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CASE REPORT

Cleidocranial Dysplasia

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Abstract

Cleidocranial dysplasia (CCD) is a rare autosomal dominant dysplasia with an estimated incidence of 1:1,000,000. Prominent features include large head with delayed suture closure, persistent metopic suture, Wormian bones, hypertelorism, small face, cleft mandible, dental dysplasia, hypoplasia or aplasia of the clavicles, hypoplastic scapula with a small glenoid, cleft sternum, coxa vara or valga, a narrow pelvis, delayed pubic ossification, and several varieties of spinal abnormalities. We report a case of a 30 year old male patient with this syndrome.

Key Words

Cleidocranial Dysplasia, Autosomal Dominant, Wormian Bones

Introduction

Cleidocranial dysplasia, is a hereditary congenital disorder due to haploinsufficiency caused by mutations in the CBFA1 gene also called Runx2, located on the short arm of chromosome 6. Cleidocranial dysplasia is a condition that primarily affects the development of the bones and teeth with no predilection of genre or ethnic group. The absence of clavicles occurs in 10% of cases. These individuals are usually short. They have persistent fonticulus of the cranium or late closure of the same. The principal oral manifestations are a delayed exfoliation of primary teeth, delayed or multiple impactions of the permanent dentition, and multiple impacted supernumerary teeth. Radiographic examination is a critical confirmatory diagnostic tool.[1] The purpose of this article is to review the oral manifestations with an outline of genetics and general manifestations.

Case Report

A 30 year old male patient reported with a chief complaint of pus discharge from the upper posterior tooth on the right side. Patient was moderately nourished. He was mentally retarded with short stature. There was stunted growth of hands and short legs. Past medical and family history was not significant. Extraoral examination revealed frontal bossing, slanting oculars and brachycephalic head. He had a narrow chest with narrow shoulders. There was hypermobility of shoulders and both could be approximated in the centre when he was asked to do so (*Fig 1*). Intraoral examination revealed poor oral hygiene with generalized gingivitis. High arched palate, severe malocclusion with abnormal spacing and numerous missing and erupting teeth were seen (*Fig 2*). Panoramic radiograph showed severe malocclusion with multiple supernumerary teeth (*Fig 3*). Lateral

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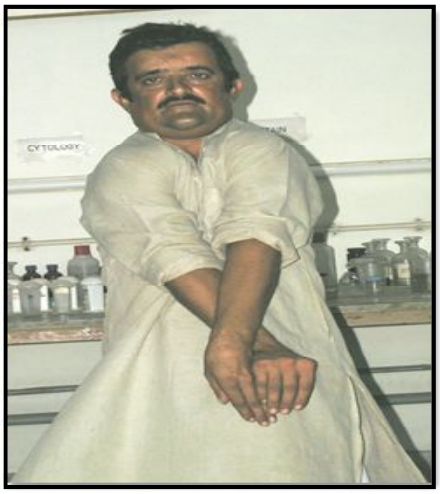

Fig 1, Hypermobility of Shoulders

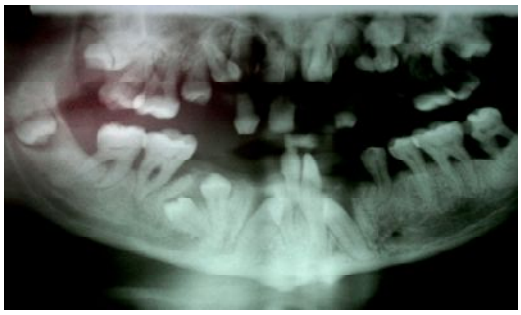

Fig 3. Severe Malocclusion Seen in OPG, with Multiple Supernumerary Teeth

cephalometric radiograph showed wormian bones (*Fig 4*). Chest x-ray showed hypoplastic clavicles (*Fig 5*). Based on the clinical and radiographical features a diagnosis of cleidocranial dysplasia was given.

Discussion

Cleidocranial dysplasia (CCD) (cleido = collar bone, + cranial = head, + dysplasia = abnormal forming), is an autosomal dominant disease with complete penetrance, but variable expressivity, first described by Marie and Sainton in 1898. It is also known as Rubber Man, Scheuthauer-Marie-Sainton or Marie-Sainton disease and Cleidocranial Dysostosis or Mutational Dysostosis. CCD is present at a frequency of one in one million individuals. It affects all ethnic groups. It is a relatively uncommon disorder with the prevalence being 0.5 per 100,000 live births. One third of cases are considered to be due to sporadic mutations .[2] A variety of mutations in RUNX2 cause CCD but no clear genotype-phenotype correlation has been established in CCD patients. Overexpression of RUNX2 results in growth arrest through p27 (KIP1)-induced inhibition of the s-phase cyclin complex, followed by dephosphorylation of the RB1 protein. Novel mutation of RUNX2 also disrupts the protein-DNA interaction. [3] The early history of CCD goes back to prehistorical times, with a case of CCD in a Neanderthal skull, which was documented by Greig in 1933. [4] In Greek mythology, the ugly hero Thersites was described by Homer as being able to oppose his shoulders in front of his chest. [5] A skeleton of an affected male who died of tuberculosis in 1809 in Vienna, shows the classical manifestations of CCD. [6] The earliest recognizable report of CCD in the

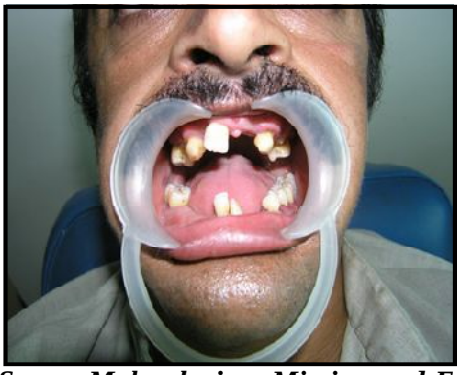

Fig 2. Severe Malocclusion, Missing and Erupting Teeth

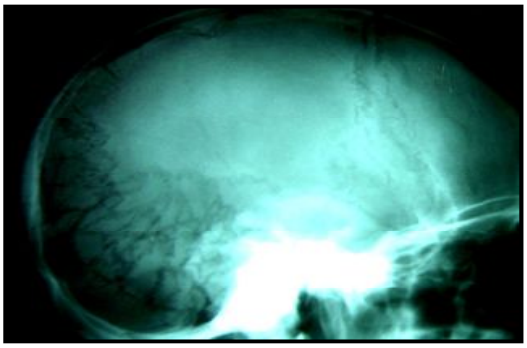

Fig 4. Lateral Cephalometric Radiograph Showing Wormian Bones

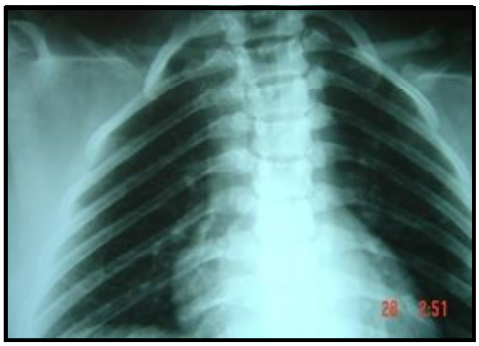

Fig 5. Chest X-Ray Showing Hypoplastic Clavicles

medical literature has been attributed to Meckel in 1760. [7] Clinically, the diagnosis is often made at birth but may not occur until later, when persistence of the widely open anterior fontanelles and sutures or short stature incites parental concern. Skeletal abnormalities include clavicular aplasia/hypoplasia, bell or cone shaped thorax, enlarged calvaria with frontal bossing and open fontanelles, wormian bones, brachydactyly with hypoplastic distal phalanges, hypoplasia of the pelvis with widened symphysis pubis, severe dental anomalies, and short stature. Our patient showed wormain bones, frontal bossing, and hypoplastic shoulders, short stature and was mentally retarded. Patients with normal clavicles have also been described in previous studies. Muscle attachments to the clavicles may also be dysplastic, leading to distortion of the neck. Hand and wrist radiographs often show pseudoepiphyses of the metacarpal and metatarsal bones resulting in characteristics lengthening of the second metacarpal. Hypoplastic and pointed phalanges can also be seen. Frequently, genua valga and pes planus are found in children younger than 5 years of age. [8]

Other clinical features include short, tapered fingers and broad thumbs, short forearms, flat feet, knock knees, flat nose, hypertelorism, small upper jaw and scoliosis. [8]

Hyperdontia is the major oral manifestation of CCD. It can involve either or both the primary and secondary dentition and leads to dental impaction, overcrowding, and malocclusion. Articulation and mastication may be compromised, and the cosmetic appearance of the dentition is also affected. Supernumerary teeth may be arranged uniformly as a double row or placed randomly on the jaws. Morphologically, these teeth may be fully formed, bifid, or represented by small tuberosities on the maxillary alveolar ridges. [9]

Delayed eruption and retention of both the dentition is also seen in CCD. The crowns of the teeth sometimes appear abnormal, the enamel may be hypoplastic, and dentigerous cysts and taurodontia are frequent findings. Radiographic manifestations in children include rounded gonion angles, kyphotic sphenoid bones, and wormian bones in the cranial sutures. The face appears small in relation to the cranium with hypoplastic maxillary, lachrymal, nasal, and zygomatic bones. The paranasal sinuses may be underdeveloped. The maxillary sinuses may be small or missing, and the maxilla is underdeveloped, causing a Class III skeletal relationship and a maxillary retrusion (a relative mandibular prognathism). The palate may be abnormally high, and, occasionally, a cleft palate has often been reported. Association with mental retardation has been shown, but most patients apparently possess normal intelligence .[10]

Conclusion

CCD is characterized by abnormalities of the skull, teeth, jaws, and shoulders girdles as well as by occasional stunting of the long bones. Large fontanelles, broad sutures, numerous wormian bones and brachycephalic head is pathognomic for the condition. The radiographic evaluation of patients is the most reliable means to confirm the diagnosis. Genetic counselling is appropriate for prospective parents with a family history of CCD. A multidisciplinary approach to treatment of these patients utilising a pedodontist, an orthodontist and an oral surgeon is recommended.

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