ORAL FEATURES IN CORNELIA DE LANGE SYNDROME

Ambika L[1*] & Raghavendra B Nayak[2]

1. Lecturer, Department of Oral Medicine and Radiology,
   School of Dental Sciences, Krishna Institute of Medical Sciences University,
   Karad-415110, Maharashtra (State), India.
2. Assistant Professor, Department of Psychiatry,
   JN Medical College and Hospital, KLE Deemed University,
   Belgaum-590010, Karnataka (State), India.

***Corresponding Author:** ambikamds@gmail.com OR rbn.psych@gmail.com

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ABSTRACT

Cornelia de Lange syndrome (CdLS) is a multisystem malformation syndrome. There is wide clinical variability in this disorder. This disorder is relatively uncommon and characterised by series of malformations which includes skeletal, craniofacial, gastrointestinal and cardiac malformations. The main clinical feature includes growth retardation, limb abnormalities, mental retardation, developmental delay and facial discrepancies. Here we report Cornelia De Lange syndrome in an adult patient with characteristic limb abnormalities and distinctive oral features.

Keywords: Cornelia de Lange; facial features; limb defects; oral features

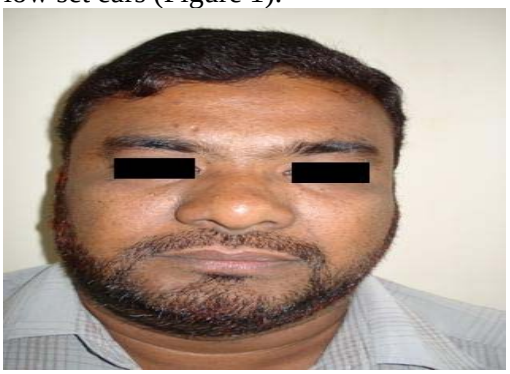
1. INTRODUCTION

Cornelia de Lange Syndrome (CdLS) is also known as Brachmann-De Lange Syndrome, De Lange Syndrome, Typus degenerativus amstelodamensis etc. It was first described by Cornelia De Lange (1933).[1] The incidence of CdLS reported to vary from 1:30,000 to 1:50,000 of live births, without any known racial predilection.[2] It is a multisystem malformation syndrome recognized primarily on the basis of characteristic facial dysmorphism, including low anterior hairline, arched eyebrows, synophrys, anteverted nares, maxillary prognathism, long philtrum, thin lips and 'carp' mouth, in association with prenatal and postnatal growth retardation, mental retardation and in many cases, upper limb anomalies.[3] About 50 to 60% of the cases of CdLS are due to mutation in the NIPBL gene. [3,4] The diagnosis of this disorder is based solemnly on clinical grounds. There are no biochemical markers or chromosomal marker1.

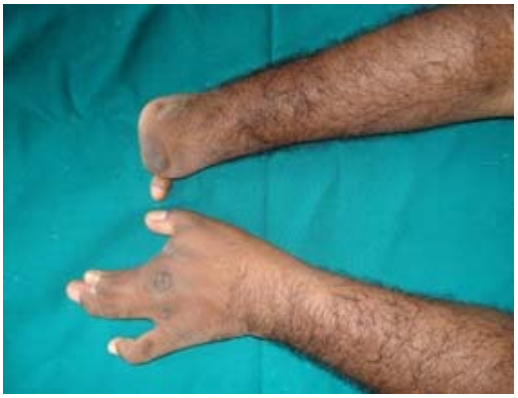
2. CASE REPORT

A 45- year old male patient reported to the Department of Oral Medicine and Radiology with the complaint of decayed teeth. Patient was known hypertensive since 2 years for which he was on regular medication. There was no significant family history. There was no history of consanguineous marriage of her parents. His mother was not exposed to any teratogenic drugs during pregnancy. He was the only son with normal natal and perinatal histories. He had mild delay in developmental milestones. General physical examination revealed short stature (Height–5 feet 2 inches) and stocky built. His weight was 80 kgs and head circumference was 56 cms. There were no cardiac, gastrointestinal and other systemic abnormalities. Psychiatric evaluation of the patient was done with the help of psychiatrist and patient was found to be normal. Extra oral examination revealed brachycephaly, hirsuitism, thick curly hair, round face, short neck, low hair line, thick

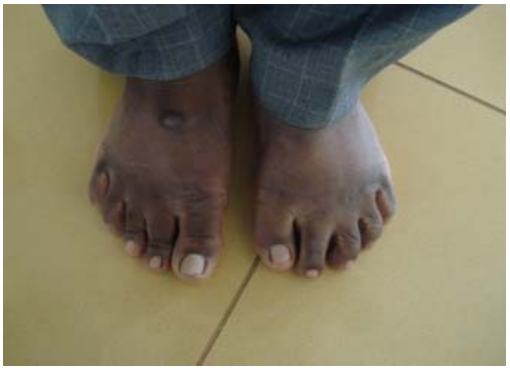
eyebrows, synophrys, hypertelorism, presence of epicanthal fold, short nose and low set ears (Figure 1).



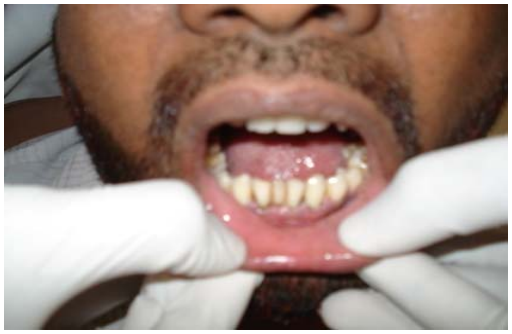
His upper limb defects included presence of only rudimentary thumb in the right hand, absence of forefinger and syndactyly of 3rd and 4th digits on the left hand (Figure 2).



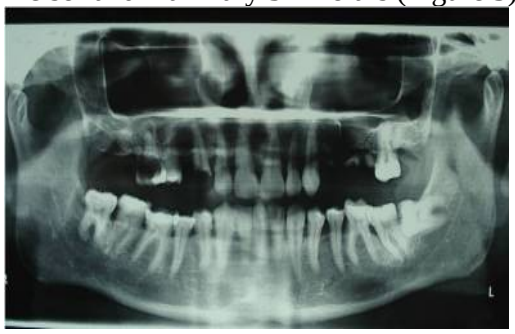
Patient also had lower limb defect such as short and wide feet with grayish black pigmentations (Figure 3).



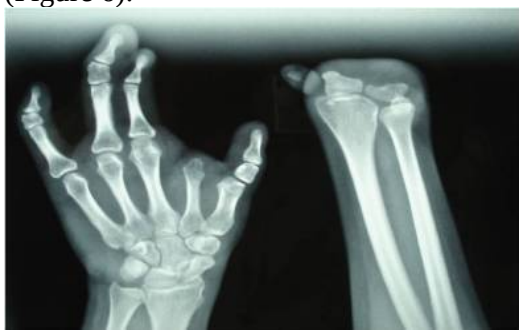
On oral examination patient had generalized microdontia, multiple carious teeth, absence of maxillary 3rd molars and mandibular central incisor, high arched palate and partial ankyloglossia (Figure 4).



Later patient was sent for other biochemical investigations which were found to be normal. On radiological investigation, Orthopantomogram showed generalized microdontia, curved roots, multiple carious teeth, impacted mandibular 3rd molar, missing mandidular incisor and maxillary 3rd molars (Figure 5).



Hand wrist radiograph showed complete absence of carpals and presence of only two metacarpals and rudimentary thumb in the right hand, short phalanges and missing middle phalanges in the left hand (Figure 6).



Based on history, clinical features and investigations a diagnosis of Carnelia de Lange syndrome was given. Further patient‘s dental problems were managed with regular dental treatment.

3. DISCUSSION

CdLS is a multisystem malformation syndrome involving congenital malformations, growth retardation, and neurodevelopmental delay. The characteristic features reported earlier were round face, short neck, growth failure, anomalies of development of hands and feet, short stature, excessive growth of hair, heavy eyebrows, synophrys, long eyelashes, low set ears, thick curly hair, low hair line, small nose with anteverted nares, hirsuitism etc.[1] These features were characteristic in the current case.

Ireland et al. (1993) in a review of 31 cases previously diagnosed as having de Lange Syndrome concluded that the facial findings were at of greatest diagnostic value. These were the combination of the characteristic eyebrows, long philtrum, thin lips, and crescent-shaped mouth. According to his review these combination of anomalies was absent in postpubertal males but not in postpubertal females.[5] Our patient is postpubertal and because of which long philtrum, thin lips and crescent-shaped mouth absent.

Van allen et al (1993) proposed a classification system for CdLS. Type I ‘classic’ patients have the characteristic facial and skeletal changes of CdLS. Type II ‘mild’ CdLS patients have similar facial and minor skeletal abnormalities that were noted in the type I; however these changes may develop later or partially expressed. Type III ‘phenocopies’ CdLS includes the patient who have the phenotypic manifestations of CdLS, which are casually related to chromosomal aneoploidies and teratogenic exposures.[1] Based on the given classification our case belongs to Type I.

Braddock et al. (1993) presented a review of the radiologic features of de Lange syndrome. The classic radiographic manifestations include microcephaly, limb and digital anomalies, delayed skeletal maturation.[6] In the present case characteristic upper limb anomalies (phocomelia, syndactaly) and partial lower limb defects were evident.

Jackson et al. (1993) in his clinical review of 310 cases of CdLS demonstrated a higher proportion of mildly affected cases. In only his study 27% of the cases had the upper limb deficiencies commonly associated with the syndrome. [7] Ullrich (1951) observed short feet without any malformation of lower limbs in his study.[8] In the current case severe upper limb abnormalities were noticed and lower limbs defects were in agreement with Ullrich.

Schlesinger et al. (1963) described radiologic anomalies associated with CdLS. 'The characteristic flat spade-like appearance of hands and short tapering fingers, inward curving of fifth finger were described in his study.[9] Here short tapering fingers and clinodactyly of little finger was observed in the left hand.

Other findings reported in the previous literature were microcephaly, strabismus, hypoplastic nipples, flexion contractures of elbows, high arched palate, macroglossia, etc.[1,2] Examination of our patient revealed high arched palate and partial ankyloglossia which were the additional features.

Dental findings reported earlier were delayed eruption, spacing of teeth, micro- or macrodontia, partial anodontia, carious teeth etc.[1] In the current case microdonts, conical shaped teeth, dental caries, missing mandibular incisor and maxillary third molars were recorded. Other dental findings which were evident in the Orthopantomograph were generalized

curved roots and impacted mandibular 3rd molar which were not quoted in the previous literature.

CONCLUSION:

Cornelia De Lange syndrome is a rare disorder with distinctive clinical and oral features. This congenital disorder includes series of malformations. In the current case the characteristic facial features, limb defects and distinctive oral features helped us in diagnosing this rare entity and helped us for further management. The features like partial ankyloglossia, dilacerated roots and impacted mandibular 3rd molar which were noted may of clinical importance in diagnosing this rare entity.

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