

JUVENILE HYALINE FIBROMATOSIS

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Juvenile hyaline fibromatosis is a rare connective tissue disorder, characterized by skin tumours, gingival hypertrophy and flexion contractures of the acral joints. Other associations include stunted growth, osteolytic changes and muscle weakness. The mental development is normal. Juvenile hyaline fibromatosis probably has an autosomal recessive inheritance¹, characterized by increased synthesis of glucosaminoglycans by the fibroblasts².

CASE REPORT

A male child of 26 months was seen at the department of Dermatology at Jinnah Postgraduate Medical Center, Karachi, in February, 1988. The patient was the first child of consanguineous parents of Indian origin. The child was born full term, by a lower segment caesarean section due to transverse lie. He weighed 2.6 kg at birth and no obvious abnormality was found. A few days later the mother noticed that the child cried when the nappies were changed. At 2 months flexion contractures of the hip joints developed, followed by the knee joints. At 8 months of age shoulder and the elbow joints were also involved. The skin changes were first noticed at 8 months, comprising of papules, nodules and subcutaneous swellings. The papules present on the neck (Figure 1),

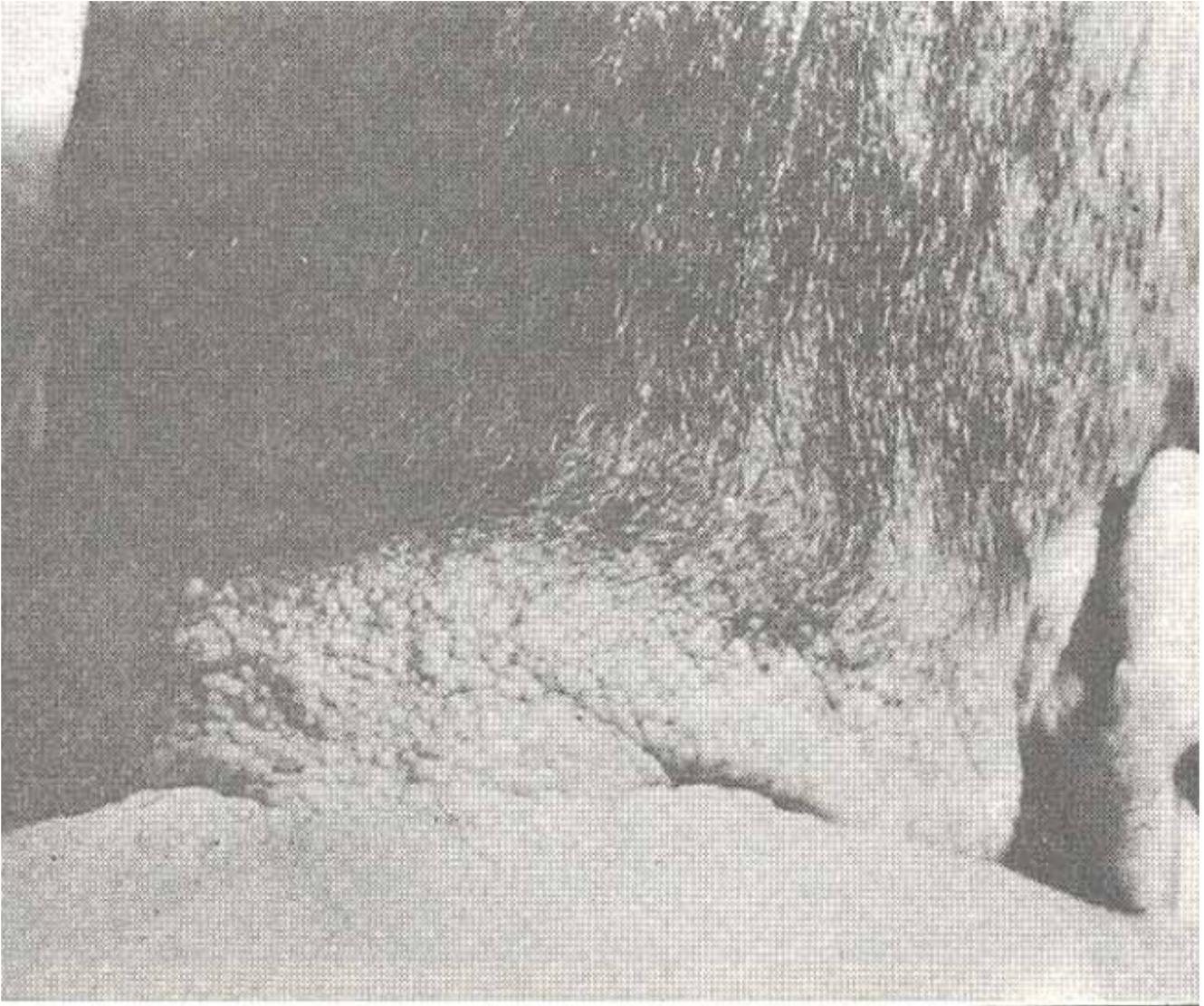


Figure 1. Papules on the neck.

ear, nose and the perianal area were firm and pearly white. Some of the nodules on the ear (Figure 2)

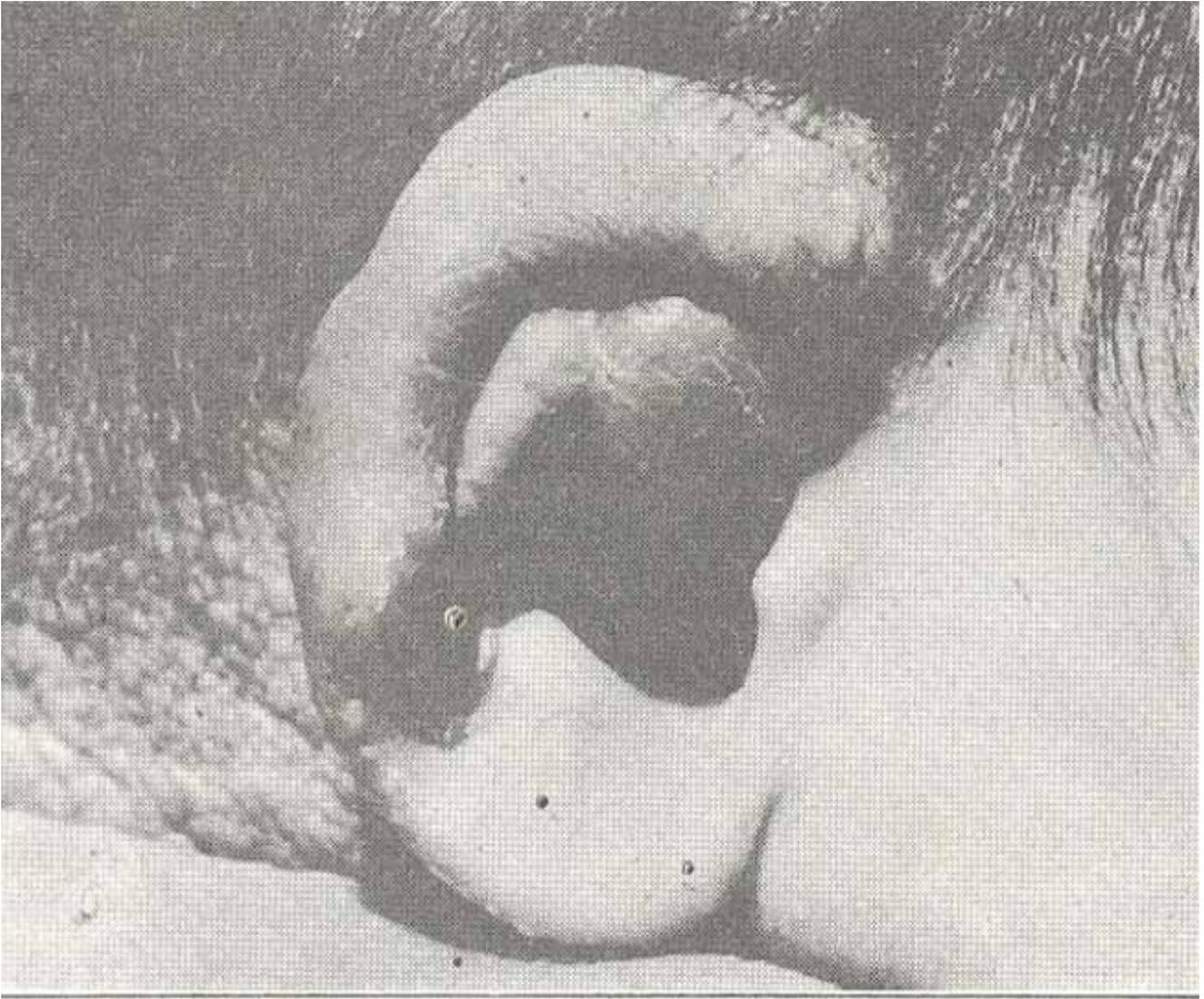


Figure 2. Nodules on the ear.

were soft and reddish which often bled and ulcerated. Others were translucent and gelatinous in consistency. The subcutaneous swellings present around the ankles and spine, were soft and fixed to the underlying structures. The gingival hypertrophy (Figure 3)

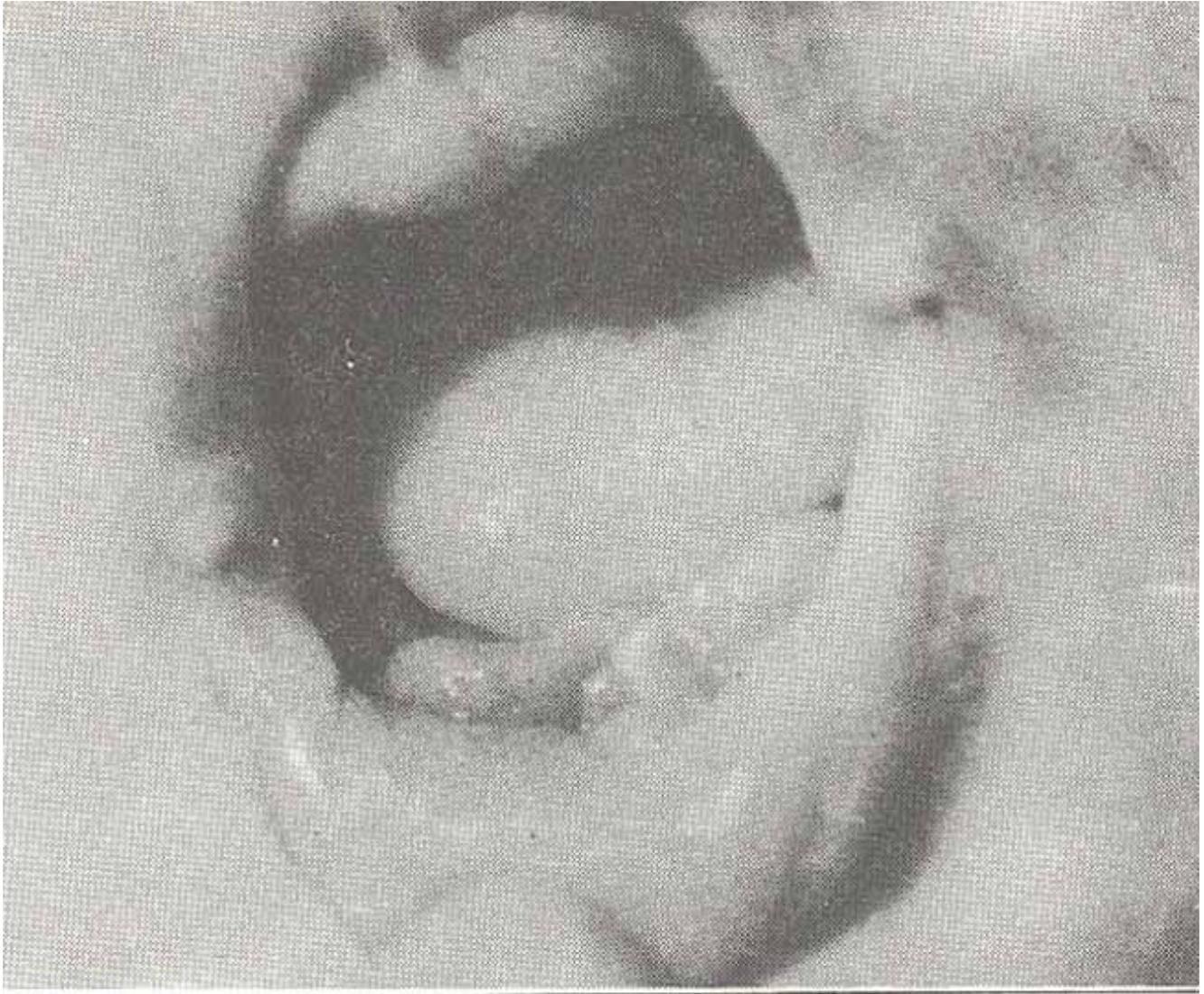


Figure 3. Hypertrophic gingivae.

was noticed at 1 year, the gingivae almost covering the teeth. The teeth erupted at the normal age, but the lower two incisors became loose and fell at the age of two. The child has not grown much since the age of one. His present height is 60cm and weight 10kg. Systemic examination revealed no cerebral, cardiovascular respiratory, renal or gastrointestinal abnormality. The child's intelligence is normal. Haemoglobin, white cell count, ESR, blood film, blood sugar, liver function tests, I^3 and T₄, urea, creatinine, calcium, phosphorus, electrolytes, lipids and cholesterol were within normal range. Rheumatoid and antinuclear factors were negative. Urine examination showed aminoaciduria, consisting of amino N butyric acid, cystine, glycine, histidine and hydroxyproline. Urinary mucopolysaccharide excretion was normal. X-ray chest was normal and radiological examination of the bones showed no evidence of osteolytic changes. Electrocardiogram and electromyogram were normal. Corneae were normal under slit lamp examination. Biopsy showed the characteristic changes of hyalinised eosinophilic material in the dermis, with fibroblasts having oval or elliptical nuclei. The hyaline material was PAS positive and diastase resistant. The overlying epidermis was atrophic. Two Eumours were removed for biopsy, the larger one showing more ground substance, and the smaller one more cellularity (Figure 4)



Figure 4. Histology showing tumors cells embedded in homogeneous ground substances.

a finding similar to Kitano¹. Oral steroids given for 6 months showed no improvement. New papules continued to appear and a subcutaneous swelling developed around the right wrist. However, the bleeding and ulceration were less. One more tooth fell after loosening. Capsulotomy is being considered for the joints.

DISCUSSION

Juvenile hyaline fibromatosis is a rare disease. A total of 17 cases were reported by Fayad et al by 1987³. Most of these have been Asians¹⁻⁶. Skin tumours, gingival hypertrophy and joint contractures have been present in all of them. The disease occurs early in life between 2 months to 4 years of age. In our case it manifested at 2 months. Consanguineous marriage has been reported in 6 cases. Siblings were affected in 5. It is likely that the mode of inheritance is recessive¹. Muscle weakness and osteolytic changes have been reported in 5 and 9 cases respectively^{1,6}. The bones commonly affected are the phalanges, humerus and tibia. Tissues taken from bone and muscle show hyaline infiltration of the collagen, similar to that of skin. Our case did not show any sign of involvement of these tissues. The other reported associations have been rectal bleeding⁵, hoarseness, faecal incontinence anal

prolapse¹, and nasal block. The mucosa was normal in our case. In most cases the laboratory investigations were normal, except for hypochromic anaemia in a few^{1,7}. Aminoaciduria occurring in our case is not reported previously. Its significance remains unclear. In all cases a similar histology is seen i.e., deposition of eosinophilic, hyaline substance in the dermis, which is PAS positive and diastase resistant. Epidermis only shows secondary changes. Juvenile hyaline fibromatosis should be differentiated from the other connective tissue disorders. In mucopolysaccharidosis there are skeletal and cutaneous abnormalities, and stunting of growth. Mental retardation, clouding of cornea and hepatomegaly, so characteristic of mucopolysaccharidosis, are absent in juvenile hyaline fibromatosis. Urinary mucopolysaccharide excretion is normal in the latter disorder. Winchester syndrome, a new child mucopolysaccharidosis, an association recently challenged by Hollister et al⁸, has the histological features almost identical to juvenile hyaline fibromatosis³. Clinically the syndrome has many similarities to juvenile hyaline fibromatosis e.g. joint contractures, stunted growth, gingival hypertrophy, bony changes, normal intelligence and normal urinary acid mucopolysaccharide excretion. The skin lesions in Winchester syndrome are in the form of cutaneous thickenings with hypertrichosis and hyperpigmentation. In juvenile hyaline fibromatosis multiple cutaneous tumours are characteristic and the corneal opacities present in Winchester syndrome are absent. Congenital generalized fibromatosis is another connective tissue disorder, characterized by proliferative activity of the fibroblasts. It involves not only the skin and joints, but also the viscera and muscles⁶. The cutaneous tumors are present at birth and the condition is fatal within the first few weeks of life. In juvenile hyaline fibromatosis the tumours are not present at birth and skin and joints are mainly involved. The initial prognosis is poor but the disease is not fatal. Flexion contractures, the most disabling feature of juvenile hyaline fibromatosis, are not found in congenital generalized fibromatosis. Neurofibromatosis is characterized by cutaneous neurofibromas which appear in childhood and increase rapidly at puberty. When present at birth the condition is often fatal⁹. The other manifestations of neurofibromatosis as cafe-au-lait spots, axillary freckling and Lisch nodules are absent in juvenile hyaline fibromatosis. Treatment is unsatisfactory. Steroids and cytotoxic drugs have been tried. Capsulotomy only helps the joints temporarily. The patient with the longest survival is 33 years old², but severely handicapped. Only one death is reported¹. The post mortem examination of the patient showed infiltration of the eosinophilic, amorphous substance in the tongue, thymus, spleen and lymphnodes. Fulminating hepatitis and oedema of the brain were the direct causes of death.

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