Juvenile Hyaline Fibromatosis

Abstract
Syndromes, especially if they occur early in the growth phase can be very debilitating and cause severe restriction of function. Juvenile hyaline fibromatosis is one such disorder. Our case report presents a girl child who was diagnosed with this debilitating condition. The case presented with both – general characteristic features of this condition such as subcutaneous nodules and flexion deformity of the joints, as well as local manifestation in the form of gingival overgrowth. Thorough clinical and radiological investigations were done to arrive at the diagnosis which was supported conclusively with histopathology of the biopsied gingival lesions. A combination of both medicinal as well as surgical therapeutic modalities was used. This case report is an effort to comprehensively document the etiology, clinical features, and prognosis of this syndrome. There being no permanent cure for the condition, the case report highlights the unique management protocol employed by us in the form of intralesional steroid therapy with endocrine consultation and the local surgical gingival excision carried out in an attempt to optimize the quality of life for our patient.

Keywords: Dexamethasone injections, infantile systemic hyalinosis, juvenile hyaline fibromatosis, Murray-Puretic syndrome, steroid therapy, surgical excision

Introduction
Juvenile hyaline fibromatosis is a rare disorder of glycosaminoglycan synthesis which is characterized clinically by skin papules or tumors, gingival enlargement, osteolytic lesions, and joint contractures and histologically by deposition of amorphous hyaline material.[1] The cause is unknown, but increased chondroitin synthesis has been demonstrated in skin fibroblasts cultured from the tumor tissue. The disease is very rare and occurs sporadically, but it has occurred in siblings. The exact incidence in classic juvenile hyaline fibromatosis is not known. Genetic predisposition has been suggested in that the disease is autosomal recessive in nature.[2,3] The gene involved however is unknown.

Our case report, “Juvenile Hyaline Fibromatosis” describes in detail this rare syndrome; the manifestations of which are morbid, debilitating, and disfiguring. In addition, it highlights the incidence, probable causes, and clinical features in a child patient who presented with the syndrome and our unique management protocol for the same which consisted of intralesional steroid injections with due endocrine consultations and excision of the gingival lesions.

Case Report
A 3½-year-old girl was brought to the Department of Oral and Maxillofacial surgery, Nair Hospital Dental College, Mumbai with the chief complaint of swollen gums [Figure 1] since 2 years. History revealed that she had similar episodes 6 months back for which she had undergone gingival excision by a dentist. In addition, it was noted that she had a peculiar facies and was very irritable to handling. A detailed history revealed astonishing facts about her general health. The girl was the second child of her parents and was delivered prematurely at 7 months by cesarian section. She weighed 3½ kg at birth and was found to be in good health. However, it was found at 1 month of age that, she was irritable to handling and cried when her hand was lifted. This was accompanied by sleep disturbances and frequent bouts of crying.

Orthopedic consultation was done, and radiographs were taken which revealed that she had sustained hairline fracture at the shoulder joint which was suspected to be a result of the birth injury. It was

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managed with cloth bandage which included the chest, both hands and the shoulders. She was also started on the course of antibiotics, details of which are not known. After 20 days, it was seen that she flexed her legs to the back and cried when an attempt was made to stretch them. Orthopedic consultation was done, and once again she was started on antibiotic regimen. Furthermore, physiotherapy was started which spanned for 4 months. However, her condition deteriorated, and she suffered from diarrhea and passed 10–12 stools/day. Her sleep pattern was disturbed, and she cried frequently. She lost weight drastically and her chances of survival were deemed to be grim. She underwent further orthopedic consultations at Mumbai and plaster of paris dressing was done for both hands and feet, but no improvement was seen. She was then started on homeopathic therapy which is said to have temporarily and marginally improved her systemic condition.

At the age of 6 months, she was operated on both knee joints following which physiotherapy was continued, and splints were used. She started walking at the age of 2 years and 3 months with the help of splints. She could only walk to a distance of 50 ft without splints. During a physiotherapy session, she fell and sustained a hairline fracture in the thigh for which she was again given a plaster of paris dressing. Subsequently, she developed swellings on the forehead and back of the ears for which she underwent surgery. However, the lesions recurred in 4 months. In addition, she developed swelling on her knees, her walking slowed, her flexion severed, and she could not walk without splints. She developed swelling on her lips which increased at a fast rate and also, she developed swelling on her gingiva at the same time.

Examination revealed that she was resistant to handling and cried when an attempt was made to do so. Her skin was of a stiffer and thicker consistency than normal. Her height was short and she walked with difficulty in slow steps. Mobility of the fingers of the hands, the toes of the feet and the joints in general was restricted and limited. Joint contractures were evident at the elbows, knees, and fingers of the hands and toes with flexion deformity at these joints [Figures 2 and 3]. The swelling was evident at the elbows and knee joints [Figure 3]. All this led to a characteristic frog position on standing. Diffuse skin papules and subcutaneous nodules were seen occupying the scalp, forehead, ears, and the perianal region [Figures 4 and 5]. Her facial features were coarse with a flattened nose. Her lower lip had a swollen and nodular appearance [Figure 1].

Intraoral examination revealed a diffuse swelling of the maxillary and mandibular gingivae [Figure 1]. It covered the teeth almost to the incisal surfaces and were tender on palpation. At some regions, the occlusal and incisal surfaces of the teeth were completely covered with the overgrowth. She was referred for whole body radiographic examination [Figure 6] which revealed enlarged joints of the hands, feet, toes and elbows, and osteolysis of the tibia. Osteolysis of the carpals was evident along with acral defects and camptodactyly was seen.
Based on the above findings, a clinical diagnosis of “Juvenile Hyaline Fibromatosis” was made, and it was planned to perform an excision surgery for the enlarged gingivae. After a formal endocrine consultation, the patient was prepared for general anesthesia with nasal intubation. 6cc of 1:300,000 saline adrenaline was infiltrated into the maxillary and mandibular labial and buccal vestibules. The enlarged fibrous gingivae were excised with electrosurgery and bleeding was controlled with cautery. The raw area was curetted and irrigated. The lip nodules were also
excised. After an in-depth review and study of the patient’s condition, an endocrine consultation was done, and it was decided to start the patient on local steroid therapy. Serum cortisol, calcium, and alkaline phosphatase levels were vigilantly monitored during the period of therapy. She was started on a regimen of 10 mg dexamethasone at an interval of 3 months for 1 year. Intralesional steroid therapy was used for the lesions of her lip and knees. Tremendous improvement has been noted in her condition with these measures [Figure 7]. A regular long-term follow-up has been advised for the patient. With endocrine consultation, she will be started on systemic low-dose steroid therapy after the age of 6 years as advised by them.

Discussion

Juvenile hyaline fibromatosis is a rare hereditary disorder named by Drescher et al. in 1969.[4] Skin lesions are present at birth or develop in early childhood. There may be small pearly papules or nodules, particularly on the face or the neck. Large subcutaneous tumors may also occur, particularly on the scalp. They may be hard or soft, fixed or mobile and they may ulcerate. Gingival hypertrophy is commonly present and finger contractures of the fingers, elbows, hips, and knees may be present. Osteolytic lesions can occur in the skull, long bones, and phalanges. The musculature is poorly developed. The condition persists into adult life, and the joint contractures are disabling.

The features listed for Murray-Puretic syndrome[5,6] (juvenile hyaline fibromatosis) include, acro-osteolysis/acral defects, camptodactyly/hammer toes, coarse facial features, enlarged joints, facial tumors, flexion deformity of the knees, gum hypertrophy, joint contractures (including arthrogryposis), late puberty in males, limited movement of fingers, lytic lesions of bone, metaphyseal dysplasia, muscle hypertrophy, muscle weakness/myopathy, nasal tumors/polypos/papillomata, ossification defects of skull, osteolysis of carpal, tumors of skin, pedunculated skin lesions/skin tags, scalp tumors, short stature, and skeletal cysts or tumors. Histopathologically [Figures 8 and 9], the skin lesions contain chondroitin cells embedded in amorphous eosinophilic ground substance in the dermis. In the early lesions, this consists of glycosaminoglycan, but in the later lesions the matrix is mainly composed of chondroitin sulfate.[7-8] The dermal collagen is decreased, and the collagen fibrils are fewer and thinner than in normal skin. The hyaline material may also be present in the muscles and bones.[2]

It is essential to differentiate this condition from infantile systemic hyalinosis which has a more widespread involvement of the viscera and a failure to thrive. Death usually occurs within 20 months of age.[9]

Joint contractures may respond to intralesional steroid injections in the early stages, and they may also respond to systemic steroid. Physiotherapy has been advocated for a lifetime.

In our case, the patient was comprehensively managed with surgery, i.e., local excision of the tumor masses and the enlarged gingival lesions and local steroid injections. This brought about a significant remission in the severity of the disease. In addition, regular follow-up, and monitoring of the patient was undertaken. Based on her favorable response to the local intralesional steroid injections, she was sent for systemic steroid therapy with endocrine consultation.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Conflicts of interest

There are no conflicts of interest.

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