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Case Study

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CORNELIA DE LANGE SYNDROME: ONCE IN A BLUE MOON

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ABSTRACT

Background: A congenital scare genetic disorder is Cornelia de lange syndrome. Prevalence of cornelia de lange syndrome is variable, ranging from 1:10,000 and 1:100,000 birth in different population. There is no gender predilection of cornelia de lange syndrome. Diagnosis of this syndrome based on distinctive facial and other systemic features. **Case:** Here for the first time a case of Cornelia de lange syndrome from Western Maharashtra, 15 year male who was reported to department of oral medicine and radiology with chief complaint of decayed teeth and on the basis of family, past medical history and clinical examination master boy was diagnosed with CDLS. **Conclusion**: This was first case report of Cornelia de lange syndrome in Western Maharashtra.

KEYWORDS: Cornelia de lange syndrome, Hirsutism**,** Cutis marmorata**,** Micromelia and macromelia polydactyl.

INTRODUCTION

Brachmann de-Lange syndrome is other name for cornelia de lange syndrome (CDLS). A congenital scare genetic disorder is Cornelia de lange syndrome. Epidemiology of cornelia de lange syndrome is variable, ranging from 1:10,000 and 1:100,000 birth in different population. There is no gender predilection of cornelia de lange syndrome. CDLS is a multisystem congenital syndrome shows characteristic features of growth retardation, congenital anomaly, developmental delay etc.[1,3,4,5]

CASE REPORT

A male age 15 year reported to the Department of Oral Medicine and Radiology, with the chief complaint of decayed teeth since a year. Master boy was asymptomatic a year back; he never underwent a dental visit. His parents asked for a dental consultation because they thought the child felt decayed teeth and swelling in upper and lower arch. The child was not able to chew or swallow. Drooling was present in child. **Past family & medical history:** On taking the history, He was a child of **nonconsanguineous marriage,** it was known that after delivery the baby did not cry. He was kept under observation in neonatal intensive care unit for 30 days. He was admitted with compliance of delayed developmental milestones. He was diagnosed to have several developmental defects and left mild hearing loss.

Past dental history: He never underwent a dental visit.

Oral hygiene Habit: Parents reported daily brushing and no fluoride assumption. Behavior management of the child was a challenge as the child was hyperactive and had low attention span.

GENERAL, EXTRAORAL EXAMINATION

General examination revealed….

- **Facial symmetry**: Symmetrical (Fig.1)
- **Growth:** Retarded osseous maturation.
- **Development:** Mental retardation, grossly delayed milestones, Initial hypertonicity, low pitched, weak, cry in infancy.
- **Cranium**: Microbrachycephaly. (Fig.2)
- **Ocipit view**: flat (Fig.2)
- **Eyes**: Bushy eyebrows and synophrys, long, curly eyelashes, history of epiphora of left eye till age of 12 years. (Fig.1)
- **Nose**: Depressed nasal bridge**, anteverted nares.** (Fig.1)
- **Mouth**: Long philtrum, thin upper lip, and, narrow-arched palate, delayed eruption,
- **Mandible**: Micrognathia
- **Skin**: Hirsutism**,Cutis marmorata (on lower abdomen)**
- **Hands and arms**: Micromelia and macromelia polydactyl of fifth fingers. (Fig.4)

- **Feet**: Micromelia
- **Other**: Myopia and nystagmus, low posterior hairline,short neck, partial hearing loss,

INTRA ORAL EXAMINATION

Hard tissue examination: (Fig.3)

- Grossly decayed with 32,43,44
- Root piece with 16,26,36,46
- Missing with 41,42
- Over-retained root with 55,65
- Maxillary anteriors proclination
- Intraoral examination revealed low narrow arch palate with soft bone. Tongue movement appears to be normal. (Fig.5)

Fig no. 1: Facial profile

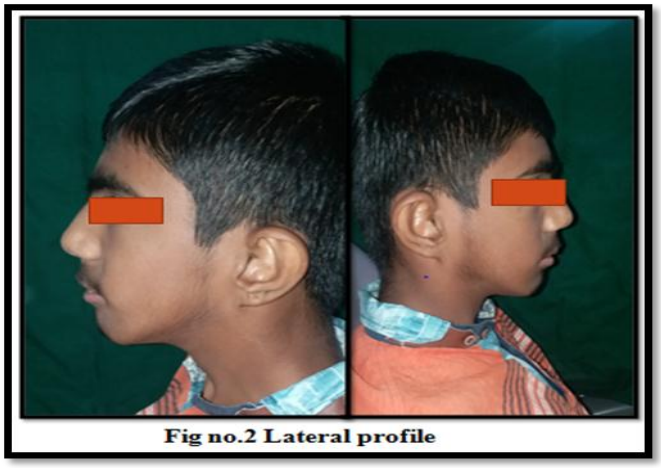

Fig no.2 Lateral profile

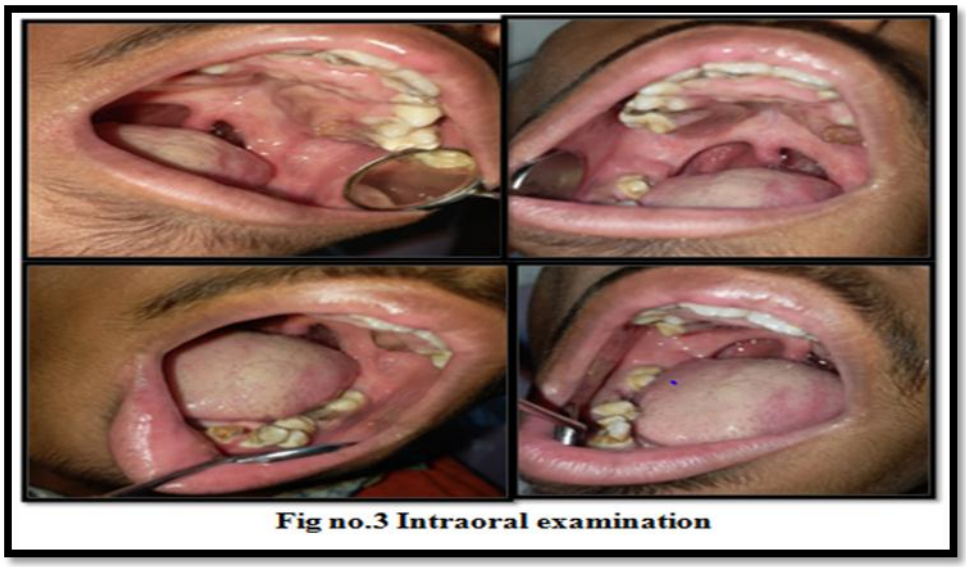

Fig no.3 Intraoral examination

Fig no 4: polydactyl of fifth fingers.

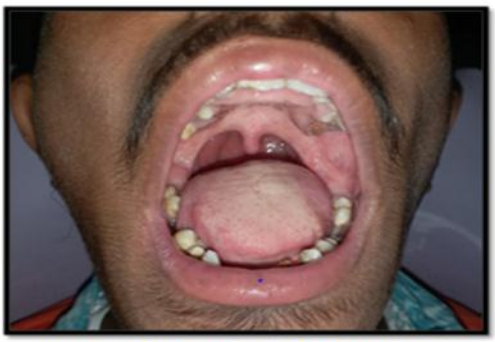

Fig no. 5: Low arched palate

Provisional diagnosis

1. Grossly decayed with 32,43,44
2. Root piece with 16,26,36,46
3. False partial anodontia with 41,42
4. Over-retained root with 55,65
5. Maxillary anterior proclination

On the basis of family and past medical history, our working diagnosis was **Cornelia de Lange Syndrome.**
After all confirmatory findings, our final diagnosis were

1. Grossly decayed with 32,43,44
2. Root piece with 16,26,36,46
3. False partial anodontia with 41,42
4. Over-retained root with 55,65
5. Maxillary anteriors proclination
6. **Cornelia de Lange Syndrome.**

Differential diagnosis

1. Coffin-Siris syndrome
2. Rubinstein-Taybi syndrome
3. Ruvalcaba syndrome

Treatment

A comprehensive dental treatment was planned and all grossly carious root pieces, over retained and teeth were extracted. Oral prophylaxis and fluoride application was done and regular 6 months follow-up was scheduled. Management of such patients is done by team approach including dental surgeon, cardiologist, ophthalmologist, ENT specialist, speech therapist and occupational therapist. Family support is also essential, especially at the time of diagnosis. It is important to provide the family with information on the syndrome, which could help parents to cope emotionally and cooperate with regard to the child's Treatment.[2,3,4]

DISCUSSION

It was first described by Vrolik in 1849. The first ever documented case was in 1916 by Brachmann followed by Cornelia de Lange, a Dutch Pediatrician, in 1933 after whom the disorder has been named.[1,5,6,7]

Various synonyms for this syndrome are

- Brachmann-de Lange syndrome
- de Lange syndrome.
- Bushy syndrome,
- Amsterdam dwarfism.

Van Allen *et al. proposed a classification system based* on the presentation of different types[8]

- 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 as that of Type I; however, these changes may develop later or may be partially expressed.
- Type III phenocopies CdLS includes the patients who have phenotypic manifestations of CdLS, which are causally related to chromosomal aneuploidies or teratogenic exposur.

LIMITATION OF CASE REPORT: RADIOGRAPHIC INVESTIGATIONS

Master boy was uncooperative for x ray and any investigation. In literature radiographic features of bushy syndrome are as follows….

- Schlesinger *et al. 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.[5,6]
- Brylewski reported that a large majority of such patients have IQ below 50.

- Radiographic examination included orthopantomogram, lateral cephalogram, and hand wrist X-ray. Orthopantomogram revealed partial anodontia. Cephalogram revealed protruded maxillary incisors, horizontal growth pattern, and delayed growth spurt Hand wrist X-ray revealed short first metacarpal, clinodactyly of fingers, and hypoplastic appearance of epiphyseal center in relation to ulna.[5,6,7]

CONCLUSION

In conclusion, preventive revisions starting in infancy and coordination with the pediatrician are necessary. Routine reviews for every 6 months facilitate the changes in orofacial growth, detection of pathologies, and maintenance of good oral hygiene at home.

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Conflicts of interest: There are no conflicts of interest.

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