Congenital Pseudoarthrosis of the Clavicle: A Diagnostic Challenge

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Introduction

Congenital pseudoarthrosis of the clavicle (CPC) is a rare clinical defect first described by Fitzwilliams in 1910.1 This condition is more common in females than in males.2 CPC is mostly unilateral, on the right side, while bilateral involvement has been reported in some cases.3 In rare cases, CPC associated with bifurcation has been described.4

The characteristic feature of CPC is the presence of a painless swelling over the right clavicle. The distance from the sternum to acromion is shortened in CPC. The swelling is considered to be due to enlarged ends of the bones and the overlapping of the fragments.5

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 clavicle fracture or child abuse as has been in our case.

Case

We present the case of a 9-month-old male patient with a chief complaint of a painless, localized mass over the right shoulder, which has been present since birth. According to the patient’s parents, he was delivered via cesarean section after failure to progress with difficult vaginal delivery. According to the parents, at the time of delivery, the obstetrician documented the swelling and associated it with lymphadenopathy. No further workup was performed at that time. As the patient aged, the parents grew increasingly concerned about the prominence as it appeared to them to have grown in size. As a result, they took the patient to the emergency department for further evaluation. X-rays of the affected shoulder were obtained and a right midshaft clavicle fracture nonunion was diagnosed. At that time, child protective services (CPS) was consulted to evaluate the patient and the family for possible child abuse. The patient’s parents were interviewed in the emergency department and 2 home visits were conducted.

On physical exam, the patient was well developed and well nourished. The patient’s clothes were removed for the physical examination. There was no evidence of external trauma. No skin lesions or areas of hyperpigmentation were appreciated. The patient exhibited an elevated prominence over the area of the right midshaft clavicle. No erythema or warmth was appreciated. The entire length of the clavicle was palpated and no tenderness was appreciated. During the exam, the patient raised his right arm with no apparent discomfort and he reached out and grasped using the right hand. Sensation was intact to the right hand and grip strength was symmetric. The remainder of the examination was unremarkable.

X-rays of the right shoulder demonstrated 2 distinct, separate portions of clavicle: a medial portion with a sternal attachment and a lateral portion with an acromial attachment. There was no evidence of reactive bone formation and both ends were corticated with a smooth appearance (Figure 1).

Discussion

Congenital pseudoarthrosis of the clavicle is a very rare skeletal condition. The true incidence is not well reported, and it is rarely reported in medical literature. The infrequent presentation of this condition presents diagnostic challenges for clinicians. In cases of difficult vaginal deliveries, pseudoarthrosis may be confused with fracture secondary to birth trauma. Cases that present later in infancy may raise suspicions of child abuse, prompting unnecessary CPS investigations as is detailed in our case. It is important for clinicians to recognize the clinical presentation and natural history to appropriately diagnose and treat this condition.

Classically, congenital pseudoarthrosis has been described as unilateral, most often affecting the right clavicle. In one series, there also appeared to be a

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predilection to female gender.6 There have been reports of bilateral involvement, although this is exceedingly rare. Involvement of the left clavicle is usually associated with dextrocardia and cervical ribs. Most often, it is discovered at birth with a painless mass over the area of the midshaft clavicle. The pathophysiology is not entirely understood and several theories exist. The clavicle is an example of a flat bone. It forms in utero by intramembranous bone formation. It is the first bone in the body to ossify, while it is the last to fuse and mature. Some authors believe that the pseudoarthrosis represents an embryologic failure of the 2 ossification centers of the clavicle to appropriately fuse. 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 dominance of right sided pathology. There have been case reports of familial association suggesting the possibility of inheritance.3,7

Radiographically, plain films are usually sufficient to make a diagnosis when history and physical examination are consistent. On x-ray, there are 2 distinct portions of the clavicle that are separated, each having a smooth and intact cortex. In contrast to a healing fracture, there will be no radiographic evidence of callus formation or reactive. Most commonly, the pseudoarthrosis 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 (Figure 2).

In the neonate, differential diagnosis should include clavicle fracture resulting from difficult vaginal birth. This may prove to be difficult to differentiate at initial presentation, because fracture callus may have not yet formed. In this case, correlation of history and physical examination is critical. Pain with palpation should also be present with a fracture. In the case of infant or toddler, clavicle fractures secondary to trauma heal reliably. Abundant callus formation should be present on plain radiographs. In these cases, a thorough history is important to screen for abuse.

It is important to differentiate pseudoarthrosis of the clavicle from other, more serious syndromes. Cleidocranial dysostosis is an autosomal dominant condition that affects the 6p21 gene, encoding the RunX2 transcription factor. Hallmarks of the syndrome include 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, and coxa vera have been reported. Hypoplasia of the clavicles is a hallmark of the syndrome and results from failure of enchondral bone formation and there is bilateral involvement. In similar fashion to pseudoarthrosis of the clavicle, patients may present with painless mass over the midshaft of the clavicle. A thorough physical examination is important to evaluate for the other typical findings of this syndrome. Patients will have hypermobility of the shoulder joint and will have bilateral involvement. Radiographically, there is incomplete formation of the clavicles. Often, the lateral portion of the clavicles are not present, however, in rare cases the clavicles may be completely absent.

The treatment for pseudoarthrosis of the clavicle is somewhat controversial. 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. Parents or older patients may be concerned with the physical...
appearance of the mass over the clavicle. In these cases surgical intervention may be considered, although a scar may be ultimately less pleasing. Indications for surgery include cosmetic concerns, pain, or functional limitations. Of particular note, there have been case reports of thoracic outlet syndrome in patients with untreated pseudoarthrosis of the clavicle.\textsuperscript{9,10} In these cases, more immediate surgical intervention may be warranted.

Pseudoarthrosis of the clavicle represents an extremely rare diagnosis. Due to the difficulty in diagnosis, possible child abuse was suspected in our case, which resulted in parent interviews and home visits by CPS.

Therefore, it is highly important for clinicians to recognize the physical examination findings, radiographic hallmarks, and differential diagnosis to differentiate from other more serious syndromes as well as to avoid unnecessary involvement of the CPS.

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References

1. Fitzwilliams DCL. Hereditary cranial cleidodysostosis. \textit{Lancet}. 1910;2:625.
2. March HC. Congenital pseudoarthrosis of the clavicle. \textit{J Can Assoc Radiol}. 1982;33:35-36.
3. Price BD, Price CT. Familial pseudoarthrosis of the clavicle: case report and literature review. \textit{Iowa Orthop J}. 1996;16:153-156.
4. Magu NK, Singla R, Devgan A, Gogna P. Congenital pseudoarthrosis of the clavicle with bifurcation. \textit{Indian J Orthop}. 2014;48:435-437.
5. Kite JH. Congenital pseudoarthrosis of the clavicle. \textit{South Med J}. 1968;61:703-710.
6. Owen R. Congenital pseudoarthrosis of the clavicle. \textit{J Bone Joint Surg Br}. 1970;52:644-652.
7. Gibson DA, Carroll N. Congenital pseudoarthrosis of the clavicle. \textit{J Bone Joint Surg Br}. 1970;52:629-643.
8. Sung TH, Man EM, Chan AT, Lee WK. Congenital pseudoarthrosis of the clavicle: a rare and challenging diagnosis. \textit{Hong Kong Med J}. 2013;19:265-267. doi:12809/hkmj133648.
9. Lozano P, Doaz M, Riera R, Gomez FT. Venous thoracic outlet syndrome secondary to congenital pseudoarthrosis of the clavicle. Presentation in the fourth decade of life. \textit{Eur J Vasc Endovasc Surg}. 2003;25:592-593.
10. Watson HI, Hopper GP, Kovacs P. Congenital pseudoarthrosis of the clavicle causing thoracic outlet syndrome. \textit{BMJ Case Rep}. 2013;2013. doi:10.1136/bcr-2013-010437.