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CLEIDOCRANIAL DYSPLASIA IN A 12-YEAR-OLD MALE PATIENT: A CASE REPORT

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ABSTRACT

Cleidocranial dysplasia (CCD) is a rare congenital skeletal disorder characterized by defective intramembranous ossification, most notably involving the clavicles and craniofacial bones. It is commonly associated with mutations in the Runx2 gene. Diagnosis is primarily clinical, based on the recognition of characteristic skeletal features, and is supported by radiological imaging, particularly X-rays.

The objective of this report is to describe and report an incidental diagnosis of CCD in a 12-year-old male patient who presented with respiratory symptoms and underwent a chest X-ray for suspected chest infection at a diagnostic center in Osogbo, Nigeria.

In the case we report, patient's lungs and cardiac silhouette were unremarkable, careful image evaluation revealed bilateral absence of the clavicles. Correlation with physical findings, including excessive shoulder mobility and narrow, drooping shoulders, confirmed the diagnosis of CCD.

This case highlights the importance of radiographer vigilance, thorough image assessment beyond the primary clinical indication, and the potential to identify rare skeletal disorders during routine imaging examinations.

Keywords: Cleidocranial dysplasia, RUNX2 gene, Clavicle absence, Incidental diagnosis.

Introduction

Cleidocranial dysplasia (CCD) is a rare congenital skeletal dysplasia caused by defective intramembranous ossification, primarily affecting the clavicles and craniofacial bones [(1)]. It follows an autosomal dominant inheritance pattern, most commonly due to mutations in the *Runx*2 (CBFA1) gene on chromosome 6p21, which is essential for osteoblast differentiation and bone formation. Typical features include partial or complete clavicular absence, delayed closure of cranial sutures and fontanelles, frontal bossing, excessive shoulder mobility, and dental anomalies such as delayed permanent tooth eruption and multiple supernumerary teeth in over 90% of cases; intelligence and general health are usually preserved [(2,3,4)].

With a global prevalence of approximately 1 per million, though higher in some populations due to founder effects with clavicular defects occurring in more than 80% of cases and serve as a key radiological hallmark [(5)]. Despite these characteristics, diagnosis is often delayed or missed if features are subtle. Radiographic imaging is crucial, as clavicular hypoplasia or aplasia may appear on routine chest radiographs, yet abnormalities are frequently overlooked when imaging targets unrelated thoracic issues [(6,7)]. Incidental CCD detection remains uncommon but clinically significant. We report such a diagnosis in a 12-year-old male during chest radiography for respiratory symptoms, highlighting the need for radiographer vigilance and thorough image evaluation.

CASE REPORT

A 12-year-old male presented to the radiology department with a history of intermittent chest pain, persistent cough, and mild breathing discomfort of several weeks' duration. There was no known history of trauma, prior chest surgery, or congenital anomaly, and the antenatal and perinatal histories were reportedly unremarkable. Systemic inquiry

was otherwise non-contributory, and there was no documented family history of skeletal deformities or similar complaints at the time of imaging.

A PA chest radiograph was requested to evaluate suspected chest infection. On general observation during positioning, the radiographer noted that the patient had narrow, drooping shoulders and a relatively slender upper thorax, with unusual laxity of the shoulder girdle. The patient was able to voluntarily approximate both shoulders anteriorly in front of the chest with minimal discomfort, a feature classically reported in patients with clavicular aplasia or hypoplasia [8].

The chest radiograph demonstrated clear lung fields with normal pulmonary vascular markings and no evidence of consolidation, pleural effusion, or pneumothorax. The cardiac silhouette and mediastinum appeared within normal limits for age. However, scrutiny of the bony structures revealed bilateral absence or marked hypoplasia of the clavicles: only small medial remnants were suspected, and the typical continuous bony contour from the sternum to the acromion was not visualized. The visualized ribs and thoracic vertebrae appeared grossly unremarkable. No obvious skull, rib, or vertebral anomalies could be assessed on this projection alone.

In the context of the radiographic appearance and the physical findings of sloping, narrow shoulders and excessive shoulder mobility, a provisional diagnosis of Cleidocranial dysplasia was made. The findings were promptly communicated to the referring clinician, and further evaluation, including detailed musculoskeletal and dental examination, family screening, and genetic consultation, was recommended. The patient was advised to return for follow-up imaging of the skull, pelvis, and dentition, but these investigations were beyond the scope of the present report.

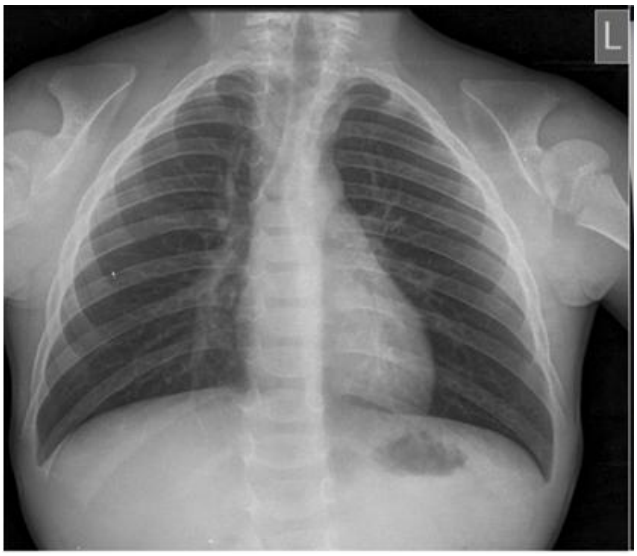

Fig.1: Postero-anterior chest radiograph of the 12-year-old patient demonstrating bilateral absence of the clavicles with normal lung fields and cardiac silhouette, consistent with cleidocranial dysplasia

DISCUSSION

Cleidocranial dysplasia (CCD) is a rare skeletal disorder with an estimated prevalence of approximately 1 per 1,000,000 individuals [9]. It is typically inherited in an autosomal dominant pattern due to mutations in the *Runx2* gene on chromosome 6p21, though up to 40% of cases arise from de novo mutations [10]. The hallmark radiographic feature of CCD is partial or complete absence of the clavicles, which enables the characteristic ability of affected individuals to approximate their shoulders anteriorly; an observation first described by Pierre Marie and Paul Sainton in 1898 [7, 11,12].

Additional skeletal manifestations often include delayed closure of fontanelles, widened cranial sutures, hypoplastic mid-facial structures, and supernumerary teeth, underscoring the multisystem nature of the condition.

In the present case, a 12-year-old male presented with chest pain, persistent cough, and mild breathing discomfort, prompting a postero-anterior (PA) chest radiograph to evaluate for suspected chest infection. On the radiograph, the lungs and heart appeared unremarkable, aligning with the absence of primary pulmonary pathology. However, comprehensive image review incidentally revealed bilateral absence of the clavicles (Figure 1), a finding not initially considered given the respiratory-focused clinical presentation and lack of musculoskeletal complaints. This scenario exemplifies how incidental findings on imaging can unveil unrelated pathologies, emphasizing the importance of scrutinizing all visible anatomy beyond the referral indication [13, 14].

Radiographers are pivotal in detecting such incidental findings. During patient positioning, physical cues like shoulder hypermobility or subtle dysmorphic features can alert the radiographer to anomalies. In this instance, routine scrutiny of the apices and superior mediastinum disclosed the clavicular hypoplasia, prompting immediate flagging to the reporting clinician. Literature reviews confirm that up to 20% of skeletal dysplasia are first identified on chest radiographs ordered for unrelated symptoms [15].

Early diagnosis of CCD holds substantial clinical value. Untreated, patients face complications including recurrent otitis media, dental overcrowding requiring extractions, short stature, and orthopedic issues like scoliosis or hip dysplasia [16]. Multidisciplinary management encompasses genetic counseling for familial risks, orthodontic interventions by adolescence, and orthopedic surveillance improves quality of life and mitigates morbidity.

For this patient, the incidental discovery facilitated prompt report to the referral, enabling comprehensive assessment. In resource-limited settings like Nigeria, where access to genetic testing remains challenging, such vigilant imaging practices bridge gaps in proactive screening. This case reinforces holistic image evaluation and incidental finding protocols.

CONCLUSION

This case report describes the incidental discovery of cleidocranial dysplasia in a pediatric patient during routine chest radiography. The rare skeletal disorder was identified through meticulous image review and correlation with physical findings, despite the exam being requested for respiratory symptoms with no pulmonary pathology evident. In patients undergoing routine chest radiography, absent or hypoplastic clavicles should prompt suspicion for cleidocranial dysplasia. Meticulous image review and correlation with physical findings can enable early diagnosis of this rare condition.

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