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ORIGINAL ARTICLE

Congenital pseudarthrosis of the clavicle: a report on 27 cases

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Hypothesis: We aimed to report our experience in treating congenital pseudarthrosis of the clavicle (CPC).
Materials and methods: A retrospective search of the archive of our institute was performed; 27 cases (12 male and 15 female patients) affected by CPC were recorded. Among these patients, 19 underwent surgical intervention for cosmetic appearance between 1960 and 2015. Of 19 patients, 18 were treated by pseudarthrosis resection and stabilization with a Kirschner wire, whereas in 1 case, the osteosynthesis was performed with a plate. Iliac crest bone autograft was used in 15 patients, whereas 4 patients were treated with a fibular allograft.
Results: The mean follow-up period was 36.3 ± 49.1 months. Bone healing was achieved in 14 of 19 operated cases (74%); none of the patients had complaints regarding cosmetic abnormalities or unesthetic appearance. All the operated patients were pain free, range of motion was complete, and no other subjective anomalies were found. No vascular or neurologic complications were observed. However, the use of allograft was associated with high rates of nonunion in this case series ($P = .037$).
Conclusion: CPC can be satisfactorily treated by K-wire fixation and autologous iliac crest bone grafting, which showed better results in terms of functional and cosmetic outcome.
Level of evidence: Level IV; Case Series; Treatment Study

Keywords: Congenital pseudarthrosis of clavicle; surgical procedures; bone graft; shoulder girdle; congenital; pseudarthrosis; clavicle

First described by Fitzwilliams[7] in 1910, congenital pseudarthrosis of the clavicle (CPC) is a rare condition with about 200 cases described in the literature.[4] Female patients seem to be more frequently affected.[18] The lesion is usually unilateral, and it is localized in the middle part of the clavicle. The right side is the most commonly affected; when found on the left side, it is usually associated with dextrocardia. Bilateral involvement occurs in approximately 10% of cases. Often, cervical ribs or abnormally elevated first ribs are present

This study has been approved by the Ethical Committee of the Rizzoli Orthopedic Institute (study protocol 0006176, Bologna 2/20/2012), and the material in this report has been acquired according to modern ethical standards.

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along with the abnormal clavicles.[15,18] Familial cases, CPC in combination with other abnormalities such as cleidocranial dysplasia or neurofibromatosis, and possible involvement of the *RUNX2* (*CBFA1*) gene mutation have been recorded in the literature.[14,15,18]

Although the etiology remains unknown, the pathogenesis is probably related to the embryology of the clavicle.[3,17,20] The clavicle is the first fetal bone to undergo primary ossification. Several studies have shown that the clavicle appears during the fourth week of gestation as a mesenchymal bar that lies under the precoracoid area.[3,17,20] Starting from the seventh week, 2 centers of primary ossification start to fuse to form the clavicle.[17] According to Lloyd-Roberts et al,[12] the failed union of these 2 centers is determined by pressure exerted by the subclavian artery that runs posteriorly and inferiorly to the midportion of the clavicle. This theory seems to be supported by the fact that the few left-sided cases of pseudarthrosis are associated with dextrocardia or situs inversus viscerum.[12]

The deformity is rarely identified at birth and is more frequently identified during the first few years of life. It usually looks like a subcutaneous swelling over the middle third of the clavicle and tends to increase in size as the child grows up. Range of motion of the shoulder is usually normal and not painful.[17]

The most common clinical presentation of this condition is a patient who remains asymptomatic during his or her entire life. A complication that might occur in adulthood is thoracic outlet obstruction, but it is a rare sequela.[6] In this view, treatment choice is controversial. Surgical treatment consists of removing the pseudarthrosis tissue, bone grafting, alignment of the fragments, and stabilization with a plate or pins. Some authors also describe the use of an external fixator that yields good cosmetic results and avoids the need for further surgical procedures.[3] This paper is a retrospective review of 27 cases of CPC treated at our institution.

Materials and methods

Case series

A retrospective search of the archive of our institute was performed. The study included 27 consecutive patients (12 male and 15 female patients) who were followed up in our department between 1960 and 2015. Cases associated with cleidocranial dysplasia, neurofibromatosis, and trauma were excluded. We did not observe any case of familial CPC. One patient had a chromosomal deletion of the second and seventh chromosomes. In 3 cases, CPC was associated with a hypertrophic transverse process of the seventh cervical vertebra. Apart from 1 patient who presented to us for assessment at the age of 47 years, the mean age at first clinical examination was 7.0 ± 3.2 years (range, 1-13 years), but in most of the cases, the parents noticed the defect at birth or in early childhood.

In all patients, the clinical manifestation was just a firm and usually visible painless bulge in the midportion of the clavicle. Shoulder and upper limb function was normal and painless in all but 3 patients. No skin, vascular, or nervous problems were noticed.

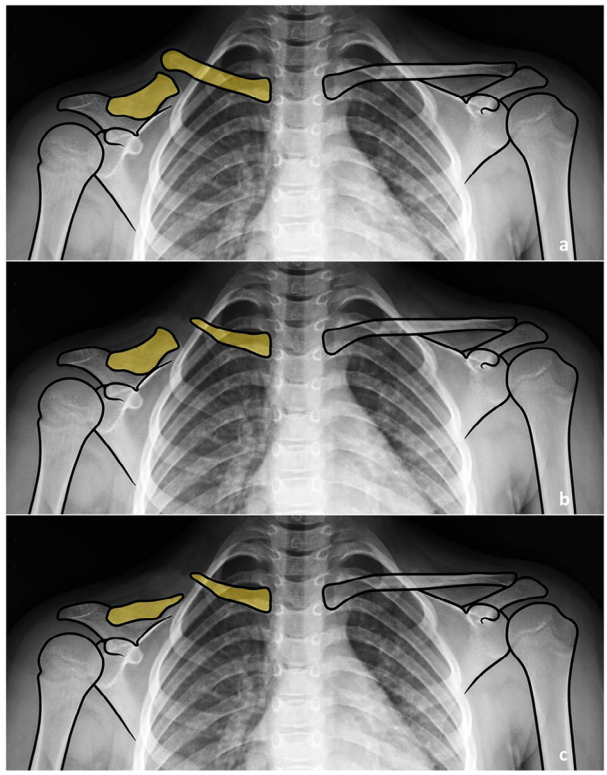

Figure 1 Main radiographic pattern of congenital pseudarthrosis of the clavicle, based on shape of clavicle segments: rounded–elephant foot (**a**), tapered–elephant foot (**b**), and tapered-tapered (**c**).

We evaluated the radiographic characteristics of every case; shape, position, and distance between the 2 clavicular segments were measured and recorded. The radiographs showed a typical pattern consisting of a pseudarthrosis of the midportion of the clavicle with lack of callus formation causing an interfragmentary gap. The distance between the segments ranged from 2 to 28 mm, with a mean gap of 9.5 ± 5.6 mm, which represented 9% ± 4% of the entire length of the clavicle.

In 10 cases, the clavicular segments were horizontal; in 11 cases, they pointed sharply upward to an abnormal high position; and in 6 cases, the medial segment pointed upward while the acromial segment was horizontal. The end shape of the clavicular segments was rounded, tapered, or enlarged with an elephant-foot appearance (Fig. 1).

Nineteen cases underwent surgery for cosmetic issues. The mean age at surgery was 8.1 ± 2.7 years (range, 4-13 years). The remaining 8 patients refused surgical treatment because the defect was slight and did not significantly compromise the esthetic appearance.

Surgical technique

The surgical technique was previously described by one of the authors,[2] was similar for the majority of patients, and was maintained throughout the years. A longitudinal skin incision was made over the palpable prominence, and the periosteum was longitudinally incised over both bony ends of bone. The fibrous tissue, bridging the sclerotic clavicular ends, was sharply dissected. The medullary canal was opened with a sharp curette, and an intramedullary Kirschner wire was inserted to align the 2 ends of the clavicle under