# THE EHLERS-DANLOS SYNDROME
## Discussion and Case Presentation

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### Introduction

The Ehlers-Danlos Sydrome (Syn. Cutis hyperelastica) is a rare syndrome, the characteristic features of which are hyperelasticity and fragility of the skin, hyperextensibility of joints and fragility of blood vessels. Musculo-skeletal, ocular and internal manifestations are present in some cases (Day and Zarafonetis, 1961)

### Pathology

There is a generalised defect of connective tissue in which the collagen fibres are scanty and arranged in a disorderly fashion, whilst the elastic fibres are increased. The essential defect is a quantitative deficiency of collagen. Light microscopy studies reveal collagen fibres which appear normal but they form an inadequate weave of loose texture in the dermis, subcutaneous tissues and joint capsules. Electronmicroscopy shows an abnormal periodicity of the striations of the collagen fibres. The elastic fibres are qualitatively normal on histochemical and electronmicroscopic examination but they are increased in number.

Defects in the adventitia of small arteries and inadequate support from surrounding connective tissue account for the vascular vulnerability (Barabas, 1966) which may be a conspicuous clinical feature as in the case reported below. Blood platelets are defective in most cases (Lisker *et al.*, 1960). This defect may contribute to the abnormal beeding tendency.

The inheritance of this syndrone is of two types. In the majority of cases, it is determined by an autosomonal dominant gene .The expression of the gene is variable and incomplete forms are frequent. In some cases inheritance seems to be sexlinked (Beighton, 1968).

### Clinical Features

The condition is usually first noticed when the child begins to move about freely, but the milder forms are often overlooked.

### The Skin

(a) *Hyperelasticity*

The synonym of this syndrome, *cutis hyperelastica,* is self explanatory. The hyperelasticity of the skin varies in degree and extent. The skin feels soft and velvety. It is hyperextensible and this can be demonstrated by pulling skin out in a fold *(Fig. I).* On release, the skin springs back to its original position. On the palms and soles, the skin tends to be redundant and may flatten out like a loose glove on pressure .The skin is not otherwise lax until later in life when redundant folds may form at the elbows.

(b) *Fragility*

Because of the reduced bulk of collagen present, the skin is fragile and may split even on minor trauma. Lacerations form gaping wounds which heal very slowly to leave broad ,paper-thin atrophic scars. In these scars, spongy tumors con-

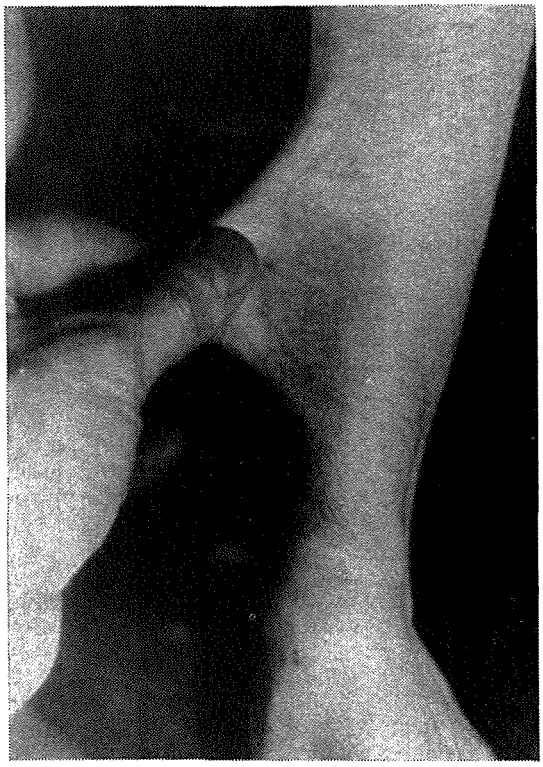

**Fig. I: Showing skin hyperextensibility.**

sisting of fat and mucoid material may develop. These in turn may calcify and feel like hard, mobile grains of rice under the skin. Sutures may tear out completely.

### The Joints

Hyperextensibility of the joints is another of the features of this syndrome *(Fig. 2)*. The hyperextensibility is variable. There may be merely double jointed fingers but in some cases walking is difficult and the gait is waddling and stumbling. Subluxation of the larger joints may occur spontaneously or through slight trauma. Due to hyperextensibility of joints and weak ligaments, kyposcoliosis is often present *(Fig. 3)*. Muscle tone is often poor and hernias develop.

### The Blood Vessels

There are two factors which cause an abnormal bleeding tendency. These are friability of the blood vessels due to the abnormal connective tissue which supports them, and to defect in the platelets.

Minor injuries not breaking the skin may induce haematoma formation and easy bruising may be a presenting symptom. Fatal haemorrhage has followed trauma to large vessels.

### Other Concomitants

Defects in other organs do not occur regularly but anomalies of the heart and dissecting aneurysms have occurred. Angioid streaks in the fundus and angiomas of the skin have been noticed in several cases.

The Ehlers-Danlos syndrone has occurred with osteogenesis imperfecta (Biering and Iverson, 1955) pseudo-xanthoma elasticum and Marfari's syndrome.

General physical and mental development are usually normal. The expectation of life is normal except in individuals with cardiac defects, but some deaths have occurred in youth from rupture of large arteries.

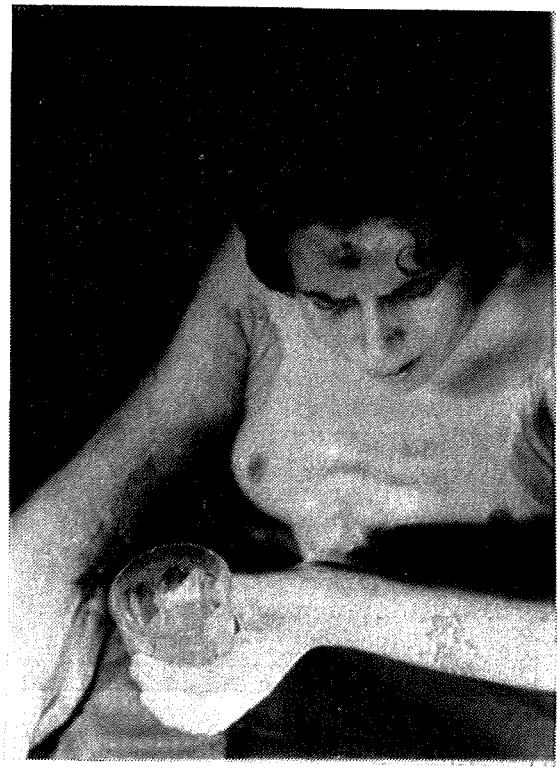

**Fig. 2: Hyperextension of the wrist joint.**

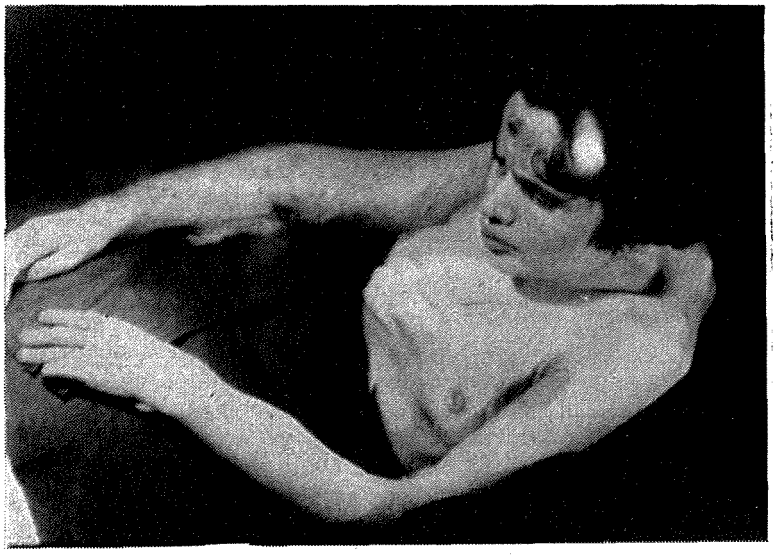

**Fig. 3: Prominent sternum secondary to Kyphoscoliosis.**

No treatment is known. Surgical procedures may present a problem as the tissues are friable and difficult to suture. Meticulous techniques and pressure dressing are desirable.

### Case Presentation

A nineteen year old girl was admitted to the Casualty Ward at York County Hospital suffering from shock. She had first attended the Casualty Department six hours previously after sustaining a small 2″ incised wound in her right forearm following minor injury. This wound had been cleaned and sutured and after the application of a dressing, the patient had been discharged.

Now she was in clinical shock. Her pulse was 140/min. and of small volume; her B.P. was 90/50 mm.Hg.; her R.R. was 30/min .and she was covered with cold sweat. Her right forearm had swollen enormously and it was quite painful. The bandages were removed and examination of her right forearm showed gross swelling and vast bruising over the part. It was obvious that the injury sustained previously has caused bleeding inside the limb, that this had continued and had now caused shock. A blood transfusion was set up. Further pressure bandages were applied and the limb was elevated. Six pints of fresh blood were given over a period of sixteen hours, by which time the bleeding had stopped and the shock had been overcome.

The girl was suffering from the Ehlers-Dunlos Syndrome. The main characteristics of the syndrome were all present:

1) Hyperextensibility of the skin *(Fig. 1)*
2) Hypermobility of the joints *(Fig. 2)*
3) Severe bleeding from minor trauma — the main features of this case
4) Gross Kyphoscoliosis *(Fig. 3)*.

*Fig. 4* shows the relative girths of the right and left forearm. This photograph was taken two days after admission. The swelling in the right forearm *(Fig. 5)* took three weeks to subside. The photograph also shows the marked Kyphoscoliosis. Angiomas are present in her arms.

The girl had a history of easy bruising from childhood but had never had such episodes of severe bleeding. The Kyphoscoliosis was first noticed in childhood but had become worse in her teens and she had evidence of respiratory insufficiency secondary to the marked chest deformity.

Two other members of her family had incomplete forms of the syndrome mainly

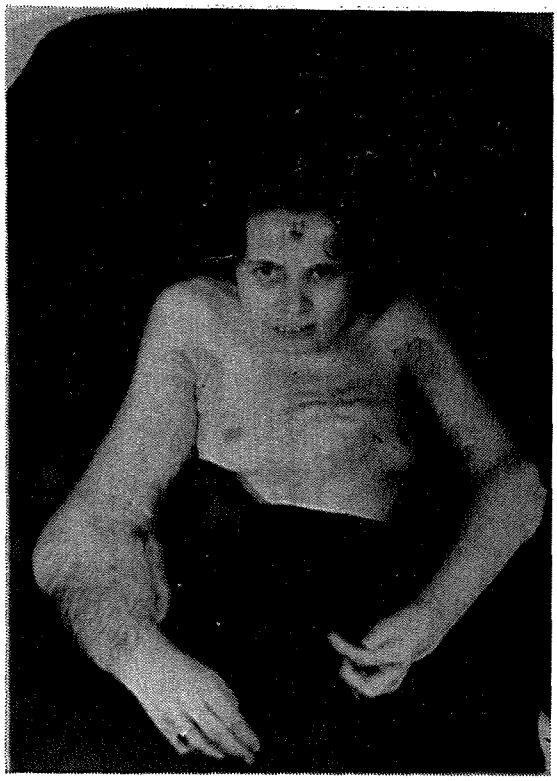

**Fig. 4: Showing relative girths of right and left forearm and marked Kyphoscoliosis.**

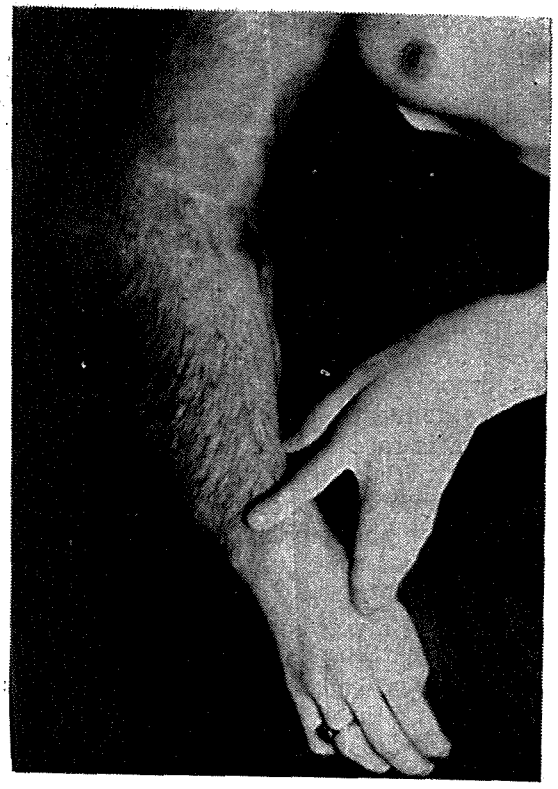

**Fig. 5: Swelling of right forearm.**

hyperextensibility of joints and hyperelastic skin but were otherwise normal.

## Comment

This case illustrates the main features of the Ehlers-Danlos Syndrome. Severe bleeding causing shock had followed minor trauma.

A review of the world literature reveals no fewer than fifteen reports of death due to bleeding, in this Syndrome. The bleeding almost always followed trauma which would not, under normal conditions, have caused more than minor bruising. In a few cases, death had resulted from spontaneous gastro-intestinal bleeding, perforation of the bowel or spontaneous intra-abdominal and other large arterial rupture (McFarland and Fuller, 1964).

Less serious bleeding had been reported in several other cases. A particular type of bleeding reported is rectal bleeding following passage of hard stools. This is probably caused by splitting up of the anal mucosa caused by hard stools, owing to the underlying tissue friability.

The cause of the bleeding is the abnormally weak connective tissue which supports the blood vessels. A platelet defect also contributes to the abnormal bleeding tendency.

## Acknowledgement

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