# SPINAL DEFORMITY IN EHLERS-DANLOS SYNDROME

## FIVE PATIENTS TREATED BY SPINAL FUSION

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**Five patients with classical Ehlers-Danlos syndrome developed severe spinal deformities. Two were shown to have type-VI collagen abnormalities. Three had a double structural scoliosis of the thoracic and lumbar regions, one had a single thoracic scoliosis and one had a thoracic kyphosis.**

**The curves first developed before the age of four years, and were not controlled by bracing. Major corrective surgery with posterior fusion was performed at a mean age of 11 years 8 months. Excessive blood loss could be controlled and although wound haematoma and dehiscence were common, they did not provide major problems. The spinal fusions healed satisfactorily.**

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Ehlers-Danlos syndrome (EDS) is a rare hereditary disorder of collagen metabolism which has recently been classified into subgroups on the basis of the underlying defect in collagen fibril formation. The characteristic clinical features of all types of EDS are hypermobility of joints, which may sublux or dislocate, hyperextensibility of the skin and general friability of the soft tissues which are easily bruised and heal poorly with 'tissue-paper' scars.

Spinal deformity may occur in EDS, but very little is known about its type and incidence (Beighton and Horan 1969). Leatherman and Dickson (1988) stated that patients with EDS are more likely to develop spinal deformities, but that there was little evidence to suggest that these could become severe; surgical treatment should not be advised.

This paper describes the patterns of spinal curvature in five patients with EDS and reports the results of surgical treatment.

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## PATIENTS

From 1976 to 1991, five patients with classical EDS presented with spinal deformities to the Scoliosis Clinic at the Princess Margaret Rose Orthopaedic Hospital, Edinburgh. All were girls with hypermobile joints, severe ligamentous laxity, and hyperextensible skin. They all had a tendency to prolonged bleeding and evidence of poor soft-tissue healing with tissue-paper scarring.

The syndrome had been diagnosed between the ages of one and nine years as a result of recurrent subluxation or dislocation of shoulders, hips, sternoclavicular or patellofemoral joints. Since tissue typing became available the last two patients (cases 4 and 5) have been diagnosed as having type-VI collagen disorders. They were sisters, but there was no relevant family history in the other three patients.

Spinal deformity had first been recognised before the age of 4 years. Three patients were treated in braces, but the curves deteriorated and all five patients required major corrective spinal surgery at a mean age of 11 years 9 months (10 years 5 months to 13 years 7 months). All had posterior spinal fusion performed by the author, two with Harrington instrumentation and three with Luque L rods.

**Case 1.** A girl presented shortly after birth with Erb's palsy, hyperextensible skin, general ligamentous laxity and dislocatable hips and shoulders. A spinal deformity was noted, but was not treated until the age of 2 years 6 months, when she had an 89° smooth thoracic kyphosis extending from T3 to L3 with its apex at T9. A Milwaukee brace was worn until the age of 10 years 4 months when the kyphosis measured 45°. The patient then refused to wear the brace and the kyphosis deteriorated rapidly to 95° at the age of 13 years 7 months. She also developed poor respiratory function with a vital capacity of less than 50%.

A posterior spinal fusion was performed with Luque L-rod instrumentation extending from T1 to L2 and correcting the kyphosis to 41°. Postoperatively, an underarm plaster jacket was used for three months, then removed because of a pressure sore in the fragile skin over her kyphosis. At the age of 14 years 8 months she became increasingly dyspnoeic, her general condition deteriorated and she died of cardiorespiratory failure. Post-mortem examination revealed an empyema of the



Four of the five developed a significant scoliosis and

fusion and correction with instrumentation is possible.

right lung and infarction of the left lung. The spinal fusion had healed satisfactorily.

**Case 2.** A girl known to have a scoliosis at the age of three years, but untreated, was diagnosed in Edinburgh at the age of nine years as a case of EDS with a major left thoracolumbar scoliosis from T11 to L4 measuring 97°, and a minor right thoracic scoliosis from T3 to T11 measuring 66°. The vertebrae at the apex of the thoracolumbar curve were markedly rotated resulting in an

right thoracic curve of 68° extended from T4 to T11 and a left lumbar curve from T11 to L4 measured 50°. There was an apparent kyphosis at the junction of the curves.

Posterior spinal fusion using Luque L-rod instrumentation from T3 to L4 corrected the deformity; autografts and allografts were used as before. The thoracic scoliosis was corrected to 22°, the lumbar scoliosis to 18° and the sagittal profile to normal. An underarm jacket for three months allowed solid fusion with thoracic and lumbar



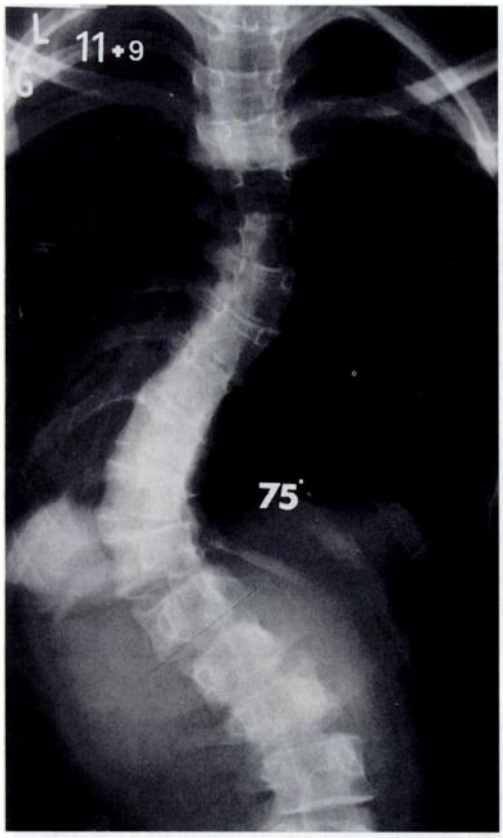



Fig. 6

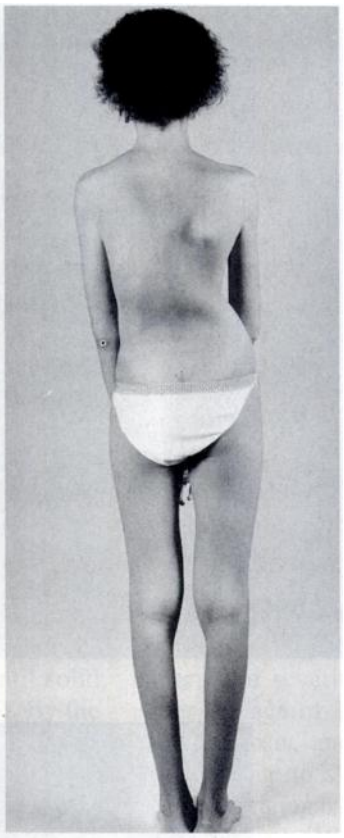


Fig. 7

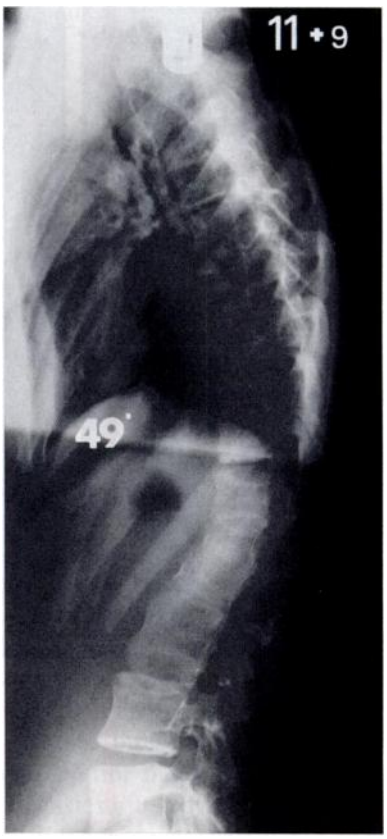
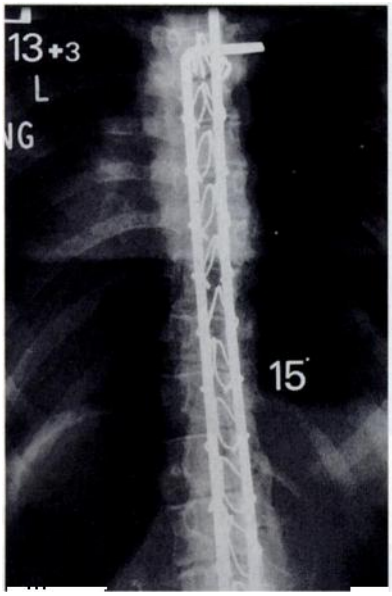
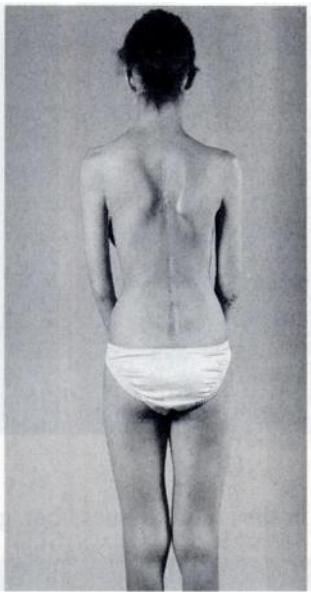
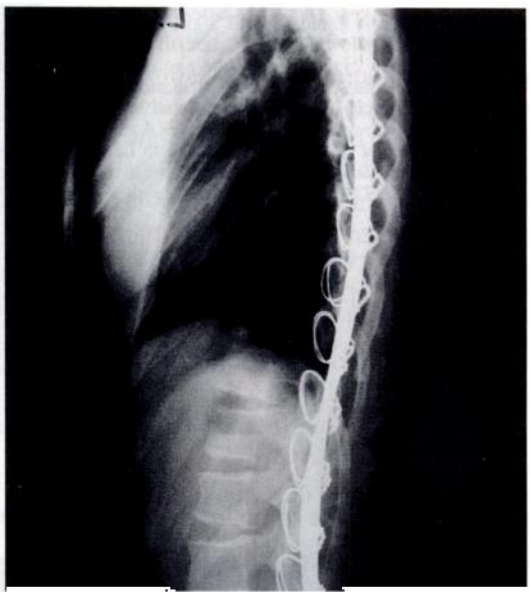



Fig. 8





matomas were controlled by repeated aspiration. Wound dehiscence was also common but was usually superficial and healed satisfactorily after secondary suture. The bone tended to be slightly osteoporotic, but there was no apparent delay or failure in bone healing. Both allografts and iliac-crest autografts were used; all patients developed a solid and stable fusion.

Severe spinal deformities may develop relatively early in patients with EDS. Major corrective spinal surgery may be needed and is possible with careful management of bleeding and soft-tissue problems. Spinal fusions heal satisfactorily and provide stable correction.

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