



Familial pseudarthrosis of the clavicle : does it need treatment ?

Edmund O'LEARY, Sherief ELSAYED, Ashok MUKHERJEE, Keith TAYTON

From the Royal Gwent Hospital, Newport, Gwent, U.K.

The eighth family with multiple cases of congenital pseudarthrosis of the clavicle is described. This usually presents as a clavicular lump and is distinct from more common conditions such as birth fractures, craniocleidal dysostosis and neurofibromatosis. There has so far been no clear indication on whether familial pseudarthrosis of the clavicle should be treated operatively. We recommend that when a suspected case of congenital pseudarthrosis of the clavicle is diagnosed the parents and siblings be examined also. If other family members are affected, we advise that the treatment should be conservative.

Keywords : pseudarthrosis ; clavicle ; familial ; treatment.

INTRODUCTION

Congenital pseudarthrosis of the clavicle (CPC) is a rare condition that was first described in association with craniocleidal dysostosis (CCD). It was later distinguished as a distinct entity from birth fractures, CCD and neurofibromatosis (4). It usually presents as a lump over the clavicle and may be asymptomatic. Over 100 cases have been reported in the literature and it predominantly affects the right side. In approximately ten percent of cases there is bilateral involvement. Left-sided lesions are even rarer (7). Although the majority of cases of CPC are unilateral and have no family history, seven families have been reported who have more than one member affected (1,2,3,6,7,8). In these cases, the mode of inheritance appears to be

autosomal dominant. There are no clear guidelines in the literature of how familial pseudarthrosis of the clavicle should be treated. When such cases were treated surgically a significant proportion continued to experience symptoms (2,3,7,8).

CASE 1

A one-month-old boy was referred to hospital for assessment of a bony lump over his right clavicle. His mother was concerned about this, but it was not symptomatic. There was no history of any birth trauma. On examination, there was a prominent, painless lump over his right clavicle, at the junction between the outer third and middle section. Other than for widened skull suture lines appropriate for his age, he had no other features of CCD or

■ Edmund O'Leary, Registrar.

Department of Trauma and Orthopaedics, Mona Vale Hospital, Sydney, New South Wales, Australia.

■ Sherief Elsayed, Specialist Registrar.

Department of Trauma and Orthopaedics, Queen's Medical Centre, Nottingham, U.K.

■ Ashok Mukherjee, Associate Specialist.

■ Mr Keith Tayton, Consultant.

Department of Trauma and Orthopaedics, Royal Gwent Hospital, Newport, Gwent, U.K.

Correspondence : Mr Edmund O'Leary, Unit 7, 1 Waruda Street, Kirribilli, New South Wales 2061, Australia.

E-mail : edmundoleary@hotmail.com

© 2008, Acta Orthopædica Belgica.



Fig. 1. — Anterior radiograph of case 2 showing bilateral pseudarthrosis of the clavicles.

neurofibromatosis. Radiographs of his clavicles showed a right sided pseudarthrosis of the clavicle.

CASE 2

After further enquiry, the above described child's mother, aged 18, said that she had experienced aching in her shoulders after exercise. She gave no history of birth or any other trauma. Examination revealed no lump and she had no clinical signs of CCD or neurofibromatosis. Radiographs of her clavicles revealed bilateral pseudarthrosis of the clavicle (fig 1).

CASE 3

Further questioning revealed that the boy's maternal aunt who was aged 22 also had problems with her shoulder. She had been seen three times in the previous 11 years with right sided shoulder pain after minor injuries. Radiographs each time revealed a "fracture" of the right clavicle. On the first two occasions she had been treated conservatively and thought to have achieved a fibrous union. After the third presentation she was referred to our department and was offered surgery to correct this, but she declined. She had had a normal birth and was re-examined by the main author and found to have no lump. Again, there were no clinical signs of CCD or neurofibromatosis. A new radiograph, as well as a review of her previous films, showed a right sided pseudarthrosis of the clavicle. There was no evidence of callus formation, but there was a rounded appearance to the ends of the two fragments indicating that this was a congenital pseudarthrosis and not an unhealed fracture.

OTHER CLOSE RELATIVES

Three out of four of the boy's maternal great grandparents, as well as his maternal grandfather (aged 45) and grandmother (aged 46), half-aunt (aged 14) and cousin (aged 2) were all examined and radiographs of their clavicles taken. Again, there was no evidence of CCD, neurofibromatosis or pseudarthrosis of the clavicle. The other maternal great grandfather died thirty years before in a road traffic accident, but had not had any problems with his shoulders. No surgical treatment was offered to the patients as they had no functional loss and were not concerned about the cosmetic appearance of the lumps. The father's lineage was not traced. A family pedigree is shown in figure 2.

DISCUSSION

The aetiology of congenital pseudarthrosis of the clavicle remains obscure. Embryologically the clavicle is probably formed by the fusion of two ossification centres (4). It was proposed that pseudarthrosis developed from a failure of these two ossification centres to fuse (2). This theory was supported by work demonstrating cartilaginous caps adding new bone on the proximal and distal bony ends of the pseudarthrosis, which is thought to be the equivalent to that of developing physes (4). Unfortunately this theory is not supported by Gibson, whose work on five embryological clavicles supported a view that in the early stages of ossification there was a single ossification centre in the middle of the bone (3).

An explanation for the lesion and its marked right sided predominance has been put forward by Lloyd-Roberts *et al* (5). They noted that during development, the right subclavian artery and the right clavicle are more closely related than those on the left. They suggested that, as a consequence, anything affecting this difference would have a greater effect on the right side, and therefore the right clavicle. Such variations include elevation of the first ribs, or the presence of cervical ribs. The cases illustrated here do not show either of these features. Lloyd Roberts' theory explains why left sided lesions are seen in patients with dextrocardia,

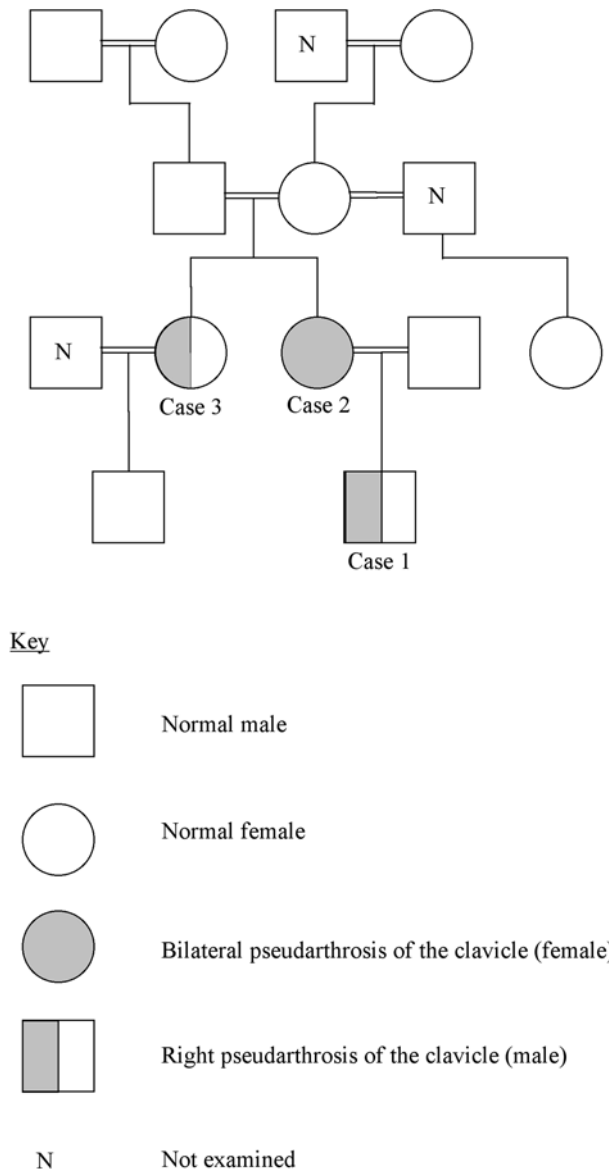


Fig. 2. — Family tree

where the positions of the great vessels are reversed (3,5). Unfortunately, despite this attractive theory, most unilateral cases of congenital pseudarthrosis of the clavicle have no family history and no other bony abnormality, an observation which supports the view that there are other environmental factors involved.

When present, the differential diagnosis of CPC is between a birth fracture, (in which there is normally prompt healing and copious callus formation), neurofibromatosis, (where there is tapering of the bone ends) and craniocleidal dysostosis. CCD is an autosomal dominant condition of proportionate dwarfism affecting bones formed intramembranously. The most common clavicular defect is loss of the central portion with rudimentary medial and lateral portions remaining. It is associated with other abnormalities, including a large skull, small facial bones and crowded teeth, wide fontanelles and suture lines and minor abnormalities in the feet, hands, pelvis and spine.

This is the eighth reported family with CPC. The consensus in the literature is that the genetic transmission of this condition is via an autosomal dominant gene. As far as can be ascertained in this family, none of the four maternal great grandparents exhibited any clavicular problems and neither did the two maternal grandparents. With both mother and maternal aunt sharing the lesion, then the only explanation would be a spontaneous mutation in one of the maternal grandparents, whose phenotype is in the form of a mosaic and the lesion did not exhibit. However, assuming that inter-family relationships are as stated, then there is little doubt that this mode of inheritance is almost certainly via an autosomal dominant gene.

In the literature a significant proportion of the cases of familial pseudarthrosis of the clavicle that were treated operatively continued to be symptomatic (2,3,7,8). In our family the mother and aunt were not aware that they had a problem with their clavicles. It is therefore recommended that when a suspected case of CPC is diagnosed the parents and siblings be examined also. If other family members are affected, we advise that the treatment should be conservative.

REFERENCES

1. **Alberink A.** Congenital pseudarthrosis of the clavicle. *Diagn Imaging* 1980 ; 49 : 94-97.
2. **Aldred A.J.** Congenital pseudarthrosis of the clavicle. *J Bone Joint Surg* 1963 ; 45-B : 312-319.
3. **Gibson DA, Carroll N.** Congenital pseudoarthrosis of the clavicle. *J Bone Joint Surg* 1970 ; 52-B : 644-652.

4. **Gomez-Brouchet A, Sales de Gauzy J, Accadbled F et al.** Congenital pseudarthrosis of the clavicle : a histopathological study in five patients. *J Pediatr Orthop* 2004 ; 13-B : 399-401.
5. **Lloyd Roberts GC, Apley AG, Owen R.** Reflections upon the aetiology of congenital pseudarthrosis of the clavicle. *J Bone Joint Surg* 1975 ; 57-B : 24-29.
6. **Owen R.** Congenital pseudarthrosis of the clavicle. *J Bone Joint Surg* 1970 ; 52-B : 644-652.
7. **Price BD, Price CT.** Familial congenital pseudarthrosis of the clavicle : case report and literature review. *Iowa Orthop J* 1996 ; 16 : 153-156.
8. **Toledo LC, MacEwen GD.** Severe complications of surgical treatment of congenital pseudarthrosis of the clavicle. *Clin Orthop* 1979 ; 139 : 64-67.