TAJ December 2008; Volume 21 Number 2

ISSN 1019-8555
The Journal of
Teachers Association
RMC, Rajshahi

Case Report

Cleidocranial Dysplasia : A Case Report

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Abstract

Cleidocranial dysplasia is an autosomal dominant disorder with a wide range of expression, characterized by clavicular aplasia or hypoplasia, retarded cranial ossification, supernumerary teeth, short stature and a variety of other skeletal abnormalities. We are reporting a case of Cleidocranial dysplasia with literature is review.

TAJ 2008; 21(2): 166-169

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Introduction

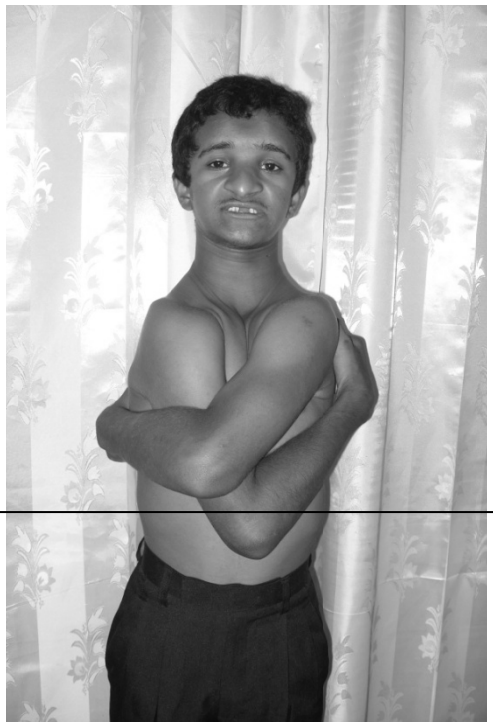
Cleidocranial dysplasia (CCD) is an autosomal dominant bone disorder with a wide range of expression, characterized by clavicular aplasia or hypoplasia, retarded cranial ossification, supernumerary teeth, short stature and a variety of other skeletal abnormalities[1]. A possible example of this disorder was detected in the skull of a Neanderthal man. The condition was first reported by Meckel in 1760 and later reported by Martin in 1765. The more obvious features of the disease like defect in the clavicle and cranium prompted Marie and Sainton to use the term Cleidocranial Dysostosis for this condition[2]. Later on, it was discovered that generalized dysplasia of bone occurs in this condition and the term Cleidocranial Dysostosis depicts only a portion of the abnormal development. Hence the term "dysostosis" has been abandoned in favor of "dysplasia"[3]. Well over 1000 cases have been reported. Rarity of this disease prompted us to report the case.

Case study

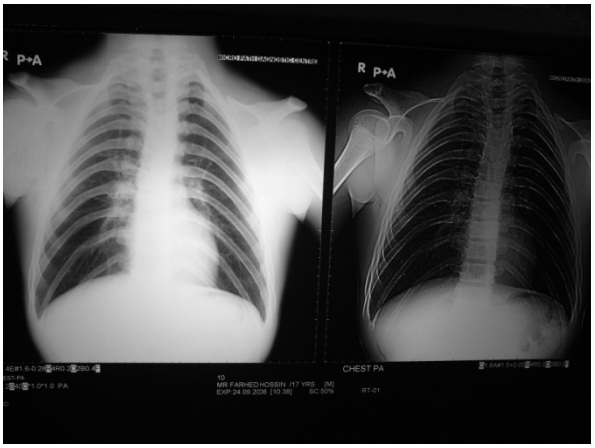
A 17 year old male patient attended medicine outpatient department with the complaints of growth retardation. On examination he was noted to have short stature (height 147 cm), bilateral absence of clavicles as demonstrated by patient's ability to bring his shoulders together, defective dentition in the form of superumery teeth and failure of eruption of upper and lower incisors. He had frontal and parietal bossing. His anterior and posterior frontalle were still open. His chest was narrow and shoulders were sloping. Examination of all other system revealed no abnormality. None of his family member has similar illness. Chest X Ray showed absence of clavicle in right side and hypoplasia in left side, scapula and glenoid cavity was small, and ribs were directed obliquely downwards. X Ray skull both view revealed widened skull sutures, presence of wormian bones, frontalles are open, some of the teethes were found missing and not erupted. His clinical features and investigation findings matches the classical description of Cleidocranial Dysplasia.

Discussion

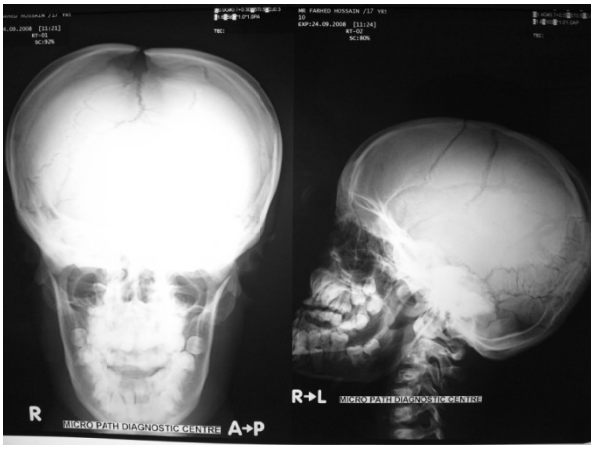
The CCD gene has been mapped to chromosome 6 p 21[4] within a region containing the *CBFA1* gene, a member of the Runt family of transcription factors[5]. *CBFA1* controls differentiation of precursor cells into osteoblasts and is thus essential for membranous as well as endochondral bone formation, which may be related to delayed ossification of the skull, teeth, pelvis and extremities in CCD[6] Typical clinical findings are shown in table 1.



Patient can bring his shoulders together because of absence of clavicles



CXR Showing aplastic right clavicle and hypoplastic left clavicle



X Ray Skull Showing widened sutures, open frontanelles, wormian bones, and defective dentition

Craniofacial growth is affected in many ways. Head circumference is usually at the upper limit without being macrocephalic. There is a broad forehead with frontal bossing and some degree of hypertelorism. Closure of the anterior frontanelles and sagittal and metopic sutures is delayed, often for life. Many patients with hypoplastic or even absent clavicles have gone through life, even working as manual laborers, without disability resulting from this defect. Depending on the degree of clavicular hypoplasia, appearance can range from a dimple in the skin to sloping, almost absent shoulders and the ability to voluntarily bring the shoulders together. According to most observations, a complete absence of the clavicle(s) is rare, whereas hypoplasia of the acromial end is common[7]. Final height is significantly reduced in patients with CCD. Previous investigations indicate that birth length is normal but that height drops below or around the 2nd centile between the ages of 4 and 8[8,9]. This is concordant with our patient, who has a height of only 147 cm. In a study by Jensen,[10] female patients were more affected than male patients. Patients usually have a mildly disproportionate short stature with short limbs compared to the trunk, more apparent in the upper limbs than the lower. Dental changes occur frequently and are very characteristic of CCD[11]. Retention of the deciduous dentition with delayed eruption of the permanent teeth is a relatively constant finding. Dental disability begins in late youth with the progressive morbidity and loss of the deciduous dentition. Many patients remember living “without teeth” for some years until the permanent teeth eventually erupted. Permanent teeth show a delay of root development and a lessened but not entirely absent eruptive potential. The situation is further complicated by the presence of multiple supernumerary teeth that displace the developing permanent teeth and obstruct their eruption.

Table 1: Clinical and radiological features of Cleidocranial dysplasia[7]

Clinical	Radiological
Skull	Multiple wormian bones
Brachycephaly	Segmental calvarial thickening
Frontal and parietal bossing	Un-ossified sutures and patent frontanelles
Open sutures and frontanelles	Dysplastic changes in the basiocciput
Delayed closure of frontanelles	Hypoplasia of maxilla
Relative prognathism	Delayed mineralization

Soft skull in infancy	Calcification of nasal bone delayed or missing
Depressed nasal bridge	Hypoplastic sinuses (paranasal, frontal, mastoid)
Hypertelorism	
Thorax and shoulders	Hypoplastic, aplastic, or discontinuous clavicles
Ability to bring shoulders together	Cone shaped thorax
Narrow, sloping shoulders	Cervical ribs, missing ribs
Respiratory distress at early age	Hypoplastic scapulae
Increased mobility	
Pelvis and hips	Delayed ossification of pubic bone
Caesarean section	Hypoplasia of iliac wings
	Widening of sacroiliac joints
	Large femoral neck, large epiphyses
Spine	Hemivertebrae, posterior wedging
Scoliosis	Spondylolysis and spondylolisthesis
Kyphosis	Spina bifida occulta
Hands	Short middle phalanges and metacarpals/tarsals III–V
Brachydactyly	Hypoplastic distal phalanges
Tapering of fingers	Accessory epiphyses especially of 2nd metacarpal
Nail dysplasia/hypoplasia	Long 2nd metacarpal
Short, broad thumbs	
Clinodactyly of 5th finger	
Dentition	
Normal deciduous dentition	Impacted, supernumerary teeth
Supernumerary teeth	
Delayed eruption	
Crowding, malocclusion	

Conclusion

CCD is very rarely encountered in clinical practice. This case is presented to show the significance of thorough clinical examination of patients with short stature including palpation of clavicles.

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