

**Case Report****CLINICAL SPECTRUM OF CLEIDOCRANIAL DYSPLASIA: A CASE REPORT****P Sakhi<sup>1</sup>, P Yadav<sup>1</sup>, R Susmitha<sup>1</sup>, A Chawla<sup>1</sup>, CJ Yadav<sup>1</sup>, J Gupta<sup>2</sup>**<sup>1</sup>Department of Radio diagnosis, Sri Aurobindo Institute of Medical Sciences, Indore <sup>2</sup>R. D. Gardi Medical College, Ujjain**ABSTRACT**

**Background:** The developmental anomaly involving skeleton and teeth is Cleidocranial dysplasia. It is an autosomal disorder with equal sex distribution. It is also known as Marie and Sainton disease, Mutational dysostosis and Cleidocranial dysostosis. Skeletal defects of several bones are noted in this disorder in the form of partial or complete absence of clavicles, late closure of the fontanels, presence of open skull sutures and multiple wormian bones. **Case presentation:** In this case report, we describe an otherwise healthy 28-year-old male with a chief complaint of chronic pus discharge from the mandibular left canine and premolar edentulous region. **Conclusion:** Cleidocranial dysplasia is very rare in occurrence, incidence being 1: 1,000,000. Since early diagnosis of Cleidocranial dysplasia is essential for initiating the appropriate treatment approach, clinicians should be aware of the characteristic features. We report a case of Cleidocranial dysplasia because of its rarity.

**Keywords:** Autosomal Dominant, Marie and Sainton disease, Mutational dysostosis, Cleidocranial dysostosis.

**BACKGROUND**

Cleidocranial dysplasia is a rare congenital anomaly having multiple skeletal defects, the most striking of which are partial or complete absence of clavicles, late closure of the fontanels, presence of open skull sutures and multiple wormian bones.<sup>1,6</sup> Basal cell nevus syndrome and Crouzon syndrome also show late closure of fontanels but they can be easily differentially diagnosed from Cleidocranial dysplasia by considering other characteristic features. Frontal bossing, Brachycephaly and Hypertelorism is seen in this disorder as a result of delayed closure of anterior fontanel and metopic sutures and reduced growth of dysplastic skull base.<sup>5</sup>

Cleidocranial dysplasia was first described by Pierre Marie and Paul Sainton in 1898,<sup>4</sup> since then more than 1000 cases have been documented in the medical literature.<sup>5</sup> It is also known as Marie and Sainton disease, Mutational dysostosis and Cleidocranial dysostosis.<sup>3</sup> This disorder primarily affects bones showing intra-membranous ossification, i.e. calvarial bones and clavicles.<sup>1,2</sup>

Excessive mobility of the shoulder girdle is noted as clavicles are underdeveloped to varying degrees and are completely absent in approximately 10 percent of cases. Bell shaped small thoracic cage is noted having short ribs.

Maxilla and paranasal sinuses are underdeveloped. Other bones may also be affected including long bones, the vertebral column, the pelvis and the bones of hands and feet.<sup>7</sup> Characteristically, Adults with Cleidocranial dysplasia have mixed dentition in their oral cavities as there is prolonged

retention of deciduous dentition and delayed eruption of permanent teeth in them. Frequently they show a large number of unerupted supernumerary teeth<sup>8</sup>

**CASE REPORT**

A 28 year old male patient reported to the department of Oral and Maxillofacial Surgery, Sri Aurobindo college of Dentistry, Indore with the chief complaint of chronic pus discharge from the mandibular left canine and premolar edentulous region.

On examination, there was active pus discharge from the region of partially erupted canine and from the alveolus in the premolar region. There was a edentulous region on the right side of the mandible. The maxillary arch had a few missing teeth and some partially erupted premolars. The palate was very narrow and deep arched. The remaining teeth were affected with caries and generalised periodontitis.

The examination of the panoramic view of the jaws showed clusters of impacted supernumerary teeth in the edentulous areas, mostly resembling premolars. This is described as one of the classical features of the syndrome, Cleidocranial dysplasia. There were total of thirty seven teeth present and the patient also gave a history of extraction of two teeth in the maxillary arch. The mandibular arch showed the absence of bilateral gonial angles (banana shape).



Figure 1. Oral view showing numerous over-retained deciduous and some missing teeth.

Figure 2. Panoramic view of the jaws showing multiple unerupted supernumerary teeth mimicking premolar, missing gonial angles

On clinical examination, the patient had a severe concave profile and midline face deficiency. The maxilla Figure 3:- Under developed Maxilla and shorter resulting in mandibular pseudo prognathism. Bossing of frontal bone. Facial profile view of the patient demonstrating

hypermobility of the shoulder girdles and frontal bosselation was under developed and shorter resulting in mandibular pseudo-prognathism. Bossing of frontal bone and hypertelorism were also observed.



The approximation of humeral head and hypermobility of shoulders was noticed. The patient was of the short stature. The dental findings, the short stature, typical facial and skeletal anomalies of the skull and clavicle are pathognomic of Cleidocranial dysplasia and a diagnosis of CCD was established.

The patient was then referred to the Department of Radiology, Sri Aurobindo Institute of Medical Sciences for whole body skeletal survey for diagnosis. On radiographic evaluation the following findings were noted.

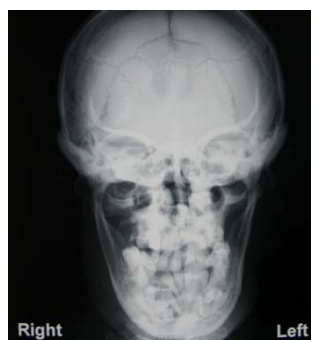


Figure 4 & 5 : - Radiograph of skull antero-posterior and lateral shows a) Widened frontanel with presence of wormian bones b) Nonfusion of saggital, coronal, and lambdoid suture of skull bones, c) Persistent metopic sutures, d) Large mandible with delayed and defective dentition.

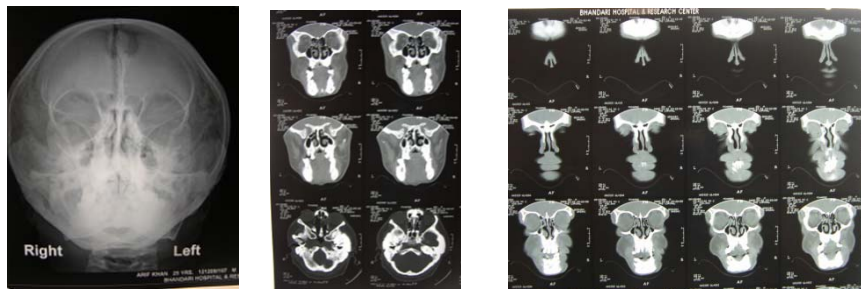


Figure 6, 7 & 8 : -Radiograph & CT scan of PNS shows a) Hypoplastic maxillary sinuses, frontal sinus and sphenoid sinus, b) There is high and narrow palate.



Figure 9 & 10- Radiograph of chest shows a) Hypoplastic lateral as well as medial end of left clavicle b) Hypoplastic lateral end of right clavicle c) Pseudo arthrosis of lateral end of right clavicle with scapula d) Shallow glenoid fossa of scapula bilaterally, e) Non fusion of acromian processes bilaterally. f) Bell shaped rib-cage.



Figure 11 : -Radiograph of pelvis with both hips showing a) Delayed ossification of bones forming symphysis pubis with diastasis of pubic symphysis, b) Hypoplastic iliac bones, c) Bilateral short femoral neck causing coxa vera deformity. Figure 12:-Radiograph of both hands shows a) Bilaterally elongated second metacarpal bone, b) Short hypoplastic distal phalanges of hand bilaterally, c) Pointed terminal tufts. Figure 13: - Radiographs of both feet shows a) Varus deformity of right foot is noted b) Short hypoplastic distal phalanx, c) Pointed terminal tufts.

A diagnosis of Cleidocranial dysplasia was confirmed and the patient was again referred to the department of Oral and Maxillofacial Surgery, Sri Aurobindo college of Dentistry, Indore. After the final diagnosis the surgical treatment was planned for the chronic problem that the patient was suffering from.

The OPG revealed a cluster of supernumerary teeth in the region of left mandibular canine and pre molar region. There was frank pus discharge from the distal crevicular surface of canine and severe tenderness. The patient was started on a course of antibiotics and the surgery was planned.

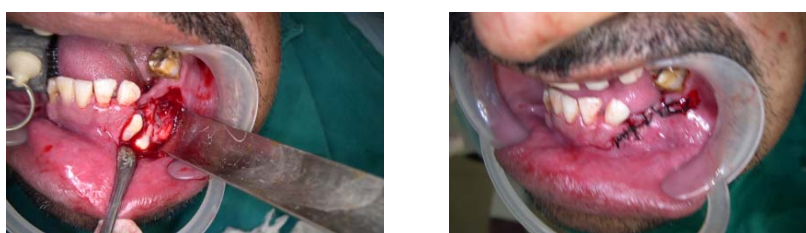


Figure 14 Surgical removals of infected impacted teeth.

## DISCUSSION

Frontal bossing and excessive mobility of shoulder girdle are the most significant clinical findings of Cleidocranial dysplasia.<sup>1,3</sup> Hypertelorism<sup>9</sup> is a common finding. Maxilla and paranasal sinuses are underdeveloped.<sup>10</sup>

Most important and reliable tool to confirm the diagnosis is radiographic evaluation of patients. Broad sutures, large fontanelles persisting into adulthood, numerous wormian bones and numerous unerupted supernumerary teeth<sup>1,3,11</sup> are the pathognomonic radiological findings of Cleidocranial dysplasia. Vertebral defects with scoliosis, kyphosis or lordosis, pelvic bony abnormalities and anomalies of phalangeal, tarsal, metatarsal, carpal and metacarpal bones are also present.<sup>3</sup>

Characteristic dental findings are numerous supernumerary teeth, particularly in mandibular premolar and maxillary anterior regions.<sup>12</sup> The removal of primary or supernumerary teeth does not usually promote eruption of unerupted permanent teeth.<sup>3</sup> The permanent molars generally erupt at proper time.<sup>1,3</sup>

## CONCLUSION

Cleidocranial dysplasia is generally missed or diagnosed at a much later time. This is generally diagnosed incidentally. Family history, excessive mobility of shoulders and radiographic pathognomonic findings of the chest, skull and jaws are useful in confirming the diagnosis.

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