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...
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
Robinson later reported two more cases in 1903 and suggested the disease be recategorized as multiple fibromata, but it was not until 1972 when this condition was given the current name of juvenile hyaline fibromatosis by Kitano et al. Thomas et al. (2004), Lim et al. (2005), Slimani et al. (2011). An autosomal recessive mode of inheritance is accepted, but sporadic cases can occur (Yayli et al., 2006). The gene that causes JHF has been mapped to 4q21 and mutations in the capillary morphogenesis (factor -2 gene) have also been described (Altug et al., 2009; Karacal et al., 2005; Thomas et al., 2004). The exact pathogenesis is unknown but several theories have been proposed, most attributing JHF lesions either to aberrant synthesis of glycosaminoglycans by fibroblasts or to disordered collagen metabolism (Lim et al., 2005; Tzellos et al., 2009) (see Table 1).

The disease is defined clinically by a constellation of findings including pearly skin papules or subcutaneous firm nodules, joint contractures, acral osteolytic lesions, gingival hypertrophy and normal intelligence (Yayli et al., 2006). Skin lesions can be polymorphous papules, small, rosy, clustered and located in the face and neck, particularly around the nostrils, ears, paranasal folds and the chin along with nodular lesions or plaques on the scalp, limbs and perianal regions (Ribeiro et al., 2009). They are slow growing and painless and have a tendency to recur following excision (Krishnamurthy and Dalal, 2011). Gingival...
hyperplasia is a common finding that may be severe enough to interfere with feeding, and may result in poor oral hygiene, infection and dental caries (Nofal et al., 2009).

Musculoskeletal involvement in JHF is frequent, and flexion contracture of large joints is the most debilitating problem; most adolescents and adults become bedridden and die of infection (Slimani et al., 2011). It has been hypothesized that contractures result from infiltration of the capsules of the joints (S, 1992). Osteolytic bone lesions are commonly observed in the distal phalanges, skull, and long bones and they have the same histological features as the skin lesions (Yayli et al., 2006). The skin and soft tissue lesions are often the first signs of presentation; however, in other patients, joint manifestations may be primary (Karacal et al., 2005).

A similar condition, infantile systemic hyalinosis (ISH), is characterized by the above findings, with further involvement of the viscera (gastrointestinal, cardiac, hepatic, splenic and thyroid) and an inevitably fatal outcome. Many postulate that JHF and ISH are the same condition with differing penetrance and phenotypic expression (Thomas et al., 2004). Hyaline fibromatosis syndrome (HFS) is a recently introduced term to include both disorders. This term was introduced because of the many similarities between JHF and ISH, including clinical features, histopathological patterns, and the same gene mutation (Raak SM, 2013).

The diagnosis of JHF can be confirmed by histology. The tumors are poorly circumscribed and consist of cords of spindle-shaped cells embedded in a homogeneous eosinophilic matrix. They are often found in the dermis, subcutis and gingiva, although the bone and joints may also be involved (Krishnamurthy and Dalal, 2011).

No specific treatment is available for JHF (Bharambe, 2012). The treatment is only esthetic and its aim is to limit orthopedic disability (Krishnamurthy and Dalal, 2011). Early surgical removal of skin lesions may help, but recurrences are common (Tehranchinia and Rahimi, 2010; Quintal and Jackson, 1985). Joint contractures may respond to intralesional systemic steroid physiotherapy, capsulotomy and oral d-penicillamine (Denadai et al., 2012; Krishnamurthy and Dalal, 2011; El-Maaytah et al., 2010). Gingivectomy can improve the amount of oral intake, but when it is not enough to recover patient’s nutritional deficiency, parenteral feeding such as tube feeding, gastrostomy and placing a central line should be considered (Park et al., 2010). Frequent visits for periodontal treatment and maintenance of good oral hygiene are important factors in decreasing the growth rate of the gingivae in patients with JHF (El-Maaytah et al., 2010). Genetic counseling is of great importance as the recurrence risk is 25% in any future pregnancy.

4. Conclusion

JHF is a rare hereditary disease with progressive course that should be highly suspected in a patient with early onset papulonodules, joint contractures and gingival hypertrophy. Early diagnosis and proper multidisciplinary management are crucial in an attempt to slow the progression of this rare disabling disease.

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Conflict of interest

We have no conflict of interest to declare.
References


