EHLERS-DANLOS SYNDROME: A CASE REPORT

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Introduction

Originally described by Van Meekeren in 1682, this condition was further clarified by Ehlers in 1901, and Danlos in 1908 (Smith; 1976). More than 100 cases have been described in literature; this is probably the first reported case in Pakistan.

Other syndsomes of this condition are Cutis Hyperelastica, India-Rubber Man, and Derma-torrhexis.

This is a generalised disorder of connective tissue which primarily affects skin and ligaments, but other parts of the body may also become, involved. The defects are the result of single defective gene. McKusick (1966) in an exhaustive review, described various clinical types of the syndrome with distinctive clinical, genetic and biochemical features.

The classical syndrome has been found to have the following features:

(1) Skin manifestations: Velvety, hyperelastic and fragile skin with poor wound healing. There are "cigarette paper" scars called molluscoid pseudotumour, over knees and shin in older children. The skin bruises easily. Minor traumas are likely to produce gaping "Fish mouth" wounds which hold sutures poorly. Friable tissues create difficulties at operation.

(2) Joints and Skeletal Defects: Extreme laxity of joints leads to genu recurvatum, habitual dislocation of various joints, flat feet and kyphoscoliosis. Other skeletal defects include small stature, down sloping, ribs and talepes equinovarus.

(3) Internal Defects: Diaphragmatic hernia, diverticuli of gastro-intestinal and respiratory tract, spontaneous pneumothorax, dissecting aneurysm of the aorta.

(4) Eye Defects: Myopia, microcornea, kera-tocornea, glaucoma, retinal detachment.

(5) Facial features: Narrow maxilla, wide nasal bridge.

(6) Biochemical changes: Some patients have collagen with an abnormally low hydroxylys ine content. Measurement of activity of the enzyme lysyol-proto hydroxylase in cultured fibroblasts, in some patients, revealed approximately l/8th of normal value. In others the enzyme procollagen peptidase has been found to be defective.

The biochemical defects are not universal and it seems that the underlying basic defect varies from patient to patient, which would explain various clinical types and modes of inheritance.

Case Report

K.K., a male child, age 1-1/2 years, reported on 4th August, 1980, with complaints of generalised weakness of limbs and difficulty in standing and walking. The baby was born at home after full term. The first stage of labour was said to be prolonged. Birth weight was not recorded but he was described as 'small' at birth. He is the fifth and youngest child. No other similar case was seen in the family. Parents were not related. Developmental motor milestones were delayed but mentally he seemed to be within normal limits, (Neck control: 6 months, sat alone: 1-1/2 years, unable to stand at the time of interview; speech: vocabulary of 3-4 words).

Examination: Height: 76.6 cm, Weight: 7.3 Kg., Head circumference: 47", anterior fontanelle present. The skin was hyperelastic and could be pulled 2" beyond the surface but returned promptly to its normal position on release (Fig. 1).

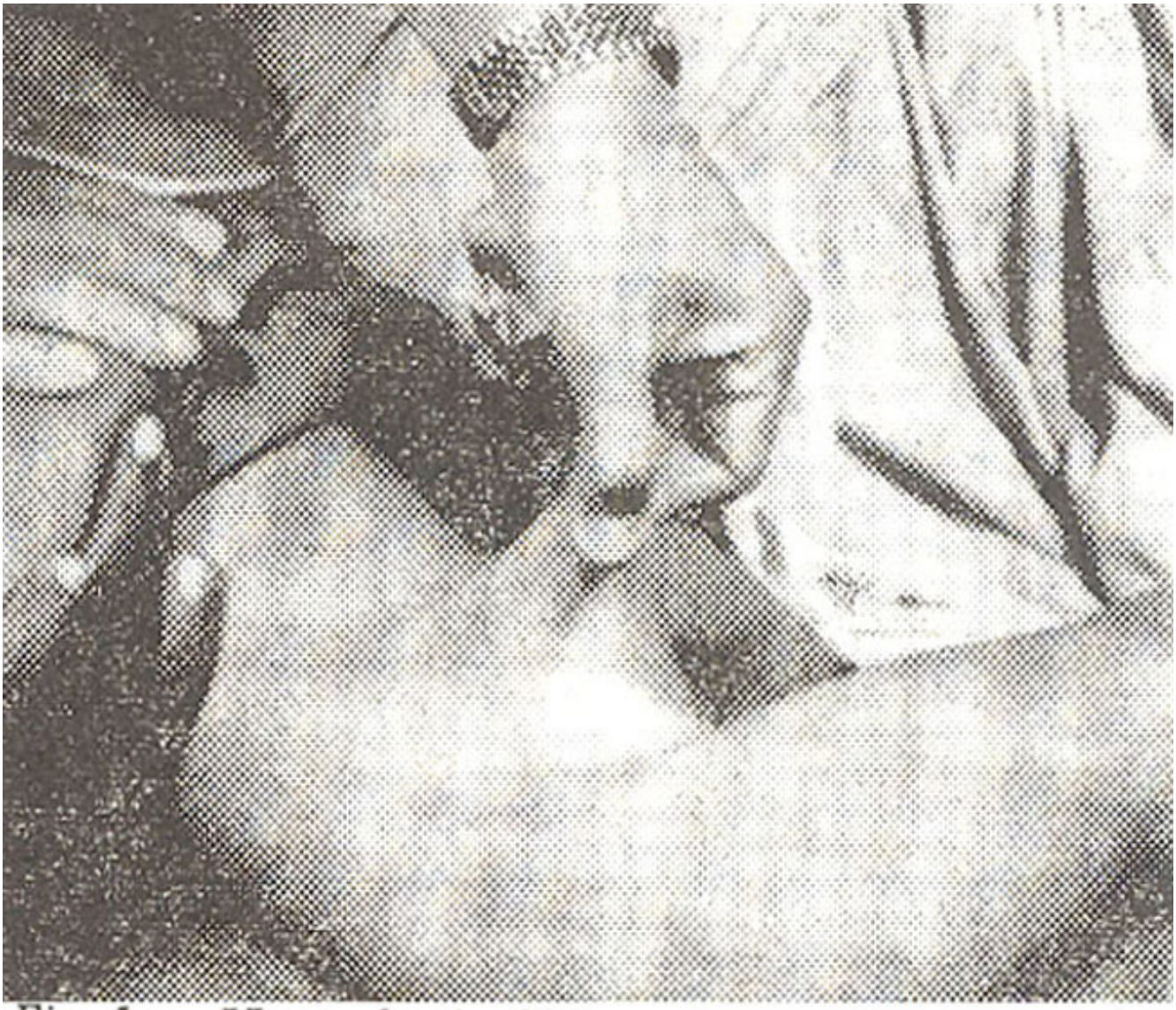

Fig. 1 : Hyperelastic Skin

There was no bruising or scarring. The joints were very lax leading to abnormal position and posture of different joints of the body. He had bilateral genu recurrvatum, flat feet and prominent heels. Passive dorsiflexion and palmar flexion of the wrist joints lead to approximation of surfaces between the hand and mid-arm (Fig. 2).

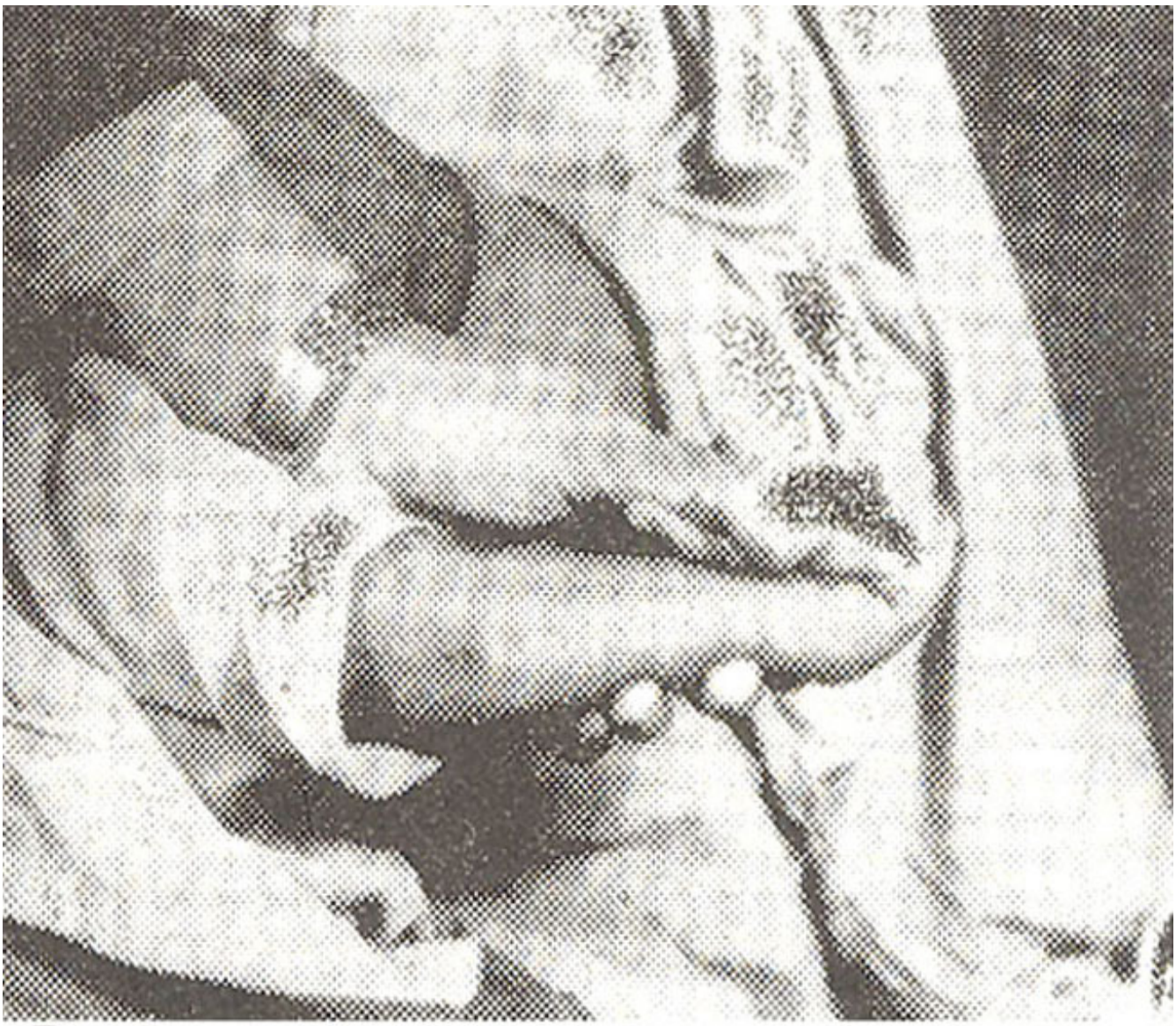

Fig. 2 : Extreme Laxity of Joints

Similarly the dorsal surfaces of both feet could be passively made to touch the anterior surface of lower legs (Fig. 3).

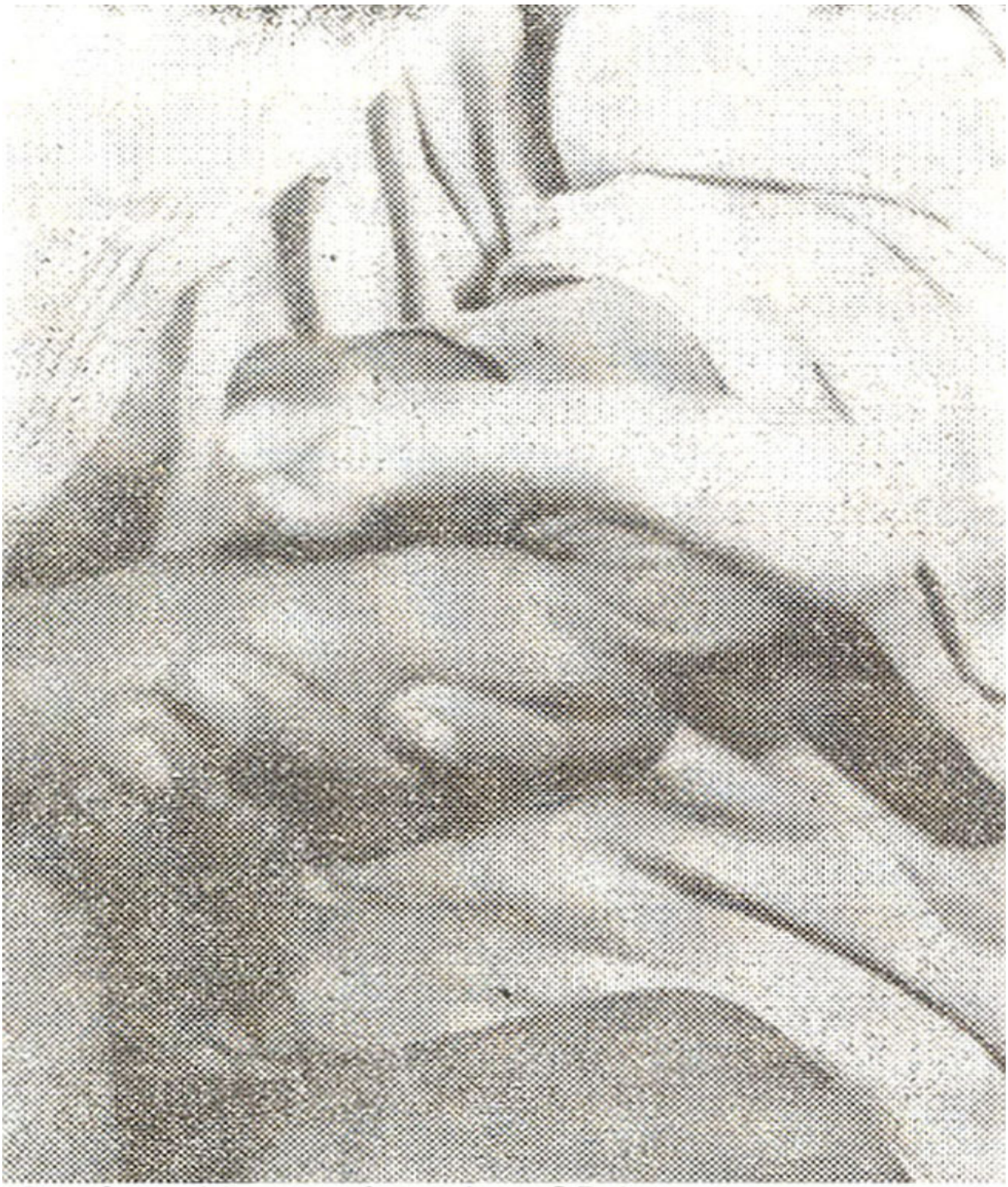

Fig. 3 : Abnormal Laxity of Joints

There was marked postural kyphosis at the lower dorsal region. Inguinal hernia was present on the right side. He also had an undescended testis on the same side. Examination of the heart, nervous system and eyes revealed no abnormality. Other examination was also within normal limits. X-Ray spine, skull, hands, feet and lungs revealed no abnormality. ECG-Normal; Blood, CP, ESR were normal.

Discussion

Brabas (1966) discovered that most patients, with Ehlers Danlos Syndrome were born before term due to the early rupture of the membranes, which, be thought, is an indication of defective tissue in these cases. Wound healing is delayed and the resultant scar is inadequate. Prolonged bleeding may occur following injuries. Surgical procedures on these individuals should be raken up with extreme caution. The patients should be instructed to avoid traumas. Gastrointestinal haemorrhage or haemoptysis may be a problem. Affected females are liable to post-partum haemorrhage. There is a tendency to chilblains and peripheral cyanosis. In time there develops subcutaneous calcification in areas of repeated trauma. Our patient had the typical laxity of joints, and elasticity of skin; however there was no evidence of bruising, nor were there any scars. But he was only 1-1/2 years old at the time of examination and had not learnt to walk. This would explain the absence of scars and bruises. Internal manifestations of the condition were not detected; again perhaps due to his being too young. These manifestations develop with the passage of time. He did not show any eye defect either.

References

1. Brabas, A.P. (1966) Ehlers-Danlos syndrome: Associated with prematurity and premature rupture of the membrane; possible increase in incidence, Br. Med., J., 2:682.
2. McKusick, V.A. Heritable disorders of connective tissue. 4th ed. Saint Louis, Mosby, 1972, p. 179.
3. Smith, D.W. In recognisable pattern of human malformation Philadelphia Saundres, 1976, p. 284.