Cleidocranial Dysplasia

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Cleidocranial dysplasia (CCD), is characterized by short stature, typical facial features and variable degree of pan-skeletal anomalies affecting skull and clavicle(l). It is a relatively underdiagnosed entity and due to its clinical features can be mistaken for a number of other conditions *(e.g.,* Noonan syndrome, Turner's syndrome, hypothyroidism and other skeletal dysplasia), before the diagnosis is confirmed radiologically(2). Three cases of CCD have been reported in Indian literature where a variety of conditions have been considered and diagnosis was finally confirmed radiologically(2,3). One more case is being described who had been diagnosed and treated twice for rickets before he was referred to us. This case is being reported to highlight that CCD should be considered in the differential diagnosis of short stature with features of skeletal immaturity. Although in all the reported conditions diagnosis was made after radiological investigations, yet retrospective examination revealed the presence of classical features(2,3). Therefore, although there is a chance of missing the diagnosis clinically, yet a high index of suspicion and confirmation by characteristic radiological features could help in making the diagnosis. [b]

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Case Report

A 4-year-old first born male child of non-consanguineous Hindu parents was brought with the complaints of short stature and abnormal head shape. On examination weight was 13 Kg (25th centile), height 87.5 cm (<10th centile) and head circumference was 50.7 cm (between 75th to 90th centile). The child had brachycephalic skull; pronounced frontal and parietal bossing; widely open anterior fontanella and sutures; small face; hypertelorism; depressed nasal bridge; high arched palate and malar hypoplasia. He had broad neck, sloping of shoulders *(Fig.* 7) and hypermobility of shoulders (mother was aware of it). The child had 20 primary teeth without any abnormality or history of delayed tooth erruption. His physical development, intelligence, hearing, spine and rest of the systemic examination were normal. His radiological examination showed missing of lateral half of clavicle on both sides; spina bifida (Fig. 2); multiple wormian bones with widely open sutures; underdeveloped paranasal sinuses *(Fig.* 3), absent pulic rami and symphysis; and hypoplastic ischial bones *(Fig. 4).*

Discussion

Cleidocranial dysostosis was first

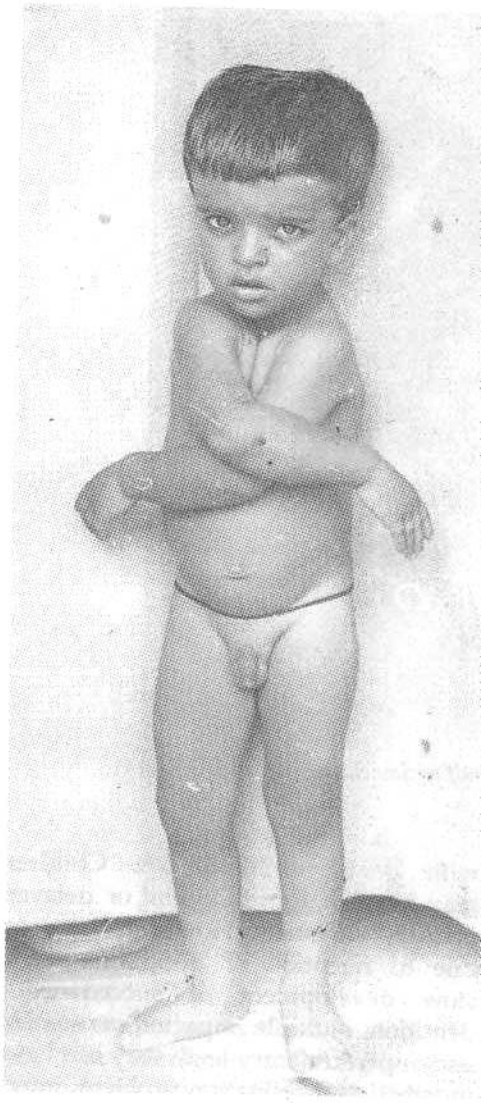

Fig. 1. Clinical photograph showing typical face, bossing of skull, sloping of shoulders.

reported in a patient with congenital defect of clavicles(4), and the classical features were described later on(1). The terminology has been changed from dysostosis meaning defective bone formation, to dysplasia meaning abnormal tissue development, thereby reflecting the more generalized nature of the disease process(5). It affects bones of intramembranous origin and endochondral bone formation of long bones in most of the skeleton are affected(6). There is a generalized failure of midline ossification resulting in patent fontanella, metopic suture, wormian bones, nasal deformity, non-union of mandibular symphysis, high arched palate, cleft palate, hypoplasia or absence of clavicles, spina bifida and delayed closure of pubic symphysis(7). All these features were observed in the present case.

The most characteristic and pathognomic skeletal feature is that one or both clavicles are frequently partially or in 10% cases completely absent(8,9). Usually rudimentary sternal and acromial stubs are present and the midclavicular position is absent. Clavicular deformity along with the dysplastic muscle attachments give rise to elongated neck, narrow drooping and hypermobility of shoulders with tendency to approximate shoulders anteriorly(8). Clavicle is the first bone to be ossified in the 6th week of fetal life and is thus the most often affected bone(8).

Other abnormalities can be short cranial base; hypoplastic maxillary, lacrimal and zygomatic bones and subsequent paranasal sinuses. Reported vertebral abnormalities are scoliosis, kyphosis, lordosis, and vertebral synostosis(8,9). It has been reported that syringomyelia should be considered in any patient of CCD with any neurological symptoms, progressive scoliosis not responding to conservative surgery, or

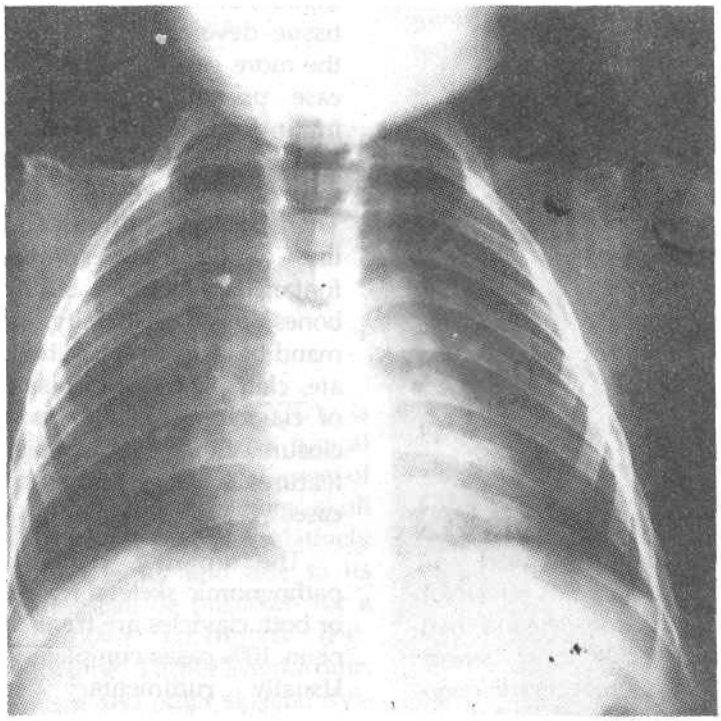

Fig. 2. X-ray chest showing missing half of clavicles and spina bifida.

progressive scoliosis after skeletal maturation(9). These features were not present in this case.

It is a relatively benign condition and the patient can seek medical help for short stature; dental abnormalities; hearing loss; complications during child birth and dislocation of joints. The final height attained by these children is significantly lower than their normal counterparts(7). Although psychosocial disorders associated with abnormal facial and body features may occur, patients have normal intelligence, with a overall good prognosis and normal life expectancy(8,9).

Often these patients initially present with dental abnormalities. Children may have an initial normal or delayed primary dentition. However, later on due to retention of deciduous teeth, slow development of succedaneous dentition, multiple impacted permanent and supernumerary teeth they can have prosthetic rehabilitation problem, infections of tooth and jaw with tendency of pathologic fractures(8). In the present case no dental abnormalities were present.

The etiology of CCD is unknown. It can have dominant and recessive pattern of inheritance and 16% patients are reported to be sporadic(10).

Because of its benign nature, CCD is

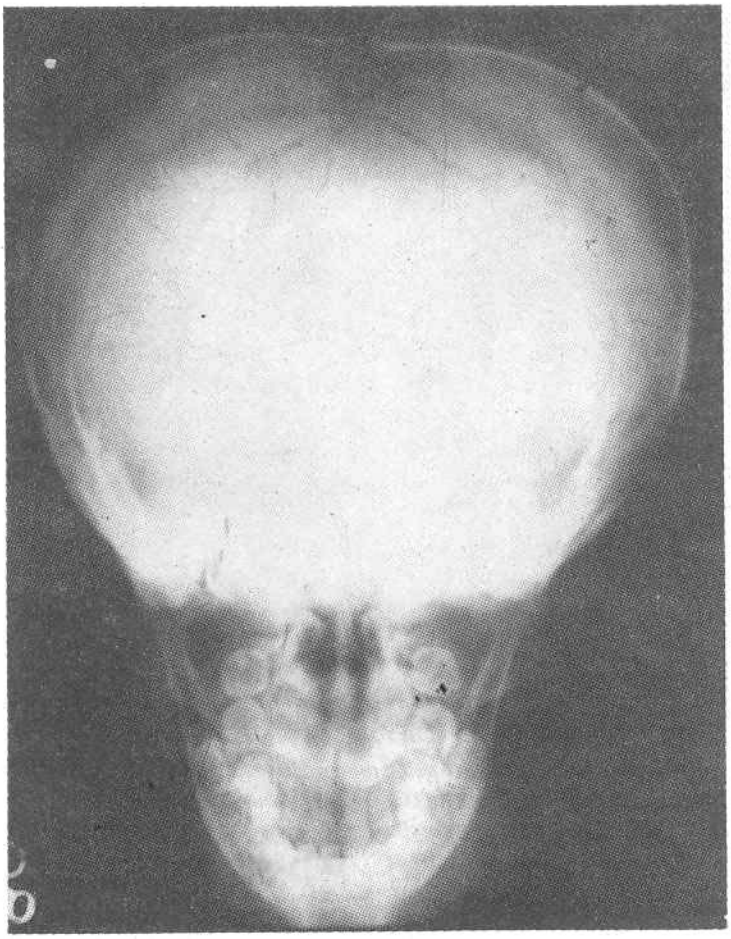

Fig. 3. *X-ray skull (lateral) showing widely open sutures and wormian bones*

perhaps an under reported entity(2). It should be considered in the differential diagnosis of short stature with skeletal immaturities like abnormally large fontanella and wormian bones. Relevant clinical examination should include palpation of clavicles.

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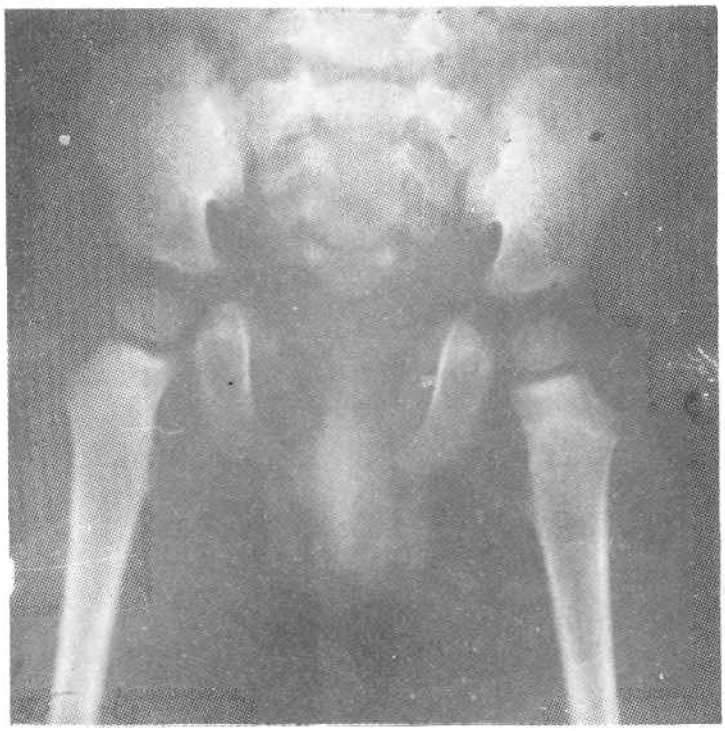

Fig. 4. X-ray pelvis (AP) showing absent pubic rami and symphysis with hypoplastic ischial bones.

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