CASE REPORT

Ectrodactary, Ectodermal Dysplasia, and Cleft Lip-Palate Syndrome

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Summary

The EEC syndrome (ectrodactyly, ectodermal dysplasia and cleft lip-palate) is a rare disorder inherited as an autosomal dominant trait or can occur sporadically. We describe a case of this syndrome with chronic ocular surface disorder secondary to abnormalities of ocular adnexia.

Key Words: EEC syndrome, Ectrodactyly, Ectodermal dysplasia

Introduction

The EEC syndrome, was coined by Rudiger et al in 1970. Early diagnosis allows parents to get accurate counseling and in particular obtain reassuring information regarding the low risk of mental handicap. Visual difficulties are potentially the most serious clinical feature of this syndrome. Early recognition of the ocular problems can prevent ocular complications that require surgical intervention.

Case Report

A 36-year-old Chinese man presented to the Eye Clinic, Hospital Kuala Lumpur complaining of foreign body sensation with frequent irritation and repeated episodes of redness in both the eyes. Vision in both eyes was decreasing gradually as well.

He had undergone re-constructive surgery for a cleft lip and palate. He had very slow growing hair which was sparse and dry. His skin was scaly and dry, with brittle and deformed nails. Almost all his teeth were replaced with dentures. He appeared to be bashful and spoke with a nasal voice. The fingers were deformed in a claw-like manner (Figure 1a). There was a central cleft on both feet, kept well hidden by high socks and shoes (Figure 1b). His developmental milestones had been normal. He had four other normal siblings. There was no history of similar problem in any of his paternal and maternal ancestors. He had no known medical ailments.

Ocular examination revealed a visual acuity of 6/24 in the right eye and 6/9 in the left eye. Both ocular surfaces were moderately inflammed. Meibomian gland orifices were absent in both the upper and lower eyelids. (Figure 2a) The left lower lid punctum was absent. In the right eye, there was pseudopterygium, symblepharon formation and superior palpebral conjunctival fibrosis, (Figure 2b) with a central corneal opacity present, involving the visual axis. The left eye was affected to a lesser degree. There was minimal corneal opacity just above the visual axis. Peripheral corneal vascularization was present superiorly in both eyes. There was also superficial punctate keratopathy in both eyes. Intraocular pressure was 14 mmHg in both eyes and fundus examination did not show any abnormalities.

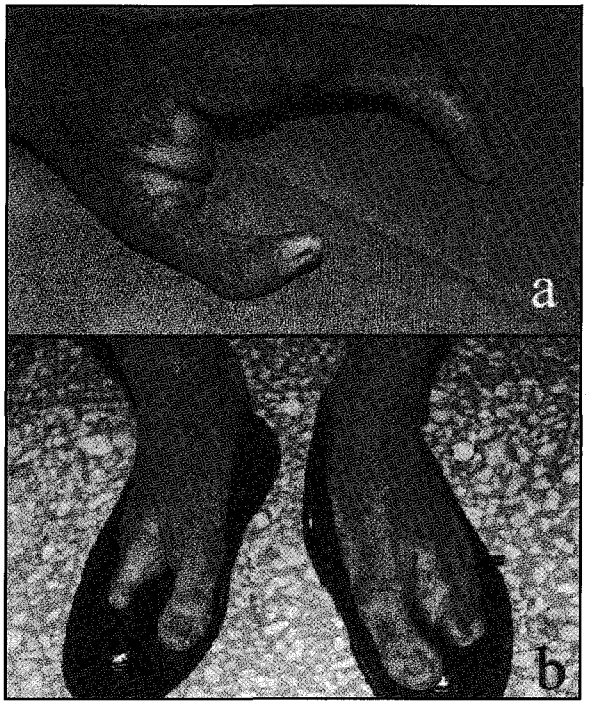

Fig. 1a : Showed the claw-like hands.
Fig. 1b : Showed the central cleft in both feet.

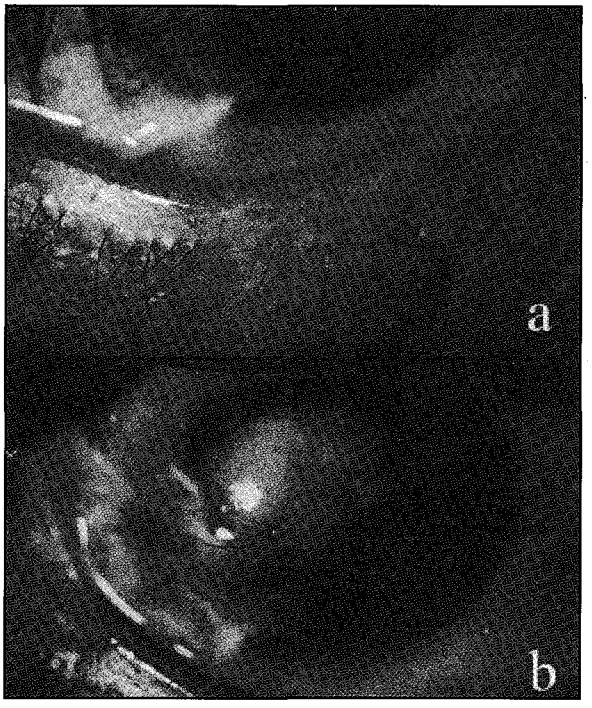

Fig. 2a : Showed absent of Meibomian gland orifices.
Fig. 2b : Showed pseudoptergium, syblepharon symblepharon formation and central corneal.

Based on the above clinical findings, a diagnosis of ectrodactyly, ectodermal dysplasia and cleft lip-palate syndrome was made. The patient was maintained on low dose steroid drops to both eyes and lubricants in the form of preservative free artificial tears drops. The ocular surface inflammation and superficial punctate keratopathy gradually resolved over a period of one month. He is currently under follow-up at the Eye Clinic Hospital Kuala Lumpur.

Discussion

EEC syndrome is a multiple congenital anomaly syndrome characterized by ectodermal dysplasia, distal limb anomaly, cleft lip and palate and ocular adnexia anomalies. In this patient, his ocular abnormalities include absence of meibomian glands orifices, lower lid punctum and lower lid tarsal plates, blepharitis, chronic ocular surface inflammation, abnormal tears film and stem cells deficiency.

EEC syndrome has been reported as early as 1970 by Rudiger et al, where they described a girl with trimelic ectrodactyly, ectodermal features involving hair, teeth, nails and bilateral cleft lip and palate[1]. The inheritance of this condition is autosomal dominant, although sporadic and autosomal recessive traits have been reported[2].

There exists numerous case reports of EEC and phenotypically related syndromes[2,3]. In a study in the United Kingdom by Buss et al, they reported 24 cases (16 males and 8 females) with EEC syndrome[2]. All cases showed evidence of ectodermal involvement. Distal limb deficits were present in 87.5%, facial clefting in 58% and lacrimal duct anomaly in 87.5%. The teeth were universally affected, although the primary dentition was usually complete but not always morphologically normal. Hair was affected in all cases. Generally they are light in colour (as were their eyelashes) and

coarse and dry in 78%. Seventy-nine percent had nail dystrophy. The nails were slow growing, transversely ridged and pitted, showing varying degrees of concavity. The skin was involved in 87% of cases. The most common manifestation was dry, scaly skin on the extremities and occasionally around the neck.

87% of these patients had limb anomaly. Majority had tetramelic involvement, 6 cases (25%) had central ray deficit in the upper limbs combined with syndactyly of the toes, asymmetry limb defects, pre-axial anomalies, unilateral absent thumb and small proximally placed thumb. The digits most commonly involved in syndactyly were digits 3 and 4 in both upper and lower limbs. Functional impairment was usually not a significant problem provided opposition could be attained. Surgery is usually required to close wide cleft hands or to separate a syndactyly.

Orofacial clefting is present in more than 50% of cases. These include bilateral or unilateral cleft lip and palate, or central cleft palate. All patients had nasal quality of speech, indicating velopharyngeal incompetence.

In 87% of cases, lacrimal duct problems will be encountered and are usually bilateral. The most common problems are epiphora and recurrent infections reflecting varying degrees of nasolacrimal duct obstruction, meibomian gland dysfunction and reduced tear production.

Other problems encountered by patients suffering from this rare syndrome include hearing loss, which is often a conductive deficit rather than sensorineural in origin. Some of these patients also suffer from genitourinary anomalies such as glandular hypospadias, ureteric reflux, hydronephrosis, recurrent urinary tract infections, dysuria, frequency and presence of thick walled, very small volume bladder. There is usually no history of mental retardation or developmental delay.

The management of these cases requires a multi-disciplinary approach. Early diagnosis will allow parents to get accurate counseling and in particular obtain reassurance regarding the low risk of mental handicap. The multidisciplinary management team should include the plastic surgeon, ophthalmologist and nephrologist. Early audiological assessment is necessary especially in children.

Managing these cases is difficult from both practical and psychological viewpoints. Severe ectodermal manifestations can be helped by the use of wigs and cosmetics. Expert dental advice may preserve primary teeth. Simple emollients may be satisfactory for dry skin.

Visual complications are particularly difficult to manage and visual difficulties are potentially the more serious clinical feature of the syndrome. Anticipation of recurrent infections in the early years is necessary, and artificial tears may also be protective if reduced lacrimal secretions or poor tears film quality is found. Absence of Meibomian secretions leads to lipid deficiency and thus tear film instability. Meibomian gland dysfunction increases tear film evaporation and increases tear film osmolarity, leading to the development of the ocular surface disease elements of keratoconjunctivitis sicca. These include subconjunctival fibrosis, decreased tear secretion by damaging accessory lacrimal gland tissue in the conjunctiva, limbal stem cell deficiency, conjunctivalization of the corneal surface, recurrent corneal erosion, persistent corneal epithelial defect, recurrent infective keratitis and corneal perforation.

Treatment of this case is aimed at moistening the eye with preservative free lubricating drops and to reduce the ocular surface inflammation with topical steroids. These measures will help reduce the ocular surface inflammation and thus minimize the ongoing destruction of limbal stem cells. However, if the corneal neovascularization progresses and impairs vision, then stem cells transplantation with or without amniotic membrane graft is an option to reconstruct the ocular surface.

References

1. Roswitha A. Rudiger, Wolfgang Haase, and Eberhard Passarge. Association of Ectrodactyly, Ectodermal Dysplasia, and Cleft Lip-Palate. The EEC Syndrome. Amer J Dis Child, 1970; 120: 160-63.
2. Buss, P.W.; Hughes, H.E.; Clarke, A. Twenty-four cases of the EEC syndrome: clinical presentation and management. Journal of Medical Genetics, 1995; 32(9): 716-23.
3. Moshegov, Con N., Daya, Sheraz M. Ectrodactyly-Ectodermal Dysplasia-Clefting (EEC) Syndrome. Arch Ophthalmol. 1996; 114: 1290-291.