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Case report

A case report of juvenile hyaline fibromatosis

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Abstract

Juvenile hyaline fibromatosis (JHF) is a rare, autosomal recessive disease characterized by early onset papulonodular skin lesions, soft tissue masses, joint contractures, gingival hypertrophy, stunted growth and osteolytic bone lesions. Histopathological examination of the cutaneous lesions is unique and characterized by an accumulation of an amorphous, hyaline material in the dermis with increased number of fibroblasts. Herein, we report an 11 year-old girl who presented with papulonodular lesions on the scalp, chin, ears, elbows, knees, back and perianal skin. She had gingival hypertrophy and contractures of the elbows, hips, knees and ankles.

Keywords: Juvenile; Hyaline; Fibromatosis; Autosomal; Recessive

1. Introduction

Juvenile hyaline fibromatosis (JHF) is a rare autosomal recessive disease with an onset in infancy or early childhood (Yayli et al., 2006). Less than 70 cases have been reported worldwide (Park et al., 2010; Uslu et al., 2007). It is characterized by papulonodular skin lesions, gingival hyperplasia, joint contractures and bone lesions (Ribeiro et al., 2009). The histological findings of cutaneous lesions in JHF are characterized by the varying degrees of fibroblasts and amorphous hyaline ground substance in the extracellular spaces of the dermis and soft tissues (Tehranchinia and Rahimi, 2010). The etiology of JHF is still unknown but capillary morphogenesis protein 2 and mutation in a gene on chromosome 4q21 are considered to be causative factors (Altug et al., 2009; Karacal et al., 2005; Thomas et al., 2004).

2. Case report

An 11-year-old girl presented with multiple asymptomatic skin lesions at different body sites. She was a product of a full term normal pregnancy for a third-degree consanguineous parents. Other family members (parents, one brother and three sisters) are healthy except for her older brother who is affected by the same disease. She was well till the second month of age when her mother started to notice difficulties in moving her limbs with progressive painless contractures. At the age of two, the first skin lesion appeared in the perianal area which was complicated by painful defecation. At the age of three, difficulty in feeding developed as a result of progressive swelling of the gums that almost covered her teeth. Multiple skin lesions appeared on the face and digits along with swellings on the scalp, back, elbows and knees at the age of 6 and increased gradually in number and size. Apart from skin lesions, joint contractures and her failure to thrive, there

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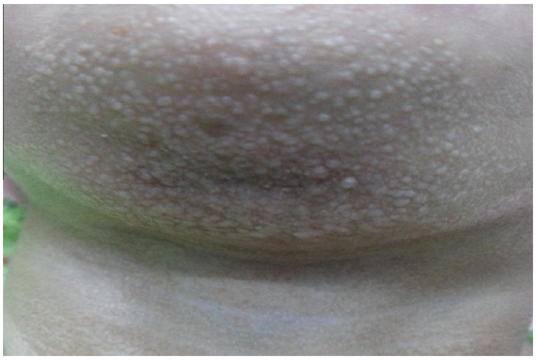

Fig. 1. Pearly papules over the chin.

were no systemic symptoms and the patient was otherwise healthy with normal mental function. They sought medical advice many times but accurate diagnosis has not been made.

Skin examination showed whitish-pinkish papules and nodules on the chin, ears, digits (Figs. 1–3), and perianal region and multiple skin colored tumors on the scalp, back, elbows and knees, largest measuring 10 × 4 cm (Figs. 4–7). Oral examination revealed extensive gingival hyperplasia (Fig. 8). Joint contractures were evident in the wrists, hips, knees and ankles (Fig. 9).

Skeletal radiographs showed joint contractures and osteolytic bone lesions (Fig. 10). Routine laboratory tests showed normal results except for iron deficiency anemia. Histopathological examination with hematoxylin–eosin stain showed dermal deposits of eosinophilic hyaline material with increased fibroblasts (Figs. 11 and 12). Based on the characteristic clinical and histopathological findings, a diagnosis of JHF was made.

3. Discussion

Juvenile hyaline fibromatosis (JHF) was originally described by Murray in 1873 under the name “molluscum fibrosum” (Denadai et al., 2012). At that time, it was considered a variant of neurofibromatosis. Whitfield and

Fig. 2. Whitish–pinkish papules and nodules involving the right ear.

Fig. 3. Right hand showing multiple pinkish nodules.

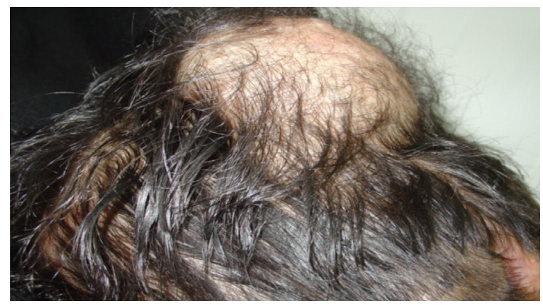

Fig. 4. Large subcutaneous tumor over the scalp.

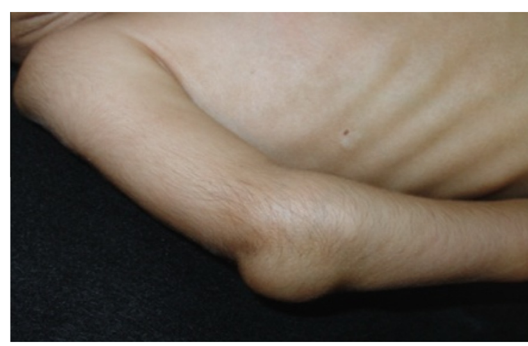

Fig. 5. Large subcutaneous tumor over the right elbow.

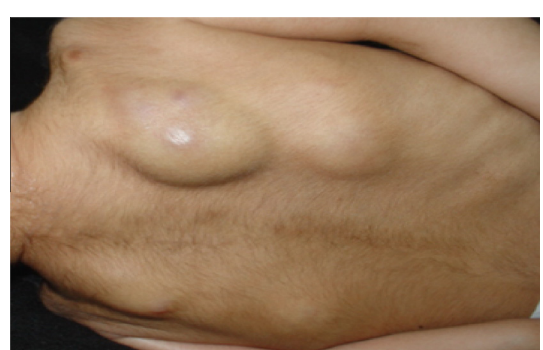

Fig. 6. Large subcutaneous tumors over the back.