DOI: 10.7860/JCDR/2022/56010.16661

A Rare Case of Ectrodactyly Ectodermal Dysplasia and Cleft Lip Syndrome

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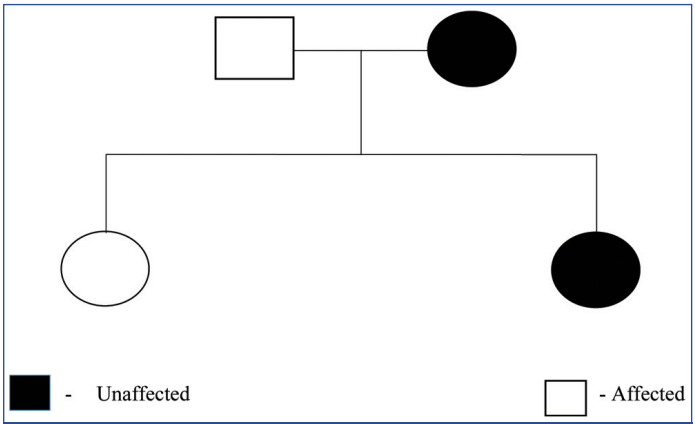
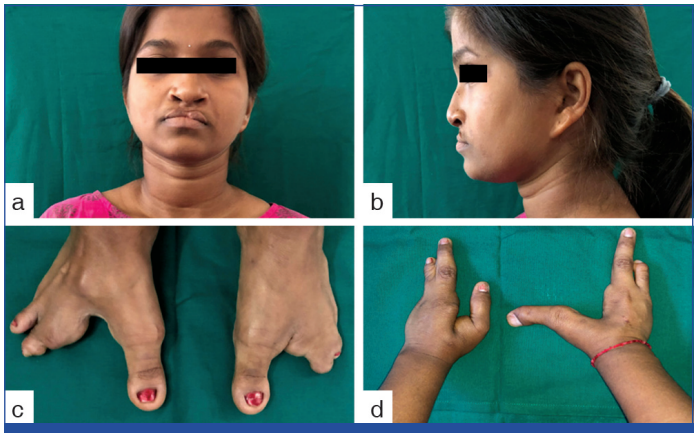
Keywords: Autosomal dominant disorder, Genetic, Multidisciplinary

A 23-year-old female patient visited the Oral Medicine and Radiology Department with the complaint of irregularly placed upper and lower front teeth since childhood. The patient was a known case of cleft lip, and alveolus and had undergone repair of lip when she was 2 months old. She was born to non consanguineous parents. Her father was a known case of cleft lip. Neither her maternal nor her paternal ancestors had a similar anomaly. [Table/Fig-1] shows an elaborate family history of the patient. Informed consent was taken from the patient before the examination.

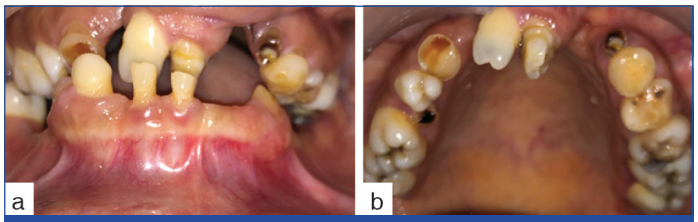


[Table/Fig-1]: Pedigree chart.

The skin had xerosis with anhidrosis. Scalp hair appeared to be hypopigmented and dry. Body hair was sparse [Table/Fig-2a]. Eyelashes were also sparse. The patient had a mesocephalic head shape, a slightly depressed nasal bridge, a retrognathic maxilla, and a prognathic mandible [Table/Fig-2b]. A surgical scar on the left side of the upper lip was observed unilaterally that extended up to the philtrum [Table/Fig-2a]. The canthal index was normal [1]. Presence of V-shaped clefts in all four limbs was observed. Feet revealed the absence of a second toe and fusion of third, and fourth toes with a cleft between the fused toes and first toe [Table/Fig-2c]. The hands of the patient showed an absence of index and middle fingers [Table/Fig-2d]. The mouth opening was adequate (38 mm). No abnormalities were evident in the temporomandibular joint and ear region. Notching of maxillary central incisors was observed. Multiple carious teeth were present. Oligodontia in respect to (wrt) 12, 13, 15, 18, 22, 25, 28, 31, 32, 33, 34, 35, 38, 41, 42, 43, 44, 45, 46, 48 with multiple retained deciduous teeth wrt 53, 55, 63, 65, 71, 73, 81, 83, 84 was evident. The total number of teeth present was 20. The number of deciduous teeth present was 9 and permanent teeth were 11 [Table/Fig-3a]. Grade III mobility was noted in the mandibular left first molar. Root stumps were observed in relation to 36. Anterior crossbite was detected. Unilateral cleft was observed in the premaxilla. [Table/Fig-3b] A provisional diagnosis of Ectrodactyly-Ectodermal Dysplasia and Cleft (EEC) syndrome syndrome was given. Differential diagnoses considered were Ankyloblepharon ectodermal defects cleft lip/palate (AEC) syndrome, Limb Mammary Syndrome (LMS), and Acro-dermato-ungual-lacrimal-tooth (ADULT) syndrome.



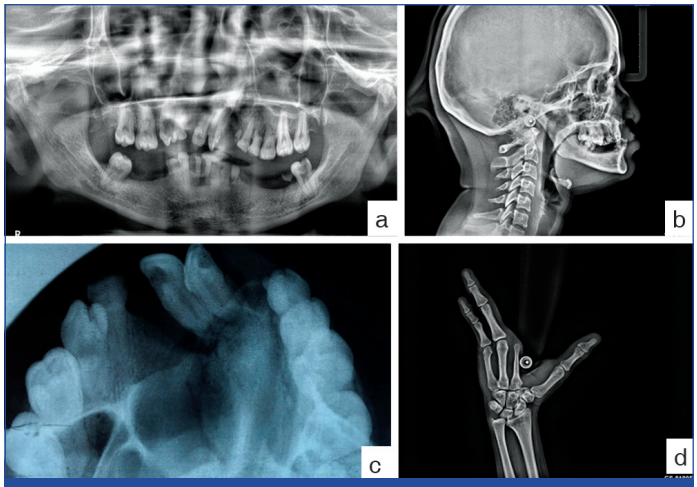
[Table/Fig-2]: Extraoral view depicting; a) depressed nasal bridge, sparse eyelashes, a surgical scar in upper lip region; b) midface hypoplasia; c) and d) ectrodactyly of toes and fingers.



[Table/Fig-3]: Intraoral view depicting; a) oligodontia with multiple retained deciduous teeth; b) cleft of the maxillary alveolus.

Radiographic analysis was done which included maxillary occlusal radiograph, Orthopantomogram (OPG), lateral cephalogram, and hand-wrist radiograph. OPG revealed oligodontia with multiple over-retained deciduous teeth, root stump was present wrt 36 and deep dentinal caries wrt 37. Also cleft was observed in the left maxillary alveolus extending up to the medial wall of maxillary sinus [Table/Fig-4a]. Lateral cephalogram revealed anterior crossbite, hypoplastic maxilla [Table/Fig-4b]. Maxillary occlusal radiograph revealed unilateral cleft of alveolus with rotated central incisors, missing lateral incisors bilaterally [Table/Fig-4c]. Hand wrist radiograph revealed the aplasia of second phalanges, metacarpals, the aplasia of third phalanges, and the hypoplasia of third metacarpals [Table/Fig-4d]. Based on the clinical and radiographic features a final diagnosis of ectrodactyly ectodermal dysplasia cleft lip syndrome was made. The treatment plan included extraction of retained deciduous teeth, root stumps; root canal treatment or restoration depending on the depth of the carious lesion; prosthodontic rehabilitation.

Ectrodactyly-Ectodermal Dysplasia and Cleft syndrome is a congenital dysplasia that consists of any one of the cardinal signs in a variable expression [2]. It has an incidence rate of 1 in 90,000 of the general population. Synonyms of this syndrome are split hand/split foot syndrome, lobster claw syndrome, and cleft hand syndrome [2-4]. The three different types of EEC syndrome with gene loci are: EEC syndrome type 1 (Mendelian inheritance in man (MIM) 129900)-7q11.2-q21.3; EEC syndrome type 2 (MIM 602077)-chromosome 19; EEC syndrome



[Table/Fig-4]: a) OPG reveals oligodontia with multiple over-retained deciduous teeth; b) Cephalogram revealed anterior crossbite, hypoplastic maxilla; c) Maxillary occlusal radiograph revealed unilateral cleft of alveolus with rotated central incisors, missing lateral incisors bilaterally; d) Hand wrist radiograph revealed the aplasia of second phalanges, metacarpals, the aplasia of third phalanges, and the hypoplasia of third metacarpals.

type 3 (MIM 604292)-3q27 [2]. Other syndromes associated with the same gene mutations are ADULT syndrome, Rapp Hodgkin syndrome (RHS), AEC syndrome, LMS [5].

A multidisciplinary approach is necessary to treat this syndrome, including a Dentist, Ophthalmologist, Dermatologist, Audiologist, Nephrologist, and Plastic surgeon. Reassurance and counselling to parents regarding the low risk of mental retardation need to be done [6]. From a dental point of view, in managing this syndrome, the steps to be followed are restoring carious teeth, prosthesis to replace the missing teeth, cleft lip and palate reconstruction, preserving the dentition, and cosmetic use, use of salivary substitutes if xerostomia persists [6,7]. Limb malformation can be repaired through surgery along with management of renal dysfunction, if required. Artificial tear supplements can be used, if xerophthalmia persists. Emollients may be used for dry skin treatment. Ocular inflammation may be treated with topical steroids. Speech and audiology therapy can be beneficial. Genetic counselling is necessary [6].

Acknowledgement

Authors are responsible for the completeness of the information. No source of funding was taken for this report. The authors declared they do not have anything to disclose regarding the conflict of interest concerning this manuscript.

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PLAGIARISM CHECKING METHODS: [Jain H et al.]
- Plagiarism X-checker: Mar 08, 2022
- Manual Googling: Apr 27, 2022
- iThenticate Software: May 09, 2022 (7%)

ETYMOLOGY: Author Origin

AUTHOR DECLARATION:
- Financial or Other Competing Interests: None
- Was informed consent obtained from the subjects involved in the study? Yes
- For any images presented appropriate consent has been obtained from the subjects. Yes

Date of Submission: **Mar 01, 2022**
Date of Peer Review: **Apr 05, 2022**
Date of Acceptance: **May 11, 2022**
Date of Publishing: **Jul 01, 2022**