CORNELIA DE- LANGE SYNDROME: A CASE REPORT

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ABSTRACT
Cornelia de Lange syndrome (CDLS) is a congenital disorder involving skeletal, craniofacial deformities together with gastrointestinal and cardiac malformations. Here with, dental management of a case with CDLS syndrome is described.

KEYWORDS: Cornelia de Lange syndrome (CDLS), Dental management, Disorder.

INTRODUCTION
Cornelia de Lange syndrome (CDLS), first described in its full clinical presentation by Dr. Cornelia de Lange in 1933, is a congenital disease, basically characterized by psychomotor retardation associated with a series of malformations, including mainly skeletal, craniofacial deformities together with gastrointestinal and cardiac malformations1. Incidence of this entity is variable, ranging from 1:30,000 to 1:50,000 in different population groups2. Although exact incidence is unknown, CDLS likely affects 1 in 10,000 newborns. There is no racial predilection. It is slightly more common in females as compared to males, (F: M: 1.3:1)3. It has characteristic abnormalities, including microcephaly, growth failure, anomalies of development of the hands and feet, short stature, excessive growth of hair, heavy eyebrows, synophrys (growth of eyebrows across the midline to form one large confluent eyebrow), long eyelashes, strabismus, small nose with anteverted nares, long philtrum, microglossa, downturned mouth, hypoplastic nipples and umbilicus, flexion contracture of elbows, micromelia and hirsutism. The clinical diagnosis of this syndrome is based mainly on a group of these features.4 The list of head and oral manifestations are presented in Table 1.

Case report
In this particular instance, we did have a 5-year-old male patient with developmental disorder and speech impediment, who came to our office for dental pain. Initially, we noted that our patient was the son of a 29-year-old mother with a history of premature births. The parents of this patient are biologically related. During the physical examination of the patient, we recorded this data as follows: Weight: 12.3 kg; Height: 110 cm Head circumference: 47 cm Body temperature: 36°C Pulse: 108 b/m (beats/minute)

Dental findings were as follows:
- Dentition
  - Prognathic maxilla
  - Cleft palate
  - Dental caries in 62,63,64,71,81,75,85.
  - Grossly decayed in 51,52,61,54,64,74,84.

Based on our findings, we performed a general anesthesia in order to extract the teeth (51,52,61,54,64,74&84) that had initially been diagnosed with the extraction indications followed by a removable functional appliance for the upper arch, pulpectomy followed by stainless steel crown was cemented on 85, restorations were done on 71,75 &81. There were no complications throughout the anesthesia and/or intubation process. Furthermore, we confirmed that there were no complications during the postoperative period.
Fig. 1 Preoperative photographs

Fig. 2 Postoperative photographs
Allen et al. biochemical test nor any other diagnostic test exist for CDLS. This syndrome is related to mental retardation, skeletal defects (including brachycephaly, hypoplastic mandible and cleft palate), ocular defects, epilepsy and varying degrees of hirsutism. The eye brows may be joined across the bridge of the nose (synophrys) in addition to hypertelorism and antimongoloid slant of the eyes, upward-facing nostrils, and thin lips, which made us become aware of the CDLS.\textsuperscript{7,8,9} In our opinion, the patient presents the typical facial characteristics of CDLS. The clinical findings of the reported case closely confirm the classical picture of CDLS.\textsuperscript{10,11} Since neither a biochemical test nor any other diagnostic test exist for CDLS, the physical diagnosis of individuals who are mildly affected, may be difficult. Beck\textsuperscript{2}, discussed the postmortem examination of the patients and revealed various congenital malformations of the internal organs including cardiac defects, pulmonary hypoplasia, diaphragmatic hernias, gastrointestinal and genitor-urinary anomalies. The features of this disorder vary widely among affected individuals and range from relatively mild to severe. Based on the clinical variability in CdLS, Van Allen et al.\textsuperscript{12} proposed a classification system. Type I, or classic, CdLS patients have the characteristic facial and skeletal changes of the diagnostic criteria established by Preus and Rex\textsuperscript{13}. They have prenatal growth deficiency, moderate to-profound psychomotor retardation, and major malformations, which result in severe disability or death. Type II, or mild, CdLS patients have similar facial and minor skeletal abnormalities to those seen in type I; however, these changes may develop with time or may be partially expressed. They have mild-to-borderline psychomotor retardation, less severe pre- and postnatal growth deficiency, and the absence of (or less severe) major malformations. Type III, or phenocopy, CdLS includes patients who have phenotypic manifestations of CdLS that are causally related to chromosomal aneuploidies or teratogenic exposures. Allanson et al.\textsuperscript{14,15} in 1997 showed that, in the mild phenotype, the characteristic facial appearance may not appear until 2 to 3 years of age, while it is always present at birth in the classic phenotype. They also noted that the characteristic facial appearance decreased with time in the mild phenotype. In our case the patient comes under type II. Some dental abnormalities reported earlier include delayed eruption, spacing and macro- or microdontia.\textsuperscript{12} Yamamoto et al.\textsuperscript{16} have reported two cases with delayed tooth eruption and microdontia, with one of these cases being a partial anodontia.

CONCLUSION

Once it has been researched, we have realized that there were only a few citations on the dental and oral findings of the Cornelia de Lange syndrome. Since the literature regarding the CDLS was not so informative, it appears that the relationship between the oral manifestations of this syndrome and other syndromes must be further investigated. Cornelia de Lange Syndrome is a rare but well characterized syndrome. The key diagnostic features are distinctive facial features, limb anomalies and growth retardation.

References


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