Juvenile hyaline fibromatosis in one Turkish child

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We describe a case of juvenile hyaline fibromatosis (JHF) in a Turkish child. Only about 40 cases of juvenile hyaline fibromatosis had been reported in English literature as of March 1998, and it had not been reported in English literature from Turkey as of November 1998. Juvenile hyaline fibromatosis characterized by multiple cutaneous masses is a rare hereditary disorder. This disease is usually found in children, and a malfunction of collagen synthesis is considered as the pathogenetic cause. In the presented case, light microscopy demonstrated an abundance of a homogeneous, amorphous, eosinophilic extracellular matrix in which fibroblasts were embedded. Well-formed collagen fibers could not be demonstrated with Gieson's method or with reticulin preparation. The hayalin material periodic acid-Schiff-positive and diastase-resistant, whereas the Congo red method was negative. Immunohistochemically, the spindle-shaped cells were actin (smooth muscle) negative.

Key words: juvenile fibromatosis, hyaline fibromatosis, fibromatosis.

Juvenile hyaline fibromatosis (JHF) is a rare hereditary disease with only about 40 cases reported in English literature as of March 1998[1]. It had not been reported in English literature from Turkey as of November 1998. This disease is usually first noticed in children between two and five years of age and it typically consists of multiple cutaneous papules, nodules, or tumor masses that vary in size from 1 mm to about 5 cm, that grow slowly and are painless, and that are found mainly in the regions of the head, back, and extremities, with a predilection for the nose, ears, scalp, back, and knees[2]. This disease affects all races[3]. We describe a case of JHF that exhibited two small dermal nodules located exclusively on the occipital region and mental region which were characterized by painless and slow growth. This case seems to be the first one reported in English literature from Turkey, as of November 1998.

Case Report

A four-year-old boy presented in August 1997 with the diagnosis of tonsillitis and a five-month history of two painless, slowly growing tumors in the mental and occipital regions. The family history was negative. He was born of healthy but related parents. In the physical examination, a painless, firm mass 2 cm in diameter was found on the scalp in the occipital region. In addition, a firm, painless subcutaneous nodule 1.5 cm in diameter was found in the mental region. The skin covering the two masses was normal. Other findings of the general physical examination were unremarkable except for tonsillitis. There was no hyperplasia of the maxillary or mandibular gingiva. The complete blood count, chest radiography, abdominal ultrasound, biochemical data and radiological skeletal survey were within normal limits except for leukocytosis. The two masses in the mental and occipital regions were excised totally (14 August 1997). The postoperative course of the patient was uncomplicated. All the samples of the excised masses were fixed in 10 percent buffered formalin prior to the routine processing of the paraffin-embedded block. Multiple sections were obtained with use of a standard microtome and were stained with hematoxylin and eosin (H&E). Several sections were also stained with Congo red, reticulin, Van Gieson, and periodic acid-Schiff (PAS) with and without diastase. In addition, sections from formalin-fixed and paraffin-embedded tumor tissue were examined by a previously described avidin-biotin-peroxidase complex (ABC) method[4]

using actin [monoclonal mouse anti-actin (smooth muscle), clone:1A4, Cat.No.08-0106, isotype:IgG2a, ready-to-use, ZYMED, San Francisco, California, USA], which is an immunomarker of smooth muscle neoplasm.

Macroscopically, the mass on the scalp was firm, yellowish-white in color, with ill-defined margins, and measured 2 cm in diameter. The mass in the mental region was firm, yellowish-white in color, with ill-defined margins, and measured 1.5 cm in diameter. No focus of hemorrhage, necrosis, calcification, or areas of cystic change was observed in either mass. Light microscopy examination of both tumors showed identical histology: an abundance of a homogeneous, amorphous, eosinophilic extracellular matrix in which fibroblasts were embedded. They consisted of parallel-arranged cells with spindle nuclei and elongated cytoplasm, and epithelioid cells with a round, vesicular nucleus and clear cytoplasm that had a chondroid appearance (Fig. 1). Large amounts of hyalinized collagen-like material were recognized in some areas. Neither mitotic figure, hemorrhage, necrosis or calcification was seen. Well-formed collagen fibers were not demonstrated with the Van Gieson's method and reticulin preparation. The hayalin material was PAS-positive and diastase-resistant (Fig. 2), whereas results with the Congo red method were negative. Immunohistochemically, the spindle-shaped cells were actin negative. On the basis of the above gross findings, light microscopic findings, and histochemical and immunohistochemical findings, we considered the histopathologic diagnosis of these tumors to be JHF.

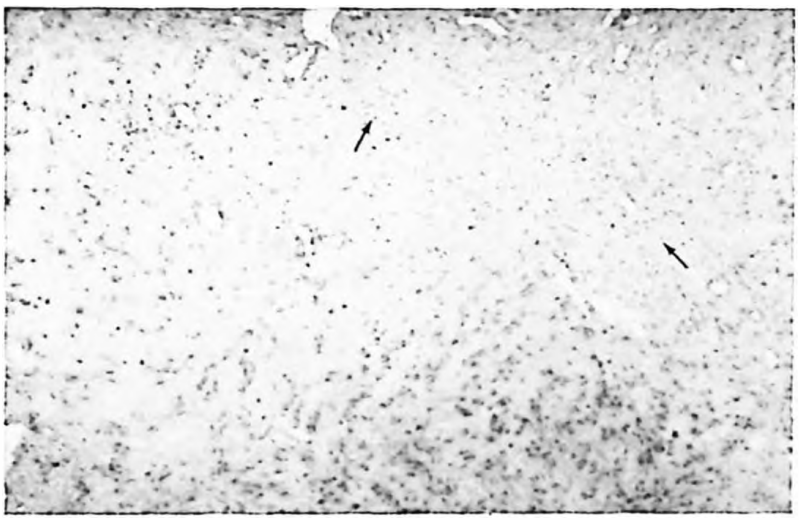

Fig. 1. Photomicrograph of the tumor characterized by scant strands of fibroblasts and epithelioid cells embedded in an abundant, homogeneous, amorphous matrix (H.E.X25).

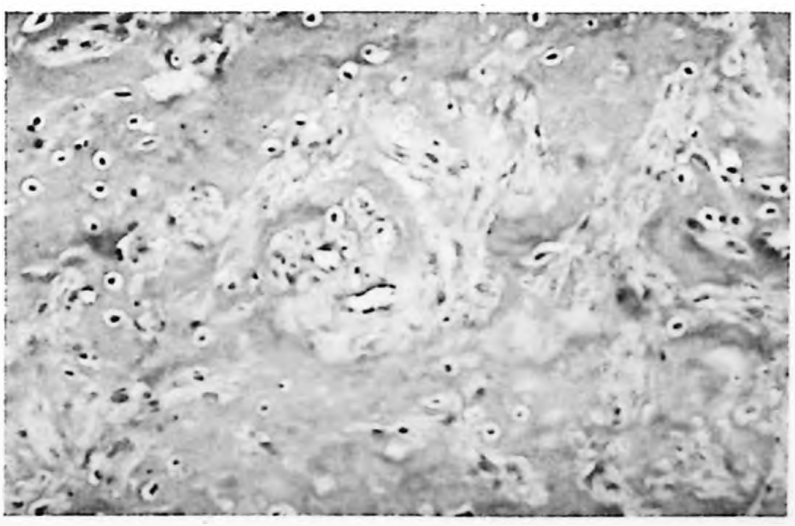

Fig. 2. The hyalinized collagen-like material is PAS-positive and diastase-resistant (PAS with diastase X 50).

Discussion

Juvenile hyaline fibromatosis is inherited as an autosomal recessive trait. It usually affects more than one sibling of the same family and there is no predilection for either sex[2]. Clinically, JHF is characterized by multiple or, rarely, single skin lesions. These lesions may appear as small, fleshy papules, translucent nodules, or as large subcutaneous masses of variable consistency. The largest masses are predominantly located on the scalp. Other frequent clinical manifestations of JHF are gum hypertrophy, flexion contractures of various joints, osteolytic lesions of bones, and undefined muscle illness[5]. These findings were not observed in our case. Histologically, the tumors consist of cords of spindle-shaped cells embedded in a homogeneous eosinophilic matrix in dermal deposition. Some cells have an epithelioid appearance with an oval or round vesicular nucleus and clear cytoplasm simulating a chondroid appearance. This appearance was observed in our case. In general, the smaller and younger lesions tend to be more cellular, while the larger and older lesions contain more ground substance[6].

The differential diagnosis of JHF includes neurofibromatosis, gingival fibromatosis, cylindromas, nodular amyloidosis, leiomyoma with sclerosis[6], infantile systemic hyalinosis, congenital generalized fibromatosis, multicentric infantile myofibromatosis, lipoid proteinosis (hyalinasis cutis et mucosae[2], and Winchester's syndrome[3], but particularly the latter five.

Multicentric infantile myofibromatosis is present at birth or occurs during the first year of life, and the lesions are better circumscribed, and are found not only in the subcutis but also in muscle, bone, and viscera, Microscopically, they consist of broad, interlacing bundles of plump myofibroblasts, often with pericytoma-like areas in the center of the lesion[2]. Congenital generalized fibromatosis is present at birth. Histologically, there is high fibroblastic cellularity and infiltrative borders. Infantile systemic hyalinosis appears at birth or during early infancy and invariably leads to death in early childhood. Winchester's syndrome is characterized by short stature, stiffness and contractures of small joints, and corneal opacities. The generalized osteoporosis with erosion of the metacarpal bones and progressive resorption of the carpal bones are characteristic radiological findings of Winchester's syndrome[3]. Lipoid proteinosis lacks the spindle cell proliferation and consists of hyalin material. In nodular amyloidosis, an abundant hyalin material is present in the dermis. This material stains positively for amyloid, whereas the same stain is always negative in JHF. In cutaneous leiomyomas, staining with smooth-muscle actin is positive, whereas the same stain is always negative in JHF[6]. In this disease, the basic defect appears to be a localized metabolic disturbance in the formation of collagen, presumably caused by incomplete alignment of precollagen or protocollagen, possibly due to increased or faulty synthesis of glycosaminoglycans by fibroblasts[2]. Mayerda-Silva et al.[7] suggested that JHF represents a disease of the connective tissue with progressive abnormal differentiation to chondroid tissue. Breier et al.[8] suggested that the band for type II collagen chain is absent in Western blot studies of clinically unaffected JHF skin. Type I collagen synthesis and degradation are increased in JHF fibroblasts compared with control fibroblasts. In contrast, type III collagen overall metabolism is significantly reduced (by 36%) compared with controls. The clinical course is variable: it may be characterized by the progressive appearance of new skin lesions or by progressive enlargement of old skin lesions over the years. The only effective treatment for JHF is surgery[6].

REFERENCES

1. Senzaki H, Kiyozuka Y, Uemura Y, Shikata N, Ueda S, Tsubura A. Juvenile hyaline fibromatosis: a report of two unrelated adult sibling cases and a literature review. Pathol Int 1998; 48: 230-236.
2. Enzinger FM, Weiss SW. Soft Tissue Tumors. St. Louis: Mosby-Year Book; 1995: 245-248.
3. Jacyk WK, Ventzel LF. Juvenile hyaline fibromatosis in two South African black children. Intern J Dermatol 1996; 35: 740-741.
4. Hsu SM, Raine L, Fanger H. Use of avidin-biotin-preoxidase complex (ABC) in immunoperoxidase techniques. A comparison between ABC and unlabeled antibody (PAP) procedures. J Histochem Cytochem 1981; 29: 577-580.
5. Woyke S, Domagala W, Markiewicz C. A 19-year follow-up of multiple juvenile hyaline fibromatosis. J Pediatr Surg 1984; 19: 302-304.
6. De Rosa G, Tornillo L, Orabona P, D'Antonio A, Staibano S, Boscaino A. Juvenile hyaline fibromatosis, a case report of a localized form? Am J Dermatopathol 1994; 16: 624-627.
7. Mayer-da-Silva A, Poiares-Baptista A, Rodrigo FG, Teresa-Lopes M. Juvenile hyaline fibromatosis. A histologic and histochemical study. Arch Pathol Lab Med 1988; 112: 928-931.
8. Breier F, Fang-Kircher S, Wolff K, Jurecka W. Juvenile hyaline fibromatosis: impaired collagen metabolism in human skin fibroblasts. Arch Dis Child 1997; 77: 436-440.