

EEC Syndrome with Urogenital Anomalies and Compound Naevus

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Summary

A case of ectrodactyly ectodermal dysplasia clefting (EEC) syndrome is presented; the congenital anomalies included ectrodactyly (cleft of the hands and feet), ectodermal dysplasia, and cleft lip and palate and also compound pigmented naevus and anomalies of the genito-urinary tract.

Key words: Ectrodactyly ectodermal dysplasia.

Introduction

The association of ectrodactyly (lobster claw deformity or cleft of the hands and feet) and dacryocystitis was first reported by Cockayne in 1936¹. It was, however, Rudiger and others² who stressed the association of atypical ectodermal dysplasia with ectrodactyly and cleft lip and palate and suggested the name EEC syndrome. This is a rare congenital syndrome which shows both a familial autosomal dominant inheritance as well as sporadic occurrence³.

We present a patient with typical features of EEC syndrome.

Patient Report

A female infant was born on 20th December 1983, as a second child to a Malay couple who were second cousins. She was referred to us by a paediatrician for cleft management. The mother denied any similar defect in other members of the family. She also denied any exposure to drugs or irradiation during the pregnancy. The child was delivered normally without any complication. Her birth weight was 2.2 kg. At 3 months, when we first saw her, she weighed 3.2 kg and measured 53 cm in length (both below 30th percentile) and the head circumference was 36.5 cm (below the 10th percentile).

We found multiple congenital abnormalities in this child. A complete bilateral cleft lip and palate was present (Fig 1a). There were also clefts of both the hands and feet (Figs 2a and 2b). There was sparse hair growth on the scalp which was silky, short and hypopigmented (Fig 1c) and the eye-brows and eye-lashes were also scanty. On the forehead there was a pigmented naevus (Fig 1b). The skin was thin, dry and light-coloured and there was hypoplasia of the nails. The primary dentition was delayed and the teeth were hypoplastic.

She complained of photophobia and was referred to the ophthalmologist, who found atresia of the lower punctum with blepharitis and dacryocystitis, but there was no corneal ulceration or scarring seen.



Fig 1a: Patient at 3 months of age showing the bilateral cleft lip and palate.



Fig 1b: Front view of patient at the age of 5 years showing result of lip repair; and the pigmented naevus on the forehead.

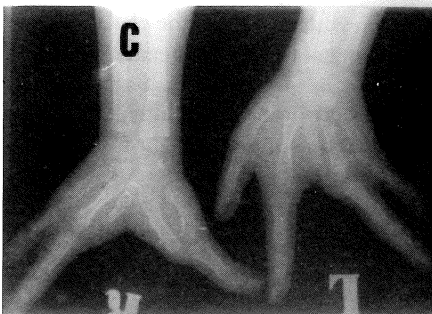
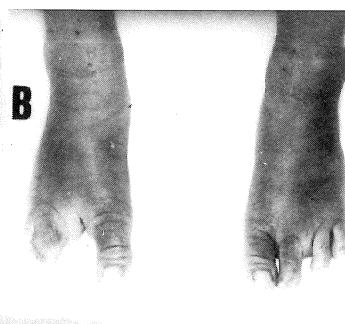
Fig 1c: Lateral view of patient also at 5 years of age showing result of nose repair; and showing scanty scalp hair-growth.

Fig 2a: Cleft of the hands with absent index and middle fingers of the right hand and absent middle finger on the left.

Fig 2b: Cleft of the feet with absent second and third toes and syndactyly of the fourth and fifth toes on the right; and absent third toe on the left.

Fig 2c: Radiological appearance of hands.

Fig 2d: Radiological appearance of feet.



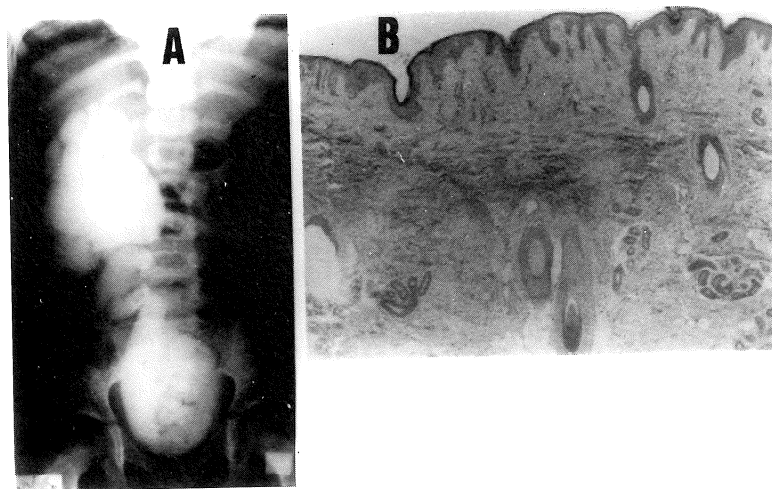


Fig 3a: Intravenous urography showing right hydronephrosis and hydroureter and non-visualisation of the left kidney.

Fig 3b: Histological section of the skin from edge of the naevus, showing presence of junctional and dermal naevus cells, scanty hair follicles of normal maturation, severe hypoplasia of the sebaceous and apocrine glands and normal eccrine glands; these features are consistent with ectodermal dysplasia and compound pigmented naevus.

She was assessed by the ear nose and throat surgeon as part of the cleft palate management, and was found to have serous otitis media on the right side and perforated ear drum on the left, with bilateral conductive deafness.

Gynaecological examination disclosed the presence of imperforate hymen.

At subsequent follow-up examinations, she was found to have attained relatively normal physical and mental development; walking was at 1 year and speech was at one and a half years. She was able to attend regular school.

Investigation

Routine haematological and biochemical tests including renal profile were normal. Radiological examination of the upper and lower limbs showed that the long bones were normal. There were bilateral abnormalities of the small bones of the hands and feet, with absence of the phalanges of some of the digits of the hands and feet and hypoplasia of some of the metacarpals and metatarsals as in Figs 2c and d.

Intravenous urography (Fig 3a) revealed a grossly hydronephrotic right kidney and right hydroureter. The left kidney was not visualised. An ultrasound examination confirmed right hydronephrosis and absence of the left kidney.

A biopsy of the edge of the pigmented naevus showed several foci of naevus cells in the junctional area as well as in the dermis, typical of a compound naevus. Hair follicles were present with normal maturation and histology but were slightly fewer in number. There was definite severe hypoplasia of the sebaceous

glands and apocrine glands. The eccrine sweat glands were normal (Fig 3b). Chromosome analysis showed normal 46xx karyotype.

Treatment and follow-up

We performed lip adhesion on this child at 4 months of age and definitive bilateral lip repair at 7 months. Palatoplasty was done at 1 year and 3 months. Columella lengthening was done at 4 years of age. She was treated with antibiotic eye drops for the blepharitis.

Gromet tube insertion for the right ear was done at one and a half years of age for the serous otitis media.

She was referred to the urologist for the hydronephrosis and hydroureter and successfully underwent right ureteric reimplantation at 6 years of age. Her cleft hands and feet were fully functional and surgical correction was considered unnecessary. The compound naevus will be excised before puberty to avoid the possibility of malignant change. She was fitted with hearing aids for the bilateral conductive deafness. She required frequent check-ups for her condition. She will require a wig to camouflage the scanty hair when she is older.

Discussion

EEC is a very rare syndrome. Rudiger and associates² estimated that the incidence could be as low as 1.5 per 100 million. About 20 or so cases have been reported in the literature⁵. The aetiology remained obscure. Many cases showed familial inheritance, but some were sporadic. In the familial cases, dominant as well as recessive transmission has been postulated⁷. The patient described in this report showed all the characteristic features of EEC as described by Rudiger. In addition, she also demonstrated abnormalities of the genito-urinary tract (including imperforate hymen), otitis media and compound pigmented naevus. These abnormalities were not present in the case described by Rudiger. However, Maisels⁴ described a patient with EEC syndrome who had congenital absence of the right kidney with hydronephrosis of the left kidney. In fact, urinary tract anomalies, including cryptorchism in the male, are possible supplementary features of EEC syndrome⁷. As such, it is important that every patient with EEC syndrome should have a complete examination and investigation of the genito-urinary tract, including an intravenous urography⁷. Pigmented naevi have also been reported in association with EEC syndrome⁶.

The otitis media and conductive deafness found in this patient were features of the cleft palate abnormality.

One might assume that the cleft of the lip and palate and of the hands and feet would be the most disabling features of the syndrome⁵. However, the cleft lip and palate can be successfully corrected surgically, as in this patient. The cleft hands and feet did not appear to have interfered with normal function and were left uncorrected. The most serious problem in this patient was the urinary tract anomaly. Correction of the vesico-ureteric obstruction was necessary and had to be done as soon as possible to prevent further renal damage leading to renal failure, especially in a patient with only one functioning kidney. The other abnormality in this patient which could have potentially given rise to serious disability was atresia of the lacrimal ducts, which predisposes to recurrent infection, corneal ulceration and scarring and reduced visual acuity⁵. Patients with EEC therefore should be seen and treated by an ophthalmologist early in the treatment plan.

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