

Cleidocranial Dysplasia - Report of A Case

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ABSTRACT

Cleidocranial dysplasia is a rare autosomal dominant disorder characterized by hypoplastic or absent clavicles, large fontanelles, dental anomalies and delayed skeletal development. A case of an eleven year old boy with this syndrome is reported having multiple supernumerary teeth, retained deciduous dentition, non-eruption of permanent dentition along with hypoplasia of orofacial and skeletal structures.

Key Words: Cleidocranial dysplasia, Supernumerary teeth, Retained teeth, Impacted teeth.

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Cleidocranial dysplasia (CCD), also known as Marie and Sainton's disease; Scheuthauer - Marie - Sainton syndrome; Mutational Dysostosis, is a rare congenital defect of autosomal dominant inheritance affecting both sexes with equal frequency.(1) The etiology of CCD is not known. Cytogenic abnormalities on chromosome no. 6 is suggested.(2-4) The condition has been mapped to a microdeletion of chromosome band 6p21, t(6;18) (p12; q24) translocation and pericentric inversion of chromosome.6 The most striking features of this syndrome are partial or complete absence (in about 10% of cases) of clavicles causing unusual mobility of the shoulders and late closure of fontanelles resulting in frontal bossing. It is of clinical significance to the dentist due to involvement of facial bones, altered eruption patterns and presence of multiple supernumerary teeth.(5)

Case Report

An eleven year old boy along with his mother reported to the department of Pedodontics with the complaint of delay in eruption of lower anterior permanent teeth. His medical and family histories were non-contributory. On general examination the patient had normal height, weight and I.Q. Brachycephaly, frontal bossing, depressed mid-face profile and hypertelorism were noticed. The right shoulder was markedly drooped. He was asked to attempt to bring his shoulders forward towards midline to check for incomplete clavicle formation; and he was able to easily perform this procedure (Fig. 1). The mother was surprised that his son could do this. Oral examination showed prolonged retention of deciduous teeth and failure of permanent mandibular lateral incisors to erupt (Fig. 2).

Following dentition with bilateral Angle's Class III malocclusion was present:

16, 55, 54, 53, 52, 51	61, 62, 63, 64, 65, 26
46, 85, 84, 83, 41	31, 73, 74, 75, 36



Fig. 1: Characteristic view of CCD patient, adduction of shoulders



Fig. 2: Intraoral photograph showing facial view of dentition presenting prolonged retention of primary teeth

The patient was subjected to critical radiographic examination of chest, skull, pelvis, spine, major joints and long bones. Cephalometric and panoramic radiographs with supplementary periapical and occlusal views were done. The X-rays revealed the following positive findings:

Skull (Fig. 3 & 4): Open sutures, large wormian bones, calvarial thickening and characteristically sunken sagittal suture giving the skull a flat appearance.

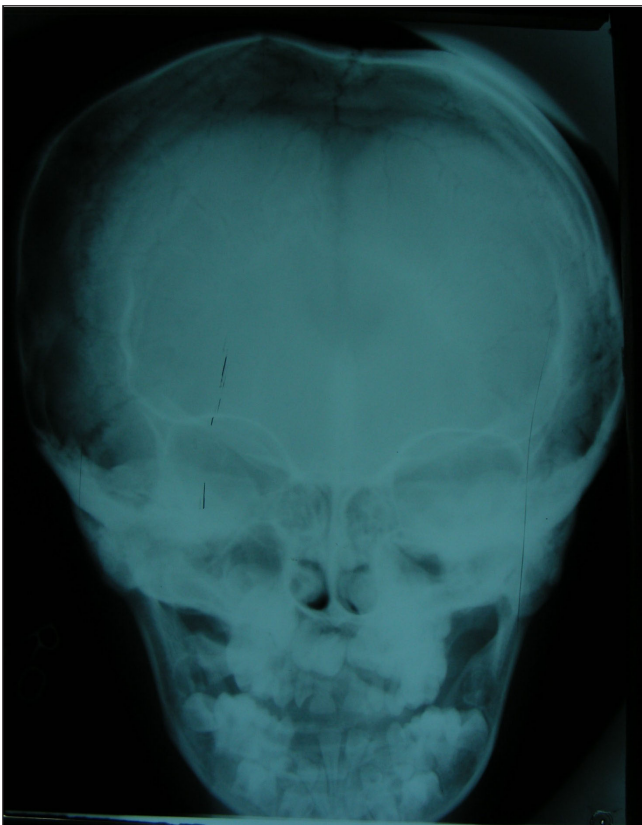


Fig. 4: Anteroposterior view of skull showing sunken sagittal suture

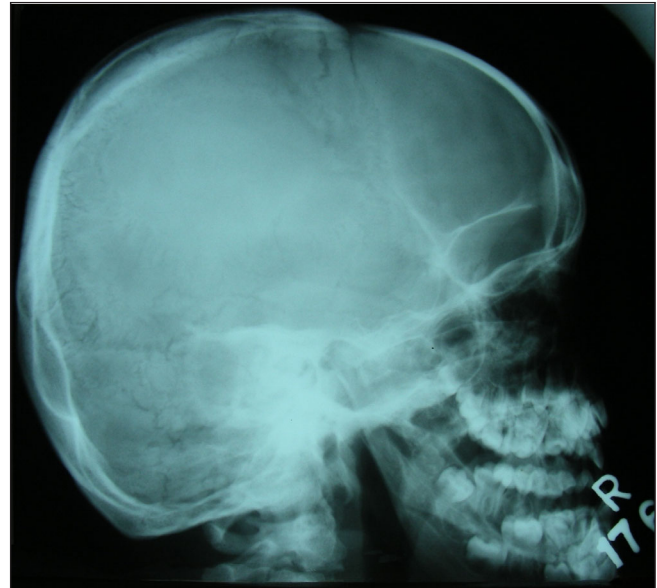


Fig. 3: Lateral view of skull showing broad sutures and wormian bones

Chest (Fig. 5): Right clavicle was absent while left clavicle was hypoplastic.

Hands (Fig. 6): Metacarpal pseudoepiphyses

Pelvis (Fig. 7): Wide symphysis pubis, broad femoral heads with short femoral necks and osteosclerosis.

Dental Findings

Cephalogram (Fig. 8) showed hypoplastic nasal bone and maxilla while the mandible was prognathic having characteristic obtuse mandibular angle. OPG (Fig. 9) and intraoral (occlusal and periapical) radiographs revealed abnormally retained



Fig. 5: Chest radiograph demonstrating partial absence of left clavicle, right clavicle absent

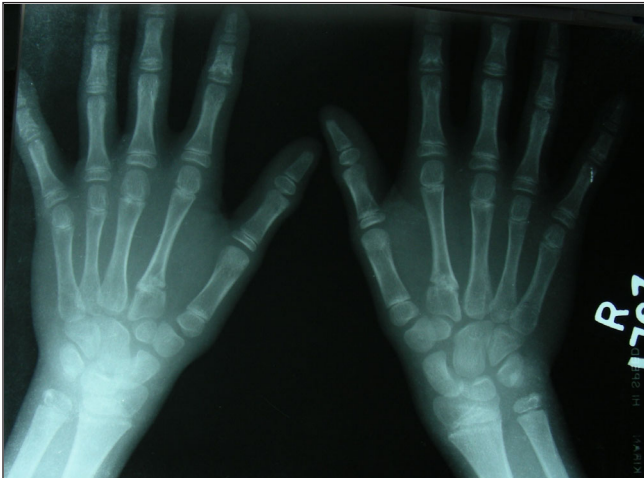


Fig. 6: Hand and wrist radiography showing pseudoepiphyseal changes of second metacarpal

primary teeth, nearly thirty six impacted permanent and supernumerary teeth (excluding second and third molars). There were eighteen supernumeraries in all, nine in each jaw.

Discussion

The clinical findings of CCD, although present at birth, are often either missed or diagnosed at a much later date. The radiographic evaluation of patients is the most important and reliable means to confirm the diagnosis. CCD is characterized by abnormalities of the skull, teeth, jaws, and shoulders girdles as well as by occasional stunting of the long bones. Large fontanelles, broad sutures, numerous wormian bones and brachycephalic head is pathognomic for the condition.

Patients with CCD exhibit a high, narrow, arched palate and an actual cleft palate appears to be common. One of the outstanding oral finding is prolonged retention of the deciduous teeth and subsequent delay in eruption of the



Fig. 7: Pelvic bone anomalies seen on radiographic examination

succedaneous teeth.(1) Scientists have postulated various views regarding the etiology of non-eruption, such as lack of cellular cementum,(6) defectiveness in post cementum formation,(7) presence of thick connective tissue between oral epithelium and dental follicle,(8) delayed tooth formation and maturation.(9) Furthermore, it is characteristic for numerous unerupted supernumerary teeth to be found by roentgenographic examination. These are most prevalent in the mandibular premolar and incisor areas.(1) The reason for the formation of multiple supernumerary teeth is still unknown. Skeletal Class III tendency / mandibular prognathism in CCD can be attributed to its uninterfered growth due to hypoplastic maxilla and upward and forward mandibular rotation.(10)

A disease known as pycnodysostosis or the Maroteaux - Lamy syndrome, which has most of the features of CCD has been described. However, patients with pycnodysostosis are also affected by dwarfism, their bones are dense and fragile and they have partial agenesis of the terminal phalanges of the hands and feet.(1)

Genetic counselling is appropriate for prospective parents with a family history of CCD or where one or both parents are



Fig. 8: Cephalometric radiograph showing mandibular prognathism

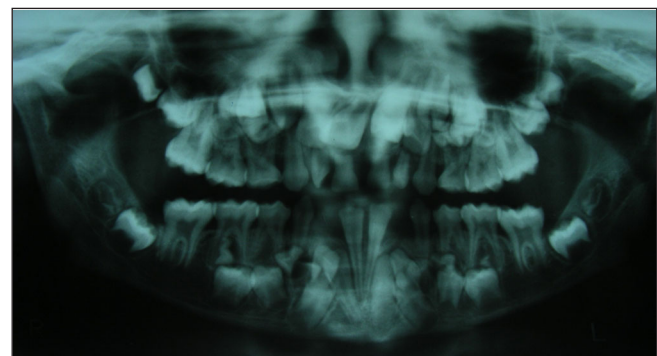


Fig. 9: OPG showing impacted permanent and supernumerary teeth

affected. There is no specific treatment for CCD, as the bony abnormalities cause little problem although care of the oral condition is important. The dental problems are the most significant complications. A multidisciplinary approach to treatment of these patients utilising a pedodontist, an orthodontist and an oral surgeon is recommended. The retained primary teeth should be restored if they become carious since extraction does not necessarily induce eruption of the permanent teeth.(1) The current mode of therapy for the dental anomalies is:

- Planned removal of nonresorbing primary teeth
- Surgical removal of supernumerary teeth
- Surgical exposure of permanent teeth
- Orthodontic alignment
- Growth modulation of maxilla and mandible with Delaire's face mask and chin cup, as per the requirement.
- When growth is complete consideration of orthognathic surgery in severe skeletal Class III malocclusion cases.

Treatment obviously extends over many years and clinicians should be aware of the child's compliance. Once the full permanent dentition has been brought into the mouth and into good alignment and function, follow up radiographs should be made to check for the development of new supernumerary teeth which seems to occur in the early to mid teens.(11)

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