Original Research Paper

Volume - 11 | Issue - 08 | August - 2021 | PRINT ISSN No. 2249 - 555X | DOI : 10.36106/ijar

Pediatrics

CLEIDOCRANIAL DYSPLASIA (CCD): A CASE REPORT

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ABSTRACT It is a rare autosomal dominant disorder and affects 1 in 200,000 (1.) to 1 in 1,000,000 live births 2). The term cleido refers to clavicle; cranial refers to head; and dysplasia means "ill formed" One-third of the cases are due to sporadic mutations, but a rare recessive form has also been reported. Males and females of all ethnic groups are equally affected. Most of the cases have been found in Caucasians, Hindus, Sudanese, Chinese, African Americans, Matzos, and Jews.(3) Patients with this morbidity are healthy, intelligent, capable of hard physical labor, and have a normal life span (Short,1979). Phenotypically the condition is characterized by dysplastic bone formation of the face, skull, pelvis, and thoracic region. Main clinical features include hypoplastic or absent clavicles, open fontanelles and sutures, supernumerary teeth, short stature, and other skeletal anomalies.

KEYWORDS : Cleidocranial dysplasia (CCD), Autosomal dominant

INTRODUCTION

Cleidocranial dysplasia is caused by a mutation in the core-binding alpha one.

(CBFA1) gene located on chromosome 6p21 that causes faulty ossification of endochondranous and intramembranous bones.(1) Affected children with CCD have one intact copy of CBFA1 and one mutated copy that may be a deletion, insertion, or missense mutation. The type determines the degree of phenotypic expression and may be classified as classic, mild, or isolated primary dental anomalies (Zhou et al., 1999).(4) Any bone can be affected, but Feldman describes the bones most affected as the classical triad of cranial, clavicular, and pelvic anomalies.

Case-1

This infant eight months old a full term normal delivery at our center presented to us with lower respiratory tract infection & the diagnosis of CCD was incidental. Examination of the infant revealed frontal bossing, large head with evidence of macrocephaly (head circumference 47.5 cm), brachycephaly and prominent forehead. He had hypertelorism, depressed nasal bridge and narrow high-arched palate,

Chest X-ray revealed a narrow thorax with oblique ribs and partial agenesis of left clavicle. His CT scan skull and MRI head revealed syringomyelia and *presence of* spina bifida*. Other radiological findings included scoliosis without any associated vertebral anomalies. A diagnosis of cleidocranial dysplasia was considered. The general and systemic examination was essentially normal. Management in the form of counseling and regular follow up was advised.*

Figs 1 & 2-Macrocephaly,frontal bossing, wide open sutures and hypertelorism

Fig-4:Partial agenesis of rt clavicle

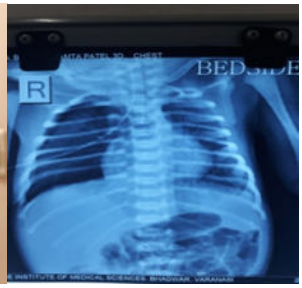

Fig- 3-Multiple syrinx & spina bifida on MRI

DISCUSSION

Cleidocranial dysplasia is a rare autosomal dominant disorder with generalized dysplasia of bone and teeth. Till now 500 cases have been reported.(6) The main clinical presentation includes absent or severely hypo plastic clavicles ,frontal and parietal bossing, , persistent open anterior fontanelle, delayed closure of the sutures, macrocephaly incomplete development of accessory sinuses and mastoid air cells, small sphenoid bones, calvarial thickening and wormian bones. Facial bones are small, mid-face hypoplasia with low nasal bridge, narrow high-arched palate, hypertelorism and conductive deafness may be seen. The primary dentition appears late and is frequently incomplete. The secondary dentition is similarly delayed and often mal-aligned with some teeth malformed and hypoplastic. Supernumerary teeth are common, especially in the premolar area. There may be associated enamel hypoplasia. Proportionate short stature may be seen. There may be syringomyelia or spina bifida occulta. The morbidities in the head and neck and include: brachycephaly, a large head with frontal bossing; small face due to small maxillary and zygomatic bones;prognathism,; open fontanelles; open cranial sutures; poor development of the foramen magnum; and dysplasia of the paranasal sinuses and mastoids. The eyes are wide set and the nose broad with a depressed, low bridge. The mouth and oral structures have a highly arched palate, delayed tooth eruption, tooth abnormalities, and absent or extra teeth (1).

Spinal abnormalities include defects such as scoliosis, kyphosis, spina bifida, syringomyelia, and spondylolysis or spondylothesis. The thorax is small and the chest is narrow with small, oblique ribs. Shoulders droop and the scapulae may be overgrown or undergrown; clavicles can be partially or fully absent. The pelvis is narrow with a wide symphysis pubis, and the extremities may have a deformity of the hip such as coxa vara or coxa valga. All joints are loose and may easily dislocate, hands have asymmetric fingers, and the feet are flat. Stature is short, but dwarfism is uncommon.(7) Because of these physical characteristics, individuals with CCD may have multiple oral, facial, or orthopedic surgeries; orthopedic complications; neurological and vascular problems (9) respiratory problems, and an increased cesarean

rate . The diagnosis is usually made at birth but may be delayed until adolescence (1), especially if the individual has a mild form, or if the condition does not seriously affect the individual. Occasionally CCD may go undiagnosed. Three dimensional computer tomography is helpful in diagnosing older children with CCD (Shen, 2000), and prenatal diagnosis with ultrasound is possible if there is a known positive family history.(11).

Management includes a multidisciplinary approach towards the patient with involvement of the primary care physicians, radiologists, neurophysicians , neurosurgeons and dental surgeons, orthopedic & vascular surgeons, physiotherapists and medical and genetic counselors.

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