Recurrent instability of the elbow in the Ehlers-Danlos syndrome

A REPORT OF THREE CASES AND A NEW TECHNIQUE OF SURGICAL STABILISATION

A. S. Cole, G. A. Hill, M. Abela, A. J. Carr

From the Nuffield Orthopaedic Centre, Oxford, England

We present three cases of recurrent instability of the elbow in association with the Ehlers-Danlos syndrome. The pattern of instability has not previously been reported. We describe our procedure for achieving stability using a bone graft to the olecranon fossa which gave a functional range of movement.

J Bone Joint Surg [Br] 2000;82-B:702-4.

Received 4 June 1999; Accepted after revision 22 December 1999

The Ehlers-Danlos syndrome (EDS), first described by Ehlers in 1899 and Danlos in 1908, is a heterogeneous disorder of connective tissue of which at least ten subtypes are recognised. It is the most common inheritable disorder of connective tissue¹ and has a variety of phenotypic presentations. In most forms transmission is autosomal dominant. The phenotype does not always fit exactly into one of the described subtypes and the features are age-dependent. A revised classification has recently been proposed² in which there are six major types as follows: classic (types I and II), hypermobility (type III), vascular (type IV), kyphoscoliosis (type VIA), arthrochalasia (types VIIA and B) and dermatopraxis (type VIIC).

Recent evidence has suggested that type-V collagen is involved in human EDS type I. The COL5A1 mutation³ results in substitution of the most 5' cysteine residue by serine within a highly conserved sequence of the pro (alpha)1(V) C-propeptide domain and causes reduction in collagen V by preventing incorporation of the mutant chains in the collagen V trimers. The resultant abnormal collagen is associated with impaired structure and function

of skin, joints, eyes and blood vessels and predisposes to dislocation of the joints. There are a number of orthopaedic manifestations including hypermobility, club foot, tendon ruptures, scoliosis and dysplasia of the hip. A review of the literature, however, has shown only one report of recurrent instability of the elbow in this condition.⁴

Patients and Methods

Case 1. A 38-year-old woman with type-I EDS (classic) presented with multiple episodes of ulnohumeral subluxation of the left, non-dominant elbow after a traumatic dislocation eight years previously. There was joint hypermobility and skin laxity, fragility and bruising. The subluxations occurred in extension (Fig. 1) and interfered with daily life. Examination revealed hyperextension of -30° with positive apprehension at 20° from full extension. She underwent stabilisation with a bone block to limit extension. One year after operation she had a range of movement from 35° to full flexion with no instability.

Cases 2 and 3. A 34-year-old woman with type-I EDS (classic) presented with recurrent dislocation of the left elbow of spontaneous onset. She had multiple involvement of joints which had required previous arthrodesis of both

Fig. 1

Radiograph showing hyperextension of the elbow with anterior subluxation.

A. S. Cole, BSc, FRCS (Tr & Orth), Specialist Registrar in Trauma and Orthopaedics

G. A. Hill, FRCS (Tr & Orth), Specialist Registrar in Trauma and Orthopaedics

M. Abela, MD, FRCS, Specialist Registrar in Trauma and Orthopaedics

A. J. Carr, ChM, FRCS, Consultant Orthopaedic Surgeon
Nuffield Orthopaedic Centre, Windmill Road, Headington, Oxford OX3 7LD, UK.

Correspondence should be sent to Mr A. S. Cole at 25 Windmill Heights, North Leigh, Near Witney, Oxon OX8 6ZD, UK.

©2000 British Editorial Society of Bone and Joint Surgery
0301-620X/00/510248 \$2.00

Fig. 2a

Fig. 2b

AP (a) and lateral (b) radiographs of the elbow showing a bone graft to the coronoid fossa to limit full extension and to prevent anterior subluxation.

shoulders and there was associated skin laxity and fragility. The degree of hyperextension of the elbow had gradually increased during the previous five years. There was subluxation of the ulnohumeral joint at full extension and hyperextension of 30°. Stabilisation was performed with a bone block. Six months later she developed similar symptoms on the right with symptomatic instability in extension. A similar procedure was undertaken on this elbow. At 18 months and one year after these operations she had loss of extension of 40° with a full range of flexion and no instability.

Operative technique. The patient is placed in the lateral position. Under tourniquet control the distal humerus is exposed through a triceps-splitting approach. A block of corticocancellous graft from the iliac crest is placed in the olecranon fossa and held by two AO cortical screws (Fig. 2). The graft is shaped to allow adjustment of its size and thus the limitation of extension induced. After operation the elbow is placed in a thermoplastic splint for one month and a hinged cast brace for six weeks.

Discussion

Recent clinical and genetic studies have defined the range of variation and molecular defects in the various subtypes of EDS. Type I is the most common and severe form and is an inherited disorder of type-V collagen.

Recurrent dislocation or subluxation of the elbow is rare. The aetiology is poorly understood and several surgical procedures to achieve stability have been described, which include the insertion of a bone block in the coronoid fossa

to limit flexion,⁵ repair of the capsule and insertion of brachialis,⁶ the formation of an intra-articular sling using biceps and triceps⁷ and transfer of the biceps tendon to the coronoid process.⁸ Osborne and Cotteril⁹ have described the pathological findings in 18 cases of recurrent instability and have advised posterolateral capsulorrhaphy.

Most cases reported are post-traumatic, in patients with normal connective tissue. Both of our patients presented with multiple episodes of instability, which occurred in hyperextension and interfered with daily activities. We feel that the stabilisation in these patients requires a different approach because of the underlying abnormalities of collagen and the likelihood of recurrence after soft-tissue procedures.

There is only one reported case of recurrent instability of the elbow in Ehlers-Danlos type I.⁴ This occurred in a skeletally immature patient and subsequent repair by the method of Osborne and Cotteril⁹ resulted in a stable joint 24 months after operation. Instability of the elbow with subluxation in hyperextension has not been previously described in EDS and differs from post-traumatic instability which is due to posterolateral capsular laxity.⁹ Reports of soft-tissue procedures and osteotomies to treat instability of other joints in EDS have been unsatisfactory.^{10,11} Arthrodesis is not a realistic option in these patients.

In all three cases the instability was abolished with a bone graft to the olecranon fossa. This causes loss of full extension while maintaining a functional range of movement. The loss of extension was less of a handicap than the recurrent subluxation and both patients were happy with the

outcome. The follow-up of these cases is short but there is no evidence of degenerative changes within the elbow at the latest review.

No benefits in any form have been received or will be received from a commercial party related directly or indirectly to the subject of this article.

References

1. **Beighton P, Horan F.** Orthopaedic aspects of the Ehlers-Danlos syndrome. *J Bone Joint Surg [Br]* 1969;51-B:444-53.
2. **Beighton P, DePaepe A, Steinmann B, Tsipouras P, Wenstrup RJ.** Ehler-Danlos syndromes: revised nosology, Villefranche 1997: Ehlers-Danlos National Foundation (USA) and Ehlers-Danlos Support Group (UK). *Am J Med Genet* 1998;77:31-7.
3. **DePaepe A, Nuytinck L, Hausser I, Anton-Lamprecht I, Naeyaert JM.** Mutations in the COL5A1 gene are causal in Ehlers-Danlos syndromes I and II. *Am J Hum Genet* 1997;60:547-54.
4. **Rames RD, Strecker WB.** Recurrent elbow dislocations in a patient with Ehlers-Danlos syndrome. *Orthopaedics* 1991;14:707-9.
5. **Milch H.** Bilateral recurrent dislocation of the ulna at the elbow. *J Bone Joint Surg* 1936;18:777-80.
6. **Dryer RF, Buckwalter JA, Sprague BL.** Treatment of chronic elbow instability. *Clin Orthop* 1980;148:254-6.
7. **Kapel O.** Operation for habitual dislocation of the elbow. *J Bone Joint Surg [Am]* 1951;33-A:708-10.
8. **Reichenheim PP.** Transplantation of the biceps tendon as a treatment of recurrent dislocation of the elbow. *Brit J Surg* 1947;35:201-4.
9. **Osborne G, Cotteril P.** Recurrent dislocation of the elbow. *J Bone Joint Surg [Br]* 1966;48-B:340-6.
10. **Jerosch J, Castro WH.** Shoulder instability in Ehlers-Danlos syndrome: an indication for surgical treatment? *Acta Orthop Belg* 1990;56:451-3.
11. **Giunta C, Superti-Furga A, Spranger S, Cole WG, Steinmann B.** Ehlers-Danlos syndrome type VII: clinical features and molecular defects. *J Bone Joint Surg [Am]* 1999;81-A:225-38.