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EC OPHTHALMOLOGY
Case Report

EEC Syndrome with Ophthalmic Manifestations – A Report of Two Cases

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Abstract

Purpose: To report two rare cases of EEC (Ectrodactyly, Ectodermal Dysplasia, Clefting) Syndrome.

Methods: Through clinical evaluation and radiologycal imaging technique confirm the diagnosis of Ectrodactyly, Ectodermal Dysplasia, Clefting Syndrome.

Results: EEC Syndrome is also known as "Split hand- Split foot- Ectodermal Dysplasia-Cleft Syndrome". EEC is characterized by the triad of ectrodactyly, ectodermal dysplasia and facial clefts. It is a multiple congenital anomaly syndrome which requires a multidisciplinary approach to manage these patients. A report of two cases of (one was 22 years and another was 37 years) old male patients with EEC was presented with photophobia, microdontia, anodontia, hypernasality in speech, dry and scale-like skin, sparse and almost blonde scalp and body hair, Ectrodactyly, Syndactyly and conductive hearing loss.

Conclusions: Early diagnosis allows parents to avail counseling and in particular obtain reassurance regarding the low risk of mental handicap.

Keywords: *Ectrodactyly; Ectodermal Dysplasia; Clefting*

Introduction

EEC (Ectrodactyly, Ectodermal Dysplasia, Clefting) Syndrome is a multiple congenital anomaly syndrome presented with ectrodactyly, ectodermal dysplasia, distal limb anomaly, cleft lip, palate and ocular anomalies. EEC is also known as "Split hand-split foot-ectodermal dysplasia-cleft syndrome" [1]. The syndrome results from a developmental abnormality that simultaneously affects the ectodermal and mesodermal tissue. Ectodermal dysplasia results from the abnormal morphogenesis of cutaneous or oral embryonal ectoderm (hair, nails, teeth, eccrine glands) [2,3]. In some forms, mesodermal abnormalities are also present. The frequency of the different ectodermal dysplasias is highly variable. The prevalence of hypohidrotic ectodermal dysplasia, the most common variant, is 1 case per 100,000 births in United States of America [4].

EEC is characterized by the triad of ectrodactyly, ectodermal dysplasia and facial clefts. Ocular features may present with lacrimal drainage system anomalies (punctal or canalicularatresia/hypoplasia, nasolacrimal duct obstruction) and consequent constant epiphora, blepharitis, conjunctivitis, corneal opacities and photophobia [2,3,5].

We attempt to describethe two rare cases of EEC (Ectrodactyly, Ectodermal Dysplasia, Clefting) Syndrome. Every case need proper physical, clinical, laboratory and imaging (if needed) evaluation to confirm the diagnosis.

Methods

Two case reports were evaluated in National Institute of Ophthalmology, Dhaka and Bangladesh eye hospital, Dhaka, Bangladesh. Through clinical evaluation and radiological imaging techniques confirm the diagnosis of Ectrodactyly, Ectodermal Dysplasia, Clefting Syndrome.

Results

Case 1 was a 22 year old male patient and case 2 was a 37 years old male patient. Both patients presented with common physical features like as photophobia, microdontia, anodontia, hypernasality in speech, dry and scale-like skin, sparse and almost blonde scalp and body hair, Ectrodactyly, Syndactyly. Conductive hearing loss was present in case 1. Ocular features were different in two cases. There was no positive family history and consanguinity in both cases. However, they had limb abnormalities as well as features of ectodermal dysplasia. Both patients did not give history of taking topical medications for ocular problems. I.Q (105 and 106) which indicates patients intelligence was average.

A detailed examination of the patient was performed, and the findings are described below.

Skeletal Abnormalities: Ectrodactyly and syndactyly of both hands and feet. A typical claw-hand abnormality was noted.

Face and Oral cavity: Speech was characterized by hypernasality. Teeth abnormality like microdontia, partial anodontia and sensitivity of teeth due to enamel hypoplasia was evident. Moderate hearing loss was evident in only case 1.

Skin: Diffuse hypopigmentation of skin and hair was present. Skin had a dry, scaly appearance. Scalp hair was dry, sparse and hypopigmented. Eyebrows were sparse laterally. Body hair ranged from sparse in some regions, to absent in others. The nails showed marked abnormality (small, brittle). Xerostomia was present.

Ophthalmological Findings

Case 1: The patient had best corrected visual acuity 6/12 in right eye and 1/60 in left eye. All eyelids were oedematous and erythematous. Blepharitis was present bilaterally. Subtarsal scarring was seen in both eyes. There was trichiasis of both upper and lower lids of both eyes. Conjunctival congestion with active discharge was present in both eyes. Punctate epithelial erosions, superficial punctate keratitis, corneal epithelial defect and corneal oedemawas present in both eyes, but exhibited greater severity in left eye. Corneal vascularization, approximately 180 degrees in right eye and 270 degrees in left eye was present in association with bilateral corneal scarring.

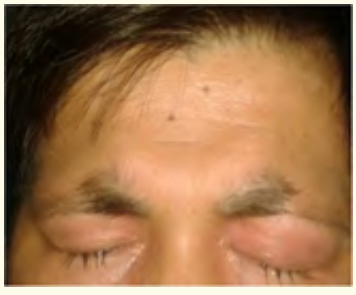

Figure 1: *Constant tearing with matted eyelashes.*

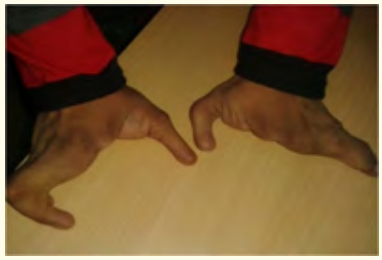

Figure 2: *Ectrodactyly of Right and left Hands.*

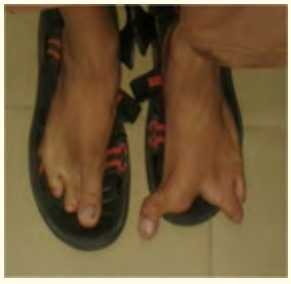

Figure 3: *Syndactyly of right foot and Ectrodactyly of left foot.*

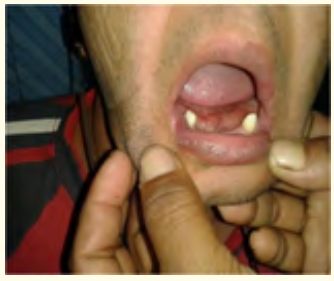

Figure 4: *Hypodontia and microdontia.*

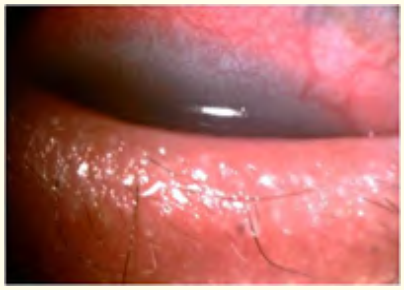

Figure 5: *Vascularization with scaring of the cornea.*

Case 2: Best corrected visual acuity 6/6 in right eye and 5/60 in left eye. Eyelids were normal. Moderate corneal opacity (macula) was present in his left eye. Watering and discharge was present in left eye. Left Naso-lacrimal duct was blocked on sac patency test.

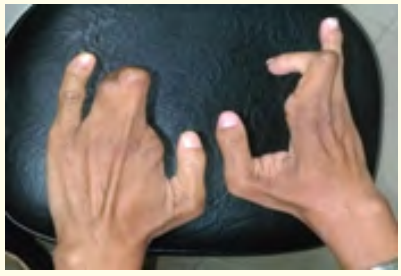

Figure 6: *Ectrodactyly of Right and left Hands.*

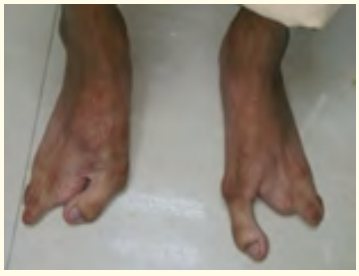

Figure 7: *Syndactyly and Ectrodactyly of both feet.*

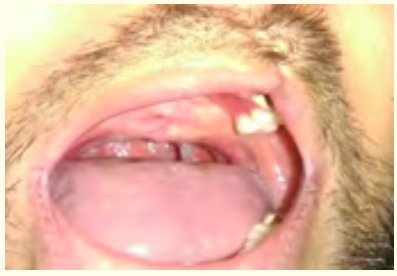

Figure 8: *Cleft Lip and Plate.*

Laboratory Investigations: CBC, urine analysis, blood sugar and blood urea were unremarkable. Serum IgG, IgM and IgA were within normal limits. Ultra sonogram of whole abdomen revealed normal study. Chest X-ray was normal.

Clinical examination, X-ray hands and foot confirmed the diagnosis EEC syndrome with rare ophthalmic manifestations.

Management: Management of EEC syndrome requires a multidisciplinary approach. The multidisciplinary management team should include the plastic surgeon, dental surgeon, ophthalmologist, dermatologist, nephrologist and speech therapist. Early audiological assessment is necessary especially in children.

Managing these cases was difficult from both practical and psychological standpoint. However, we could only manage the ocular problems of our patient. Blepharitis was managed with conservative treatment. Patient had bilateral entropion which caused trichiasis and we had to manage that problem by doing bilateral entropion surgery. Corneal manifestations like superficial punctate keratitis, corneal vascularization, corneal epithelial defect, corneal oedemain both eyes was managed conservatively by taking valuable opinion from cornea specialist. Case 2 had under gone Dacryocystorhinostomy (DCR) surgery in his left eye and waiting for keratoplasty surgery in his left eye.

Discussion

The ectodermal component of this syndrome involves the hair (hypotrichosis and hypopigmentation with sparse, fair and dry scalp hair, eyebrows and eyelashes); teeth (hypodontia, microdontia and enamel dysplasia) and nails (dystrophic in most cases). Hypohidrosis is variable. Other manifestations include urogenital anomalies (kidney and ureter malformations, urinary reflux, hypospadias and cryptorchidism), mental retardation, choanal atresia, a reduced number of meibomian orifices, conductive hearing loss, variable ear anomalies, breathy voice, hypopituitarism and isolated growth hormone deficiency [4-6].

The inheritance of this condition is AD with incomplete penetrance and variable expressivity. Genetic locusis TP63 (3q27). Sporadic and AR cases may also occur [7].

EEC Syndrome is a very rare condition, and the current cases represent very interesting presentations. Features of EEC syndrome are ectrodactyly (missing central digital rays of hands and or feet give the characteristic appearance of split hand or split foot. Other limb anomalies include thumb hypoplasia/aplasia, syndactyly and brachydactyly. Ectodermal dysplasia (Dry skin with decreased sweating. Sparse, brittle and hypopigmented hair. Sparse eyebrows and eye lashes. Dystrophic nails. Small/peg shaped/missing teeth.) Clefting (Cleft lip and/or cleft palate), obstruction of lacrimal drainage system and reduced tears leading to infections and corneal scarring and other features include renal anomalies, conductive hearing loss and choanal atresia [7-10].

Management of EEC syndrome is very difficult needs multidiscipline approach. Early surgical repair of cleft lip or palate may lessen facial deformities and improve speech. Other mid facial defects or hand/foot deformities may be surgically corrected in order to improve function and reduce physical disfigurement. Medical Care of affected patients depends on which ectodermal structures are involved.

The following measures should be taken:

- Air conditioning for home, school, and work for patients with anhidrosis/hypohidrosis.Maintaining adequate hydration and thermoregulation need frequent consumtion of cold liquids and better to wear cool clothing.
- Advise early dental evaluation and intervention and encourage routine dental hygiene for for patients with dental defects.
- Topical emollients may benefit for the patients with xerosis or eczematous dermatitis
- Wigs can help to improve appearance and use of topical minoxidil to enhance hair growth in case of severe alopecia.
- Scalp erosions should be treated with topical and systemic antibiotics as needed. General scalp care may involve the use of weekly dilute bleach baths or acetic acid soaks to minimize bacterial colonization of the scalp.
- Artificial tear is helping to prevent damage to the cornea in patients with dry eye.
- Protect nasal mucosa with saline sprays followed by the application of petrolatum.
- Immunodeficiency should be monitored for infection and treated with therapeutic and/or prophylactic antibiotics when appropriate. Allogeneic stem cell transplantation has been performed in a small number of patients [6,11-12].

Both patients had ectrodactyly, syndactyly and features of ectodermal dysplasia. Clefting of the lip, palate and tear duct abnormalities were found in case 2. But we think the difference of these features does not compromise the diagnosis of EEC Syndrome which had variable expression.

The presence of perioral papillomatosis, classically seen in Goltz Syndrome, has been reported only once before in EEC Syndrome and not found in our patients.

Hearing loss which is not so frequently found in patients with EEC Syndrome was present in our patient case1. It was severe conductive type of hearing loss for which the patient uses hearing aid.

Some unusual and rare ophthalmological findings like periorbitaloedema, erythema and subtarsal scarring was found in this patient. These findings have so far not been reported in any case of EEC Syndrome.

Conclusion

This syndrome requires a multidisciplinary approach for management. Early diagnosis allows parents to avail counseling and in particular obtain reassurance regarding the low risk of mental handicap.

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