

## CASE REPORT

### Cleidocranial Dysplasia, Radiological Findings in a New-Born (a case report)

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#### ABSTRACT

**BACKGROUND:** Cleidocranial dysplasia (CCD) is a rare autosomal dominant skeletal disorder presenting with a variety of clinical and radiological features that can prove to be a diagnostic challenge.

**CASE PRESENTATION:** We report a case of CCD in a female new-born from a well-followed pregnancy, without prenatal ultrasound, presenting a too soft skull. The diagnosis was made with radiographs. There were no major complication and this presentation of

CCD was compatible with life.

**CONCLUSION:** The diagnosis of CCD is based on clinical and radiographic findings, confirmation is possible if molecular genetics test available.

**Key words:** Cleidocranial dysplasia; Radiography; New-born

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#### Abbreviations

CCD: Cleidocranial dysplasia; CT: Computed tomography; 3D: Three dimension.

#### BACKGROUND

Cleidocranial dysplasia (CCD) is a rare autosomal dominant skeletal disorder presenting with a variety of clinical and radiological features that can prove to be a diagnostic challenge<sup>[1,2]</sup>. The diagnosis of CCD is based on clinical and radiographic findings<sup>[3]</sup>. The early history of CCD goes back to prehistorical times, by virtue of a possible example of CCD in a Neanderthal skull, which was documented in 1933 by Greig, a Scottish surgeon who became curator of the Museum of the Royal College of Surgeons of Edinburgh<sup>[4,5]</sup>. Several other historical cases are reported<sup>[5]</sup>. The earliest recognizable report of CCD in the medical literature date from 1760<sup>[5]</sup>. By the end of the last millennium, mutations in the RUNX2 gene were identified as the molecular aetiology of this this rare autosomal dominant inherited disorder<sup>[1]</sup>. We report a rare case of CCD without major complication, compatible with life.

#### CASE PRESENTATION

A first female new-born of 3-weeks from a non-consanguineous couple, borne by vaginal delivery without incident after 40 weeks



**Figure 1** Skull anteroposterior (a) and lateral (b) radiographs showing absence of ossification of both parietal bones (*arrow*), too wide fontanels and cranial sutures.

of well-followed pregnancy, without any prenatal ultrasonography performed. She was 3150 g weight and 54 cm length on birth. She was brought to the paediatrician's consultation for too soft consistency of the skull. The examination of the head shows a normal outer appearance with an intact skin but felt soft when palpated.

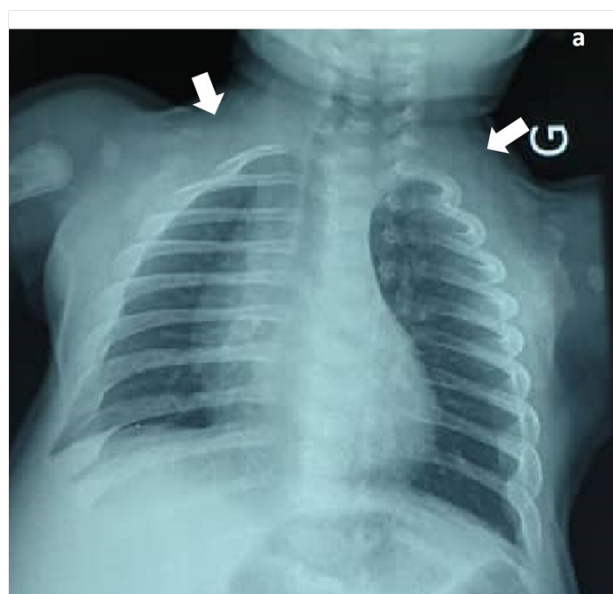
A transfontanellar ultrasound performed was normal. An anteroposterior, lateral skull radiographs and whole-body radiographs were performed showing absence of ossification of both parietal bones (Figure 1) and bilateral agenesis of clavicles (Figure 2). There were no limbs and spine bone's abnormalities (Figure 3). CCD was suspected by the clinical geneticist. The family investigation identified no other members with bone abnormalities.

## DISCUSSION AND CONCLUSIONS

Cleidocranial dysplasia (CCD), also known as Scheuthauer Marie-Sainton Syndrome, is a rare autosomal dominant inherited disorder, characterized by general retardation in bone ossification, hypoplastic clavicles and various craniofacial and dental abnormalities<sup>[6]</sup>.

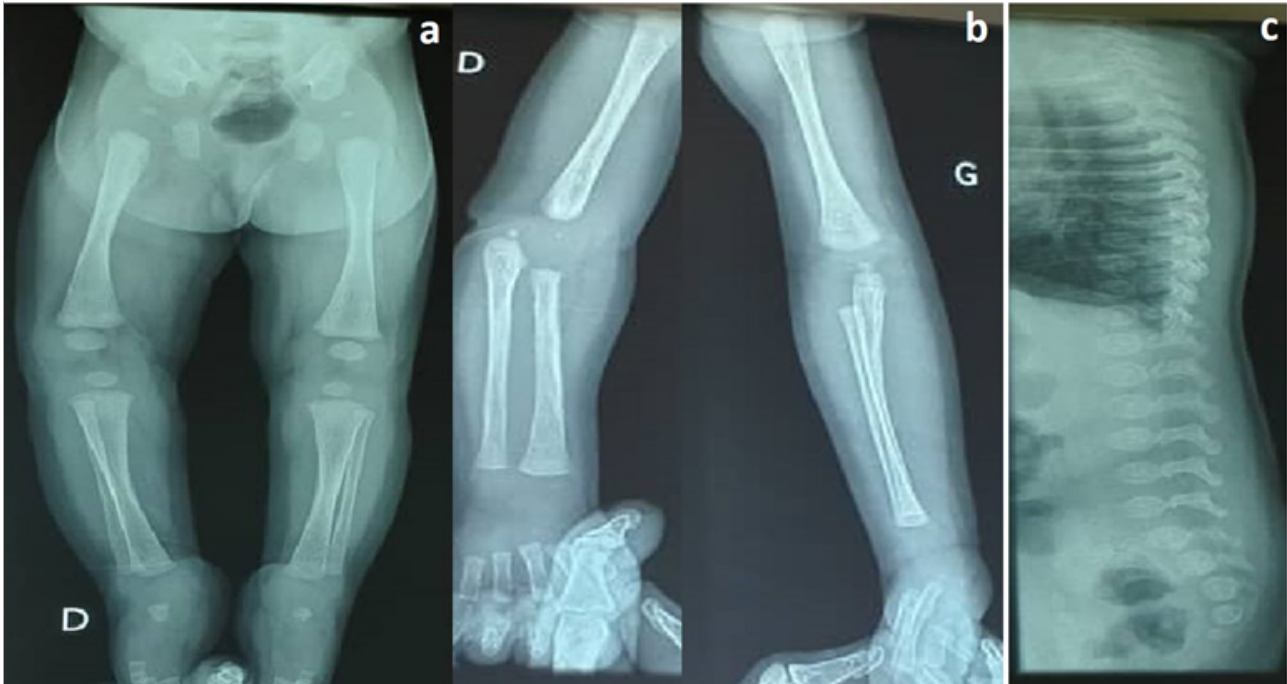
The variety of clinical and radiological features of CCD can be a real diagnostic challenge. The combination of hypoplastic clavicles and delayed closure of fontanels were suggestive of CCD<sup>[3]</sup>. The presence of clavicular hypoplasia is strongly suggestive of CCD, but this anomaly can also occur as an isolated nonsyndromic entity, which is usually unilateral<sup>[5]</sup>.

Complete absence of both clavicles may suggest the Yunis-Varon syndrome but in this rare genetic disorder, intellectual dysfunction and anomalies of the hands and feet are associated with malformations in other systems<sup>[7]</sup>. Delayed closure of fontanels,



**Figure 2** Chest radiograph presenting bilateral clavicles agenesis (*arrows*).

Wormian bones, and hypoplastic clavicles are also features of pyknodysostosis, mandibuloacral dysplasia and Yunis-Varon syndrome<sup>[1]</sup>. The finding of increased bone density (osteosclerosis, with risk of fractures) and dwarfism distinguishes pyknodysostosis from CCD. Defective cranial ossification leading to patency of the anterior fontanels and Wormian bones in the sutures is an important feature of CCD. Similar manifestations occur in osteogenesis imperfecta (frequent fractures), pycnodysostosis (skeletal density),



**Figure 3** Pelvic and low limbs (a), upper limbs (b) and lateral spine (c) radiographs, absence of bones abnormalities of limbs and the spine.

and congenital hypothyroidism (disturbed thyroid metabolism)<sup>[5]</sup>.

Radiologic findings are important in the diagnostic management of CCD. In postnatal, a whole-body radiograph of an infant (babygram) is required for any live-born infant, preterm foetus or stillborn with a suspected constitutional disorder of bone<sup>[8]</sup>. Some bone abnormalities are best appreciated on 3D whole-body CT images<sup>[9]</sup>. The CT should be carried out in low dose<sup>[3]</sup>. In families with a parent affected, the risk is 50% for each pregnancy. Further studies have shown the contribution of prenatal ultrasound in the diagnosis of CCD between 12 and 15 weeks of pregnancy<sup>[10-14]</sup>.

## ACKNOWLEDGMENTS

### Authors' contributions

MT and PBL designed the study and wrote the initial draft of the manuscript. PG, BK, LS contributed to the design of the study, data collection and assisted in the preparation of the manuscript. LA and KA contributed to the interpretation and critically reviewed the manuscript. All authors approved the final version of the manuscript and have agreed to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved. The authors would like to thank Dr BRUCE and Dr RIVOAL, paediatricians for their support in the diagnosis

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