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Dr. Olatunji Oladapo Babalola
MBBS, FWACS, MRCS England, MPH Consultant Trauma & Orthopaedic Surgeon, The Comforter Hospital, Lagos, Nigeria

Dr. Ifeanyi Agwulonu
MBBS, FMC Ortho, Consultant Orthopaedic & Trauma Surgeon, Department of Orthopaedic & Trauma Surgery, Benjamin Carson (Snr.) School of Medicine, Babcock University Ilishan Remo, Ogun State, Nigeria

Corresponding Author:
Dr. Olatunji Oladapo Babalola
MBBS, FWACS, MRCS England, MPH Consultant Trauma & Orthopaedic Surgeon, the Comforter Hospital, Lagos, Nigeria

Cleidocranial dysostosis: Orthopaedic presentation as a shoulder deformity

Dr. Olatunji Oladapo Babalola and Dr. Ifeanyi Agwulonu

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Abstract
Cleidocranial dysostosis is a rare form of skeletal dysplasia inherited in an autosomal dominant fashion with high penetrance and variable expressivity. It is characterized by a range of skeletal abnormalities affecting mostly bone that undergoes intramembranous ossification. Here we present a case report of CCD in a male with no family history, who complained of reduced ability to raise heavy object with the left upper limb and deformity of the left shoulder. There is an absence of the medial half of the ipsilateral clavicle. He was counselled on the aetiology and nature of the disease and had sessions of physiotherapy with satisfactory outcome.

Keywords: Cleidocranial dysostosis, skeletal dysplasia, absent clavicle, case report

Introduction
Cleidocranial dysostosis (CCD) is a rare skeletal dysplasia marked by a range of skeletal abnormalities, including short height, malformed clavicles, patent sutures and fontanelles, and too many teeth [1]. First described in 1898 by Mane and Sainton [2], other names for it include Cleidocranial dysplasia, Mutational dysostosis, Scheuthauer-Marie-Sainton syndrome, and Marie and Sainton disease [3].
It has a prevalence of 0.5 per 100,000 live births and is an uncommon congenital disorder with autosomal dominant inheritance [4, 5]. The CCD gene has been mapped to chromosome 6p21, the region contains α1 a core binding factor (CBFA 1) gene [6]. CBFA1 is required for both membranous and endochondral bone development and it also regulates the differentiation of precursor cells into osteoblast, this may be connected to delayed ossification of the teeth, pelvis and the skull [6]. It primarily affects bones that undergo intramembranous ossification [2]. It is established that CCD is caused by mutations with high penetrance and considerable variability [7], nevertheless the phenotype is often incomplete, and the clinical picture varies, even within families [1]. Since it presents with a range of clinical and radiological characteristics, diagnosis can be difficult [7]. Many people who have hypoplastic or even missing clavicles have managed to live normal lives and even work as manual labourers without experiencing any handicap because of their condition [1]. A familial case has been reported in a Chinese family [8].

Case Report
History
A.J. an 18-year-old right hand dominant male university undergraduate presented with a 5-year history of inability to lift heavy objects with the left arm, left shoulder deformity and upper chest wall asymmetry (with the abnormality on the left side). The onset of the symptoms was insidious, and the patient was concerned that it made him look awkward and uncomfortable when removing his clothes among his peers. There was no pain, no history of trauma, infections or tumour that could lead to an early arrest of growth of structures around the left shoulder girdle. There were no delayed eruption of the teeth nor supernumerary tooth. He is the third in a non-consanguineous marriage and there was no positive family history. Maternal age was 33 years at his birth. Antenatal period, labour and delivery were uneventful. Developmental milestones were achieved at the appropriate time with no delay. There were no ear symptoms, his father confirmed the presence of the deformity around the

left collar from birth except that they were subtle, and the parent thought that the abnormality would disappear with growth.

Physical Examination

His overall growth was not affected, he was 5 feet 8 inches tall. He had a triangular face and no demonstrable cranial nerve palsy. The deltoid contour on the ipsilateral side was attenuated with a corresponding reduced deltoid muscle bulk. There was also a reduction in the bulk of the left pectoralis muscles compared to the right. There was obvious asymmetry of the shoulder with the left at a lower level than the right. There was no Sprengel shoulder on the left, and no discrepancy in the length of both upper limbs. However, there was a slight reduction in the muscle bulk of the left arm compared to the right. The left clavicle was not palpable in its medial half, and there was no palpable left sternoclavicular joint. The left shoulder girdle (Clavicular length) was shorter than the right by about 3cm. Power in the left upper limb group of muscles were 5/5 with normal sensation and reflexes. There was no ptosis, no myosis and no anhidrosis. There were no abnormalities in the vertebral column and lower limbs. His teeth were normally formed, and he had normal secondary sexual characteristics.

Figures 1 to 3 are clinical photographs.

Fig 1: Clinical photograph

Fig 2: Clinical photograph

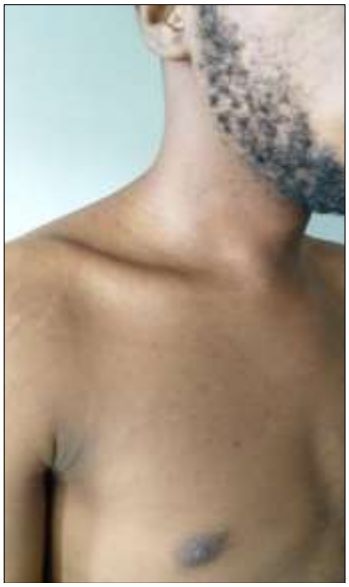

Fig 3: Clinical photograph

Radiology

A plain radiograph done showed absence of the medial half of the left clavicle. This is shown in figure 4.

Fig 4: X ray shows absence of the medial half of the left clavicle

A CT scan with three-dimensional reconstruction revealed the complete absence of the medial half of the left clavicle with no left sternoclavicular joint. The proximal four ribs on the left were also defective in their anterior ends with no corresponding sternocostal joints visible on the CT scan. There were no cranial or vertebra anomalies seen.

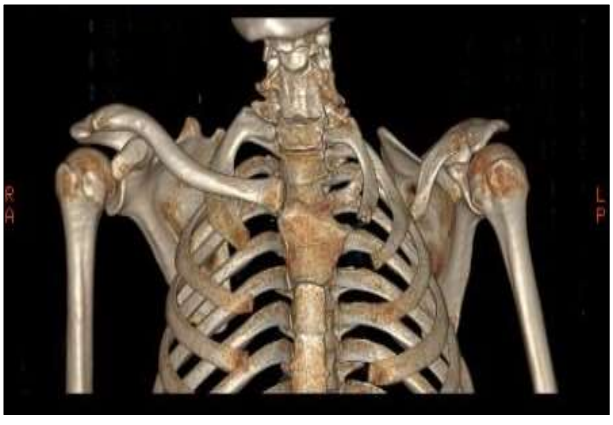

Fig 5: CT scan three-dimensional reconstruction

Treatment

The patient was counselled on the aetiology of the condition and had sessions of physiotherapy. His self-esteem improved considerably after the counselling sessions, and he remarked that "being educated on the aetiology and nature of the condition was helpful in alleviating his anxiety". He is regularly being seen at the follow up clinic.

Discussion

Skeletal hereditary illnesses are a complex and heterogeneous group of genetic disorders [1]. The underlying mechanisms of skeletal development, patterning, bone and cartilage creation, growth, and homeostasis are reflected in the various clinical presentations [1]. Since the publications of Meckel in 1760 and Martin in 1765, instances of CCD have been recorded [9]. Marie and Sainton in 1898 coined the name "dysostose cléido-crânienne héréditaire" for the condition [10]. A study described the descendants of a Chinese man named Arnold and of the 356 members of his family which were traced, 70 were affected by the "Arnold Head" [11]. A case of CCD in which the patient's father and daughter were also affected has also been described [2]. The condition's intrafamilial variances and the importance of clinical evaluation of family members following the birth of an apparently sporadic case has been emphasized [12]. CCD was found with varied presentations in three generations of a family (The mother, aunt and grandmother) after the birth of a propositus who presented with respiratory distress and hypoplastic clavicles [12]. Our patient had no family member affected.

Like any other hereditary disorder or defect, cleidocranial dysostosis might manifest all the characteristics or only a few nearly subclinical ones [13]. It has been shown that complete clavicular absence is rare whereas hypoplasia is commoner at the acromial end of the clavicle and rare at the sternal end [1]. Our patient presented with absence of the sternal half of the clavicle. It is also noted that bilaterality is the rule but not always the case [1], our patient however presented with unilateral involvement.

A range of other syndromes are associated with clavicular hypoplasia or agenesis, and they include pseudoarthrosis of the clavicle [13]. In clavicular pseudoarthrosis however, the hypoplasia or absence of one of the clavicles usually is on the right side [2]. Pyknodysostosis and Gardner syndrome are other differential diagnosis.

In conclusion CCD can present with only a few of its characteristic features, hence a high index of suspicion is important in clinching the diagnosis. Radiologic investigations are important tools for its diagnosis and in patients with only subtle abnormalities, counselling and physiotherapy may be all that would be necessary like in our index patient.

Conflict of Interest

Not available.

Financial Support

Not available.

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