

Case Report

Ectrodactyly, Ectodermal Dysplasia, and Cleft Lip/Palate Syndrome: A Case Report

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ABSTRACT

Ectrodactyly–ectodermic dysplasia-cleft lip/palate (EEC) syndrome is a rare congenital anomaly with varying clinical features. It includes clefting in hands and feet, cleft lip and palate, and defects in ectodermic derived structures such as skin, nails, hair and teeth. Management of this type of patient requires a multidisciplinary approach. The purpose of this case report is to describe a case of 16 year old female suffering from this syndrome treated conservatively in our institution.

Key-words: EEC syndrome, Ectrodactyly, Cleft lip, Ectodermal dysplasia

INTRODUCTION

Ectrodactyly–ectodermal dysplasia–cleft lip / palate (EEC) syndrome is a rare autosomal dominant disorder with high penetrance and variable clinical expression. [1] The classic phenotype comprises limb anomalies [split hand / foot malformation (SHFM) and syndactyly], ectodermal dysplasia (anomalies in hair, teeth, nail, skin, sweat gland, lacrimal duct, breast and nipple development) and orofacial clefting. [2] Genitourinary and external ear malformations and hearing loss are occasionally seen. Clinical diagnosis requires the presence of two of the three main features in sporadic cases and of at least one of the cardinal features in familial cases. [3]

CASE REPORT

A 16-year-old female patient reported to the Department of Conservative Dentistry and Endodontics with the chief complaint of congenitally missing teeth and multiple decayed teeth.

Medical history revealed that child was born after 38 weeks of gestation without any complications during pregnancy and delivery and no anomaly was detected during routine prenatal examinations. She was operated for cleft lip and palate during first year of her life. Medical investigations revealed no genitourinary or other systemic abnormality. Developmental milestones were at time with normal mental development. She complaint of recurrent eye and ear infection and was taking medication for the same. Family history was unrelated as the girl was the first child of healthy unrelated parents. She is having normal younger brother.

Extraoral examination showed ectrodactyly of both hands and feet [Figure 1, 2]. Right hand showed incompletely formed intermediate phalanx and missing distal phalanx of index finger [Figure 1]. Feet had four toes with absence of second toe [Figure 2]. There was syndactyly between third, fourth and fifth toe. Radiograph showed presence of phalanges

of all toes, except second toe, which was completely missing [Figure 2].



Figure 1: Ectrodactyly of hands (A, B)



Figure 2: Ectrodactyly of feet (A,B).

Her face was narrowed with a broad nose, flat nose tip and a prominent chin [Figure 3]. Her upper lip showed surgical scar related to cleft lip operation. She had dry skin with sparse hair on the body, scalp and sparse eyelashes [Figure 4]. Hair present were fine in texture and lighter in color [Figure 5] and she complaint of delayed hair growth and was taking hair supplements. Her skin was warm and dry.



Figure 3: Facial view showing narrow face, broad nose, flat nose tip



Figure 4: View showing sparse eye lashes



Figure 5: View showing light hair color



Figure 6: Multiple carious teeth and congenitally missing teeth in upper and lower arch (A,B), pre-operative orthopantomograph (C)

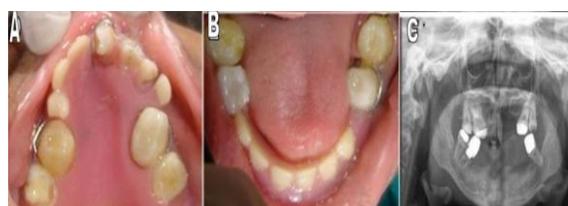


Figure 7: Postoperative view after endodontic treatment, composite build-up, removable partial denture in upper and lower arch (A,B), postoperative orthopantomograph (C).

Intraoral examination revealed congenitally missing permanent lower anteriors, upper lateral incisors, canines and premolars, upper lateral incisor was rotated, decayed D, E were present in upper arch. Upper and lower first and second molars were carious [Figure 6]. Extraction of

retained deciduous teeth was done. Root canal treatment was completed in right maxillary and mandibular first molar. Composite build-up was done in right maxillary first and second molars, left maxillary first molar, left mandibular first molar, right mandibular first and second molars. Removable cast partial dentures were prepared [Figure 7].

DISCUSSION

EEC syndrome is a rare congenital disease of autosomal dominant transmission, first described by Eckholdt and Martens in 1804. [4] In 1970, Rudiger et al [2] introduced the acronym EEC (Ectrodactyly-ectodermal dysplasia-clefting), the current name for the syndrome. Patients affected by EEC syndrome characteristically present ectrodactyly (84% of cases) also known as “lobster claw” hands and feet, which is caused by abnormal development of the hands and feet with an alteration of the central axis of the digits, absence of digits, a deep midline cleft, and fusion of some of the remaining digits. [5] There may also be abnormalities of the teeth and hair: hypoplastic or absent teeth, or early loss of the permanent teeth due to caries (77% of patients), cleft lip with or without cleft palate (68% of cases), and abnormalities of the lachrymal system (59% of cases). [5] It is believed that the classical cases of EEC syndrome are due to p63 gene mutation that is inherited as autosomal dominant trait, but sporadic occurrence is also reported. [1] Our case may be an example of sporadic occurrence as there was no family history. In our case, ectrodactyly was present with respect to right-hand index finger, along with the syndactyly of third, fourth, fifth toes of both right and left feet. Buss et al. [6] (1995) identified 24 cases of EEC syndrome as part of a nationwide study in the UK. Ectodermal dysplasia, by study definition, was present in all cases and hair and teeth were universally affected. According to Buss et al. [6] a diagnosis of EEC can be made based on ectodermal dysplasia with

one of the following features ectrodactyly, cleft lip or palate and lacrimal duct abnormalities. Our case showed similar problems as that of Bowen and Armstrong [7] showing syndactyly, ED with abnormalities of scalp hair, and teeth and hidrotic problems, but with unilateral cleft lip and palate instead of bilateral cleft lip and palate. Sparse and kinky hair with small, carious teeth, and broad nose and a prominent chin gave a typical facial appearance to our patient, similar to cases of Brill et al. [8] Mental retardation has been reported in the EEC syndrome [2] but appears to be infrequent and the same was present in our case.

Prenatal detection of specific chromosomal abnormalities of this type of syndrome marks an important step forward for those patients wishing to have children. Prenatal detection of abnormalities of the p63 gene of chromosome 3 has already been used successfully, and healthy children have been born to parents with EEC syndrome. [9]

Other syndromes with overlapping features, such as the Rapp Hodgkin syndrome and Ankyloblepharon, Ectodermal dysplasia, Cleft lip/Palate (AEC) syndrome should be considered in the differential diagnosis. Management of these patients must be multidisciplinary. This should start with the surgical correction of defects that could cause a functional deficit, such as problems of phonation and hearing due to cleft palate, difficulties walking or handling objects due to ectrodactyly, and abnormalities of the genitourinary system. Dental management should include preservation of the primary dentition with the use of fluoridated mouthwashes & salivary substitutes, restoration of carious teeth and replacement of missing teeth. Artificial tears and topical antibiotics may be required to avoid ocular problems.

CONCLUSION

The treatment of EEC syndrome is directed toward the specific symptoms that are apparent in each individual and may

require the coordinated efforts of a team of specialists depending on its phenotypic manifestations.

ACKNOWLEDGEMENTS

The authors deny any conflicts of interest related to this study.

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How to cite this article: Gupta S, Mittal S, Tewari S et al. Ectrodactyly, ectodermal dysplasia, and cleft lip/palate syndrome: a case report. *Int J Health Sci Res.* 2017; 7(4):459-462.
