

Case Report

**Incomplete Ectrodactyly Ectodermal Dysplasia Cleft Lip/Palate Syndrome-
Report of 3 Cases****Rajput Chetan¹, Patil Amit², Shameela Aafreen³, Gore Sanjay⁴**^{1,4}Assistant Professor, Department of Skin and VD, S.B.H Government Medical College, Dhule-424001, Maharashtra, India²Assistant Professor, Department of Otorhinolaryngology, S.B.H Government Medical College, Dhule-424001, Maharashtra, India³Senior Resident, Department of Skin and VD, S.B.H Government Medical College, Dhule-424001, Maharashtra, India***Corresponding author**

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Abstract: Ectrodactyly-ectodermal dysplasia-cleft (EEC) syndrome is a large heterogenous group of disorders with an autosomal dominant inheritance characterized by the triad of ectrodactyly-ectodermal dysplasia, and facial clefting of lip or palate or both along with some systemic manifestations. Presence of all the three major features in a single individual is extremely rare, incidence being approximately 1.5/100 million population. We report 3 cases of EEC syndrome having ectodermal dysplasia, ectrodactyly, syndactyly along with some systemic features that is incomplete form of syndrome.

Keywords: Ectrodactyly, Ectodermal dysplasia, Facial cleft, Syndactyly, EEC syndrome, Incomplete EEC syndrome.

INTRODUCTION

Ectrodactyly-ectodermal dysplasia-cleft (EEC) syndrome is a multiple congenital anomalies characterized by triad of ectrodactyly-ectodermal dysplasia, and cleft lip or plate or both and is a genetic disorder[1]. All these features rarely coexist, in a single individual and the incidence being 1.5/100 million population [2]. Other features may include maxillary hypoplasia, hypertelorism, conductive hearing loss, partial noncanalization of the lacrimal duct leading to obstruction defect, choanal atresia, fair skin with mild hyperkeratosis, renal agenesis, delayed developmental milestones with mental retardation.

In 1961, Rosselli and Gulienetti [2] reported 4 cases with hypohidrosis, hypotrichosis, microdontia, cleft lip/palate, dystrophic nails, deformities of limbs and malformation of genitourinary system. Syndactyly was the prominent digital deformities. Rudiger *et al.* [3] and Freire-Maia [4] in 1970 reported a very similar clinical condition and described it as EEC syndrome i.e. ectodactyly, ED and cleft lip/palate. There are very less number of published cases in India.

CASE REPORT**Case 1**

A 32 years female presented with reduced sweating all over body since birth absence of fingers of both hands and feet since birth. On clinical examination she was having bilateral hypoplastic maxilla and elongated face (Fig.1), lips and palate were normal

except voice with nasal twang. Both hands and feet showed ectrodactyly (Fig. 2). There was dry, lusterless anhidrotic skin with poor heat tolerance.

**Fig. 1: Elongated face with hypoplastic maxilla****Fig. 2: Ectrodactyly and syndactyly with absent fingers and toes**

Otorhinolaryngology and ophthalmic examinations were normal. All routine investigation including chest X-ray and ultrasonography were normal.

Case 2

A 5 year male child presented with hand and foot deformity since birth and absence of sweating since 2 years. On clinical examination there was no cleft lip or palate (Fig. 3 and 5) with hypertelorism (Fig. 6) and generalized dry and anhidrotic skin with absence of index and middle finger of both the hands and splitting and syndactyly i.e. lobster claw deformity of both the feet (Fig. 4).



Fig. 3: No cleft lips seen



Fig. 4: Ectrodactyly with syndactyly of both the hands and the feet (lobster claw deformity)



Fig. 5: No cleft palate seen with normal dentitions



Fig. 6: Hypertelorism seen on ophthalmic examinations

Otorhinolaryngology examinations was normal along with normal all routine investigation including chest X-ray and ultrasonography.

Case 3

A 30 year female presented with decreased sweating all over body and absence of fingers of both hands and feet since birth. On clinical examination there was hypoplastic maxillae with long face (Fig. 7), lips and palate were normal. Both hands and feet showed ectrodactyly with mild hyperkeratotic skin over palms and soles (Fig. 8). Rest all investigations were normal.



Fig. 7: Hypoplastic maxilla and elongated face



Fig. 8: Ectrodactyly and syndactyly of both the hands and feet

DISCUSSION

Ectrodactyly-ectodermal dysplasia-cleft syndrome exists in two forms: One with cleft lip with or without cleft palate and other with cleft palate alone. Both forms are inherited as autosomal dominant traits [6].

Ectrodactyly-ectodermal dysplasia-cleft syndrome is a form of ectodermal dysplasia with defective ectodermal structures such as hair, teeth, nail, skin, sweat and sebaceous glands etc, ectrodactyly and syndactyly as well as facial cleft [1]. Ectrodactyly (absence of all or part of one or more digits) is only constant component in this syndrome. Other features of EEC syndrome that may be present includes maxillary hypoplasia, conductive hearing loss, blue irides photophobia blepharophimosis, blepharitis, dacryocystitis, lacrimal duct abnormalities, sparse body hair, xerostomia, choanal atresia, microdontia, hypoplastic nipples, micropenis, transverse vaginal septum, renal agenesis, bladder diverticula, fair skin and mental retardation (7%) [6].

Genetic research relating to EEC has made great strides in recent years, but many findings are currently being debated in the literature. Chromosome 19, within the region of D19S894 and D19S416 has been postulated as the locus for the abnormalities found in EEC syndrome. This is supported by reports (though conflicting) regarding the association of cleft lip \pm palate on locus 19q, which suggests that EEC could be an allelic variant [11]. More recently, the p63 gene and TP73L gene has been targeted in numerous studies [5]. Interestingly, the p63 gene is a homologue of the tumor suppressor gene p53, [1] though this is not indicative that patients with EEC are more likely to develop tumors.

Here we have reported 3 cases of EEC syndrome which sans facial clefting. All 3 cases have ectrodactyly, ectodermal dysplasia as consistent features with hypertelorism and maxillary hypoplasia noted in 2 cases.

CONCLUSION

Early diagnosis, sympathetic and rational attitude and a multidisciplinary treatment approach of dentists, otorhinolaryngology, ophthalmologists and dermatologists are necessary for the physical, psychological and social rehabilitation of such patients. Management of clinical manifestations is a unique challenge.

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