Pseudartrose Congénita da Clavícula

Congenital Pseudoarthrosis of the Clavicle

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Resumo

Introdução: A pseudartrose congénita da clavícula apresenta-se como uma tumefação congénita indolor da clavícula, normalmente direita, afetando predominantemente o sexo feminino. Esta deformidade persiste ao longo da vida, não estando por norma associada a défice funcional. Descrição de um caso de pseudartrose congénita da clavícula com 12 anos de seguimento e revisão da literatura.

Caso Clínico: Recém-nascido de termo, com gravidez e parto eutóico não complicados, apresentava, à observação, tumefação óssea (1x1 cm), no 1/3 médio da clavícula direita, sem limitação funcional do membro superior ou outras alterações. A imagem radiográfica mostrava um defeito ósseo no 1/3 médio da clavícula direita, com topos arredondados, lisos e regulares, sem evidência de calo ósseo. Foi estabelecido o diagnóstico de pseudartrose congénita da clavícula.

Durante 12 anos de seguimento em consulta externa manteve-se assintomático, sem limitação funcional e desenvolvimento estaturo-ponderal normal, e nas radiografias realizadas mantinha as características descritas do defeito ósseo. Aos 4 anos, por motivos estéticos, foi proposta correção cirúrgica, que os pais recusaram.

Conclusão: Sendo rara, a pseudartrose congénita da clavícula pode ser diagnosticada por uma história clínica adequada, exame objetivo e achados radiográficos típicos. É essencial excluir diagnósticos diferenciais, como fratura ou doenças ósseas raras, e tranquilizar os pais quanto à benignidade desta condição.

Palavras-chave: Clavícula/anomalias congénitas; Criança; Pseudartrose/congénita; Pseudartrose/diagnóstico; Pseudartrose/reabilitação.

Abstract

Introduction: Congenital pseudarthrosis of the clavicle presents as a congenital painless swelling over the mid-third of the clavicle, mostly on the right side and females, that persists over lifetime with no functional limitations. We present a 12 years follow-up case of congenital pseudarthrosis of the clavicle and literature review.

Case Report: A full-term newborn boy, with uncomplicated pregnancy and vaginal delivery, presented with a firm bony protuberance (1x1 cm), in the mid-third of the right clavicle without functional limitations and no other physical examination abnormality. The X-ray identified a bony defect in the mid-third of the right clavicle with smooth, regular, intact/sclerotic cortex and without any evidence of callus formation. The diagnosis of congenital pseudarthrosis of the clavicle was performed.

Over the twelve follow-up years, he kept asymptomatic, without functional limitations and normal physical and skeletal development. Over time, the X-rays showed the same bone defect with the same characteristics. At fourth years old, surgery was proposed for aesthetic reasons but parents refused.
Conclusion: Although rare, congenital pseudarthrosis of the clavicle can be easily diagnosed through characteristic physical examination findings and radiographic hallmarks. It is essential to exclude other differential diagnosis, like clavicular fracture or rare bone diseases, and be able to reassure parents, explaining the benign nature of this condition.

Keywords: Child; Clavicle/abnormalities; Pseudarthrosis/congenital; Pseudarthrosis/diagnosis; Pseudarthrosis/rehabilitation.

Introduction

Congenital pseudarthrosis of the clavicle (CPC) is a rare entity defined as a discontinuity or cleft in the mid-third of the clavicle.1-3 Its true incidence is not well reported in literature, it is more frequent in females and usually involves the right side, with bilateral involvement in 10% of cases.4 It usually presents at birth or early childhood with a painless swelling over the right clavicle mid-third, which can be confused with a traumatic clavicle fracture. It persists over lifetime most often with no functional limitation.5,6 Regarding treatment, in many cases, the natural history is benign and no treatment is necessary.6 Surgical treatment is advocated if pain, functional limitation or thoracic outlet syndrome develops; nowadays aesthetic is also an important surgical indication.7,8

In this work we describe a clinical case of a newborn diagnosed with CPC at birth with a clinical follow-up of twelve years. With this case we pretend to illustrate the benign nature of the bone defect, its clinical and imagological hallmarks, differential diagnosis and management options.

Case Report

A newborn boy, born at term, with a birth weight of 3.805 kg, to a white, married (nonconsanguineous) 31-year-old G1 parents, who had an uncomplicated pregnancy. No family history of congenital or acquired musculoskeletal disorders registered.

The birth was an uncomplicated vaginal delivery, apgar scores were 9 and 9. Although he moved his arms freely since birth, a bulge over his right mid-clavicle made the suspicious of clavicular fracture during delivery. For this reason, he presented at pediatric orthopedics department where the examination revealed a painless, non-tender, 1x1 cm firm bony mass protuberance over the mid-third of the right clavicle. No functional limitation of the right upper arm was noted and no other physical examination alteration was observed. The X-ray identified a bony defect in the mid-third of the right clavicle, with the proximal fragment over the distal one and without any evidence of callus formation. Each bone segment ended in a bulbous, smooth and regular mass. The diagnosis of CPC was performed.

For precaution and parents’ reassurance, the boy was kept under surveillance for 12 years. He kept asymptomatic and physical and skeletal development was normal, as was right upper limb function, with no range of motion or strength limitation. There was no growth delay or abnormality registered and only a discrete scoliosis was observed with no significance or necessary treatment. The X-ray image was repeated from time to time, and the same bone defect was always present with the same appearance and characteristics previously described and no evidence of callus formation.

Figure 1 - Zero month. The X-ray identified a bony defect in the mid-third of the right clavicle, with the proximal fragment over the distal one and without any evidence of callus formation. Each bone segment ended in a bulbous, smooth and regular mass.
Figure 2 - Twelve years. The X-ray image was repeated from time to time, and the same bone defect was always present with the same appearance and characteristics previously described and no evidence of callus formation.

Figure 3 - Combined movements of shoulder at 12-years old.

At 4-years old, surgical correction of the defect was proposed for aesthetic reasons but parents refused.
Discussion

Congenital pseudarthrosis of the clavicle (CPC) is a rare clinical defect defined as a discontinuity or cleft in the mid-third of the clavicle.1-3 It is true incidence is not well reported in the literature with, in a study from 2000, just 200 cases reported.6 It is more frequent in females, unilateral and usually involves the right side, with 10% of bilateral involvement.4 Involvement of the left clavicle is usually associated with dextrocardia and cervical ribs or situs inversus.6

The pathophysiology is not entirely understood but is probably related to clavicle’s embryology. The clavicle is an example of a flat bone, it forms in utero by intramembranous ossification. It is the first bone in the body to ossify and the last one to fuse and mature.5 Some authors believe that the pseudarthrosis represents an embryologic failure of the two ossification centers of the clavicle to appropriately fuse.10,11 This two ossifications centers are connected by a fibrous bridge that is contiguous with the periosteum and surrounded by a synovial membrane; failure of ossification of this bridge may lead to pseudarthrosis.10,11

Recently, the more common belief is that pulsatile pressure from the subclavian artery on the developing clavicle is responsible for the condition. This theory also explains the right sided dominance of this pathology as the subclavian artery is normally in closer relation with the clavicle because of its higher position compared with the left side. The fact that when it’s presents on the left side it is associated with dextrocardia or situs inversus reinforces this thesis.9-11

A family history of the disorder has also been documented but no genetic data are available so far.12

Most frequently CPC presents at birth or early childhood with a painless swelling over the mid-third of the right clavicle with no functional limitation. The protrusion usually becomes more prominent with age since it is associated with shortening and “drooping” of the scapular belt.6 Later in childhood, there may be pain and functional impairment, ranging from fatigue or discomfort with strenuous activity to more severe disability.6

Although CPC is a condition that has been described in the literature, the relative infrequency of its occurrence makes it a diagnostic challenge. Since it occurs in newborns and is often presented at a young age, it could be confused with traumatic clavicle fracture.13,14

When history and physical examination are consistent, plain films are usually sufficient to make a diagnosis.6 On X-ray, there are two distinct portions of the clavicle that are separated with a gap wider than in a fracture, each having a smooth and intact/sclerotic cortex, suggesting it’s chronic nature. In contrast to a healing fracture, there will be no radiographic evidence of callus formation. Most commonly, the pseudarthrosis occurs at the junction of the middle and distal third of the clavicle. The medial portion of the clavicle is typically cephalad compared to the lateral. If there are concerns regarding an acute fracture, repeated radiographs after 10–14 days will confirm ongoing absence of callus reaction.6,13,14

Differential diagnosis includes clavicle fracture resulting from difficult vaginal birth (neonate) or trauma (infant or toddler) and this may prove to be difficult to differentiate at initial presentation, because fracture callus may have not yet formed.14 In this case, correlation with history and physical examination is critical. Pain with palpation suggests a fracture and with time, abundant callus formation on plain radiographs will confirm this diagnosis.

Other differential diagnosis is cleidocranial dysostosis, a familiar autosomal dominant condition that results in defects of membranous bone ossification with bilateral involvement.14 It includes multiple skeletal disorders affecting both the axial and appendicular skeleton, hypoplasia of the maxilla, persisting skull sutures, supernumerary teeth, frontal bossing, delayed bone ossification, scoliosis, coxa vara and hypoplasia of both clavicles. Patients will have hypermobility of the shoulder joint, bilateral involvement and incomplete formation of the clavicles on X-ray. Often, the lateral portion of the clavicles are not present and in rare cases the clavicles may be completely absent.14

Neurofibromatosis can also result in a pseudarthrosis, although these typically manifest in the long bones, particularly involving the lower extremities. Other findings such as café-au-lait spots and soft tissue tumors would help differentiate the exceedingly rare neurofibromatosis-related clavicular pseudarthrosis.14

In 1968, Kite2 classified congenital pseudarthrosis of the clavicle in two types based on anatomical, clinical and pathological differences, with implications for treatment indications.

I: Congenital failure of two segments of the clavicle to unite. This is seen at birth, and occurs because of hypoplasia of the distal fragment of the clavicle. Pressure on the prominence causes pain. On radiographs, the medial fragment is bigger than the lateral fragment, and the space between the fragments is evident. Surgical treatment should not be indicated.

II: Congenital bone deficiency. Like in cases of congenital pseudarthrosis of the tibia, the clavicle is seen at birth to have formed normally and fractures with the slightest trauma. In these cases, surgical treatment may be indicated. This is the form that presents a better prognosis.
There is controversy regarding the treatment of CPC.6,13-15 In many cases, the natural history is benign and no treatment is necessary. Most patients retain full range of motion of the shoulder and have no long-term complications.5,13-15 For those concerned with physical appearance, surgical treatment can be considered but results in an exchange of a protuberance for a scar (that could become hypertrophic, form keloid or remain painful, as well as possible scarring of the donor site, when autologous bone grafts have to be used).5,13-15 Other surgical indications may be pain, functional limitation or more rarely thoracic outlet syndrome (characterized by venous distension and a bluish discoloration of the arm) with urgent surgery needed.7,8

Many authors recommended that surgery should be delayed until the patient’s age is between 2 and 6 years.13-15 At that age, surgical management includes excision of the pseudarthrosis, with or without bone grafting, and stabilization with surgical hardware (external fixation, plates and screws, screws alone, Kirschner wires or Steinmann intramedullary pins). Postoperative immobilization consists of thoracic-brachial containment using a Velpeau sling or Desault bandage for four to six weeks.5,13,14 Few complications of surgical treatment have been described which included failure of osteosynthesis material, pain, weakness, hypotrophy in the operated limb, nonconsolidation with the need of a new approach, superficial infection, reversible neuropaxia of the brachial plexus and iatrogenic thoracic outlet syndrome.6,13-15

Conclusion

Despite being extremely rare it’s important that clinicians are able to diagnosis CPC through its characteristic physical examination findings and radiographic hallmarks. It is also essential to exclude other differential diagnosis, like clavicular fracture or rare bone diseases, to be able to reassure parents, explain the benign nature of this condition and provide appropriate care and follow-up, and referral to surgery if desired or necessary.