FAMILIAL CONGENITAL PSEUDOARTHROSIS OF THE CLAVICLE: CASE REPORT AND LITERATURE REVIEW

Bryan D. Price, B.A.*
Charles T. Price, M.D.**

ABSTRACT

Congenital pseudoarthrosis of the clavicle (CPC) is a rare condition of unresolved etiology and pathogenesis. Familial occurrence of this anomaly has been documented but the pattern of genetic transmission remains obscure. Two cases involving a father and daughter are presented here with a review of the literature. These cases add support to the genetic basis for CPC.

INTRODUCTION

Congenital pseudoarthrosis of the clavicle (CPC) is a rare clinical defect. Although most orthopedic surgeons have had experience with this abnormality, its familial nature is not commonly noted. This study offers a review of the literature and presents two cases of CPC involving a father and daughter.

The condition was initially described by Fitzwilliams as a distinct variant of cleidocranial dysostosis in 1910. CPC has since appeared in a number of studies involving more than 100 patients. This malformation appears to be 25-80% more common in females than males. Usually the condition is unilateral and occurs on the right side, but in approximately 10% of the cases involvement is bilateral. Left sided unilateral involvement is extremely rare and is often associated with dextrocardia or cervical ribs.

The malformation is fully present at birth, but most often the diagnosis is made a few months to several years later. The patient usually presents with a painless swelling over the middle third of the right clavicle. This swelling often becomes more evident with maturation. Full range of shoulder motion is present. The adult with CPC may complain of fatigue or discomfort related to strenuous activity. CPC markedly shortens the distance from the sternum to the acromion due to an absence of a small portion of the bone. This separates the clavicle into two parts. The larger sternal segment always overlies and is anterior to the proximal end of the acromial segment. There is often a small depression on the "inferior aspect of the outer end of the medial segment into which the blunted inner end of the lateral segment fits." The swelling associated with CPC is attributed to the enlarged ends of the bones, the overlapping of the fragments, and the cephalad angulation of the fragments. The bony ends are never pointed as they are in pseudoarthrosis of the tibia. With palpation, a discontinuity in the substance of the clavicle and mobility between medial and lateral fragments is evident. Often cervical ribs or abnormally elevated first ribs are present along with the abnormal clavicles.

CASE REPORT

A 9-year-old girl was seen for evaluation of congenital pseudoarthrosis of her right clavicle. Gestation, labor, and delivery were atraumatic and uneventful. Separation of the right clavicle was noted one day after birth. This was initially thought to be a birth fracture, but never healed. The patient has remained very healthy and her developmental milestones have been normal. At age 9 the mid-clavicular swelling had become more prominent and surgical correction was contemplated. She reported no pain or difficulty with normal activity, although extreme physical exertion, such as throwing a baseball, occasionally produced pain in the region of the defect.

Examination revealed a prominent mid-clavicle on the right, consistent with the diagnosis of CPC. There was no tenderness. She demonstrated full range of motion of the shoulder and had normal strength in the right upper extremity. No other musculoskeletal abnormalities were noted.

Radiographs were reviewed and revealed a 1 to 2 cm defect in the middle third of the clavicle (Figure 1).

The patient's father also had a diagnosis of a right CPC. A bone graft at the age of 10 did not successfully heal his pseudoarthrosis. He experiences right shoulder pain with strenuous activity, such as weight lifting or overhead work. Radiographs of the father's clavicle revealed a pseudoarthrosis with callus formation in the middle third of the clavicle (Figure 2).
DISCUSSION

Genetics

Gibson and Carroll were among the first researchers to document familial incidence of CPC. One patient gave a history of an aunt and grandfather who also had the same anomaly. A second family appeared to display an autosomal dominant type of transmission. However, minor bone abnormalities in this family suggest that these patients may have had a variation of cleidocranial dysostosis rather than CPC.

Some researchers suggest that environmental factors rather than genetic factors influence the development of CPC. The high prevalence of unilateral CPC exclusive of any family history or additional anatomical abnormalities support this hypothesis.

However, documented cases of familial CPC exclusive of other bone abnormalities have been reported. Owen reported a father and daughter with identical right sided lesions. Toledo noted a positive family history in two sisters whose mother also had a typical right-sided CPC. Alberink studied a man whose three sisters and father had right clavicular pseudoarthrosis. Although the majority of reported cases of CPC show no family history of this disorder, these reports suggest a possible autosomal dominant inheritance pattern in families. Clearly a familial basis for some cases of CPC exists, but a definite genetic pattern has not been established.

Pathogenesis

The anatomical basis of CPC is still questionable. Theories of development of CPC include nonunion of two ossification centers in the embryonic clavicle, exogenous or mechanical causes, and abnormalities of the vasculature surrounding the developing clavicle.
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Figure 2. Child's father demonstrates pseudoarthrosis of the same side 30 years after attempted surgical correction.

Studies of clavicular ossification centers by Mall and Wall indicated that the embryonic clavicle forms from two ossification centers. Many researchers believe that CPC results from a failure of these two centers to fuse. Gibson and others studied the embryology of the clavicle and believed that it developed from one ossification center. Ogata and Uhthoff have conducted research on embryologic ossification of the clavicle which supports previous studies claiming that the clavicle forms from two membranous ossification centers. After reviewing previous reports, they concluded that the two centers had already fused prior to the histological study by researchers claiming development from only one center. However, Ogata and Uhthoff also indicated that these two centers are more lateral than the usual location of CPC. Thus, failure of fusion of ossification centers may not explain the etiology of this malformation.

A theory based on exogenous or mechanical causes suggests that the prevalence of right sided lesions could be directly attributed to the more common left occipitoanterior presentation during birth. This position may place the right shoulder at a higher risk for trauma. Thus, CPC may result from birth fracture and nonunion of the clavicle rather than an abnormality during clavicle development.

Another theory strongly supported by Lloyd-Roberts et al. suggests that certain anatomical abnormalities are predisposing factors for CPC. It is believed that cervical ribs or “vertically disposed and elevated upper ribs” may cause compression of the subclavian artery between the clavicle and first rib. The exaggerated arterial pulsations and pressure which result could influence the development of a pseudoarthrosis. This theory suggests that the increased incidence of right sided CPC may be related to the fact that the right subclavian artery is normally elevated to a point directly underneath the clavicle. Anatomical abnormalities such as cervical ribs may influence the relationship of the subclavian artery to the right clavicle. Although not all cases document predisposing abnormalities, cervical ribs have been reported in roughly 15% of the
literature on CPC\textsuperscript{8,14,20}. Further support is offered by the fact that dextrocardia or large left sided cervical ribs were present in many reported cases of left-sided CPC\textsuperscript{7,11}. In such instances it is suggested that the level of the left subclavian artery may exceed the right\textsuperscript{13}. Rib abnormalities are known to have a familial occurrence. If abnormally elevated ribs are a predisposing factor for CPC then this would lend further support to a possible genetic etiology of CPC\textsuperscript{11}.

CONCLUSION

The father and daughter reported here add support to a genetic basis for CPC. The association of CPC with other familial anatomical anomalies also supports familial inheritance. However, the frequent occurrence of CPC without family history prevents the formation of a conclusive genetic explanation. While researchers are beginning to agree that the clavicle develops from two primary ossification centers, patterns of clavicular formation seem unrelated to formation of CPC. Associated anatomical abnormalities are infrequently documented but may provide some explanation for the formation of CPC. The etiology and pathogenesis of this condition remain obscure. It is our opinion that genetic and anatomical factors play a role in the development of CPC.

REFERENCES