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Case report

Bilateral clavicular hypoplasia and cleidocranial dysplasia: Case report

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Abstract: Clavicular hypoplasia or agenesis is common in patients with cleidocranial dysplasia (CCD). CCD is an uncommon congenital pathology with an autosomal dominant pattern affecting males and females equally. CCD presents with wide phenotypic variability, but there is no stable link between the genotype and phenotype. In addition to clavicular hypoplasia/agenesis, such patients present with thoracic deformities, iliac hypoplasia, cranial bone deformities, and delayed dentition. An early diagnosis is important to determine treatment options. However, there is no specific orthopedic treatment for this disease.

Keywords: agenesis, clavicle, cleidocranial dysostosis, hypoplasia

INTRODUCTION

Cleidocranial dysostosis (CCD) was first described in 1765 by Martin [1], and is also known as Marie-Sainton disease and mutational dysostosis. The gene responsible for the pathology is *RUNX2*, located on chromosome 6p21 [2].

CCD is a rare congenital disorder with an incidence of 1:1,000,000 and an autosomal dominant inheritance pattern with variable expression, and commonly affects several members of a family. The disease has no predilection for sex or ethnic group. It presents with variable expression and complete penetrance. Despite familial transmission, it can occur spontaneously in almost 40% of cases [3]. Bone defects in patients with CCD involve mainly the clavicles, skull and mandible, although a wide variety of anomalies can be found in other bones [4].

The muscles associated with the abnormal clavicles are poorly developed. Patients have broad necks, and the absence or hypoplasia of the clavicles leads to unusual mobility of the shoulders, which can approach/touch at the midline of the thorax. This is a characteristic and pathognomonic finding of the syndrome [5].

CASE REPORT

A 57-year-old man consulted the Shoulder and Elbow Surgery service of the Hospital Santo Antônio on January 23, 2017 with the complaint of shoulder pain that worsened after physical exertion.

He reported being diagnosed with CCD, but had no regular medical follow-up. There was no family history of dysplasia.

Physical examination revealed rib cage deformities, bilateral clavicular fossa accentuation, shoulder girdle muscular hypotrophy, and scapular hypoplasia. There was no restriction of shoulder mobility and muscle strength was grade 5 in the American Spinal Injury Association socre. There was a positive bilateral Jobe sign (Figures 1 and 2).

(Figure 1)

(Figure 2)

Considered a pathognomonic finding, the patient demonstrated hypermobility by touching the shoulders at the midline (Figure 3).

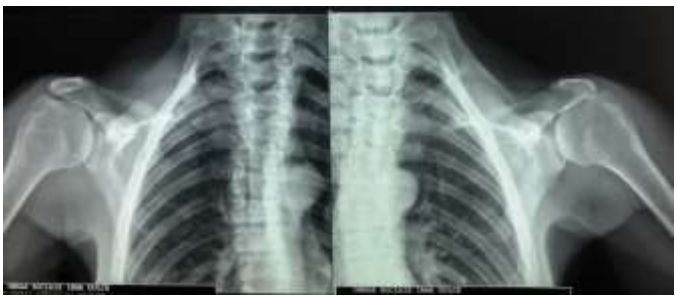
The Shoulder Pain and Disability Index (SPADI) was used for functional evaluation. The SPADI is a quality of life questionnaire designed to assess pain and disability associated with shoulder dysfunction. Initially, the SPADI was proposed as a Visual Analog Scale (VAS), and was later validated as a Numeric Rating Scale (NRS). Several studies have supported the use of the SPADI in clinical practice and research. All psychometric properties are evaluated using the SPADI, with a response format using an NRS and short questions that facilitate completion in 3 to 10 minutes. The version of the SPADI used has been translated and validated for Portuguese [6].

The patient's score on initial evaluation was 47 points.

(Figure 3)

Radiological examination showed bilateral clavicular hypoplasia (Figure 4). Shoulder ultrasonography revealed bilateral supraspinous tendinopathy. Conservative treatment was initiated with anti-inflammatory drugs and motor physical therapy.

On reassessment on March 13, 2017, the patient reported improvement of symptoms and the SPADI score was 13 points.



(Figure 4)

DISCUSSION

CCD is considered rare, with an estimated incidence of 1:1,000,000, and presents with a varied clinical spectrum. There is no clear genotype-phenotype correlation.

In most cases, this syndrome remains underdiagnosed because of the relative lack of medical complications as compared to other skeletal dysplasias [7]. CCD is usually present from birth, but is often not diagnosed or is an incidental finding [8].

Clavicular hypoplasia is one of the main findings. The shape of the shoulder girdle is modified by changes in the origins and insertions of the muscles without affecting shoulder function. Some patients can touch their shoulders at the midline.

As clavicles have varying degrees of hypoplasia in these patients and are completely absent in approximately 10% of cases, this allows for excessive shoulder mobility [2]. This finding was obvious in our patient.

Deficient cranial bone formation can cause hearing loss [9]. This is a frequent complication in patients with CCD and should be investigated. However, we did not find this complication in our patient.

Because CCD is a hereditary bone disorder that exhibits phenotypic manifestations in varying degrees, it may be confused with other bone pathologies or underdiagnosed in clinical practice [10].

There is no treatment for cranial, clavicular, or other bone abnormalities associated with CCD. However, treatment of oral conditions is important because of the associated high morbidity rate [11].

CONCLUSION

Cleidocranial dysostosis is a rare condition that causes severe hypoplasia of the clavicles but with little functional limitation. There are no studies in the literature associating clavicular hypoplasia with rotator cuff degeneration. Our patient complained of shoulder pain related to bilateral supraspinous tendinopathy, and responded well to conservative treatment.

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