei in neurofibroma are serpentine-like.

The eosinophilic material is PAS positive in lipid proteinosis and Amyloid positive in nodular amyloidosis. Whinchester's syndrome is a rare disease characterized by periarticular thickening, joint deformities and fibrous proliferation without deposition of any hyaline matrix.

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