CASE REPORT

Juvenile Hyaline Fibromatosis - Child with Scalp Swellings

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Abstract

Juvenile hyaline fibromatosis is a recessive autosomal hereditary disorder characterized by abnormal growth of hyalinized fibrous tissue. Its clinical presentation is manifested by benign skin lesions, bone lesions, joint contractures, and gingival hyperplasia. The underlying cause is an abnormal growth of hyalinized fibrous tissue with cutaneous, mucosal, osteoarticular, and systemic involvement. This report presents a case of a 4-year-old male child; with complaints of multiple painful body swellings and pus discharge from scalp swellings for the past two years admitted at Dr. Ruth Pfau Civil Hospital, Karachi, 2021. This patient had right occipital scalp swelling of 10x 10 cm due to which surgery was done and the excision was successful. Other findings included stiff joints, skin overgrowth over the scalp and back + buttocks, multiple firm cystic swellings on the scalp, back, trunk, and whole body involving the face (nose), anal region, gum hypertrophy, and various papules over the nose.

Keywords: *Fibromatosis, hyaline, gingiva, lesion, scalp.*

INTRODUCTION

A rare pediatric hereditary disorder of collagen metabolism that occurs in early childhood, associated with the deposition of hyaline material in various soft tissues of the body is termed Juvenile hyaline fibromatosis. It is a milder form of disease mostly characterized by skin lesions such as large papules and subcutaneous nodules around the perianal region, face, gingiva, scalp, wrist and elbow joints of the body. It is a slowly progressive disease leading to thickening of the gingiva, reduced mobility of the joints due to contractures and bone destruction due to osteolysis [1].

Juvenile hyaline fibromatosis, if left untreated, will progress to a disabling disease. Genetic counselling is highly recommended since there is a chance that 25% of the siblings to develop this rare disease. Moreover, it has been reported, that the incidence of Juvenile hyaline fibromatosis increases with consanguineous marriages [2].

Early referral to a dermatologist for surgical removal of subcutaneous nodules with multidisciplinary care can slow the progression of the disease. Capsulotomy, corticosteroid injections at the site of joint lesions and physiotherapy are suggested treatments for contracture of joints. Hypertrophy of the gingivae can be treated by gingivectomy [3].

CASE REPORT

A 4-year-old male child, with no history of vaccination, presented with complaints of multiple painful body swellings for the last two years. According to the mother, the child was born full-term with normal vaginal delivery. The initial two months of the child were uneventful. Later on, she noticed stiffness in his joints while moving the limbs. Delays in developmental milestones along with difficulty in feeding and multiple papules over the nose were also noted during the first year of his life. She consulted local General Practitioners (GPs), but there was no improvement. At two years of age, initially, two nodular swellings appeared on his scalp which gradually increased in size. Moreover, there was a detrimental increase in the stiffness of joints with limited mobility of limbs. More swellings appeared over the trunk region, back, limbs and face up to three years of age.

Family history revealed no consanguineous marriage of the parents. The first child of the family reported an early neonatal death due to febrile illness. The second female and third male children died at the ages of ten and eleven months respectively. According to the mother, both these children developed stiffness of joints at three months of age but died earlier without any skin lesions due to severe gastroenteritis. However, their fourth and fifth children are normal and healthy. However, their sixth child has remarkable stiffness of joints along with skin lesions. They consulted various doctors but could not reach to accurate diagnosis. Finally, this child was brought to the pediatric outpatient department with complaints of multiple painful body swellings and pus discharge from scalp swellings and was admitted to Dr. Ruth Pfau Civil Hospital, Karachi.

On examination, positive findings included stiff joints and skin overgrowth over the scalp and back. Moreover, multiple firm cystic swellings were found in the

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trunk and anal region. Gum hypertrophy and multiple papules over the nose were also noticeable. The patient was admitted with the above-mentioned complaints. Differential diagnoses included Cowden's syndrome, Juvenile Hyaline Fibromatosis and Ma fucci's Syndrome. Grossly, skin lesions appeared to be greyish-white in colour. Three scalp swellings (10x10 cm; 6x7 cm; and 7x7 cm) were noted. Right occipital swelling was firm in consistency. Left occipital swellings had soft-to-firm consistency with central ulceration (**Fig. 1A&B**).

Fig. (1): (**A):** Three scalp swellings and **(B)**: Patient with multiple papules over the nose and joint contracture.

The patient underwent excision of the right occipital swelling. Surgical removal of the subcutaneous nodule was done and whole tissue was fixed in 10% buffered formalin for 24 hours. The sample was sent to the histopathology lab and 3–5 µm sections were obtained and stained with H&E, periodic acid Schiff (PAS) and orcein stain for elastic fibres. The results of the excision biopsy of right scalp swelling revealed intense thickening of the dermis surrounded by large amounts of eosinophilic matrix (**Fig. 2A**). Moreover, fibroblast-like cells along with little vascular spaces and inflammatory cells were found embedded in the ground substance. The sample was positive for PAS staining and did not stain with orcein as it lacked elastic fibres (**Fig. 2B**).

These histopathological findings were conclusive of Juvenile Hyaline Fibromatosis [4]. The patient was kept on broad-spectrum antibiotics after surgery. Recovery was uneventful. The patient was lost to follow-up after discharge.

DISCUSSION

The current case report showed that the parents of the child were not related to each other. This was in agreement with the case conducted by Castiglione *et al.,* in Italy. He reported a case of a 10-year-old Moroccan girl diagnosed as a case of Juvenile hyaline fibromatosis with no parental consanguineous marriage [5]. However, a case of Juvenile hyaline fibromatosis was reported at Jinnah Post-Graduate Medical Centre by Haroon *et al.,* in which the parental consanguinity was reported which did not agree with the results of our study [6].

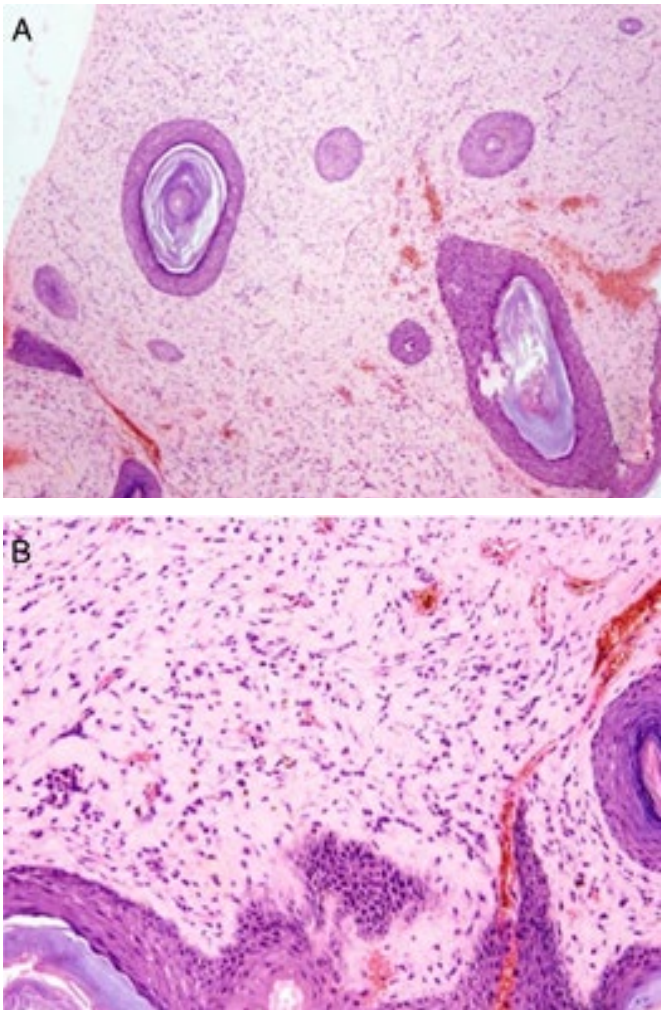

Fig. (2): (A): Biopsy showing thickened dermis along with abundant eosinophilic matrix and **(B):** Periodic Acid-Schiff (PAS) staining.

In our case, according to the mother, the patient was well at birth and later the parents noticed joint stiffness, delayed milestones and dentition. This disease is progressive and the disease course worsens gradually with time. This was inconsistent with the study conducted by Varshini *et al.,* who reported a case of a one-year-old female child in which the symptoms were unnoticeable initially at birth. At the age of three months, the parents noticed that the baby experienced some difficulty in moving her limbs due to joint stiffness. According to them, there was a delay in her milestones and dentition as well [7].

Moreover, our case reported that the child had several hyperpigmented lesions, gingival hypertrophy and progressive joint contractures. This was in contrast with the findings of the case reported by Kalgaonkar *et al*., in which a four-and-a-half-year-old female child presented with multiple soft tissue swellings with no joint contractures and gum disease [8].

In our case, the patient underwent excision of the right occipital swelling and histopathology showed hyalinized

stroma. This was in agreement with the findings of the case reported by Xia *et al.,* [9] in China and Rajendran *et al.,* [10]. They preferred surgical excision as a treatment for scalp lesions.

A Turkish study by Baltacioglu *et al.* reported the case of a 14-month-old male child with multiple papulonodular skin lesions, progressive flexion contractures of joints, and severe gingival hyperplasia. They had a follow-up of the patient for 10 years and found that the disease was progressive and there was a recurrence of gingival hyperplasia even after multiple periodontal surgeries. This was not in agreement with our case as the child was lost to follow-up [11].

In our case, histopathology of excision biopsy of right scalp swelling showed hyalinized stroma with PAS staining along with inflammatory cells. These findings were consistent with Ravikumar *et al.* study in which the histological findings included cords of the oval to spindle-shaped cells within the PAS-positive amorphous eosinophilic matrix, containing abundant hyaline material [12].

Along with the surgical intervention, our patient was given fusidic acid (Antibiotic) for topical application of the skin nodules and physiotherapy exercises were advised for muscle contractures. These treatment modalities were similar to a case conducted in Qatar by Braizat *et al.*, who also treated a patient with Juvenile Hyaline Fibromatosis with a combination of surgical excision, topical corticosteroid therapy and physiotherapy [13].

CONCLUSION

As Juvenile Hyaline Fibromatosis (JHF) is a recessive autosomal disorder, early surgical intervention and appropriate multidisciplinary care are of prior importance, to decelerate the progress of the disease.

CONSENT FOR PUBLICATION

Consent was taken from both the parents of the child and was assured about maintaining confidentiality, by not sharing the personal information in the final publication.

CONFLICT OF INTEREST

The authors declare no conflict of interest.

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Declared none.

REFERENCES

1. Mantri MD, Pradeep MM, Kalpesh PO, Pranavsinh RJ. Hyaline fibromatosis syndrome: a rare inherited disorder. Indian J Dermatol 2016; 61(5): 580.
DOI: https://doi.org/10.4103/0019-5154.190129 PMID: 27688461

2. Abduljabbar MH. A case report of juvenile hyaline fibromatosis. J Dermatol Dermatol Surg 2014; 18(1-2): 38-42.
DOI: https://doi.org/10.1016/j.jdds.2014.06.003

3. Lyra ALCO, Razo LM, Estrella RR, Pantaleão L. Juvenile hyaline fibromatosis: an unusual clinical presentation. Dermatol Online J 2019; 25(7): 13030/qt75082292.
PMID: 31450278

4. Pereira TDSF, Sales JF, Travassos DV, Lanza CR, Castro WH, Gomes CC, *et al*. Hyaline fibromatosis syndrome: A case report. Oral Surg Oral Med Oral Pathol Oral Radiol 2020; 130(6): e328-e335.
DOI: https://doi.org/10.1016/j.oooo.2020.06.022 PMID: 32771412

5. Castiglione D, Terranova MC, Picone D, Lo Re G, Salerno S. Hyaline fibromatosis syndrome (juvenile hyaline fibromatosis): whole-body MR findings in two siblings with different subcutaneous nodules distribution. Skeletal Radiol 2018; 47(3): 425-31.
DOI: https://doi.org/10.1007/s00256-017-2799-y PMID: 29058046

6. Haroon TS, Zaidi Z. Juvenile hyaline fibromatosis. J Pak Med Assoc 1990; 40(8): 194-6.
PMID: 2126302

7. Varshini KA, Haritha K, Desai CA, Rao GR, Chowdary AP, *et al*. Juvenile hyaline fibromatosis or infantile systemic hyalinosis: Hyaline fibromatosis syndrome. Indian J Paediatr Dermatol 2016; 17(1): 38-41.
DOI: https://doi.org/10.4103/2319-7250.173155

8. Kalgaonkar PS, Wade M, Warke C, Makhecha M, Khare M. Juvenile Hyaline Fibromatosis- A Rare Autosomal Recessive Disease. J Clin Diagn Res 2017; 11(7): SD04-SD06.
DOI: https://doi.org/10.7860/JCDR/2017/25280.10293 PMID: 28892992

9. Xia L, Hu Y, Zhang C, Wu D, Chen Y. Juvenile hyaline fibromatosis: a rare oral disease case report and literature review. Transl Pediatr 2021; 10(11): 3124-9.
DOI: https://doi.org/10.21037/tp-21-169 PMID: 34976780

10. Rajendran P, Karmegaraj B, Vij M, Scott JX. Unusual cause for gum hypertrophy and skin nodules in a child. BMJ Case Rep 2015; 2015: bcr2015211506.
DOI: https://doi.org/10.1136/bcr-2015-211506 PMID: 26682835

11. Baltacioglu E, Guzeldemir E, Sukuroglu E, Yildiz K, Yuva P, Aydin G, *et al*. Juvenile hyaline fibromatosis: a 10-year follow-up. Indian J Dermatol 2017; 62(2): 210-2.
DOI: https://doi.org/10.4103/ijd.ijd_166_16 PMID: 28400645

12. Ravikumar VR, Ramamoorthi RGV, Manisankar S. Juvenile hyaline fibromatosis in siblings. Indian J Pathol Microbiol 2019; 62(2): 300-2.
DOI: https://doi.org/10.4103/IJPM.IJPM_76_17

13. Braizat O, Badran S, Hammouda A. Juvenile hyaline fibromatosis: literature review and a case treated with surgical excision and corticosteroid. Cureus 2020; 12(10): e10823.
DOI: https://doi.org/10.7759/cureus.10823 PMID: 33173631