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Original Research Paper

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# CLEIDOCRANIAL DYSPLASIA : CASE REPORT OF RARE CASE

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**ABSTRACT** cleidocranial dysplasia (CCD) is a rare congenital defect ,primarily affecting bones, undergoing intramembranous ossification.CCD presents with skeletal defects of several bones, the most striking feature of which are partial or complete absence of clavicles,late fontanels closure, and presence of open skull sutures and multiple wormain bones

**KEYWORDS :** Cleidocranial Dysplasia, Absent Clavicle,CBFA1 Gene

## INTRODUCTION:

Cleidocranial dysplasia(CCD) is a rare congenital defect ,primarily affecting bones, undergoing intramembranous ossification. CCD presents with a skeletal defects of several bones the most striking feature of which are partial or complete absence of clavicles late frontanels closure, and presence of open skull sutures and multiple wormain bones

## CASE REPORT:

A 10year old male patirnt reported with a complaint of not gaining weight. There was history of consangious marriage. Patients mother also stated that the patient mental status was not up to the mark he was dull in grasping things. The general examination revealed he was moderately built and moderately nourished patient. On the extraoral examination, the patient presented with flat face and occipital,parietal bossing, hypertelorism and sunken nasal bridge. Open fontanelle could be palpated in the anterior region of the head . Shoulders appeared narrowand showed marked drooping.

Patient was able to approximate both shoulders in the midline on the chest . Intra oral examination revealed the presence of a set of deciduous dentition and narrow high arched palate.a strawberry colored, fissuring on the dorsal anterior 2/3rd of tongue was observed.

## DISCUSSION:

CCD which is generally accepted as an autosomal disorder, was first described in 1765 by matrin. A gene for this disorder has been mapped to chromosome 6p21. The aetiology of cleidocranial dysplasia is unknown. Both dominant and recessive patterns of inheritance have been described. Spontaneous mutation occurs in 20-40% cases in the core binding factor alpha 1 (CDFA-1) gene. Located on chromosome 6p21. It may be discovered at any age ,but the cranial defeciencies noticed at birth. Both sexes are affected to an approximately equal extent. The defect often appears in several successive generations. The radiograph is the most important means by which the diagnosis of CCD can be confirmed . Chest radiograph with CCD Shows hypoplastic or aplastic clavicle

Radiographs of skull are pathognomic of the disease. The skulls show diffuse areas of rarefraction with most ossification in the frontal bones and least in the temporal and parietal bone. Cranial sutures are broad qith anomalous suture lines and the fontannels are large and persists into adulthood. Paranasal sinuses are usually underdeveloped and narrow In the majority of cases, results of blood analysis have been with in normal limits.

## CONCLUSION

CBFA1 was identified as the culprit gene underlying CCD. Cbfa1 controls the differentiation of precursor cells into osteoblasts, the cells that actually secrete bony matrix and thereby form bone. In addition, Cbfa1 plays a role in the regulation of chondrocytes differentiation during endochondral bone formation. Understanding this new ' master gene' will provide insight into the pathobiology of CCD as well as into the basic mechanisms of none formation

## MANAGEMENT:

Although there is no specific treatment for the patient with CCD, A multidisciplinary operation must be considered in the treatment of CCD and a long time approach must be planned. The time of the diagnosis of CCD may also be suggested as a guide for the choice of the necessary treatment model.

## IMAGES: