

IMAGE CASES

Touching shoulders

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Preterm male newborn of 33 weeks and 5 days, delivered by cesarian section due to non-reassuring fetal status, born with an Apgar score of 7/8 at 1^o/5^o minutes respectively, without requiring resuscitation. Chest x-ray showed hypoplastic clavicles (**Figure 1.**), confirming a diagnosis already made during the prenatal period. He currently has a multidisciplinary follow-up, including Pediatric Orthopedics consultation, where it could be observed, during examination, that the shoulders could be approximated in the midline (**Figure 2.**).

What is your diagnosis?

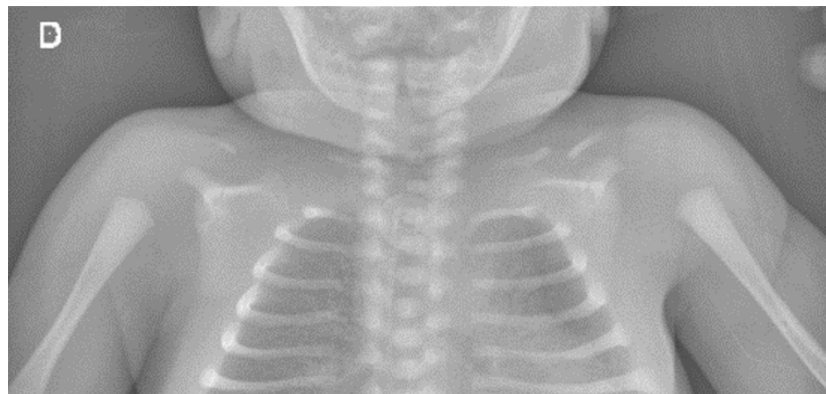


Figure 1 - Chest X-ray at birth



Figure 2 - At Pediatric Orthopedics consultation

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DIAGNOSIS

Cleidocranial dysplasia

DISCUSSION

Cleidocranial dysplasia (CCD) is a rare skeletal autosomal dominant syndrome (1: 1,000,000) with an almost complete genetic penetrance and variable clinical expression.^(1,2) It is a disorder in which the bones formed by intramembranous ossification (primarily the clavicles, cranium, and pelvis) are abnormal.⁽³⁾

All the features of CCD may vary, even among the members of the same family. However, the most prominent clinical findings in individuals with CCD include abnormally large, wide-open fontanelles at birth that may remain open throughout life, clavicular hypoplasia resulting in narrow, sloping shoulders that can be opposed at the midline, and abnormal dentition.

Patients with CCD are more likely to have other skeletal and orthopedic problems, such as flat feet, valgus, scoliosis and osteoporosis.⁽⁴⁾ They are smaller than their unaffected siblings and may have low IGF-1 levels.

Radiographic features include wide-open sutures, patent fontanelles and wormian bones; cone-shaped thorax with arrow thoracic diameter, absence or hypoplastic clavicles and hypoplastic scapulae. Other radiological findings may include delayed ossification of the pubic bones.^(4,5)

The diagnosis can be done during prenatal period through ultrasound findings along with positive family history, or after birth through clinical and radiographic features. Nevertheless, documentation of heterozygous pathogenic variant in RUNX2 (CBFA1) identified by molecular genetic testing is also a possibility for a definitive diagnosis.

The differential diagnosis includes mandibulo-acral dysplasia, dental anomalies, congenital clavicle pseudoarthrosis, Yunis-Varon syndrome, picnodysostosis, hypophosphatasia, congenital hypothyroidism, among other entities that share common clinical features.⁽²⁾

Ear nose and throat complications, such as recurrent sinus infections and other upper-airway complications are significantly more often observed in individuals with CCD than in the general population, as they present abnormally small or nonexistent maxillary sinuses, hypoplastic zygomatic bones and patency of the mandibular symphysis.⁽⁶⁾

Some children with this condition are slightly delayed in the development of motor skills such as crawling and walking, but intelligence is unaffected.⁽⁷⁾

Long-term follow-up includes a multidisciplinary care, comprising orthodontics, maxillofacial and orthopedics care, among other specialties according to the different clinical features of CCD presented.

ABSTRACT

Cleidocranial dysplasia (CCD) is a rare autosomal dominant disorder mostly affecting bone and cartilage growth, as well as tooth formation. The most common skeletal abnormality in affected individuals are hypoplastic/aplastic clavicles. Successful treatment and management patients with CCD require a multidisciplinary team of healthcare specialists.

Keywords: cleidocranial dysplasia; clavicles; cranial fontanelle; ossification

AUTHORSHIP

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