JUVENILE HYLINE FIBROMATOSIS: A RARE CASE

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ABSTRACT: Juvenile hyline fibromatosis is a very rare autosomal recessive condition resulting from aberrant biosynthesis go glycosaminoglycans and collagen type III and IV. It presents in early childhood with relapsing papulo nodular skin lesions, gingival hyperplasia and skeletal deformities. Diagnosis is confirmed by characteristic histopathology. There is no definitive treatment other than cosmetic procedures, deformity prevention and correction. The risk of having an affected child is 25% in each of pregnancies. The condition has not previously been reported from Sri Lanka.

Keywords: Juvenile, Hyline Fibromatosis, a Rare Case.

INTRODUCTION
Juvenile hyaline fibromatosis (JHF) is a rare autosomal recessive disorder with distinctive clinical and histopathological features.(2,3). Literature revealed less than 70 cases have been reported by 2014(1, 2, 3). Onset of JHF is before the age of 5 years and characterized by papular and nodular skin lesions gingival hyperplasia, joint contractures and variable degree of bone involvement (2, 3)

CASE REPORT
We report a case of a 12 years old girl, the first child of consanguineous marriage presented with multiple, recurrent, painless, variable sized nodules over the scalp, back and shoulders. Lesions first appeared at the age of 10 and several were excised but recurred. The histopathological features were consistent with JHF. Child’s growth and school performance were normal and her 9 years old brother was not affected.

On examination there were well circumscribed subcutaneous nodules of varying sizes (fig: 1, 4) which were firm and non tender. There was gingival hyperplasia(fig: 2) and molluscum like papules in the back of the neck (fig: 3). No dysmorphic features, skeletal or limb deformity, lymph adenopathy or organomegaly.

Nodule on the forehead was excised for cosmetic reasons and sent for histopathology

Pathologic findings: microscopic sections of the lesion revealed extensive deposits of amorphous hyline material with entrapped fibroblasts. Surrounding these deposits, fibrocollagenous tissue with many bland fibroblastic cells identified without atypia or necrosis were found. The masson trichome stain highlighted the fibrocollagenous tissue in consistant with JHF

The child is still being followed up in the surgical clinic at our hospital.
DISCUSSION

JHF is originally described by McMurray in 1873 under the name of molluscum fibrosum and was re named by Katino as JHF in 1972(2). It is a rare connective tissue disorder characterized by aberrant synthesis of...
glycosaminoglycans and biosynthesis of collagen III and IV due to an unknown aetiology (5). The causative gene is mapped to be 4q21 and mutations in capillary morphogenesis factor 2 gene is identified. (1, 3, 4, 5). It is inherited in autosomal recessive pattern but sporadic cases do occur (3).

It usually occurs in children below 5 years of age with one or more siblings affected unlike in present case. Condition is characterized by multiple skin nodules, papules and tumor-like masses in head, neck and upper body, subcutaneous fibromatosis, gingival hyperplasia, joint contractures and osteolytic defects (2). The disorder occurs in two forms; slow growing localized form and rapid growing diffuse form (3, 4).

Joint contractures usually precede other manifestations occurring in infancy and may be disabling unlike in present case. Skin nodules may vary in size with a maximum diameter of 5 cm, slow growing and painless (2). They have tendency to recur after excision (2) like in this patient. Cases with removal of numerous tumors in long durations of time have been reported with variable cosmetic outcome (2). The diagnosis is confirmed by histology (1, 5).

The skin lesions consist of benign fibroblastic proliferation-chondroid cells occupying the dermis and subcutaneous tissue, surrounded by an amorphous hyline or chondroid-like substance rich in chondroitin-6-sulphate; made up of glucosamine and galactosamine (1, 2, 3).

The disease has a progressive course with most patients surviving up to 4th decade and there is no specific treatment other than aesthetic procedures, deformity prevention and correction (1, 4). However there is no risk of malignant transformation according to available literature.

Genetic counseling is essential as the risk of having a diseased baby is about 25% in any pregnancy (1, 2).

REFERENCES