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Dental Management of a Rare Case of Cornelia de Lange Syndrome

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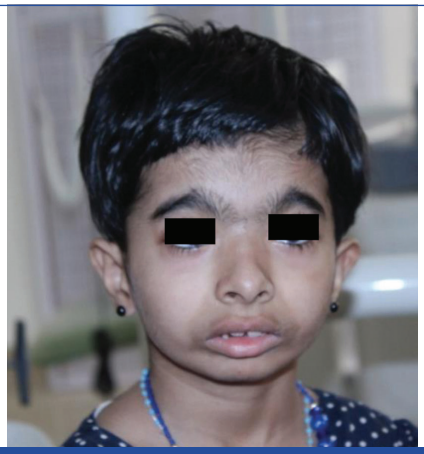
A six-year-old female patient with Cornelia de Lange Syndrome (CdLS) presented to the Department of Paediatric and Preventive Dentistry with multiple mutilated teeth and an inability to chew, resulting in low food intake. The patient complained of nocturnal pain and disturbed sleep. A behavioural analysis of the child showed that she was introverted, had difficulty in interacting with other children or strangers, and exhibited distinctly negative behaviour, as per Frankel's behaviour rating scale.

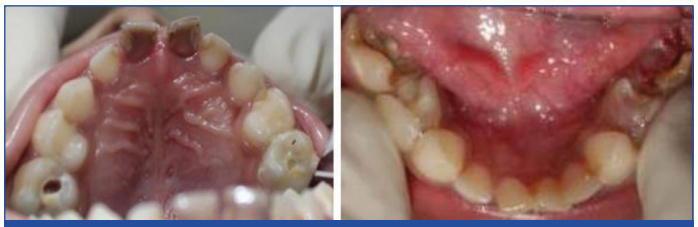
Extraoral findings included facial dysmorphism, a triangular face, low frontal hairline, thick and highly arched eyebrows, widely spaced eyes (hypertelorism) with mild epicanthal fold, a shallow and protruded philtrum, and a wide mouth with downward-slanting corners [Table/Fig-1,2]. No skeletal deformities were noted. These facial findings were suggestive of CdLS.



[Table/Fig-1]: Extraoral features: Low frontal hairline, highly arched thick eyebrows, widely spaced eyes (hypertelorism) with mild epicanthal fold, triangular face, shallow protruded philtrum and wide mouth with downwards slanting corners.

[Table/Fig-2]: Extraoral features.

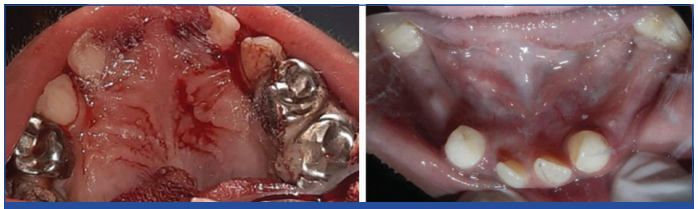
Intraoral findings included narrowed mandibular and maxillary arches, a high-arched palate, and crowding of teeth [Table/Fig-3,4]. Dental caries were observed in relation to 52, 62, and 63, with deep dental caries in relation to 55, 65, 64, 51, 52, 74, 75, 84, and 85. Due to the child's inability to cooperate during dental treatment because of her mental disability and difficulties in managing her behaviour, it was decided that full mouth rehabilitation would be done under



[Table/Fig-3]: Intraoral findings- Maxillary arch.
[Table/Fig-4]: Intraoral findings- Mandibular arch. (Images from left to right)

General Anaesthesia (GA). The major challenge in performing the procedure under GA was the child's airway features, specifically micrognathia, restricted mouth opening, receding chin, and stiff neck. These features hindered the visualisation of the vocal cords during laryngoscopy and Endotracheal Tube (ETT) placement. To manage the narrow airway of the patient, a modification in the Ryles tube was performed. An Oral/Nasal Endotracheal Tube (ONETT) was used in the present study. The family was fully informed about all clinical procedures, and the parents signed an informed consent form before the treatment.

Pulpectomy was performed on teeth 55 and 65, followed by the placement of Stainless Steel Crowns (SSCs) [Table/Fig-5]. Silver Diamine Fluoride (SDF) was applied, followed by the Hall technique in relation to teeth 54 and 64. Additionally, SDF application was done with respect to teeth 52, 53, 62, and 63, and extractions were performed for teeth 51, 61, 74, 75, 84, and 85. Immediate post-operative images of the mandibular arch could not be taken due to profuse bleeding. A follow-up image has been added as [Table/Fig-6]. The patient was followed-up after six months and one year, showing an improvement in dental hygiene and no new caries. Thus, there was an overall improvement in the quality of life and general health of the patient.



[Table/Fig-5]: Postoperative picture- Maxillary arch.
[Table/Fig-6]: Follow-up of mandibular arch. (Images from left to right)

DISCUSSION

In 1916, Dr. Brachmann initially described CdLS, also known as Brachmann-de Lange syndrome. Two cases of this syndrome were reported by Dr. Cornelia de Lange in 1933 [1]. CdLS is a genetic syndrome that affects between 1 in 10,000 and 1 in 60,000 neonates, with no racial predilection. It is more commonly observed in females (1.3:1) [2]. The molecular and genetic basis of this syndrome is not completely clear. However, it is thought to involve a dominant mutation in five genes, namely Nipped-Blike Protein (NIPBL), Structural Maintenance of Chromosomes 1A (SMC1A), Structural Maintenance of Chromosomes 3 (SMC3), Human

Homolog of Schizosaccharomyces pombe radiation-sensitive mutant 21 (RAD21) and Hstone Deacetylase 8 (HDAC8), which have been associated with CdLS [3].

Anomalies in limb structure, delayed growth and development, mental retardation, hirsutism, and distinctive facial characteristics are the main clinical features of this syndrome [4]. Regarding mental retardation, most cases fall under the profoundly disabled category. The Intellectual Quotient ranges from below 30 to 86 (average: 53), and initial hypertonicity is a common finding that hinders their performance [5].

Extraoral findings include micrognathia, down-slanted mouth angles, a long shallow philtrum, and a prominent symphysis. Intraoral examination usually reveals delayed eruption with a high-arched palate [6].

The aim of the present case report was to describe the challenges faced during the dental management of patients with CdLS, to identify the clinical features, and to demonstrate how treatment resulted in the improvement of the quality of life for these patients.

The above case presents the main characteristics of this syndrome, which have been well-documented previously. CdLS not only exhibits clinical but also genetic heterogeneity and, so far, pathogenic Deoxyribonucleic Acid (DNA) sequence variation. The genes responsible for CdLS are NIPBL, SMC1A, SMC3, RAD21, and HDAC8 [7]. Mutations in NIPBL, SMC3, and RAD21 lead to the autosomal dominant form of CdLS (AD-CdLS), while the causative genes in X-linked CdLS are SMC1A and HDAC8. Among the five genes, the NIPBL gene accounts for about 80% of the mutated cases [8].

Due to the difficulty in managing the child's behaviour and her mental disability, it was decided to perform full mouth rehabilitation under GA. The present case report mainly emphasises the dental management of CdLS.

Even for an experienced clinician, diagnosing mild cases of this syndrome is a challenging task. Complications associated with CdLS, both medically and developmentally, make dental treatment difficult. The prime concern in treating such patients is the management of the airway. Difficult airway should thus be expected in every case of CdLS, and proper preparations for airway management should be conducted. To manage the narrow airway in this patient, modifications were made to the Ryles tube. Special attention was given to the patient throughout the postoperative period. Managing patients with CdLS requires a multidisciplinary approach that includes nutritionists, cardiologists, geneticists, nephrologists, gastroenterologists, ophthalmologists, paediatric dentists, and paediatricians [9].

The CdLS is a complicated disorder in which multiple body systems are affected, and understanding the role of a paediatric dentist in treating patients with CdLS is important. It is not just limited to treating dental defects. Dental management and behaviour shaping of such children are crucial to deliver quality dental care. Thorough evaluation and early intervention are necessary in these patients, as they are at a higher risk for self-inflicting oral habits, orthodontic problems, and dental caries.

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