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.

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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. 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. 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”. 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 supernumery 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, frontalle 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 within a region containing the CBFA1 gene, a member of the Runt family of transcription factors. 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. Typical clinical findings are shown in table 1.
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. 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. This is concordant with our patient, who has a height of only 147 cm. In a study by Jensen, 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. 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

<table>
<thead>
<tr>
<th>Clinical</th>
<th>Radiological</th>
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<tbody>
<tr>
<td><strong>Skull</strong></td>
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<tr>
<td>Brachycephaly</td>
<td>Multiple wormian bones</td>
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<tr>
<td>Frontal and parietal bossing</td>
<td>Segmental calvarial thickening</td>
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<td>Open sutures and frontanelles</td>
<td>Un-ossified sutures and patent frontanelles</td>
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<td>Delayed closure of frontanelles</td>
<td>Dysplastic changes in the basiocciput</td>
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<td>Relative prognathism</td>
<td>Hypoplasia of maxilla</td>
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<td>Delayed mineralization</td>
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<tr>
<td>Soft skull in infancy</td>
<td>Calcification of nasal bone delayed or missing</td>
</tr>
<tr>
<td>Depressed nasal bridge</td>
<td>Hypoplastic sinuses (paranasal, frontal, mastoid)</td>
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<tr>
<td>Hypertelorism</td>
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**Thorax and shoulders**
- Ability to bring shoulders together
- Narrow, sloping shoulders
- Respiratory distress at early age
- Increased mobility
- Hypoplastic, aplastic, or discontinuous clavicles
- Cone shaped thorax
- Cervical ribs, missing ribs
- Hypoplastic scapulae

**Pelvis and hips**
- Caesarean section
- Delayed ossification of pubic bone
- Hypoplasia of iliac wings
- Widening of sacroiliac joints
- Large femoral neck, large epiphyses

**Spine**
- Scoliosis
- Kyphosis
- Hemivertebrae, posterior wedging
- Spondylolysis and spondylolisthesis
- Spina bifida occulta

**Hands**
- Brachydactyly
- Tapering of fingers
- Nail dysplasia/hypoplasia
- Short, broad thumbs
- Clinodactyly of 5th finger
- Short middle phalanges and metacarpals/tarsals III–V
- Hypoplastic distal phalanges
- Accessory epiphses especially of 2nd metacarpal
- Long 2nd metacarpal

**Dentition**
- Normal deciduous dentition
- Supernumerary teeth
- Delayed eruption
- Crowding, malocclusion
- Impacted, supernumerary teeth

**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.

**References**

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