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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 malformations[1]. Incidence of this entity is variable, ranging from 1:30,000 to 1:50,000 in different population groups[2]. 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, micrognathia, 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 this 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)

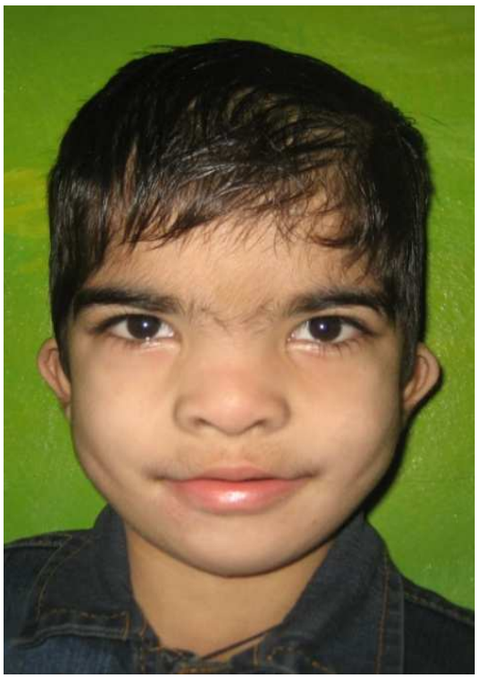
His face seemed dysmorphic; he had thick and curly hair eyebrows meeting at the midline, and he had long and curved eyelashes **(Fig. 1).** His ears were abnormally low placed, they were dysplastic, and his mandibular symphysis was bumpy. He also had anteverted nostrils, a small nose, thin lips with the downward turned angles of the mouth, micrognathia, short neck, fairly small feet and hands.

Dental findings were as follows:
Dentition

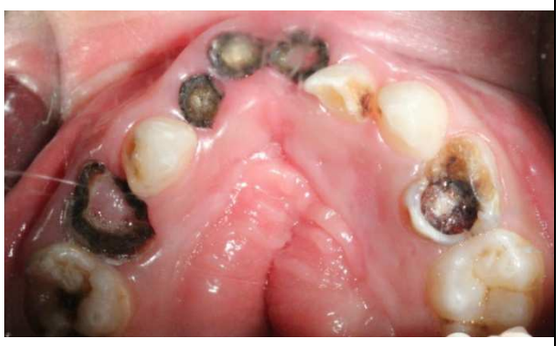
E D C B A	A B C D E
E D C B A	A B C D E

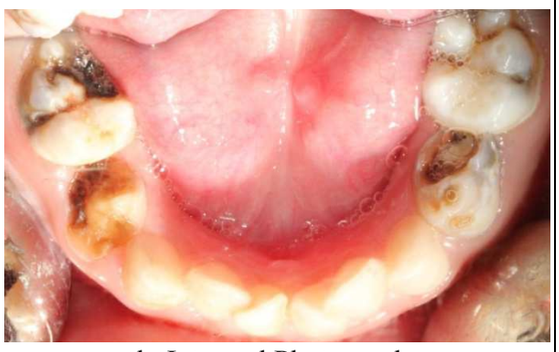
Prognathic maxilla
Cleft palate
Dental caries in 62,63,64,71,81,75,85.
Grossly decayed in 51,52,61,54,64,74,84.

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.



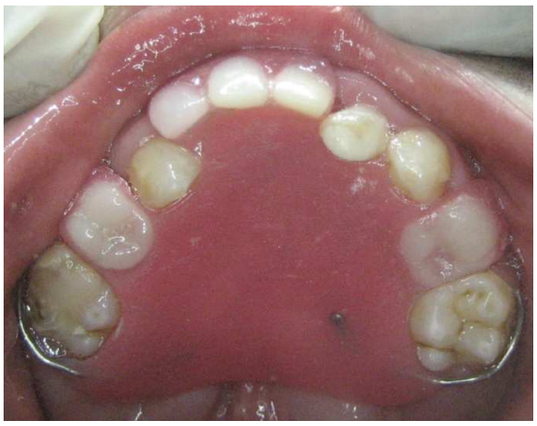
a. Extraoral Photograph





b. Intraoral Photograph

Fig.1 Preoperative photographs



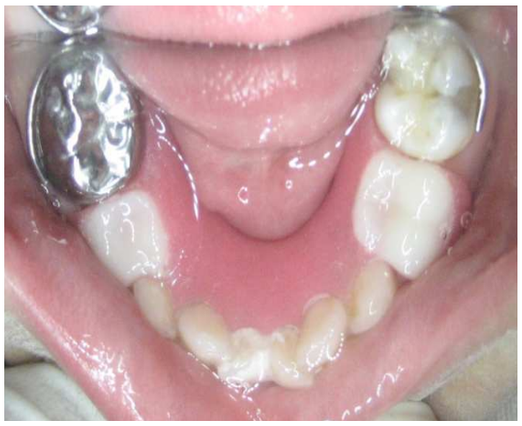


Fig.2. Postoperative photographs

Discussion

Cornelia de Lange or Brachmann de Lange syndrome is a rare congenital disorder of unknown aetiology. The possibility of diagnosing this syndrome at birth is about 1 out of 40,000.[6] 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.[7,6,8,9] In our opinion, the patient presents the typical facial characteristics of CDLS. The clinical findings of the reported case closely confirm with the classical picture of CDLS.[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[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.[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[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.[14] 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.12 Yamamoto etal.[15] have reported two cases with delayed tooth eruption and microdontia, with one of these cases being a partial anadontia.

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

1. Braddock SR, Lachman RS, Stoppenhagen CC, Carey JC, Ireland M,Moeschler JB, et al. Radiological features in Brachmann-de Lange syndrome. Am J Med Genet 1993; 47:1006-13.
2. Beck B, Fenger K. Mortality,Pathological Findings and Causes of Death in the De-Lange Syndrome.Acta Paediatr Scand 1985;74:765-9.
3. Jackson L, Kline AD, Barr MA, Koch S.de Lange syndrome: A clinical review of 310 individuals.Am J Med Genet 1993; 47:940-6.
4. Yamamoto K, Horiuchi K, Uemura K, Shohara E, Okada Y, Sugimura M. Cornelia de Lange syndrome with cleft palate. Int J Oral Maxillofac Surg 1987; 16:484-91.
5. Huang WH, Porto M. Abnormal first-trimester fetal nuchal translucency and Cornelia de Lange syndrome. Obstet Gynecol 2002; 99:956-8.
6. Aitken DA, Ireland M, Berry E, Crossley JA, Macri JN, Burn J, et al.Second-trimester pregnancy associated plasma protein-A levels are reduced in Cornelia de Lange syndrome pregnancies. Prenat Diagn1999; 19:706-10.
7. Grau Carbó J, López Jiménez J, Giménez Prats MJ, Sànchez Molins M. Cornelia de Lange syndrome: a case report. Med Oral Patol Oral Cir Bucal 2007; 12:E445-8.
8. Allanson JE, Hennekam RC, Ireland M. De Lange syndrome: subjective and objective comparison of the classical and mild phenotypes. J Med Genet 1997; 34:645-50.
9. Barrett AW, Griffiths MJ, Scully C. The de Lange syndrome in association with a bleeding tendency: oral surgical implications. Int JOral Maxillofac Surg 1993; 22:171-2.
10. Braddock SR,Lachman RS,Stoppenhagen CC,et al.Radiological features in Brachmann-De Lange Syndrome.Am J Med Genet 1993;47:1006-13.
11. Ireland M.Cornelia De Lange Syndrome: Clinical Features,Common Complications and Long Term Prognosis.Curr Pediatr 1996;6:69-73.
12. Van Alien Mi,Filippi G,Siegel-Bartelt J et al.Clinical Variability within Brachmann De Lange Syndrome: A Proposed Classification System.Am J Med Genet 1993;47:947-58.

13. Preus M,Rex AP: Definition and Diagnosis of the Brachmann-de Lange syndrome.Am J Med Genet 1983,16:301-12.
14. Allanson JE, Hennekam RC, Ireland M:De Lange Syndrome:subjective and objective comparision of the classical and mild phenotypes. J Med Genet 1997, 34:645-50.
15. Krantz ID,McCallum J,DeScipio C,Kaur M,GillisLA,Yaeger D et al.Cornelia de Lange syndrome is caused by mutations in NIPBL,the human homolog of Drosophila melanogaster Nipped-B. Nat Genet 2004, 36:631-5.

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