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CASE REPORT

Cleidocranial dysplasia with bilateral polycystic ovarian disease and Mullerian abnormality of the uterus: a case report

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We report an interesting case of cleidocranial dysplasia associated with an unusual finding of bilateral polycystic ovarian disease and bicornuate uterus with unicollis (Mullerian abnormality).
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Keywords: cleidocranial dysplasia; Mullerian abnormality; supernumerary teeth

Case report

A 22-year-old female patient came for the replacement of the missing maxillary anterior teeth (Fig. 1). The teeth were extracted by a private practitioner due to caries. The patient was short and moderately built. She was mentally alert and appeared to be of normal intelligence. Her mid-face was poorly developed with marked frontal bones, underdeveloped maxilla which made her look pseudoprognathic. The oral examination revealed 13 retained deciduous teeth that were attrited and five permanent teeth (Fig. 2). Panographic examination revealed severe dental malformation with 13 retained deciduous teeth, five erupted permanent teeth, 66 impacted permanent and supernumerary teeth along with parallel ascending rami (Fig. 3). The differential diagnosis of cleidocranial dysplasia (CCD) and Gardner's syndrome was considered. Further physical examination revealed that the patient was unable to approximate her shoulders. Detailed radiographic and medical examination was carried out. Patient's posteroanterior skull radiograph disclosed presence of wider sutures than normal and multiple Wormian bones (Fig. 4). The posteroanterior chest radiograph revealed a congenital malformation with an evidence of sprengle's shoulder, both the scapula were rotated and highly placed, right clavicle was short and hypoplastic especially on its medial end (Fig. 5). The medical history revealed that the patient was a diagnosed case of bilateral polycystic ovarian disease (PCOD) and bicornuate uterus with unicollis (Mullerian abnormality). Laboratory findings were within the normal ranges. With the above findings a diagnosis of CCD with bilateral PCOD and bicornuate uterus with unicollis (Mullerian abnormality) was made.

Detailed family examination showed that she was the fourth child among the five children. There were no similar findings of CCD in any of the family members except for the presence of a palatally erupted paramolar in the maxillary left region in the patient's elder sister whose panoramic radiograph revealed no other impacted supernumerary teeth (Fig. 6). Her thorough medical and radiological examination showed no other abnormality.

Comments

CCD is a rare autosomal dominant disorder with a prevalence of one per million, characterized by brachycephalic skull, aplasia or hypoplasia of one or both of the clavicles, delayed ossification of the fontanelles, frontal and parietal bossing, wide pubic symphysis, short middle phalanges of fifth fingers, dental and vertebral anomalies, poor development of the premaxilla and pseudo-anodontia (1).

CCD is a disorder of bone caused by a defect in *RUNX2* (CBFA1) gene that has been mapped to chromosome 6p21. *RUNX2* is a multifactorial transcription factor that controls the differentiation of precursor cells into osteoblasts and is essential for both membranous and endochondral bone formation. Hence, it is known as 'master gene' of bone development (2). *RUNX2* knockout mice have no bone, and their skeleton is composed solely of cartilage. Teeth formation is initiated, but their development is arrested after the bud stage, indicating that *RUNX2* function is necessary for bud to cap stage transition (3). However, heterogenous loss of function of *RUNX2* gene in

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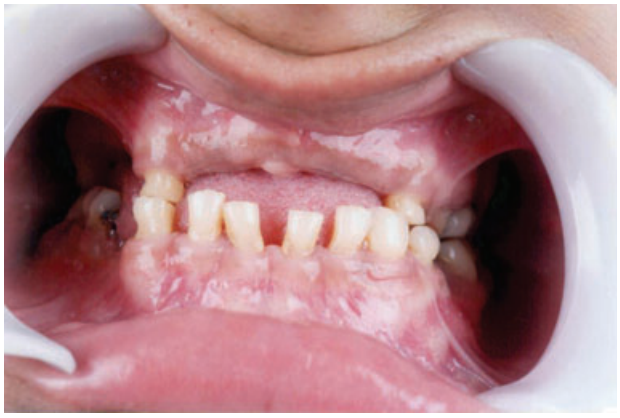

Figure 1 Intra-oral photograph showing missing maxillary anteriors and retained deciduous teeth.

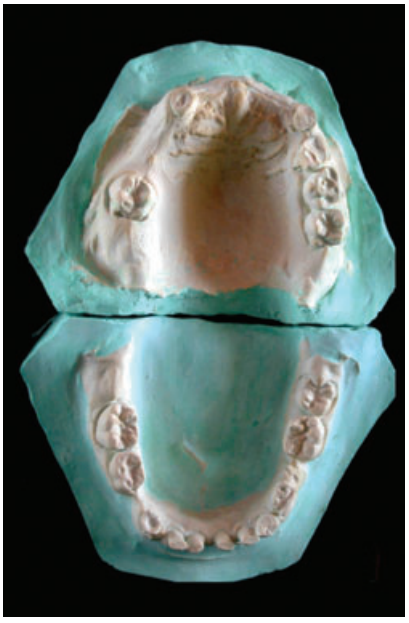

Figure 2 Casts showing retained deciduous and permanent teeth.

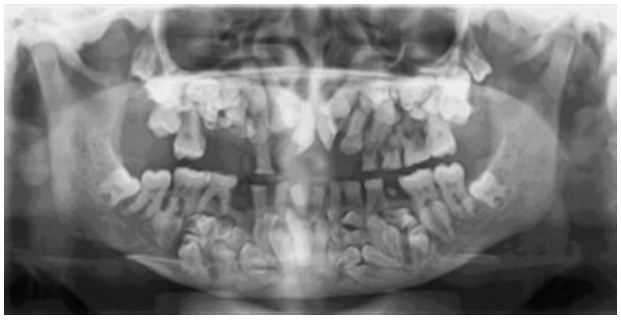

Figure 3 A panoramic radiograph showing retained deciduous and multiple impacted permanent and supernumerary teeth.

patients with CCD, leads to supernumerary teeth instead of having missing teeth. The mechanism of action of *RUNX2* gene in tooth development, in CCD remains to be clarified (4).

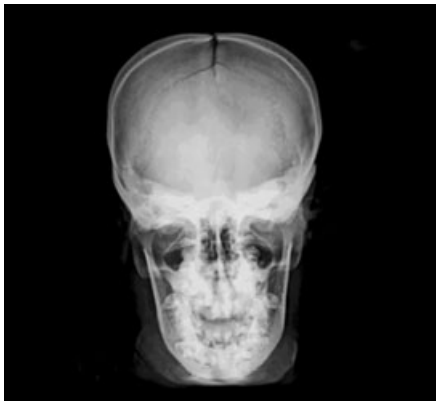

Figure 4 A posteroanterior skull radiograph showing wide sutures and multiple Wormian bones.

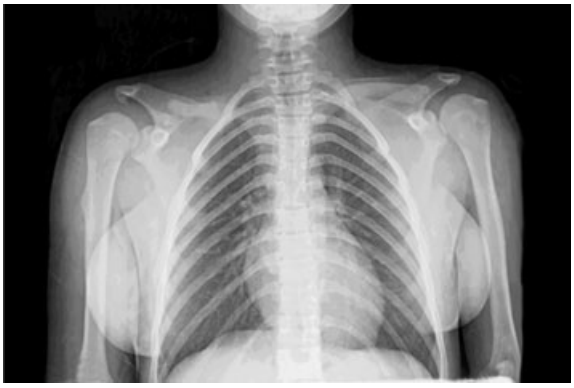

Figure 5 A posteroanterior chest radiograph showing short and hypoplastic right clavicle.

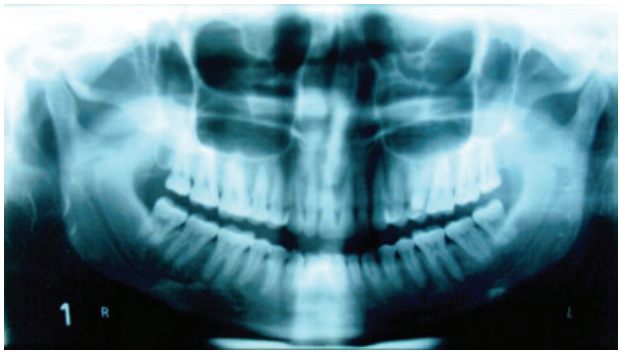

Figure 6 A panoramic radiograph showing erupted paramolar in the maxillary left quadrant of the patient's sibling.

The oral manifestations of CCD are retained deciduous teeth, multiple impacted permanent and supernumerary teeth. The failure for eruption of the permanent teeth is secondary to the mechanical obstruction by the presence of the supernumerary teeth and lack of resorption of the overlying bone (5).

The patient's with CCD are discovered incidentally in most of the cases, many of them report to the dentist for the tardy emergence of teeth as in our case. There is no

known treatment for CCD, except counseling regarding its hereditary tendency. Extraction of primary teeth does not necessarily result in the eruption of the secondary dentition, although exposure combined with orthodontic procedures may be helpful in some cases (6, 7).

This case appears to be of significance because of its association with bilateral PCOD and bicornuate uterus with unicollis (Mullerian abnormality) that has not been reported until now.

This case appears to be of spontaneous mutation because no characteristic features of CCD were evident in the other family members except for the presence of an erupted paramolar in the patient's sibling. This case also stands unique because of its presentation of one supernumerary tooth per permanent tooth, thus accounting for 84 teeth totally associated with multiple ovarian and uterine abnormalities.

We also highlight the need for the detailed family examination of any case of supernumerary teeth because of its possible familial and syndromic association as noticed in our case.

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