A NEONATE WITH ECTODERMAL DYSPLASIA ECTRODACTYLY CLEFTING SYNDROME AND VENTRICULAR SEPTAL DEFECT

S P Ram, A R Noor, N A Ariffin

ABSTRACT

A 37-week gestation male boy was born to a gravida seven para six mother by spontaneous vertex delivery at home. The baby cried at birth. On day 3 of life, he was admitted for respiratory distress. Physical examination revealed ectrodactyly, thin dry skin, anomalous tear duct with cardiomegaly. X-ray revealed absent radii, cardiomegaly and hemivertebra at L1. Echocardiogram revealed perimembranous type of ventricular septal defect. A diagnosis of Ectodermal Dysplasia Ectrodactyly Clefting Syndrome with ventricular septal defect was made. He was managed conservatively in the nursery. However, he expired on day 27 of life following short spell of fever and apnoeic episode due to neonatal sepsis.

Keywords: *ectrodactyly, ectrodermal dysplasia, cleft palate, ventricular septal defect, absent radii, neonate.*

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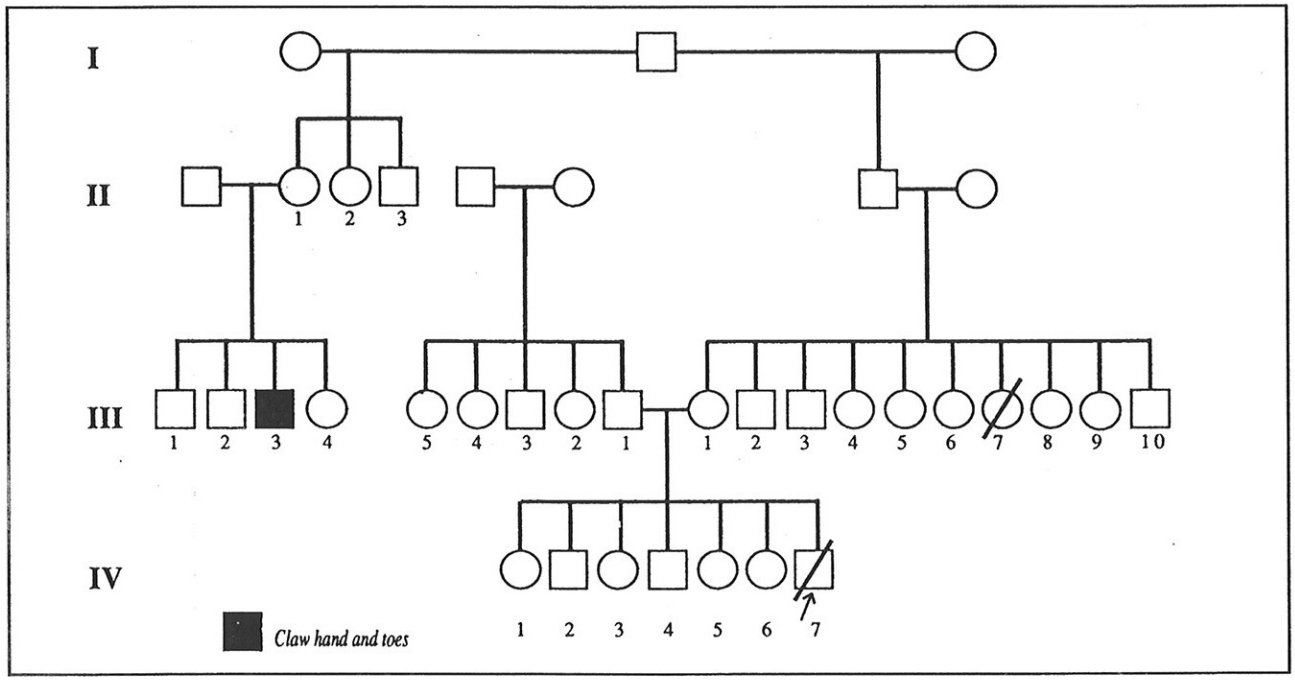
Fig 1 - Pedigree chart showing autosomal dominant with variable penetrance



INTRODUCTION

Ectodermal dysplasia, ectrodactyly and cleft lip/palate (EEC), occurring either individually or as a syndrome are phenotypically variable and genotypically a heterogeneous group of conditions (Fraser 1970)[1,2]. EEC syndrome is usually inherited as an autosomal dominant disorder. Less common features are oro-facial, skeletal and tear duct anomalies.

A neonate having an unusual association of hemivertebra, absent radii with ventricular septal defect in EEC syndrome is reported.

Department of Paediatrics
Hospital Universiti Sains Malaysia
16150 Kubang Kerian
Kelantan
Malaysia

S P Ram, MD (Paed)
Lecturer

A R Noor, M Med Sci (UK)
Lecturer

W A Ariffin, MRCP (UK)
Lecturer

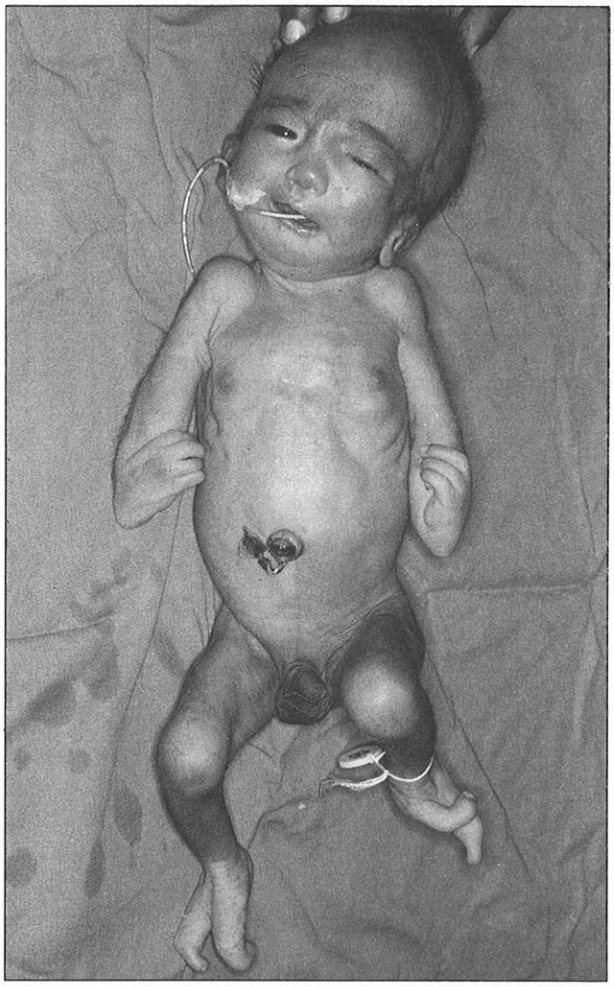
Correspondence to: Dr S P Ram

CASE REPORT

A, 37-week gestation, 2100 gm male baby was born to a gravida seven para six mother by spontaneous vertex delivery at home. The baby cried immediately at birth. The mother's antenatal history was normal and the other offsprings were normal. The pedigree chart showed one male member affected with bilateral limb anomalies (Fig 1, III-3.). The baby was admitted on day 3 of life for respiratory distress. Physical examination revealed the following: head circumference 33 cm (10 to 50th centile), chest circumference 32 cm, length 46 cm (10-50th centile), the ratio of upper segment and lower segment was 1.5:1. He was lethargic, dyspnoeic and mildly cyanosed. His skin was dry, thin and he had sparse eyebrows. Intermittent watery discharge was seen from both the eyes, with redness of the left eye. He also had low set

ears, posterior midline cleft palate with incomplete cleft lip on the right side, hypertelorism, micrognathia, lobster claw deformity of the feet, hypoplasia of the forearms, absent right thumb, arthrogryposis of elbow and knee joints (Fig 2 and 3). The heart rate was 150/minute with normal first and second heart sounds. There was grade 3/6 pansystolic murmur over the left sternal edge. A firm liver was palpable 3 cm below the right subcostal margin. His respiratory rate was 60/minute with bilateral basal crepitations of the lungs suggestive of bronchopneumonia.

Fig 2 – The baby with ectrodactyly and low set ears.



Investigations on admission revealed the following: total leucocyte count 4.4 x 10^9/L, (differential count: polymorph 15%, lymphocyte 77%, monocyte 1%, eosinophils 7%), red blood cell count 4.08 x 10^{12}/L, Hb: 15.0 g/dl, PCV: 41.9%, platelet count 95 x 10^9/L (normal platelet count 84-478 x 10^9/L) and reticulocyte count 1.8%. The chest X-ray (Fig 4) showed cardiomegaly. Skeletal survey revealed hemivertebra at L1 and bilateral absence of radii. Two dimensional echocardiogram showed perimembranous type of ventricular septal defect with normal contractility of the heart. Eye swab was positive for *Klebsiella* and *Serratia,* both were sensitive to cefotaxime, gentamicin and amikacin. The arterial blood gas analysis and the blood glucose level were normal. The blood culture was negative.

On admission, he was managed conservatively with intravenous infusion of 10% dextrose 80 ml/kg on day 3 of life. He was treated with intravenous benzyl penicillin 100,000 units /kg and gentamicin 2.5 mgm/kg 12-hourly for the bronchopneumonia. On day 6 of life, he was noted to be pale, jaundiced and had fresh blood in the gastric aspirate. The prothrombin time 21.0 sec, INR (International Normalised Ratio)

Fig 3 – The baby with ectrodactyly of the lower limbs.

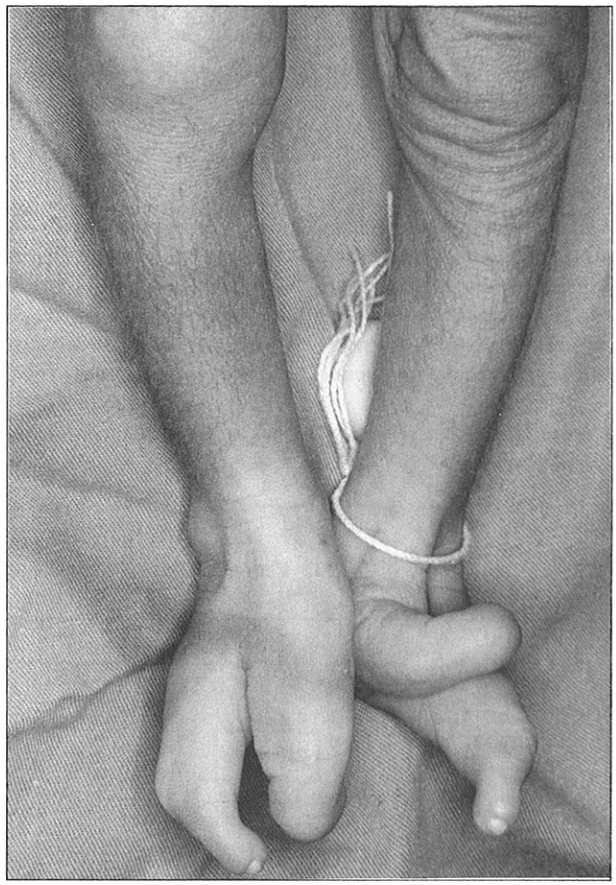
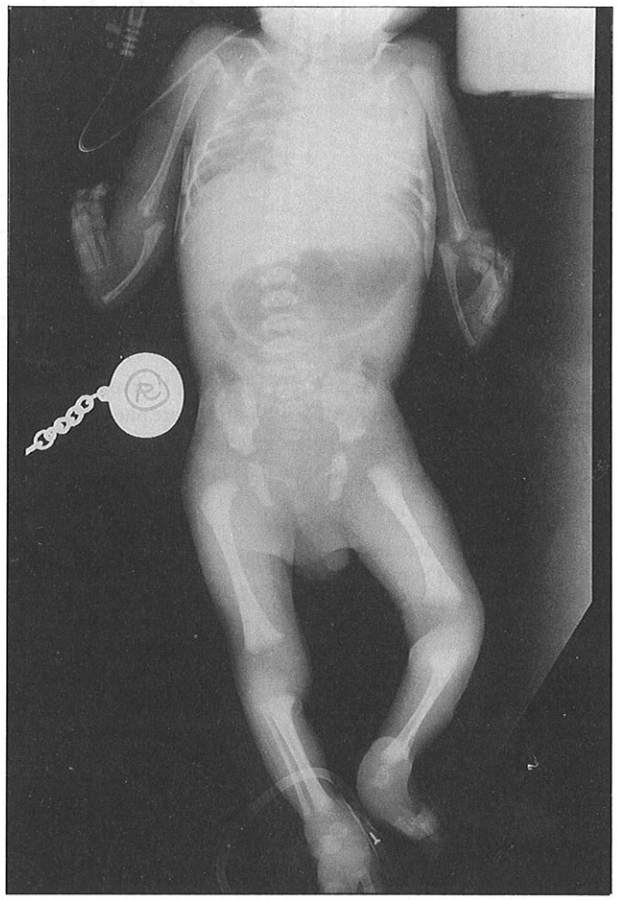


Fig 4 - X-ray of the baby showing cardiomegaly with absent radii.



1.6, partial thromboplastin time 55.0 sec and control 43.0 sec. He was given 10 ml/kg of fresh frozen plasma with intravenous vitamin K. He showed improvement within twenty-four hours. He was then given small quantities oral feeds via orogastric tube. However, on the 17th day of life, he looked dull, less active and pale. He was also dyspnoeic and was not tolerating oral feeds. His haemoglobin was 10.5 g/dl; total leucocyte count, 16 x 10^9/L and platelet count, 270 x 10^9/L. He was given packed cell transfusion 10 ml/kg to correct the falling haemoglobin. At this stage, injection cefotaxime, amikacin and furosemide in divided doses were added to the antibiotic regimen in view of the baby being unwell and the persistent lung signs. Forty-eight hours later, he appeared tachypnoeic but was able to tolerate orogastric feeds. Repeat haemoglobin was 12.5g/dl, platelet count 39.1 x 10^9/L. On day 26 of life at 6 pm, he developed high grade temperature (axillary temperature 40°C). He was treated with tepid sponging and syrup paracetamol. Six hours later, he had an episode of apnoea and became bradycardic, cyanosed and expired on day 27 of life despite cardio-respiratory resuscitation. His parents did not give consent to do postmortem examination.

DISCUSSION

The EEC syndrome is usually inherited as an autosomal dominant trait. The most common clinical manifestations of EEC syndrome are ectodermal dysplasia, ectrodactyly, cleft lip/palate and tear duct anomalities. The clinical manifestations of EEC syndrome are quite variable except for the ectodermal features[3]. The orofacial anomalies include lack of permanent incisors, adontia, enamel hypoplasia, deeply furrowed tongue, dryness of the mouth, blepharitis, dacryocystitis, keratoconjunctivitis and photophobia[4].

The diagnosis of this syndrome is essentially clinical. A skin biopsy may reveal absence of sebaceous glands and disorganised arrangement of sweat pores[4]. The dermatoglyphic studies may show dysplastic ridges with multiple irregular furrows. Dilated cardiomyopathy and cardiac arrhythmias are known to occur in ectodermal dysplasias[5]. Sudden death presumed to be due to congenital heart disease has been reported[6]. Richieri Costa et al[7] described association of ventricular septal defect with ectrodactyly, cleft lip and palate, micropenis and mental retardation. Bernstein et al have also described sudden infant death due to hypohidrotic ectrodermal dysplasia[8].

Skeletal defects in EEC syndrome are variable as are the skin, hair and teeth changes[9]. Skeletal abnormalities may include syndactyly, clinodactyly, ectrodactyly and nail dystrophy of hands and feet[4]. Such skeletal defects are considered to be dysplastic sequence rather than individual malformation[10]. Richieri Costa et al[11] described variable ulnar and radial defects due to autosomal dominant mode of inheritance. Richieri[12] also described tibial hemimelia with cleft lip and palate.

In this neonate, the primary features like ectrodactyly, ectodermal dysplasia, cleft lip and palate were present. He also had unusual association of absent radii, hemivertebra and ventricular septal defect. The features like an episode of fresh moderate gastric bleed on day 4 of life, prolonged prothrombin time and partial thromboplastin time, normal platelet count response to vitamin K and fresh frozen plasma within twenty-four hours were suggestive of haemorrhagic disease of newborn. The cause of anaemia on the 17th day of life was probably due to previous gastric bleed, gram negative sepsis and frequent blood sampling.

He developed high grade continuous fever on day 26 of life which failed to respond to conservative therapy. He had cardio-respiratory arrest following an episode of apnoea and subsequently expired. The cause of fever in this neonate was most probably due to deficient sweat glands and associated sepsis. The features suggestive of sepsis in this neonate were lethargy, bronchopneumonia, firm hepatomegaly, conjunctivitis, anaemia, moderate to severe neutropenia and moderate thrombocytopenia. Hence, the cause of death in this neonate was most probably due to neonatal sepsis associated with multiple congenital anomalies.

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