IMAGES IN CLINICAL PRACTICE

Figure 1: Patient with fronto-parietal bossing, hypotelorism, flat nasal bridge

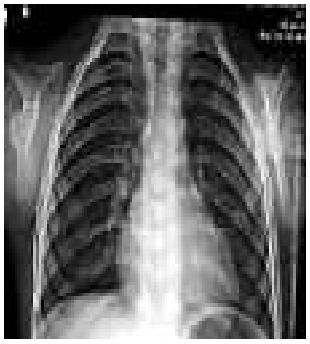

Figure 2: Chest X-ray showing absence of bilateral clavicles

OPEN ANTERIOR FONTANELLE WITH ABSENT CLAVICLES IN A 5 YEARS OLD BOY

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A 5 year old boy presented with an open anterior fontanelle. Physical examination revealed that the anterior and posterior fontanelles were open. Anterior fontanelle diameter was 4 x 3 cm. In addition, he had frontal and parietal bossing, brachiocephalic head, flat nasal bridge, retrognathism and dental deformities like malocclusion and crowding of teeth (Figure 1). Shoulders were hypermobile with the ability to bring shoulders together. Developmental milestones and other systems were normal. Bilateral clavicles were absent on chest X-ray (Figure 2).

What is the diagnosis?

Cleidocranial Dysplasia (CCD). It is caused by heterozygous mutations in RUNX2 gene, which encodes a transcription factor required for osteoblast differentiation and is located on chromosome 6p21. (1, 2) In approximately 40% of CCD patients, a genetic transition cannot be identified, and the condition develops spontaneously. (1, 3) Cleidocranial dysplasia is inherited as an autosomal dominant genetic trait. The abnormal gene can be inherited from either parent, or can be the result of a new mutation (gene change) in the affected individual. The risk of passing the abnormal gene from affected parent to offspring is 50% for each pregnancy regardless of the sex of the resulting child. The major features of CCD are aplastic or hypoplastic clavicles, dental abnormalities and delayed closure of the fontanelles. Typically, our patient had all of these findings that are pathognomonic for a diagnosis of CCD. (1) Other findings of CCD are short stature, a bell shaped thorax, hypoplasia of the pelvis, enlargement of the frontal and occipital bones, and phalangeal abnormalities. Shortened or absent nasal bones, paranasal sinus abnormalities, thickening of some segments of the calvaria, small maxillae, and delayed union of the mandibular symphysis are less common findings of CCD. Depending upon the degree of clavicular hypoplasia, appearance can range from a dimple in the skin to almost the ability to bring the shoulders together. According to most observations, clavicles are underdeveloped to varying degrees and in approximately 10 percent of cases, are completely absent. This allows excessive mobility of the shoulder girdle. Other bones may also be affected including long bones, the vertebral column, the pelvis and the bones of hands and feet. (4) In our patient there was complete absence of both the clavicles. Dental deformities like malocclusion, crowding and supernumerary teeth is one of the main features of CCD. It has been suggested that supernumerary teeth in such cases should be removed as soon as possible. (5) The main finding in our patient was an open anterior fontanelle. Delayed closure of fontanelles could be a feature of hypothyroidism, rickets, hypophosphatasia, osteogenesis imperfecta, pycnodysostosis, and other syndromes such as Apert syndrome, Dubowitz syndrome. (6) Care for (CCD) should be coordinated through a paediatrician, craniofacial or neurosurgery specialists, an ear, nose and throat (ENT) surgeon or audiology (hearing) doctor. Management of the dental problems by orthodontist before the age of 6 years has been recommended. Hearing should be screened and followed up on a regular basis.

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