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CASE REPORT

Cleidocranial Dysplasia: A Rare Case Report

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Cleidocranial dysplasia (CCD) is a rare autosomal dominant skeletal disorder, characterized by delayed closure of anterior fontanelle, absent or hypoplastic clavicles, dental problems, and short stature. Usually, the presenting complaints are open anterior fontanelle and dental abnormalities. We hereby present a 5-year-old Indian child who presented to us with the complaints of persistently open anterior fontanelle and short stature. A detailed, thorough examination and high degree of suspicion in a child presenting with persistent open fontanelle has a great importance in the diagnosis of a genetic syndrome like CCD.

Key words: Open anterior fontanelle, short stature, cleidocranial dysplasia

INTRODUCTION

Cleidocranial dysplasia (CCD) (OMIM 11,960) is a rare skeletal disorder which is characterized by delayed closure of anterior fontanelle, absent or hypoplastic clavicles, dental problems, and short stature. It usually inherited in autosomal dominant manner and caused by mutation in human osteoblast − specific, runt-related transcription factor 2 (RUNX2) gene which is located on chromosome 6p21.[1] CCD is also known as Marie–Sainton disease, mutational dysostosis, and cleidocranial dysostosis.[2]

CASE REPORT

A 5-year-old male child, second in birth order, born to nonconsanguineous couple, out of normal vaginal delivery presented to us with the complaints of open anterior fontanelle and short stature. His antenatal, natal, and postnatal period were uneventful. Developmental milestones and cognitive functions were appropriate for age. On examination, his anthropometry parameters were as follows: weight 12.8 kg (below 3rd percentile); height 98 cm (below 3rd percentile), and head circumference 50 cm (between 3rd and 50th percentile).

Physical examination revealed wide open anterior fontanelle measuring 4.5 cm × 4.5 cm and frontal bossing. There were no dental abnormalities. On asking the child to approximate both arms and shoulders, he brought his shoulders easily forward in midline [Figure 1]. Systemic examination was normal.

On laboratory investigations, thyroid profile, serum calcium, phosphorus levels, and 25-hydroxy cholecalciferol levels were normal. Chest roentgenogram revealed absent right clavicle with hypoplastic left clavicle and absent left 12th rib [Figure 2]. X-ray skull showed multiple wormian bones, widened sutures and patent anterior fontanel and occipitalization of C1 vertebrae [Figure 3]. X-ray pelvis revealed hypoplastic iliac bones and pseudo-widening of the pubic symphysis and also coxa vera [Figure 4]. Incidentally, X-ray of the left arm and forearm showed fracture of the distal end of the left humerus and resorption of terminal phalanx of the right thumb [Figure 5]. On further questioning, parents revealed child had a history of fall from height 8 months back, for that parents consulted orthopedic doctor and Plaster of Paris cast was applied, but proper follow-up was not taken.

On the basis of these clinical and radiological findings, the patient was diagnosed as a case of CCD. Genetic testing was offered but could not be performed because of poor affordability of parents.

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Figure 1: Approximation of shoulder due to absence/hypo plastic clavicles and prominent forehead

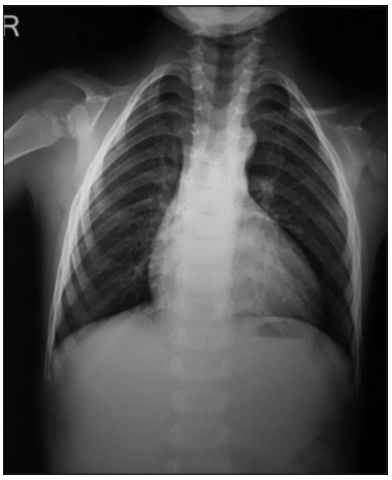

Figure 2: Chest roentgenogram showing absent right clavicle with hypoplastic left clavicle and absent left 12th rib

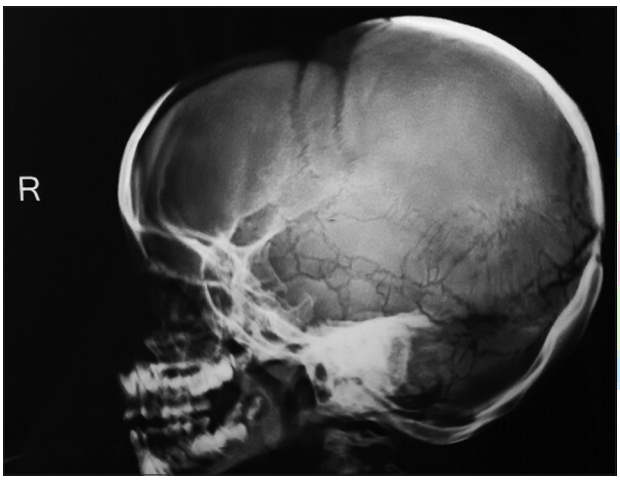

Figure 3: X-ray skull showing multiple wormian bones, widened sutures, and open anterior fontanelle and occipitalization of C1 vertebrae

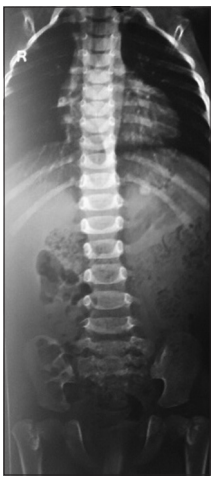

Figure 4: Hypo plastic iliac bones and pseudo widening of pubis symphysis

DISCUSSION

Delayed closure of anterior fontanelle and frontal bossing are very common symptoms and signs in children. These symptoms also have great emphasis in developing countries like India as undernutrition and Vitamin D deficiency is very prevalent in these countries.

A detailed and thorough clinical examination is essential to every child presenting with these symptoms. The diagnosis of CCD is primarily based on clinical features as these children have characteristic findings which can be easily point out.

The incidence of CCD is one per million births. CCD is caused by a heterozygous loss-of-function mutation in the RUNX2 gene, encoding transcription factor CBFA1, and located on chromosome 6p21. Approximately 40% individuals with CCD are sporadic cases, having unaffected family members or parents suggesting *de novo* mutation in the affected individual.[3,4] This child was also a sporadic case.

Consistent with features described in literature, present case also shared similar clinical features. CCD usually presented as a generalized skeletal dysplasia. The major affected bones are those which undergo intramembranous ossification such as cranial vault, clavicles, maxilla, nasal, and lacrimal bones.[5] It is characterized by the absence of the clavicles, which usually occurs in 10% of cases or the presence of hypoplastic clavicles which allow the hypermobility of shoulders that can move it up to the medial plane of the body. Absent clavicle may also present as an isolated anomaly, but this is usually unilateral and does not cause any impairment. Yunis-Varon syndrome (OMIM 216,340) also has a feature of complete absence of both clavicles but intellectual disability, anomalies of the hands and feet and malformations in other systems also present in this syndrome.[6,7]

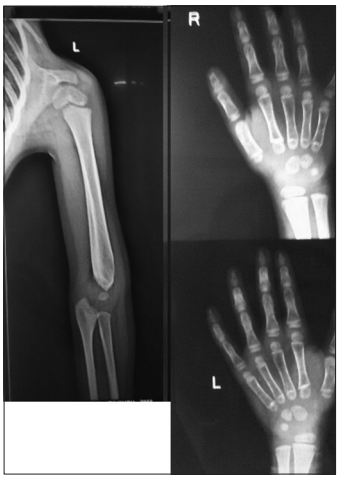

Figure 5: X-ray of left arm and forearm showed fracture of distal end of left humerus and resorption of terminal phalanx of the right thumb

The dental abnormalities described in CCD are supernumerary teeth, prolonged retention of the primary dentition, failed eruption of the permanent teeth, multiple crown and root abnormalities, crypt formation around impacted teeth, and ectopic locations of teeth.[8] At present, as this child still had primary dentition, natural history and complications of this syndrome was explained to parents, and regular follow-up ensured.

Management involves multidisciplinary approach involving dentist, orthopedician, and pediatrician. Early diagnosis is essential to prevent morbidity and subjecting the patient to early intervention program.

CONCLUSION

High degree of suspicion in a child presenting with persistent open fontanelle has a great importance in the diagnosis of a genetic syndrome like CCD. Early detection is essential for prevention of morbidity and disability. A detailed and thorough clinical examination is essential for every child to prevent the missing cases like CCD.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Nil.

Conflicts of interest

There are no conflicts of interest.

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