

Lobster-claw hand: A manifestation of EEC syndrome

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ABSTRACT

The combination of ectrodactyly, ectodermal dysplasia, and cleft lip with or without cleft palate (EEC syndrome) is a rare type of congenital anomaly. It usually occurs as an autosomal-dominant trait or less commonly in a sporadic form. In this complex, multiple congenital anomaly syndrome, any of the three cardinal manifestations may present with variable expressions.

Key Words: Ectodermal dysplasia, EEC syndrome, Ectrodactyly, Lobster claw

INTRODUCTION

The rare combination of ectrodactyly (lobster-claw deformity), ectodermal dysplasia and cleft lip with or without cleft palate has been described as EEC syndrome.^[1] It was first described by Cockayne in 1936,^[2] but the acronym was coined by Rudiger *et al.*^[1] The syndrome is inherited as an autosomal dominant trait, although it may occur sporadically.^[3] It is a complex, pleiotropic, multiple congenital anomaly or dysplasia syndrome in which any of the three cardinal signs can present with variable expression; it may also be associated with many defects not necessarily of ectodermal origin.^[4] We report on a case of sporadic EEC syndrome.

CASE REPORT

A 26-year-old male presented to our outpatient department with a history of recurrent episodes of maceration and infection of the first interdigital space of the right foot. The right foot showed an unusually

deep cleft at the first interdigital space. A linear scar caused by surgical repair was evident, extending up to the distal one-third of the foot. The second and third toes were absent on the right foot. The left foot was normal, except for a short second toe. Both palms showed a classic lobster claw deformity: the middle digit was absent and the remaining four fingers were parted, two on either side, the cleft almost dividing the palm into two halves (Figure 1). The right hand also showed syndactyly of the first and second digits.

There was no visible cleft lip. Submucous clefting of the palate was palpable as a bony defect in the central midline. Maxillary hypoplasia was clinically apparent. There were no other bony abnormalities. Both pinnae were low set. The entire skin showed moderate dryness. The hair and nails were normal. Sweating was normal. The lacrimal ducts were intact and there were no auditory and neurological defects. No other systems were clinically involved.

Routine investigations were within normal limits.

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X-rays of the hands and feet showed radiological findings corresponding to the clinical features. An X-ray of the skull (submentovertical view) confirmed the submucous cleft palate (Figure 2). Skin biopsy taken from the flexor aspect of the forearm showed normal appendages.

DISCUSSION

The term ectodermal dysplasia includes many disorders characterized by the abnormal development of embryonic ectodermal tissue, which leads to anomalies of the hair, teeth, sweat glands, and nails.^[5] EEC syndrome (OMIM No. 129900) is rare and is usually



Figure 1: Lobster claw deformity of left hand

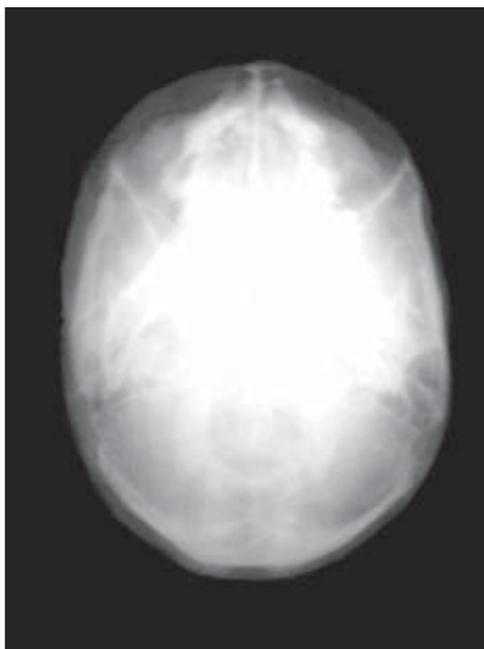


Figure 2: X ray skull showing submucous cleft palate

inherited in an autosomal-dominant manner. The trait is variable in its penetrance and clinical expressivity.^[6] In addition to ectrodactyly and cleft palate, this patient had maxillary hypoplasia and low-set ears. He did not have ectodermal dysplasia.

The term ectrodactyly denotes congenital absence of all or part of one or more fingers or toes. It is synonymous with split hand or foot deformity or lobster claw. Syndactyly indicates fused or webbed fingers or toes. A permanent deflection of one or more fingers is referred to as clinodactyly.

The features of EEC syndrome include the following:^[7]

- Ectodermal dysplasia, with varying degrees of involvement of the sweat glands, hair, nails, and teeth
- Cleft lip or palate
- Eye and lacrimal duct anomalies
- Midfacial hypoplasia
- Limb anomalies, such as ectrodactyly, syndactyly, and clinodactyly
- Auricular anomalies
- Short stature
- Genitourinary anomalies
- Central nervous system anomalies, such as hearing loss and mental retardation
- Multiple nevocellular nevi and hypopigmentation

The manifestations in our patient were consistent with an incomplete expression of EEC syndrome.

Two other conditions that resemble this condition are Rapp-Hodgkin syndrome and Hay-Wells (AEC) syndrome.^[7] The Rapp-Hodgkin syndrome manifests with ectodermal dysplasia, cleft lip or palate, a distinct facies with midfacial hypoplasia, mild frontal prominence, small mouth, and and dermatitis of the scalp. The inheritance is autosomal-dominant.^[8] The AEC syndrome, also autosomal-dominant in inheritance, is characterized by ectodermal dysplasia, ankyloblepharon, and cleft lip or palate. Associated scalp dermatitis may occur in more than 50% of the patients.^[8]

Management of cases with EEC syndrome requires a multidisciplinary approach that includes a dermatologist, plastic surgeon, ophthalmologist, and,

if needed, a speech therapist.^[9] Most patients need no surgical intervention as there is no functional problem except for the cosmetic disturbance.^[10]

REFERENCES

1. Rudiger RA, Haase W, Passarge E. Association of ectrodactyly, ectodermal dysplasia and cleft lip-palate-the EEC syndrome. *Am J Dis Child* 1970;120:160-3.
2. Cockayne EA. Cleft palate-lip, hare lip, dacryocystitis, and cleft hand and foot. *Biometrika* 1936;28:60-3.
3. Zonana J. Ectodermal dysplasias. *In: Rimoin DL, Connor JM, Pyeritz RE, editors. Principles and Practice of Medical Genetics.* New York: Churchill Livingstone; 1996. p. 1279-91.
4. Arbesfeld D, Thomas I, Janniger CK, Desposito F, Lambert WC, Schwartz RA. Ectrodactyly, ectodermal dysplasia, cleft palate syndrome: Report of a case with generalized telangiectasias. *J Am Acad Dermatol* 1993;29:347-50.
5. Sybert VP. Ectodermal dysplasias. *In: Freedberg IM, Eisen AZ, Wolff K, Austen KF, Goldsmith LA, Katz SI, editors. Fitzpatrick's Dermatology in General Medicine.* 6th ed. New York: McGraw-Hill; 2003. p. 515-22.
6. Kalla G, Anush G. Ectrodactyly. *Indian J Dermatol Venereol Leprol* 2002;68:152-3.
7. Fosko SW, Stenn KS, Bologna JL. Ectodermal dysplasias associated with clefting: significance of scalp dermatitis. *J Am Acad Dermatol* 1992;27:249-56.
8. Spiegel J, Cotton A. AEC syndrome: ankyloblepharon, ectodermal defects, and cleft lip and palate. *J Am Acad Dermatol* 1985;12:810-5.
9. Batra P, Duggal R, Parkash H. EEC Syndrome: a case report. *J Indian Pedo Prev Dent* 2003;21:75-8.
10. Thami GP, Kaur S. Split hand-foot malformation: a congenital central limb ray deficiency. *Indian J Pediatr* 1983;50:337-40.