



Case report

The diagnostic challenge of juvenile hyaline fibromatosis, a case report with literature reviews

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ABSTRACT

Introduction: Juvenile hyaline fibromatosis (JHF) is a rare genetic condition characterized by impaired collagen production or metabolism. This study aims to present a rare case of JHF.

Case report: An 11-year-old boy presented with bilateral keloid-like lesions on his ears and admitted intermittent reappearance of such lesions since he was seven. He was born to second-degree relative consanguineous parents. Physical examination revealed bilateral soft pink masses on the ears, multiple scars on the scalp, severe gingival hypertrophy, multiple small soft white papules on the anterior neck, broadly shaped enlargements on the ends of the fingers and toes, and multiple reticulated hard livedoid and hyperpigmented macules on the back and anterior lower extremities. A 5 mm biopsy was taken from the lesion on the ear and histopathological examination of the specimen revealed a normal epidermis but dermal and subcutaneous deposits of nodules composed of abundant amorphous eosinophilic hyaline material with sparse embedded fibroblast associated with areas of congestion and focal hemorrhage. The ear lesions were managed by surgical excision with intraregional steroid injections to prevent relapse. To improve eating ability and oral hygiene, a gingivectomy was planned.

Discussion: JHF presents with bone lesions, gingival hypertrophy, joint contractures, and skin lesions. The clinical features usually appear late in infancy and up to 5 years. The condition occurs mostly sporadically. A portion of the cases can be in siblings born to consanguineous parents.

Conclusion: JHF is a rare genetic disorder that can present even beyond five years. There is no standard treatment for these cases.

1. Introduction

Juvenile hyaline fibromatosis (JHF) is an exceedingly rare genetic condition of the connective tissue characterized by impaired collagen production or metabolism. Murray et al. described the disorder for the first time in 1873. In 1972, Kitano and colleagues first coined the word JHF for the disease [1]. JHF is an autosomal recessive illness that results in joint contractures, papulonodular skin lesions, bone lesions, and gingival hyperplasia [2]. The skin lesions can emerge as pink papules and plaques on the perianal region, scalp, trunk, ears, and paranasal area and can also present as large tumors in the head and neck areas [3]. The

clinical features of JHF usually appear late in infancy and up to 5 years of age [4]. Two forms of the condition have been proposed: a localized type with slow-growing tumors and a generalized type with rapidly growing tumors [3]. The incidence rate of JHF has not been established yet [5].

The current study aims to present a rare 11-year-old case of JHF. The paper was written by taking the SCARE 2020 guidelines into consideration [6].

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2. Case presentation

2.1. Patient information

An 11-year-old boy presented to our dermatology clinic suffering from repeated formation of bilateral keloid-like lesions on his ears since he was seven. The youngster was born to second-degree relative consanguineous parents. There was no history of similar conditions in any of the other siblings, nor a history of chronic illnesses or medication use.

2.2. Clinical findings

Physical examination revealed bilateral soft pink masses on the ears, multiple scars on the scalp (previous surgical tumor removals), severe gingival hypertrophy, multiple small soft white papules on the anterior neck, broadly shaped enlargement on the ends of the fingers, and toes, and multiple reticulated hard livedoid and hyperpigmented macules on the back and anterior lower extremities. The child had normal mental development.

2.3. Diagnostic approach

All the laboratory findings were normal. A 5 mm biopsy was taken from the lesion on the ear, and histopathological examination of the specimen revealed a normal dermis but dermal and subcutaneous deposits of nodules composed of abundant amorphous eosinophilic hyaline material with sparse embedded fibroblast associated with areas of congestion and focal hemorrhage.

2.4. Therapeutic intervention

The lesions on the ears were managed through surgical excision with intraregional steroid injections to prevent relapse. To improve eating ability and oral hygiene, a gingivectomy was planned.

2.5. Follow-up and outcome

The operations were uneventful, and the patient was kept under close follow-up.

3. Discussion

JHF was initially termed “molluscum fibrosum” by Murray and colleagues in 1873. The disease has an autosomal recessive mode of inheritance [7]. It may occur because of mutations in the capillary morphogenesis gene 2 (CMG2) located on chromosome 4q21 [8]. It has been suggested that JHF ensues due to impaired glycosaminoglycan synthesis by fibroblasts [4]. JHF cases are often healthy at birth. The condition usually starts to manifest early in life, especially in late infancy and up to five years, and new lesions can still develop in adulthood [1,7]. Lyro et al. diagnosed a case that showed clinical manifestations at eight years [9]. The clinical manifestations of the current presentation appeared at the age of seven. The disorder can impact the quality of life in these patients based on the progression and its severity [10].

The clinical features of the condition include painless skin lesions that can occur as pink papules and plaques on the perianal region, scalp, trunk, ears, and paranasal area and can also present as large tumors in the head and neck area [11]. Nodules can also be found on the tips of the digits [3]. Other common manifestations include gingival hypertrophy that can cause decreased eating ability and lead to malnutrition, joint contractures that can cause movement disability, and bone lesions [12]. Similar findings were observed in this study. Most studies report no difference in gender [8,13]. However, some studies point out a slightly higher male predominance [4]. Even though the disease occurs sporadically [5], one-third of the cases are siblings, and a large number

of these patients are born to consanguineous parents [3]. Our patient was a chap born to consanguineous parents, and none of his siblings were affected.

It is crucial to distinguish JHF from a similar condition, infantile systemic hyalinosis (ISH). Both disorders share similar histopathological findings and mutations. ISH usually manifests in early infancy with the added involvement of the viscera (hepatic, thyroid, cardiac, splenic, and gastrointestinal) [14]. Compared to JHF it is much more fatal, in which death often occurs within the first two years of age [10]. Meanwhile, patients with JHF can survive up to the 4th decade of life [8]. Hyaline fibromatosis syndrome (HFS) is the suggested term to cover both conditions [15].

In JHF patients, laboratory and radiological findings are nonspecific [8,10]. The condition can be defined via the previously described presentations [14]. The diagnosis can be confirmed with a histopathological examination, which can show spindle-shaped cells, fibroblast proliferation, eosinophilic substance, and amorphous hyaline material. Older lesions usually contain more ground substances and are less cellular [4].

There is currently no standard treatment for JHF. Early surgical excision has been performed for skin lesions and tumors. However, they do not respond well to radiotherapy, and they are associated with a high rate of recurrence [14]. Woyke and colleagues surgically removed over 100 tumors over 19 years [16]. Karande and associates reported that intralesional steroid injection aid in treating subcutaneous tumors [5]. Braizat et al. stated that local and systemic corticosteroids have only partial or temporary results in hand nodules [13]. Gingivectomy can be performed for gingival hyperplasia to improve feeding ability. It has been reported that joint contractures may respond to intralesional steroid therapy. Regular follow-up of these cases is often required, and genetic counseling is crucial as 25% of future pregnancies can also result in JHF [12].

In conclusion, JHF is a rare genetic disorder characterized by bone lesions, gingival hypertrophy, joint contractures, and skin lesions. The condition can present even beyond five years. There isn't a standard cure for these cases. Early surgical intervention is recommended with regular and frequent follow-up.

Consent

Written informed consent was obtained from the patient's family for publication of this case report and accompanying images. A copy of the written consent is available for review by the Editor-in-Chief of this journal on request.

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Ethical approval

Approval is not necessary for case report (till 3 cases in single report). The family gave consent for the publication of the report.

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CRedit authorship contribution statement

Abdulwahid M. Salh: major contribution of the idea, final approval of the manuscript.

Ronak S. Ahmed: physician managing the case, final approval of the manuscript.

Fahmi H. Kakamad: Writing the manuscript, literature review, final approval of the manuscript.

Alaa A. Ali, Hiwa O. Abdullah, Ari M. Abdullah, Sharo Naqar, Marwan L. Fatah: literature review, final approval of the manuscript.

Declaration of competing interest

None to be declared.

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