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## CASE REPORT

# Three siblings with juvenile hyaline fibromatosis

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Juvenile hyaline fibromatosis (JHF) is an extremely rare hereditary genetic disease of autosomal recessive transmission that is characterized by large cutaneous tumors commonly involving the scalp, papulonodular skin lesions, flexural joint contractures, gingival hyperplasia, and osteolytic bone lesions. JHF is usually diagnosed in young infants and in children younger than 5 years, and the lesions characteristic of this disorder consist of fibrous tissue and homogenous amorphous eosinophilic hyaline material. We report the case of a 9-year-old girl with severe gingival hyperplasia, nasal enlargement, mild osteoporosis, and multiple papulonodular skin lesions. Her two brothers (7 and 13 years of age, respectively) were also diagnosed as having JHF. In the patient described in this report, the maintenance of oral hygiene after gingivectomy enabled the continued resolution of gingival hyperplasia, although skin lesions recurred and nasal overgrowth persisted.

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### **Case report**

In 2003, a 9-year-old girl was referred to the Baskent University Department of Periodontology in Adana, Turkey, for the treatment of recurrent severe gingival hyperplasia. This patient, who was born to unrelated parents, demonstrated compromised mastication and facial esthetics caused by gingival overgrowth (Fig. 1A,B). Physical examination revealed nasal enlargement (Fig. 2A,B), multiple papulonodular skin lesions on the hands, and a stick-shaped appearance of the fingers. A nodule was noted in the ear (Fig. 3). The patient's disease had first been noticed in 2001 after

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gingivectomy performed at another medical center. Prior total bone mineral densitometry had confirmed mild osteoporosis. The results of hematologic and biochemical laboratory evaluations were within the reference range, except for a moderate elevation in the level of alkaline phosphatase. The results of abdominal and thoracic computed tomography scans, abdominal ultrasonography, and cervical, thoracic, lumbosacral, vertebral, pelvic, and cranial radiographs were within normal limits.

Tissue specimens from the anterior mandibular region, which were obtained during gingivectomy, were stored in 10% buffered formaldehyde for pathologic examination. The results of the histopathologic examination of those specimens via light microscopy showed an abundance of amorphous, homogeneous, eosinophilic, extracellular hyaline matrix with overlying squamous epithelium. The underlying pathogenesis of juvenile hyaline fibromatosis (JHF) has been postulated to be a disorder of collagen metabolism or the result of defective glycosaminoglycan formation. The nature of the hyaline material characteristic of JHF lesions has not been established, but research suggests that this matrix may consist of type VI collagen (1).

The diagnosis of JHF was confirmed in our patient and, after careful evaluation, that disorder was also identified in her two brothers. The patient underwent scaling, root planing, and full-mouth gingivectomy. Consistent oral hygiene was encouraged. After the patient had undergone her last surgical procedure at our hospital, she returned for re-examination every 6 months for two consecutive years. She maintained good oral hygiene throughout the follow-up period, during which no major recurrence of gingival hyperplasia was observed (Fig. 4). Her remarkable nasal enlargement was corrected by means of rhinoplasty, after which nasal overgrowth recurred.

#### Comments

To date, fewer than 70 cases of JHF have been reported in the literature, probably because that disorder has been misdiagnosed as, for example, Winchester syndrome or congenital generalized fibromatosis. The etiology of

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Figure 1 (A, B) Mild gingival hyperplasia (a characteristic of juvenile hyaline fibromatosis) in a 9-year-old girl.



Figure 2 (A, B) Nasal overgrowth in a 9-year-old girl with juvenile hyaline fibromatosis.



Figure 3 Ear nodule, a feature of juvenile hyaline fibromatosis.

JHF is unknown, but Rahman et al. provided strong

evidence for the location of a recessive JHF-predispo-

sition gene on chromosome 4q21 (2). JHF is usually

diagnosed in neonates, infants, and in children younger

than 5 years. In infants, extensive gingival hyperplasia

may cause problems with sucking and (eventually)



**Figure 4** Healthy gingival features 2 years after gingivectomy in a 9-year-old girl with juvenile hyaline fibromatosis.

mastication, which can, in turn, lead to malnutrition, recurrent infections, and even death in childhood (3). Some authors believe that the lesions of JHF resolve as the patient's age increases (4, 5), although the effect of puberty on the course of JHF has not been well documented.

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Surgical excision of and/or treatment for the lesions of JHF are the standard therapeutic regimen. Although an overview of the literature indicates that in patients with JHF, gingivectomy is usually followed by a recurrence of disease (6), our treatment regimen (which combined full-mouth gingivectomy with consistent oral hygiene during a 2-year follow-up period) was very effective in controlling gingival overgrowth in the patient described in this report. No additional excisions were required during a 2-year follow-up period, and she experienced no recurrence of JHF. Although osteolysis is a well-known pathologic process associated with JHF, no significant periodontal destruction was observed in our patient. We concluded that appropriate surgery insures a successful outcome of treatment for this disorder, both esthetically and functionally.

Further research is necessary to reveal the underlying mechanisms of gingival hyperplasia and other abnormalities characteristic of JHF. Identification of the gene mutation responsible for that disorder will also improve the outcome of treatment and the patient's quality of life, and may prolong life expectancy.

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