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Congenital pseudarthrosis of the clavicle in two siblings

İki kardeşte doğuştan klavikula psödoartrozu

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Doğuştan klavikula psödoartrozu etyolojisi bilinmeyen nadir bir durumdur. Günümüze kadar sunulan yaklaşık 200 olgu arasında aile bağının bildirildiği çok az olgu vardır. Bunlar da birinci derece aile üyeleri değildir. Dokuz yaşındaki bir kız çocuğunda, sağ kolda güçsüzlük ve sağ omuzda şişlik şikayetleri vardı. Klinik muayenede sağ klavikula üzerinde kitle izlenimi veren bir şişlik ve klavikulada patolojik hareket saptandı. Omuzlarda asimetri vardı. Omuz eklem hareket açıklığı iki tarafta da normal sınırlarda idi. Düz radyografilerde sağ klavikula diyafizinde bir defekt izlendi. Bilgisayarlı tomografi incelemesinde sağ klavikulada devamlılığın olmadığı saptandı. Hastanın üç yaşındaki kız kardeşinde de benzer klinik ve radyolojik bulgular saptandı. Her iki hastada da herhangi bir travma, zorlu doğum ya da doğum komplikasyonu öyküsü yoktu. Kas-iskelet sistemi ile ilgili başka patolojiyi düşündürecek bulguya rastlanmadı. Rutin laboratuvar incelemelerinde anormal bir değerle karşılaşılmadı. Yapılan genetik analizde patoloji saptanmadı. Bu bulgular ışığında hastalara doğumsal klavikula psödoartrozu tanısı kondu. Eklem hareket açıklıkları doğal olan ve ağrı şikayeti bulunmayan hastalar takibe alındı. Olgularımız, literatürde özkardeş olarak doğuştan klavikula psödoartrozu tanısı konan ilk olgulardır.

Anahtar sözcükler: Çocuk; klavikula/patoloji; psödoartroz/doğuştan/radyografi; kardeş.

Congenital pseudarthrosis of the clavicle is a rare disorder of unknown etiology. Among nearly 200 cases hitherto reported, only a few cases have familial coexistence, and none are first-degree relatives. A nine-year old girl had complaints of weakness in the right arm and swelling in the right shoulder. On physical examination, a mass-like lesion in the right clavicle, abnormal clavicular movement, and asymmetric shoulders were noted. The range of motion of the shoulder was in normal range on both sides. A plain radiogram showed a defect in the diaphysis of the right clavicle and computed tomography showed discontinuity of the right clavicle. Similar clinical and radiologic findings were also detected in her younger sister who was three years old. None had a history of trauma, difficult delivery, or natal complication, any abnormal findings related to the musculoskeletal system, any abnormality in routine laboratory test results and genetic analysis. The diagnosis was made as congenital pseudarthrosis of the clavicle in both siblings. Since they had normal range of joint movements without pain, they were scheduled for clinical follow-up. To our knowledge, these two siblings are the first to be reported in the literature for having congenital pseudarthrosis of the clavicle.

Key words: Child; clavicle/pathology; pseudarthrosis/congenital/radiography; siblings.

Congenital pseudarthrosis of the clavicle (CPC) is a rare disorder. The frequency is reported to be equal in both sexes.[1] As the right side is involved the most, 10% of bilateral involvement is also reported. Nine cases of bilateral involvement is reported.[2] Also, in a few patient involvement of the left side is reported. CPC is distinguished from acute neonatal fractures with positive history of birth trauma and the absence of early exuberant callus formation. In radiographies, it is different from cleidocranial dysostosis as the clavicle is in a defective appearance and characteristical bone malformations are not present in the other bones

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(cranium and pelvis). The sternal fragment is almost always bigger and revealed on the superior portion of the acromial fragment.[3]

The genetical basis and etiology of CPC is obscure. Most of the cases are diagnosed seperately. There are 2 principal theories suggested to explain the etiology. The first theory suggests that clavicula develops from 2 ossification centers and CPC is due to the failure in the fusion of these centers.[4,5] According to the other theory, the subclavian artery which is in a more cephalad location than the clavicle compresses on the clavicle and CPC occurs, also cervical ribs may cause compression.[6] The second theory seems to be more sufficient to explain why the right sided involvement is more common and the bilateral involvement.

CPC is usually diagnosed in infancy or early childhood with the presence of a painless and mobile mass in the 1/3 medial portion of the clavicle. A radiographical evaluation must be made to differentiate the diagnose from birth fractures and cleidocranial dysostosis. The deformity slowly develops in terms of cosmetic measures and the mass grows as the shoulder girdle declines.The lesion may be painful during overhead activities and when directly palpated or compressed superficially. Shoulder range of motion is usually normal and the joint is functional. In many cases it is reported that thoracic outlet syndrom developed.[7,9]

Case report

A nine years old girl applied to our outpatient clinics with the complaints of weakness in the right arm and presence of a mass on the right shoulder. The patient did not have a history of trauma. In the past, the patient did not have any problem to use her right arm and discomfort caused by the mass on the shoulder. The patient did not have a history of difficult delivery or birth complications. In clinical examination, a mass on the shoulder and pathological movement in the clavicle was observed (Figure 1a). The shoulders were asymetrical. Shoulder range of motion was normal in both sides. Any other signs about an additional pathology of musculosceletal system was not detected. Routine laboratory studies revealed normal results. A genetical evaluation was held and it was reported to be normal. Plain radiographies revealed a defect in the diaphysis of the right clavicle (Figure 1b). Residual bone fragments were seperated and sclerosed without

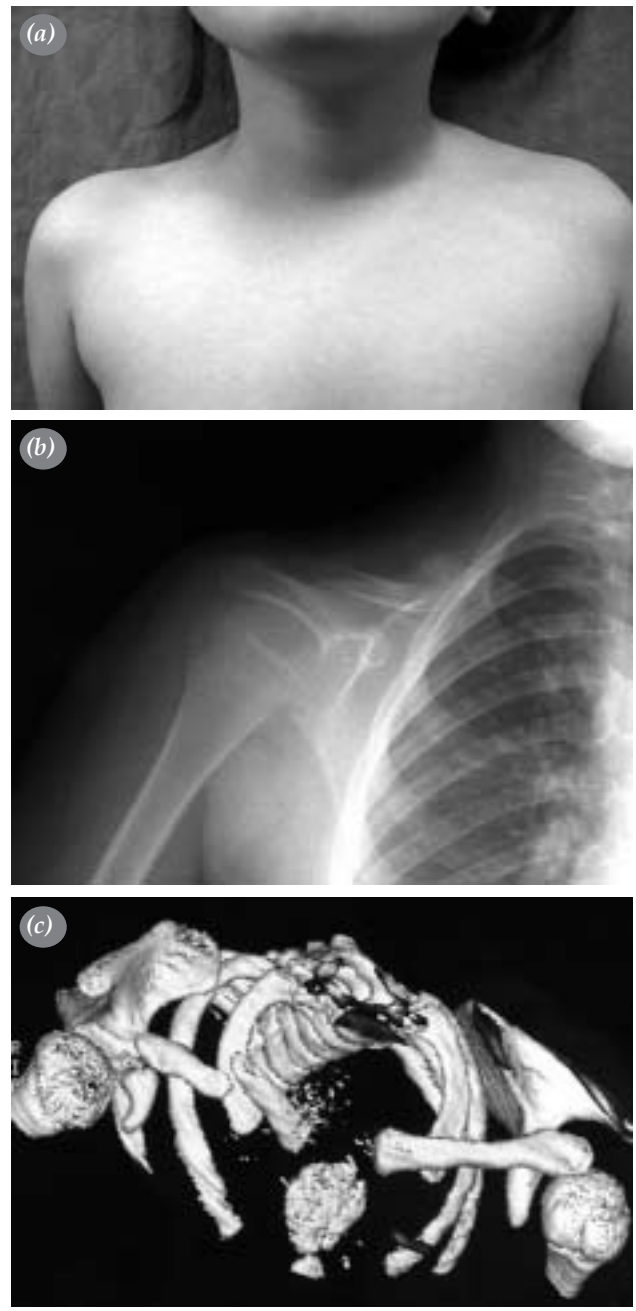

Figure 1. (a) shoulder asymetry, **(b)** discontinuity in the right clavicle revealed by bilateral AP shoulder radiography, **(c)** defect in the right clavicle revealed by computerized tomography evaluation, in the 9 years old sibling.

any callus or periosteal reaction. An overriding of the medial fragment on the lateral one and its dislocation to superior direction was observed. Computerized tomography evaluation revealed discontinuity in the right clavicle (Figure 1c). There were no signs of abnormality in cervical vertebrae, cervical ribs, dextrocardia, signs associated with cleidocranial dysostosis

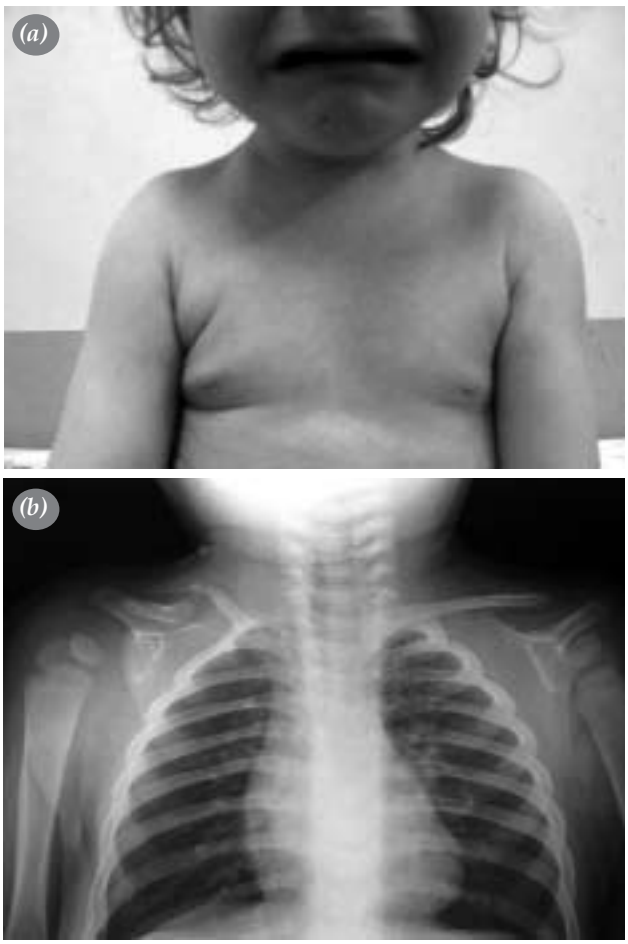

Figure 2. (a) clinical appearance, **(b)** AP shoulder radiography, of the 3 years old sibling.

in radiographical evaluation and the patient was diagnosed as CPC. When the other family members were examined, it was observed that the patient's 3 years old sister had similar clinical findings (Figure 2a). Radiographies revealed discontinuity also in this case (Figure 2b). The patient did not have a history of difficult delivery or birth complications. This patient also did not have a history of difficult delivery or trauma, pathological laboratory and genetical analysis results, so the patient was diagnosed as CPC. Surgery was not indicated for the patients who had normal joint range of motion and did not have pain and were scheduled for follow-up. Our cases are the first to be reported as the siblings with CPC .

Discussion

CPC is a rare entity and first reported by Fitzwilliams in 1910.[4] In 1963, Alldred reported 9 cases and distinguished CPC from cleidocranial dysostosis and birth fractures of the clavicle.[11] There are about 200 cases reported so far.[12] Clavicula is the first embryonical/fetal bone to be primarily ossified. There are some theories suggested to explain the pathogenesis of this disorder with unknown etiology. One of these theories suggests that the clavicle normally starts ossifying from 2 centers and there is failure in the fusion of these centers.[4,5] However this theory cannot explain why most of the cases are involved in the right side. According to the other theory suggested by Lloyd-Roberts et al [6], the right subclavian artery which is located more cephalad than the left causes the disorder by compressing on the developing clavicle. Also it is reported that cervical ribs may cause the same kind of compression on the clavicle.[6] The histological study which Hirata et al made suggested that the clavicle developed from 2 ossification centers as they could explain the presence of bilateral involvement.

As an interesting condition, almost all of the cases were involved in the right side. Our cases were also involved in the right side. In 10% of the reported cases, bilateral involvement was reported. A total of 4 cases, a case which Sakkers et al reported [14], another reported by Lloyd-Roberts [6] and 2 reported by Gibson and Carroll [4], were reported to be involved in the left side. In the cases which were reported by Lloyd-Roberts et al [6] and Owen [11], a big cervical rib and in the cases which were reported by Gibson and Carroll, dextrocardia was present. This particularity strengthen the hypothesis that the right subclavian artery causes CPC by compressing on the clavicle. The only case which is without cervical ribs or dextrocardia is the one which Sakkers et al reported. This case does not bear Lloyd-Roberts et al's theory. In the differential diagnosis of CPC, cleidocranial dysostosis, neurofibromatosis and pseudarthrosis due to trauma should be noted. CPC is distinguished from these disorders by the absence of ossification defects in the other bones of the body, osseous or fibrous callus and cafe-au-lait spots. Our cases also did not have these features. A few of the cases have familial relation. Alldred [10] reported 2 step siblings, Gibson and Carroll [4] reported 8 cases from 3 generations in the same family. The cases which Gibson and Carroll described as short and with teeth and palate problems, are probably related with cleidocranial dysostosis. The familial cases reported are probably recessively transmitted. There are no other cases which are reported to have familial relation. There is not a certain con-

cencus in the treatment of CPC. As there are articles [2,4,7,9-11,15-18] which claim that surgical applications are necessary in the early or late periods, to fix the mass like appearance on the shoulder for aesthetical concerns and to prevent thoracic outlet syndrom which is an often complication in CPC , there are some others [8,14,19] which claims that follow-up of the patients are sufficient because the disorder is asymptomathic and painless, with normal range of motion. The presence of the cases which are in adulthood and have functional joints and are pain free despite of not being operated.[8, 20] Based on the 6 patients that they operated, Lorento Molto et al [2] claims that the patients in whom surgery was indicated must be operated in the early period. Alldred, [10] Gibson and Carroll [4] suggested that the patients should be at least 4 or 5 years old for surgical intervention. Adviced technique is autografting after debridement of the pseudarthrosis site, and internal fixation with plates and screw or Kirschner wires.[1,2,7,15,16,20] Generally it is thought that bone grafting is necessary.[4,10,15] Alldred (109 suggested that bone grafting is necessary for all cases below 8 years old. Lorento Molto et al [2] first operated the left side of a case with bilateral involvement and they used half of the bone graft that is derived from the iliac crest for grafting. They conserved the other half in liquid nitrogen in strict aseptic conditions and used it for grafting the right clavicle in the second session. According to Grogan et al [15], internal fixation is not necessary in the cases younger than 3 years old and it is sufficient to close up the fragments and to fix a small bone graft between them as the site is covered with a periosteal sheet.[2,4] Lozano et al [9] operated a woman who was asympthomatical till 48 years old but in whom thoracic outlet syndrom developed then, by performing surgical decompression and resection of the sternal clavicular fragment and they gained good result. The authors suggested that this technique was better than correcting osteotomy, graft interposition and internal fixation in this age group of patients. Some complications like sepsis, nonunion and brachial plexus palsy is reported in the operated cases.[10,11,17] Toledo and MacEwen [17] performed graft interposition and internal fixation with a Steinmann nail in a patient. As acute neuropraxia developed in the early post-operative period Steinmann nail was immediately removed. The symptoms regressed and the patient was cured with minimal neurological deficit. We decided to schedule the patients for follow-up instead of performing surgery as they had full range of motion and were pain free.

In conclusion, there is no CPC case which are reported to be siblings. The cases that are reported to have familial relation [4,10] are not whole siblings and their diagnose of CPC is suspicious. Normal genetical analysis of our cases and that they do not have additional pathologies strengthen our diagnose of CDC. For this reason, these cases are the first to be reported to be siblings with CPC.

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