CASE REPORT

Congenital pseudarthrosis of the clavicle: a rare and challenging diagnosis

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Congenital pseudarthrosis of the clavicle is a rare clinical entity, first described in 1910. We report on a newborn baby girl who presented with a painless lump over mid-portion of right clavicle at her routine newborn examination, which was subsequently diagnosed as a congenital pseudarthrosis. Here we explore its pathogenesis, elaborate on its differential diagnoses in paediatric patients, and comment on its distinct radiological features.

Introduction

Birth defects of the clavicle encountered at newborn examinations are not uncommon. Clavicular fracture from birth trauma contributes to most of the cases, with a reported frequency of 15 per 1000 live births.[1] The important association with brachial plexus injuries is also well-known, and is therefore a tempting diagnosis in the presence of clavicular birth defects. Nonetheless, in the presence of conflicting clinical findings, a rare but essential differential diagnosis, namely congenital pseudarthrosis should also be considered.

Case report

A newborn baby girl presented with a focal non-tender swelling in the mid-portion of the right clavicle during routine newborn examination in October 2010. She was born by caesarean section due to high-grade maternal placenta praevia, but there was no obvious traumatic birth history. The baby was active with normal upper limb movements.

The initial chest radiograph taken on the first day of life showed an abnormal contour of the right clavicle (Fig 1). The localised view of the right clavicle demonstrated discontinuity of the mid-portion with postero-infero-medial positioning of the acromial half with respect to the sternal half. Both ends were smooth in contour and not attenuated. Follow-up radiographs showed no callus formation. Subsequent plain computed tomography (CT) of the clavicles performed 5 months later confirmed the presence of well-corticated borders with mild hypertrophy of both bone ends at the discontinuity site, but there was no obvious callus formation (Fig 2).

In the absence of local tenderness and preceding birth trauma, the overall radiographical features were more in favour of congenital pseudarthrosis of the clavicle rather than clavicular fracture with non-union. Other differential diagnosis of cleidocranial dysostosis and neurofibromatosis were unlikely, as there were no other associated structural abnormalities.

Key words
Birth injuries; Clavicle; Hong Kong; Infant, newborn; Pseudarthrosis

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Discussion

Congenital pseudarthrosis of the clavicle has been a rare entity in paediatric radiology, first reported by Fitzwilliams in 1910.[2] Its exact frequency is not well documented in the literature. Notably, there is predominance of right-sided involvement; in a review by Owen,[3] 100% of the 33 patients with congenital pseudarthrosis affected the right clavicle. A relative female predominance (70%) was also observed, but such gender predilection has not been described in other studies. Associations with other structural abnormalities in even rarer cases with left-sided or bilateral involvement have been reported, such anomalies being dextrocardia and cervical ribs, respectively.[4-7] Possible genetic transmission of the condition has also been suggested with a postulated autosomal dominant trait, as illustrated by Gibson and Carroll[8] who described nine cases in a single family.

The pathogenesis of congenital clavicular pseudarthrosis is related to the embryology of the clavicle. The clavicle is the first bone to become ossified during embryogenesis, but after birth it is the latest to attain full maturation (in the early 20s). An unusual situation of membranous diaphyseal and metaphyseal ossification in concurrence with

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先天性鎖骨假關節：一個既罕見又具挑戰性的診斷

先天性鎖骨假關節症很罕見，最早記載於1910年。本文報告一名新生女嬰在接受常規新生兒檢查時發現在右鎖骨中部有一個無痛性腫塊，後被診斷為先天性鎖骨假關節。本文續探討此症的發病機制、在小兒患者的鑒別診斷，以及其獨有的放射學影像特徵。

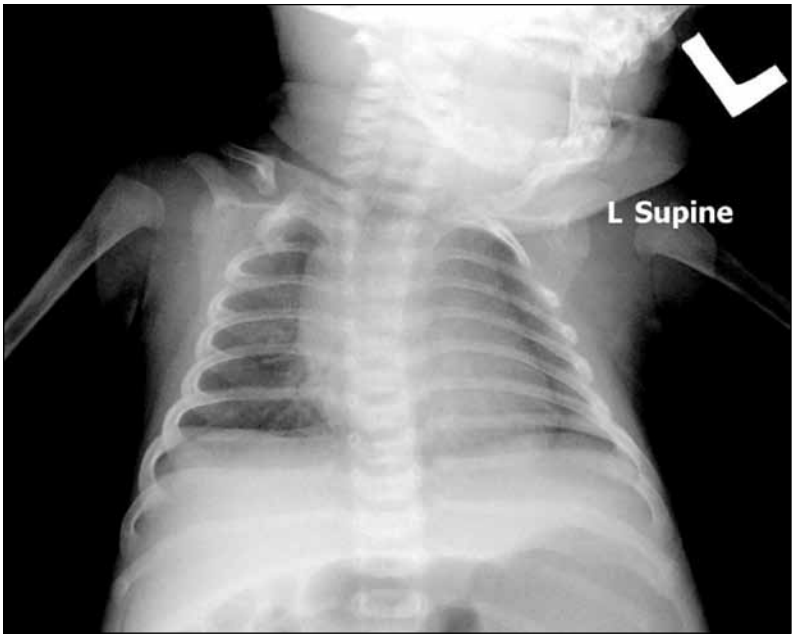

FIG 1. Plain radiograph of the chest shows an isolated defect in the mid-portion of the right clavicle

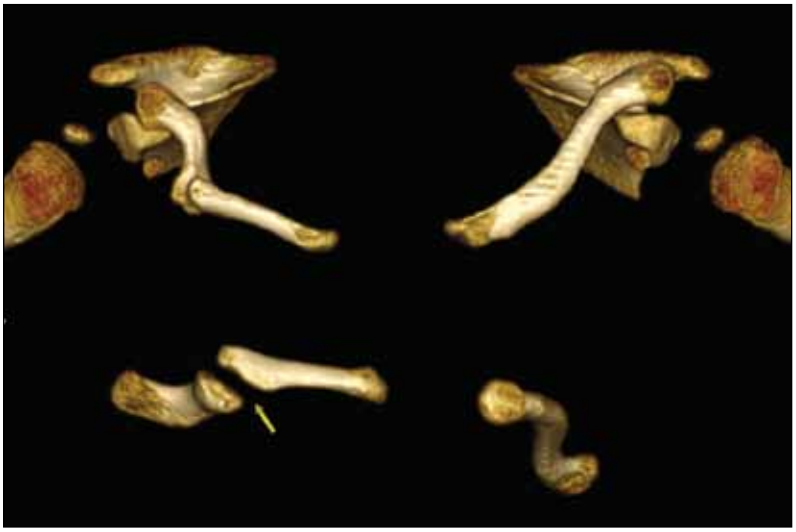

FIG 2. Subsequent computed tomography with 3-dimensional reconstruction demonstrating the well-corticated bony ends of the clavicular defect with no frank callus formation (arrow), which favoured congenital pseudarthrosis in the clinical context of our case

the endochondral longitudinal growth in the same bone has been observed in the clavicle. Two primary ossification centres are formed, one medial and the other lateral, in the fifth and sixth gestational weeks, and fuse during fetal development; endochondral ossification subsequently takes place.[9] In view of this embryological finding, it has been postulated that an interrupted formation of the two primary ossification centres could be responsible for clavicular pseudarthrosis. However, further morphological studies had provided contrary evidence. In a Japanese study by Ogata and Uhthoff,[10] the junction of the two primary ossification centres was shown to be located between the lateral and middle third of the clavicle, which does not correspond to the typical location of congenital pseudarthroses that are typically situated slightly more medially in the middle part of the clavicle. Currently, there is a general consensus that mechanical factors during embryogenesis probably contribute to the pathogenesis. Most investigators believe that the condition is caused by extrinsic pressure exerted on the budding clavicle by the adjacent pulsatile subclavian artery. The right subclavian artery is generally located at a higher level, which conforms with right-sided predominance. This theory is also supported by and neatly explains the aforementioned observation that rare left-sided and bilateral involvements are associated with dextrocardia and cervical ribs, in which higher aberrant positions of the subclavian artery are encountered.

Radiological features of congenital clavicular pseudarthrosis described in a variety of reports have been consistent. Characteristically, they include involvement of the middle part of the clavicle with a definite separation into two parts.[11] Notably, at the site of pseudarthrosis, both ends show bony hypertrophy with well-defined corticated borders; the sternal half typically lies above and anterior to the acromial half.[3] Lack of callus formation at the site of the pseudarthrosis and an uneventful delivery helps distinguishing it from the much more common differential diagnosis of clavicular fracture with non-union. The evaluation of its morphology and the presence of callus is sometimes difficult to discern, due to suboptimal radiographical projections. As illustrated in our case, the use of CT with 3-dimensional (3D) reconstruction demonstrated the underlying anatomical details, which was important in excluding other differentials including trauma, infective and neoplastic causes, and provided essential information for operative planning. Another rare yet important differential diagnosis would be cleidocranial dysostosis, which is characterised by hypoplasia/aplasia of the lateral clavicular ends, retarded cranial ossification, supernumerary teeth, and short stature. The latter's dissimilar location (lateral involvement), tapering of both bone ends,

and co-existing structural abnormalities help to distinguish itself from congenital pseudarthrosis.

Excisional biopsy of the clavicle demonstrates histological evidence of its cartilaginous structure at the ends of the pseudarthrosis.[12] Surgical treatment of the condition could be considered in patients with upper limb dysfunction, eg reduced strength or range of motion; for cosmetic needs; and to prevent future-onset thoracic outlet syndrome.[13] Excision of the pseudarthrosis and internal fixation of both bone ends with pinning or bone grafting generally yields satisfactory outcomes in terms of postoperative functioning and appearance. Yet, controversies exist about the optimal age for surgery; intervening during infancy or later have both been advocated.[8,14]

In summary, we described a patient with congenital pseudarthrosis of the right clavicle, which is a rare entity. Computed tomography with 3D reconstruction delineated the underlying anatomical details that aided making a diagnosis and subsequent operative planning. High level of clinical alertness, a detailed birth history, and physical examination for associated structural abnormalities are essential in establishing this diagnosis from a number of other differential diagnoses (clavicular fracture with non-union, cleidocranial dysostosis, and neurofibromatosis). Excisional biopsy of the clavicle showing its cartilaginous structure at the end surfaces of pseudoarthrosis can provide further histological evidence.

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