The Ehlers-Danlos syndrome has been so named from descriptions made by Ehlers in 1901 and Danlos in 1908, although it was first described by Meek’ren in 1682. In 1934 Tobias described the various conditions that may be associated with the syndrome, such as acrocyanosis, club foot, fibromata, naevi, lymphangiectasis, mental deficiency, supernumerary teeth, syringomyelia, and von Recklinghausen’s disease. In 1936 Ronchese emphasised the feature of skin fragility and reviewed the literature; since then numerous articles have appeared, notably that by Johnson and Falls in 1949.
Clinical features—The Ehlers-Danlos syndrome is usually noticed in early childhood. Delay in sitting and walking is associated with widely spaced eyes and a broad nasal bridge (Fig. 1). The skin, which has a lack of subcutaneous tissue, is lax, hyperelastic and velvety: it splits easily on minor trauma, being abnormally fragile, and, after delay in healing, leaves typical tissue-paper scars which are atrophic and telangiectatic (Fig. 2). If care is not taken to place sutures well away from the wound edges they will pull out of the skin.
Pseudo-tumours of a molluscid nature may follow minor trauma, and haematomata form easily—probably not from any abnormality in the vascular walls themselves, but from lack of adequate connective tissue support of the blood vessels.

The laxity of the joints (Figs. 3 and 4), although of advantage to the so-called india-rubber man at the circus, has many attendant drawbacks as it may, with the associated muscle weakness, resemble amyotonia in early life, and leads to sprains, spontaneous dislocations (reported in the shoulder and patella) and collapse of the longitudinal arches of the foot.

Calcified subcutaneous nodules may be present, and because of the general laxity of the connective tissues there is a definite tendency to diaphragmatic and other herniae.

There is usually a family history; genetic studies of affected individuals suggest that the syndrome is transmitted by a dominant gene.

Pathology—The typical changes are a decrease in the collagen fibres, which are less compact than normal, and which are said to be confined to the skin, connective tissue and joints. The elastic fibres seem to be increased and to have a closer grouping, with less separation by collagen than is normal. There is a generalised decrease of dermis and subcutaneous tissue, and in the latter fatty nodules may occur, which are enclosed in a fibrous tissue capsule, and which may be calcified. Radiographically these nodules are characterised by dense outer margins and translucent centres.

There is no apparent abnormality of bleeding time, clotting time, platelet count or capillary fragility.

The association of the Ehlers-Danlos syndrome with wedging and deformity of the vertebrae has not been reported. The following case report illustrates this feature and is also of interest in that some of the members of the family on the maternal side have suffered from acholuric jaundice.

CASE REPORT

Boy, aged twelve. History—Shortly after birth in February 1946, which was by breech delivery, the baby was observed to have an Erb's palsy of both arms, a thoraco-lumbar kyphosis and a right talipes calcaneus. At the age of one year the arms had recovered but the child had hypermobile joints and could not sit up. When he began walking at two years and eight months the kyphosis became more marked. Despite the provision of a back support, when the boy was eleven, the kyphosis...
continued to deteriorate and he was referred to the Robert Jones and Agnes Hunt Orthopaedic Hospital. He said he never played rough games, as cuts always opened widely and healed with difficulty. 

Family history—The boy's mother underwent splenectomy and cholecystectomy for acholuric jaundice in 1951, which was followed by normal wound healing. She was also found to have 15 degrees hyperextension of the interphalangeal joints.

The boy's maternal grandmother had normal skin, but early osteoarthritis of the hip, knees and ankles. Her husband, the grandfather, had died at the age of fifty-seven from a stroke, but was said to be normal.

Of four maternal uncles one has 15–20 degrees hyperextension of the proximal interphalangeal joints. The second is normal but has a son with a slight increase of skin elasticity, and hyperextension of all interphalangeal and metacarpo-phalangeal joints as well as 20 degrees of calcaneus of the feet. The third has had a splenectomy for acholuric jaundice with normal healing. He has poor movements in a symptomless lumbar scoliosis. The fourth has also had a splenectomy for acholuric jaundice.

The one maternal aunt is normal.

Fig. 5
Case 1. Figure 5—The kyphoscoliosis at the age of eleven years. The wedge-shaped vertebrae are most unusual in this condition. Figure 6—The abnormality in the epiphyses of the arm is slight.

The patient's sister has 60 degrees hyperextension of the metacarpo-phalangeal joints, which is less marked in the interphalangeal joints.

A cousin of the boy's mother has a son with thoracic scoliosis and bilateral Sprengel's shoulder, but he was not able to be examined. His case notes show no other record of skin or joint abnormality.

The boy's father, paternal grandparents and one surviving paternal uncle appear to be normal. Examination—The child was underdeveloped for his twelve years, being 3 feet 10 inches in height. He was myopic and stood with a fixed thoraco-lumbar kyphoscoliosis (Fig. 1) with hypermobility of the spine above and below; he could rotate the cervical spine through 220 degrees and touch his seventh cervical vertebra with his occiput. The elbows and knees appeared fusiform, and the legs were bowed. Atrophy of the pectoral muscles has left a depression below each clavicle. There were numerous tissue-paper scars on the legs and forehead. The skin felt velvety and was very elastic (Fig. 2). The wrists and metacarpo-phalangeal joints were hypermobile (Fig. 3) and the shoulders abducted to 225 degrees (Fig. 4). The elbows and hips showed slight restriction of movements but the knees, subtalar and interphalangeal joints were normal.
Investigations—The boy had a normal haemoglobin, red and white cell counts, blood urea and renal function. Other biochemical tests showed:

- Serum sodium: 280 milligrams/100 millilitres: 122 mEq/litre.
- Serum potassium: 17.0 milligrams/100 millilitres: 4.4 mEq/litre.
- Alkali reserve: 58 millilitres CO₂/100 millilitres: 26 mEq/litre.
- Alkaline phosphatase: 15 units.
- Serum calcium: 9.8 milligrams/100 millilitres.
- Serum inorganic phosphate: 4.4 milligrams/100 millilitres.
- Basal metabolic rate: +14 per cent.

Radiography—The thoraco-lumbar spine showed wedge-shaped vertebral bodies (Fig. 5). The other bones were to all intents normal with the exception of slight irregularity of the humeral, radial and lower ulnar epiphyses (Fig. 6).

Treatment—The kyphosis is being corrected and it is hoped to perform a spinal fusion ultimately.

DISCUSSION

This patient shows many of the features of Ehlers-Danlos syndrome, with the typical hyperelastic, velvety skin, loss of subcutaneous tissue, skin fragility and hypermobile joints. The unusual features are: 1) some joints show restricted movement; 2) the thoraco-lumbar kyphosis and wedge-shaped deformity of the vertebral bodies bring to mind similar changes seen in chondro-osteodystrophy. Gargoyleism and achondroplasia are excluded in this patient.

It is also unusual to find no genetic background in the family history, in which there is little abnormality except the acholic jaundice.

I should like to thank Mr Gordon Rowley and Mr Robert Roaf for permission to publish this case and Miss Norah Walker for her advice on the radiographs.

REFERENCES


