JUVENILE HYALINE FIBROMATOSIS WITH RAPID TUMOUR GROWTH AND ULCERATION

ADEL ABDULLAH BANJAR, FRCS*
FUAD KHALID HASHEM, MBBS, FRCS(C)*
SAAD MOHAMMAD ASIRI, MD**

Juvenile hyaline fibromatosis is a disease characterised by multiple skin tumours, flexion contractures of the joints, hypertrophic gingiva, osteolytic hone lesions and perianal granulomas. The skin tumours may regress, but progressive tumour growth with or without ulceration, continuous development of new skin lesions with subsequent deformity is the characteristic course of this disorder.

We describe a clinical case with rapid tumour growth and ulceration in a short time. Other features of the syndrome are discussed.

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Juvenile hyaline fibromatosis is a rare fibrous proliferative disorder. The disease has a well defined clinical, microscopic and ultrastructural features but little is known about the cause, the natural history, complications[1] and the management. The histopathological feature is charactised by fibroblastic proliferation and the presence of thin fibrillar elements embedded in an amorphous, eosinophillic hyaline material.

THE CASE

A 4 year old girl presented to the Paediatric Clinic in January 1993 with multiple skin swellings in the body and limitation of the movement of the arms and legs. The condition started at the age of 18 months as the parents noted that the movement of the elbows and knees is diminished and associated with the development of numerous skin tumours at the periarticular areas, the face, the scalp and the trunk. The tumours grew slowly and deformity and physical retardation developed.

The patient was born after an uneventful full-term pregnancy by normal delivery to non-consanguineous parents. The patient had a sibling who died of a similar condition at the age of 6 months and another healthy sibling. The family history is unremarkable. Clinically, the mental development of the patient is normal in spite of the physical retardation. Numerous skin tumours of various sizes, consistency and location are seen in the scalp, the lips, the nose and the periarticular areas with muscle wasting and flexion contracture deformity were observed (Fig 1). There were 4 tumours involving the scalp but more prominent at the right side in the occipito-parietal and frontal areas and measured 8 x 7 cm and 5 x 6 cm, respectively with firm consistency (Fig 2).

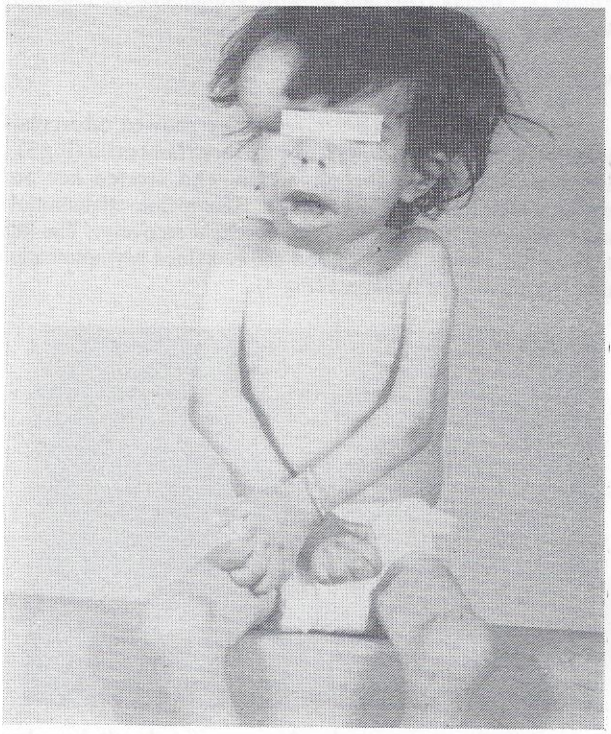

Figure 1. Multiple tumours at the scalp, nose, lips and periarticular areas. Notice the pigmentation around the ankles and flexion contracture deformity.

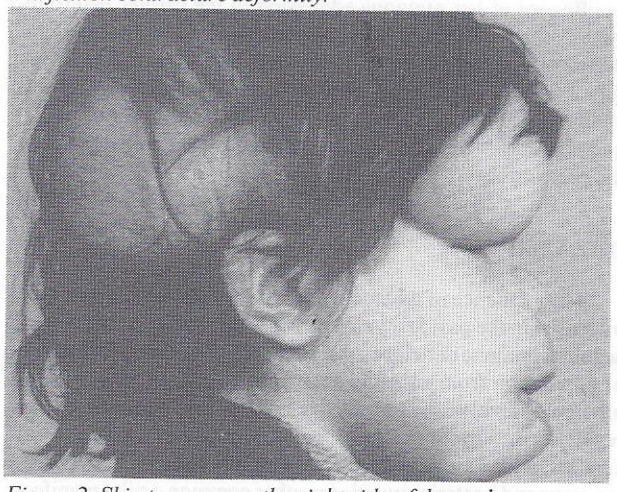

Figure 2. Skin tumours on the right side of the scalp

* Consultant Plastic Surgeon
The Section of Plastic and Reconstructive Surgery
Department of Surgery
King Faisal Specialist Hospital & Research Centre
Riyadh, Saudi Arabia
** The Section of Otolaryngology
Department of Surgery
Security Forces Hospital
Riyadh, Saudi Arabia

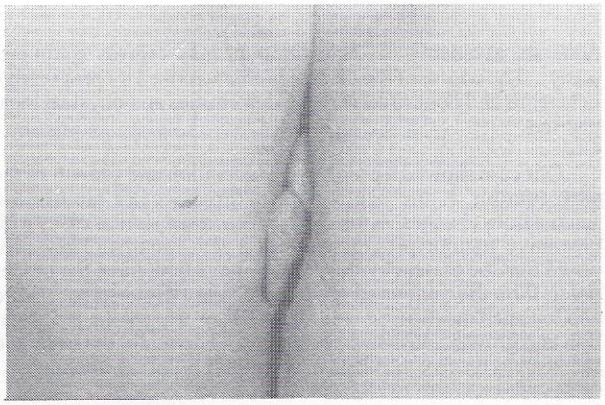

Figure 3. Perianal granulomas

The patient had hypertrophy of the gingiva, pigmentation around the ankles and perianal granulomas (Fig 3). Skeletal survey revealed soft tissue swelling and profound osteopenia of long bones and hands without obvious osteolytic lesions (Figs 4 & 5). Apart from microcytic, hypochromic anaemia, all laboratory tests were normal. The diagnosis of juvenile hyaline fibromatosis was proposed because of the combination of clinical features. Further follow-up of the patient was not available.

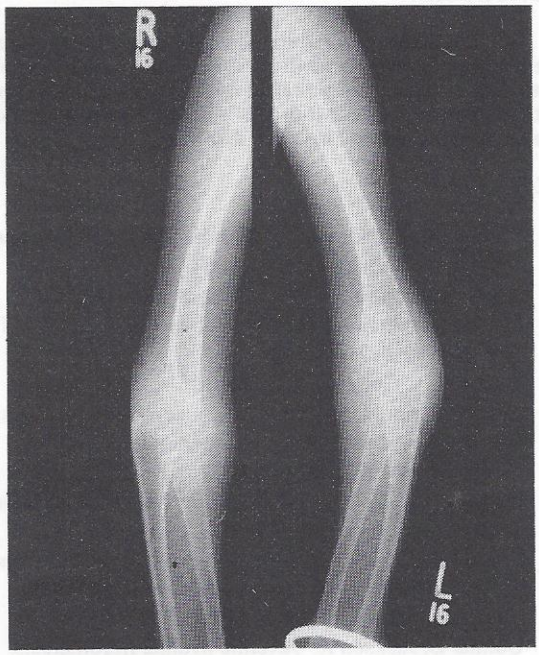

Figure 4. Roenterograms of the upper limbs show profound osteopenia

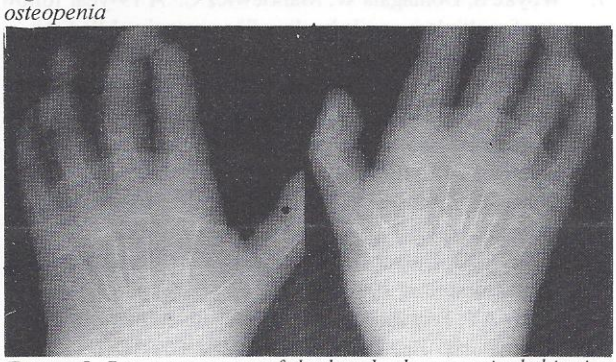

Figure 5. Roenterograms of the hands show cortical thinning and soft tissue swelling at carpal regions

Twenty months later, the patient presented again to the Emergency Department with a discharging ulcer measured about 2 x 2 cm that appeared at the right occipito-parietal tumour. The ulcerative tumour enlarged progressively and two weeks later, the ulcer measured 10 cm x 8 cm (Fig 6). CT scan of the head revealed soft tissue tumour without bone involvement or hemorrhagic or necrotic changes (Fig 7). Biopsy was taken and histopathological examination confirmed the initial diagnosis and showed hyaline fibrous tissue infiltrated by inflammatory cells with ulcerated surface. No malignant cells were seen.

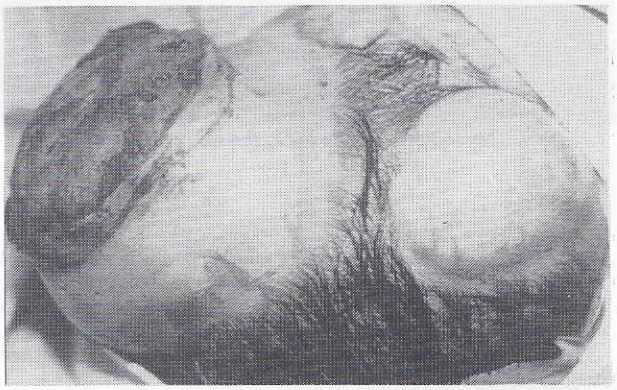

Figure 6. Skin tumours of the scalp. The largest tumour is ulcerated

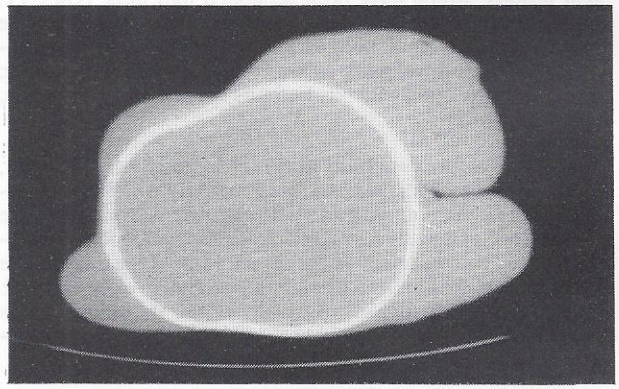

Figure 7. CT scan of the head. Huge extracranial soft tissue tumours over occipito-parietal areas. No haemorrhage, necrosis, bone erosion or intracranial abnormalities

The ulcerative tumour was excised with primary closure (Fig 8). In spite of the incomplete excision, no recurrence was observed after two years follow up. One year later, another ulcer developed on the back (Fig 9) but progressed slowly and was managed surgically in a similar fashion.

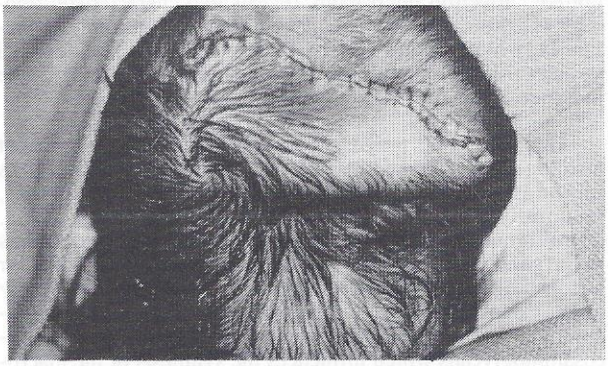

Figure 8. Excess skin facilitate the primary closure of the wound

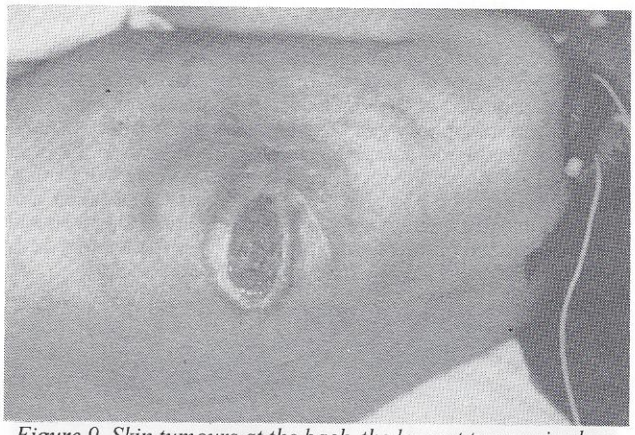

Figure 9. Skin tumours at the back, the largest tumour is ulcerated

DISCUSSION

Juvenile hyaline fibromatosis is inherited as an autosomal recessive disorder and there is an almost equal sex distribution. The disease has been described in siblings and many are born to consanguineous parents. No ethnic differences appear to exist[2]. The onset is usually noted from birth to early childhood[2-3]. Late diagnosis may occur[4].

The most obvious and near constant features are: the fibrous skin lesions, which have been separated into three types: (1) small, fleshy, pearly papules occurring especially on the face and the neck; (2) small nodules and large plaques with translucent appearance developing on fingers, ears and around the nose; (3) firm, large, subcutaneous tumours especially on the scalp, trunk and limbs[3].

Other features include gingival hypertrophy, flexion, contractures of the major joints, macrocephaly with facial asymmetry and perianal granulomas[3]. The growth is stunted and the mental development is normal[2]. Myopathy[3] and repeated infections have been reported. In the severe form of the disease, the patients usually die of overwhelming infection in early infancy[2]. Multiple osteolytic defects especially of the skull, long bones and phalanges with or without cortical erosion, soft tissue calcification are the main radiological features[3].

There is no characteristic laboratory finding for this disorder. Microcytic hypochomic anaemia, hypoproteinemia, hypoglycaemia and slight elevation of erythrocyte sedimentation rate secondary to poor nutrition and infection have been described.

An increase of hyaluronic acid and dermatan sulphate was found in one case and hyperglycinuria with increased histidinuria in two patients. No immunologic abnormalities have been reported[2]. Electron microscopy reveals two characteristic findings: (1) stromal cells with enlarged rough endoplasmic reticulum and a hypertrophic Golgi apparatus containing cystic Golgi vesicles filled with a fibrillar and granular material and, (2) the presence of this fibrillar material in the hyaline matrix[3].

Gilaberte et al[3] proposed a clinical diagnostic criteria but histological confirmation is needed. The differential diagnosis includes arthrogryposis multiplex congenita, mucopolysaccharidosis, Farber lipogranulomatosis, neurofibromatosis and inflammatory connective tissue disorders in infants[2,3,5]. Infantile systemic hyalinosis has similar histopathological features but clinically characterised by early onset, extensive internal organ involvement and poor prognosis[3]. The pathogenesis is unknown. An accumulation of glycosaminoglycans which influences the synthesis or the macromolecular organisation of collagen fibers has been proposed[3]. Follow-up reports suggest a reasonable life span if complications are avoided[1,2,6,7]. Skin tumours may regress spontaneously[7]. The course of the disease is characterised by progressive cutaneous and bone involvement, deformities and physical incapacities[1,3].

Our case showed a progressive ulceration and growth of the skin tumour at the scalp within three weeks simulating sarcomatous changes. Kitano et al[8,9] described similar cases with ulcerative tumour progressed within few months. They proposed that the mesenchymal cells of the patient have the potential to form the tumour and to produce growth substance. Mild and repeated mechanical stimuli probably trigger the proliferation and growth substance production. The scalp, the back and sacral areas receive the mechanical stimulus during infancy[8].

The treatment of this disorder is unsatisfactory. Radiotherapy is ineffective. Gum hypdertrophy has been treated by surgery with some beneficial results[10,11]. Intra-lesion steroid injection[8], anti-inflammatory agents[10], orthopaedic procedures[12] and physiotherapy may improve the pain and joints function state.

Complete excision of troublesome skin lesions is needed[2,4-7]. Hallock[4] and Woyke et al[7] advised early surgical intervention.

CONCLUSION

Juvenile hyaline fibromatosis affects different areas of the body and multiple medical specialities may be involved in the management. Further investigation and case reports with different presentation will add more information to our experience about this rare disorder.

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