

Case Report

Juvenile Hyaline Fibromatosis- A rare cause of cervical swelling

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ABSTRACT

Background: Juvenile Hyaline Fibromatosis (JHF) is a rare autosomal recessive connective tissue disorder characterized by abnormal hyaline deposition. Mandibular involvement is uncommon and may mimic malignancy.

Case Presentation: A 6-year-old female child presented with a gradually progressive left submandibular swelling for four months. Examination revealed a firm, non-tender mass measuring 6 × 4 cm. Ultrasonography demonstrated a heterogeneous lesion, while fine-needle aspiration cytology suggested a spindle-cell lesion. Computed tomography revealed extensive osteolytic changes involving the left mandibular body and ramus. Surgical exploration revealed a dumbbell-shaped mass extending into the floor of the oral cavity with associated mandibular destruction. Complete gross excision was performed. Histopathological examination demonstrated spindle-shaped cells arranged in fascicles with intervening hyaline bands, suggestive of Juvenile Hyaline Fibromatosis.

Conclusion: JHF should be considered in the differential diagnosis of paediatric submandibular swellings with mandibular osteolysis. Histopathological evaluation is essential for diagnosis, while long-term follow-up is recommended because of the risk of recurrence.

Keywords: Juvenile Hyaline Fibromatosis, Submandibular Swelling, Mandibular Osteolysis, Histopathology, Case Report.

INTRODUCTION

Juvenile Hyaline Fibromatosis (JHF) is an exceptionally rare autosomal recessive connective tissue disorder characterized by abnormal deposition of amorphous hyaline material in the skin, subcutaneous tissues, gingiva, joints, and skeletal structures. The disease typically manifests during infancy or early childhood, presenting with multiple cutaneous papules and nodules, gingival hypertrophy, joint contractures, and osteolytic bone lesions. Although histopathologically benign, progressive tissue involvement and recurrence may result in considerable morbidity [1,2].

JHF is now recognized within the spectrum of hyaline fibromatosis syndrome, which also encompasses infantile systemic hyalinosis. The disorder is associated with pathogenic variants in the ANTXR2 gene, also known as CMG2, which plays an important role in extracellular matrix homeostasis. Genetic abnormalities

result in abnormal accumulation of hyaline material within affected tissues. Clinical manifestations vary considerably, ranging from localized cutaneous lesions to extensive systemic involvement, making early diagnosis challenging [2,3]. Skeletal involvement, particularly osteolytic lesions, is an uncommon but important manifestation of JHF. Involvement of the craniofacial skeleton, especially the mandible, is unusual and may mimic aggressive benign or malignant lesions [4,5]. Submandibular swelling in children has a broad differential diagnosis, including inflammatory lymphadenopathy, salivary gland disorders, congenital lesions, and mesenchymal tumours. Radiological investigations help determine lesion extent and associated bone destruction, while histopathological examination typically reveals spindle-shaped fibroblast-like cells embedded within abundant eosinophilic hyaline material [1,4]. Management primarily involves surgical excision of

symptomatic localized lesions, although recurrence remains a concern, necessitating long-term follow-up [5]. The present case describes a 6-year-old female child presenting with a solitary left submandibular swelling associated with extensive osteolytic involvement of the mandibular body and ramus. Histopathological examination of the excised lesion revealed features suggestive of JHF, highlighting an unusual presentation and the importance of considering this rare disorder in the differential diagnosis of paediatric submandibular masses.

Case Presentation

A 6-year-old female child presented to the Department of Otorhinolaryngology with a history of gradually progressive swelling in the left submandibular region for approximately four months. The swelling was initially noticed as a localized mass and had progressively increased in size, prompting medical consultation. There were no reported inflammatory changes in the overlying skin. In view of the persistent swelling and its anatomical location, a detailed clinical examination and diagnostic evaluation were undertaken to determine the underlying pathology. On local examination, a solitary swelling measuring approximately 6 × 4 cm was observed in the left submandibular region, extending from the inferior border of the mandible to the level of the hyoid bone. The swelling was firm to hard in consistency, with a smooth surface, and was non-tender on palpation. The overlying skin appeared normal, without discoloration or visible inflammatory changes. The clinical findings suggested a deep-seated lesion involving the submandibular region, with possible involvement of adjacent anatomical structures. Routine hematological investigations were performed and were within normal limits. Considering the progressive nature and firm consistency of the swelling, further radiological and cytological investigations were undertaken. Ultrasonography of the left submandibular region revealed a well-defined heterogeneous lesion involving the left submental and submandibular regions. The underlying left hemimandible was obscured by the lesion, indicating its close anatomical relationship with the mandibular structures. These findings suggested a soft-tissue lesion requiring further evaluation to determine its extent and possible involvement of the underlying bone. Ultrasonography-guided fine-needle aspiration

cytology (FNAC) was performed for preliminary characterization of the lesion. Aspiration yielded transparent, fluid-like material. Microscopic examination demonstrated cellular smears containing spindle-shaped tumour cells arranged in small, loose clusters and scattered singly. The cells exhibited a moderate amount of cytoplasm, with occasional atypical features, against a haemorrhagic background. Based on these cytomorphological findings, the lesion was reported as a spindle-cell lesion. However, a definitive diagnosis could not be established on cytological examination alone. Consequently, tissue biopsy with histopathological examination and immunohistochemistry was recommended for further characterization. Computed tomography (CT) of the mandible was performed to assess the anatomical extent of the lesion and evaluate possible osseous involvement. CT imaging revealed a diffuse osteolytic lesion involving the body and ramus of the left mandible, associated with marked cortical thinning and areas of bony destruction. An adjacent soft-tissue component was also identified in the left submandibular region. The extensive osteolytic changes raised suspicion of an aggressive pathological process, including the possibility of malignancy. Considering the clinical presentation, cytological findings, and radiological evidence of mandibular destruction, surgical exploration and excision were planned. Surgical exploration revealed a dumbbell-shaped soft-tissue mass extending from the left submandibular region into the floor of the oral cavity along the mandibular margin. The lesion was closely attached to the body and ramus of the left mandible. Intraoperative examination demonstrated a moth-eaten appearance of the involved mandibular bone, with significant thinning of the inner cortex and a lytic cavity involving the mandibular body and ramus. These findings were consistent with the extensive osteolytic changes identified on preoperative CT imaging. Complete gross excision of the submandibular mass was performed. Following removal of the lesion, the involved mandibular surface was smoothed, and the operative field was carefully examined for residual pathological tissue. No gross residual lesion was identified at the completion of the procedure. The excised specimen was subsequently submitted for histopathological examination to establish the nature of the lesion and exclude malignancy. Gross examination of the surgically excised specimen revealed three globular tissue fragments

measuring approximately $4.5 \times 4 \times 1$ cm, $4 \times 3 \times 1$ cm, and $3 \times 2 \times 1$ cm. The external surfaces of the fragments were fibrous, while the cut surfaces appeared whitish and firm. Representative tissue sections were processed for routine histopathological examination. Microscopic examination of hematoxylin and eosin-stained sections revealed a proliferation of plump spindle-shaped cells arranged predominantly in fascicles. The spindle-cell fascicles were separated by thin bands of eosinophilic hyalinized stromal material, producing a characteristic fibrohyaline appearance. Proliferating blood vessels and areas of extravasated blood were also observed within the lesion. The spindle cells exhibited relatively uniform morphology, without evidence of significant cellular atypia or malignant features. The presence of spindle-cell proliferation associated with intervening hyaline bands was suggestive of Juvenile Hyaline Fibromatosis. Based on the characteristic microscopic findings, the histopathological impression was suggestive of Juvenile Hyaline Fibromatosis (JHF). Correlation with the clinical presentation and radiological findings supported an unusual manifestation of JHF presenting as a solitary submandibular mass with extensive osteolytic involvement of the left mandibular body and ramus. The absence of malignant features on histopathological examination was particularly significant, considering the initially aggressive radiological appearance of the lesion. Following surgical excision, postoperative clinical photographs documented the appearance of the left submandibular region and healing of the surgical incision. The surgical record confirmed complete gross excision of the lesion, with no visible residual disease at the end of the procedure. However, the available clinical records did not specify the duration of hospitalization, immediate postoperative complications, or the exact postoperative follow-up interval. Considering the possibility of local recurrence associated with Juvenile Hyaline Fibromatosis, long-term clinical surveillance was considered necessary. Follow-up assessment would focus on detecting recurrent swelling, evaluating mandibular involvement, and identifying any additional manifestations of the disorder. The present case highlights an unusual presentation of Juvenile Hyaline Fibromatosis in a young child, characterized by a solitary submandibular swelling associated with extensive mandibular osteolysis. The destructive radiological

appearance initially raised suspicion of malignancy, while FNAC demonstrated a nonspecific spindle-cell lesion. Histopathological examination of the excised tissue revealed characteristic spindle-cell proliferation with stromal hyalinization, supporting the diagnosis of JHF. This case emphasizes the importance of including rare connective tissue disorders in the differential diagnosis of paediatric submandibular masses with associated osteolytic changes and demonstrates the essential role of histopathological examination in distinguishing such lesions from aggressive or malignant conditions.

DISCUSSION

Juvenile Hyaline Fibromatosis (JHF) is a rare hereditary connective tissue disorder with variable clinical manifestations, ranging from localized soft-tissue lesions to extensive skeletal and systemic involvement. The present case involved a 6-year-old female child presenting with a solitary left submandibular swelling and extensive osteolytic changes involving the mandibular body and ramus. Bedford et al. [6] reported two severe paediatric cases characterized by joint contractures, cutaneous and oral lesions, osteolytic defects, and systemic complications, highlighting the broad clinical spectrum of the disorder. Mandibular involvement is an unusual manifestation of JHF. Altuğ et al. [7] described a 21-year-old male with a mandibular lesion associated with bone involvement and displacement of adjacent teeth. Similarly, Al-Malik et al. [8] reported an 11-year-old boy with extensive gingival hyperplasia, dental abnormalities, and restricted mouth opening. In contrast, the present patient exhibited a solitary submandibular mass with destructive mandibular changes, without documented prominent gingival manifestations. Radiological examination in the present case revealed diffuse mandibular osteolysis with cortical thinning, initially raising suspicion of malignancy. Miyake et al. [9] described progressive skeletal abnormalities in a child with JHF during five years of observation, emphasizing its potential for locally destructive behaviour despite benign histopathological characteristics. In the present case, FNAC suggested a spindle-cell lesion, while histopathological examination demonstrated plump spindle cells arranged in fascicles with intervening hyaline bands, supporting the diagnosis of JHF. Surgical excision was

performed because of the progressive swelling and extensive mandibular involvement. Dukan et al. [10] described surgical management of an atypical presentation involving the hands, illustrating the importance of individualized treatment for localized symptomatic disease. However, recurrence remains an important concern. Baltacioglu et al. [11] documented recurrent lesions despite multiple surgical procedures during a 10-year follow-up period. Therefore, long-term surveillance is essential. The present case highlights the importance of considering JHF in the differential diagnosis of paediatric submandibular swellings associated with osteolytic mandibular lesions. Clinical, radiological, and histopathological correlation is essential for accurate diagnosis and appropriate management.

CONCLUSION

Juvenile Hyaline Fibromatosis is a rare connective tissue disorder that may present as an unusual submandibular swelling with extensive mandibular osteolysis, mimicking an aggressive or malignant lesion. The present case highlights the diagnostic challenges associated with atypical presentations and emphasizes the importance of clinical, radiological, and histopathological correlation for accurate diagnosis. Surgical excision is an important treatment option for localized symptomatic lesions. Early recognition and long-term follow-up are essential for appropriate management and timely detection of recurrence.

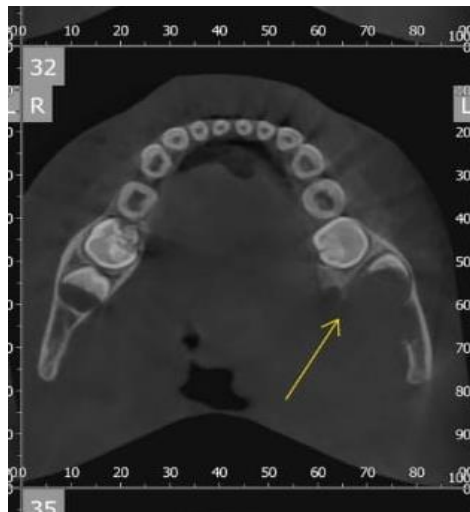


Figure 1: On Ultrasound: Well Defined Heterogeneous Lesion in Left Sub Mental and Submandibular Region with Left Hemi Mandible Obscured.



Figure 2: On CT Scan: A Diffuse Osteolytic Lesion on Left Side of Body and Ramus of Mandible was Detected



Figure 3: Dumbbell Shaped Mass: Extending into the Floor of Oral Cavity from Submandibular Region Along the Margin of Mandible and Attached to Body and Ramus of Mandible.

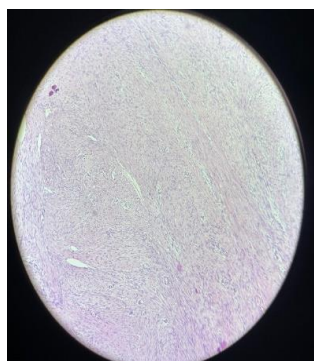


Figure 4: Moth Eaten Appearance of Mandible with Significant Thinning of Inner Cortex and Lytic Cavity in Body and Ramus of Mandible Was Noted.

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