JUVENILE HYALINE FIBROMATOSIS

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ABSTRACT

Juvenile hyaline fibromatosis is a rare, autosomal recessive disease. We report a 4-year-old female born of first-degree consanguineous marriage, presenting with gingival hyperplasia, nodular swellings involving the both pinna and right ankle joint. She had contractures of elbows, knees and shoulder joints, restricting the movements of these joints. Cutaneous examination revealed erythematous papules and plaques involving the nape of neck and the perianal region. Histological examination revealed deposition of PAS positive amorphous eosinophilic hyaline material with scattered intervening fibroblast like cells resembling chondroid cells. The patient was diagnosed as a case of Juvenile hyaline fibromatosis based on characteristic clinical and histopathological findings.

***Key Words**: Autosomal recessive disease, Gingival hyperplasia, Nodular swellings, Chondroid cells, Juvenile hyaline fibromatosis*

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INTRODUCTION

Juvenile hyaline fibromatosis (JHF) is a rare, autosomal-recessive hereditary disease with distinct clinical and histopathological features. The clinical onset is usually noted from birth up to 5 years of age. It is characterized by papular and nodular skin lesions, gingival hyperplasia, joint contractures and bone involvement in variable degrees[1]. A scan of the world literature revealed that less than 70 cases have been reported so far[2]. We report a 4 years old female child born of a first-degree consanguineous marriage, presenting with JHF. showed characteristic clinical and histopathological features of JHF.

CASE REPORT

A 4 years old female child born of consanguineous marriage presented to us with presenting complaints of gingival hyperplasia leading to almost near obliteration of the teeth, nodular swellings involving the both pinna (Figure 1) and right ankle joint. He had contractures of elbows, knees, shoulder and ankle joints restricting the movements of these joints (Figure 2). Cutaneous examination revealed erythematous papules and plaques involving the nape of neck and the perianal region (Figure 3 and 4).

Her hemogram, liver function test and urine examination were normal. Abdominal ultrasound revealed normal morphology and texture of abdominal and pelvic organs.

Histological examination of the biopsy obtained from the papular lesion revealed deposition of PAS positive amorphous eosinophilic hyaline material with scattered intervening fibroblast like cells resembling chondroid cells.

The patient was diagnosed as a case of Juvenile hyaline fibromatosis based on characteristic clinical and histopathological findings.

The patient underwent surgery for removal of tumorous growths involving the ankle joint.

DISCUSSION

Juvenile hyaline fibromatosis is a rare autosomal recessive disorder characterized by presence of tumor like lesions, particularly in the head and neck region, gingival hyperplasia and osteolytic bone lesions. Joint deformities, are the earliest presenting features which interfere with growth of the child[3-7].

The gene for the disease is located on chromosome 4q21. This gene encodes capillary

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Figure 1: Gingival hyperplasia leading to almost near obliteration of the teeth, nodular swellings involving the both pinna

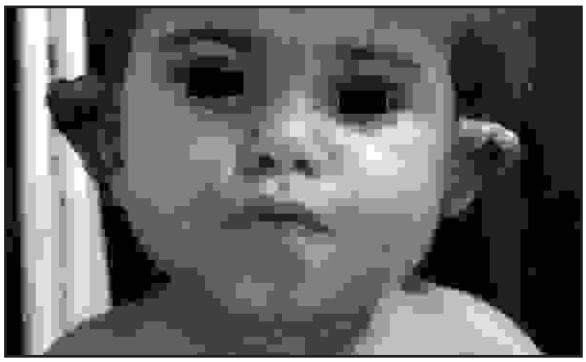


Figure 2: Developed contractures of elbows, knees, shoulder and ankle joints restricting the movements of these joints

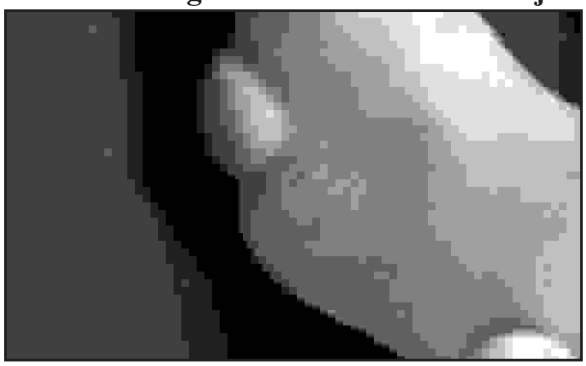


Figure 3: Nodular swellings involving the both pinna and erythematous papules and plaques involving the nape of neck

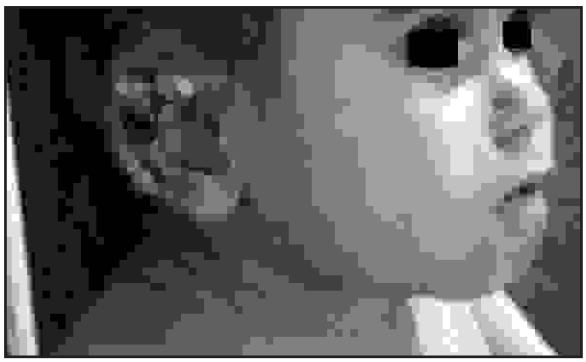
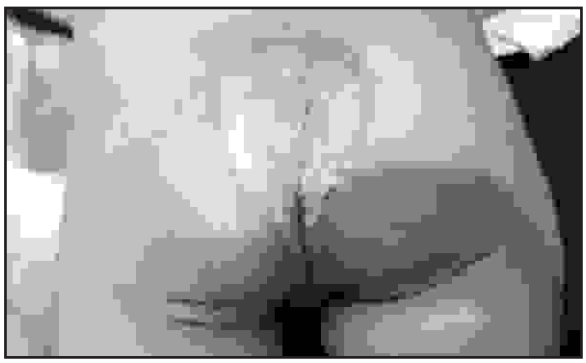


Figure 4: Erythematous papules and plaques involving the perianal region



morphogenesis protein 2(CMG2), a transmembrane protein that is induced during capillary morphogenesis. Besides mutation CMG2 gene various abnormalities in the biosynthesis of glycosaminoglycans and a defect in collagen III and VI have been described[1,2,8].

The diagnosis is confirmed by characteristic histology of the skin lesion[1, 9-12].

The skin lesions consist of benign fibroblastic proliferation-'chondroid cells' occupying the dermis and subcutaneous tissue. These fibroblasts are surrounded by an amorphous hyaline or chondroid-like PAS +ve substance[1,11,12]. This substance, rich in chondroitin-6-sulphate, is made up of glucosamine and galactosamine[1].

There is no definitive cure for JHF, however supportive treatment given in the form of physiotherapy to prevent contracture formation helps the patient to live normal life and avoid disabling contractures[1]. Genetic counseling is essential to explain to parents about a 25% chance of having a diseased baby in any pregnancy[2].

The disease has downhill course and the patient usually die in the fourth decade of life[1,2].

CONCLUSION

All symptoms of typical juvenile hyaline fibromatosis are not necessarily present in each patient. We suggest genetic counseling, supportive treatment and regular followup of these patients for the observation of complication and specific management.

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