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CASE STUDY

ECTRODACTYLY-ECTODERMAL DYSPLASIA-CLEFTING SYNDROME PRESENTING WITH PDA

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ABSTRACT

Ectrodactyly-Ectodermal dysplasia-Clefting (EEC) Syndrome is inherited as autosomal dominant disorder of ectodermal and/or mesodermal interaction with highly variable expression and reduced penetrance. It is characterized by the triad of ectrodactyly of the hands and/or feet, ectodermal dysplasia of the skin, hair, nails and teeth, with or without clefting anomalies affecting the lips and/or palate (Roelfsema *et al*., 1996). In patients with the EEC syndrome without cleft lip or palate, a characteristic facial morphology characterized by maxillary hypoplasia, a short philtrum, and a broad nasal tip has been described (Pries *et al*., 1974). We reported a rare case of 53 year old male patient with EEC syndrome presenting with PDA and heart failure with typical characteristic features of ectrodactyly. He underwent surgical closure for his PDA.

Conclusion: The presence of ectrodactyly with other congenital abnormalities is reason to consider the EEC syndrome within the differential diagnosis. While rare in itself, the association of the EEC syndrome with a Patent Ductus Arteriosus is uncommon.

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INTRODUCTION

Ectrodactyly-Ectodermal dysplasia-Clefting (EEC) Syndrome is an autosomal dominant disorder of ectodermal and/or mesodermal interaction with highly variable expression and reduced penetrance. This syndrome was first described in 1804 by Eckhold and Martens (Obel *et al*., 1993). It is characterized by the triad of ectrodactyly of the hands and/or feet, ectodermal dysplasia of the skin, hair, nails and teeth, with or without clefting anomalies affecting the lips and/or palate (Roelfsema *et al*., 1996). In patients with the EEC syndrome without cleft lip or palate, a characteristic facial morphology characterized by maxillary hypoplasia, a short philtrum, and a broad nasal tip has been described (Pries *et al*., 1974). The presence of lacrimal tract abnormalities and urogenital abnormalities have also been associated with this syndrome. (Roelfsema *et al*., 1996; Giannotti *et al*., 1995). The present case involves an adult male with isolated EEC syndrome and a concurrent Patent Ductus Arteriosus, an uncommon association.

Case Report

A 53 year old male patient presented with the complaints of breathlessness since 3 months Grade 2, aggravated with exertion, and cough since 3 months which is productive and mucoid in nature. Past history and childhood period is nil significant. No substance abuse.

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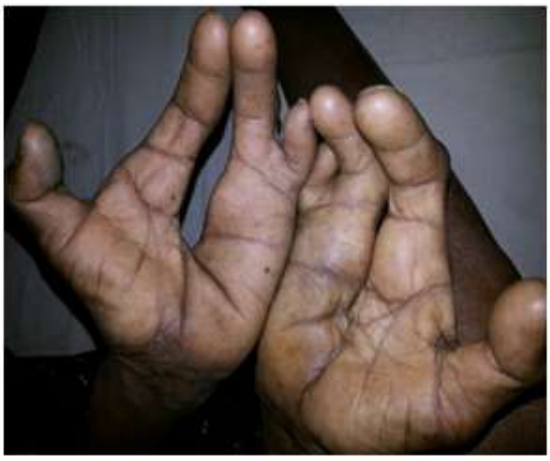
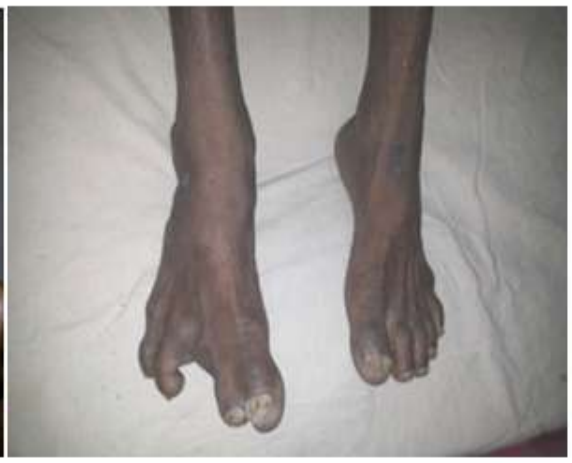
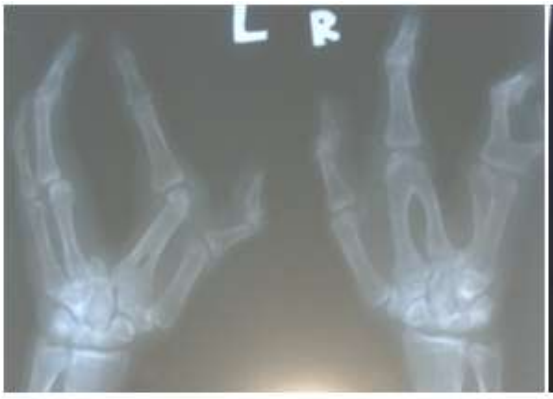
ON GENERAL EXAMINATION: There is Low set ears , sparsely populated eyebrows in lateral aspects, broad nose, short philtrum and ectrodactyly of both hands and right foot.JVP is raised.

On systemic examination, apical impulse is felt in left 5th intercostal space 2.5 cm lateral to mid-clavicular line and is diffuse with the presence of grade 3 parasternal heave. On auscultation, there is continuous murmur in left 1st intercostal space and bilateral basal crepitations are heard. Investigations showed normal blood sugars, hemogram, RFT and LFT. X rays of both hands and right foot showed ectrodactyly. Chest Xray showed prominent and enlarged right pulmonary artery and cardiomegaly.ECG showed right axis deviation with right ventricular hypertrophy. Echocardiogram showed PDA of 6mm size with moderate pulmonary artery hypertension.USG abdomen is normal.MRI brain is normal. Genetic testing not done in this patient.

DISCUSSION

The association of ectrodactyly, ectodermal dysplasia, and clefting with or without cleft palate is characterized as EEC syndrome (Obel *et al*., 1993). The presence of lacrimal tract abnormalities and urogenital abnormalities have also been associated with this syndrome (Roelfsema *et al*., 1996; Giannotti *et al*., 1995). Numerous other less common associations have been described, including choanal atresia, vesicoureteric reflux, conductive hearing loss

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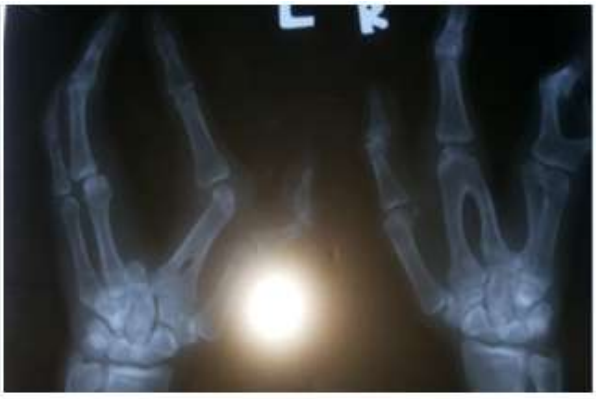
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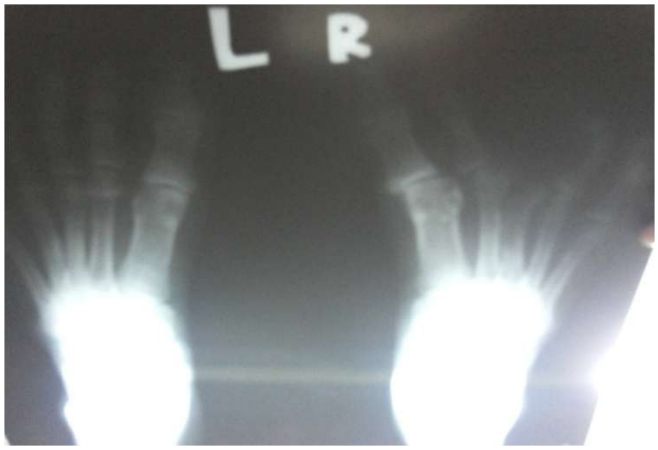

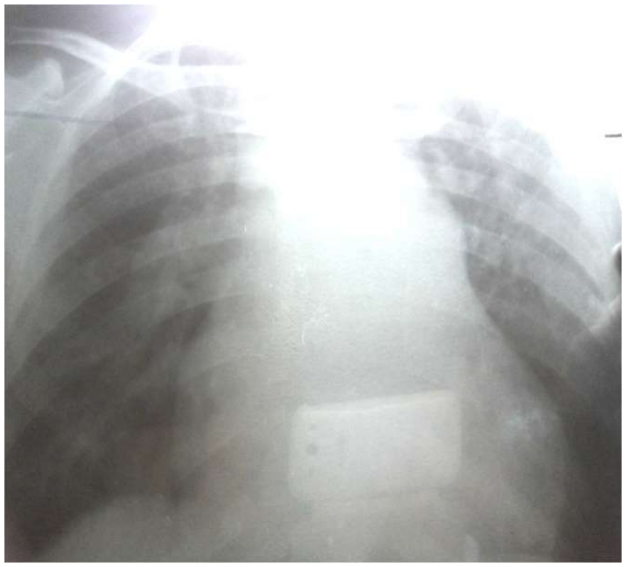

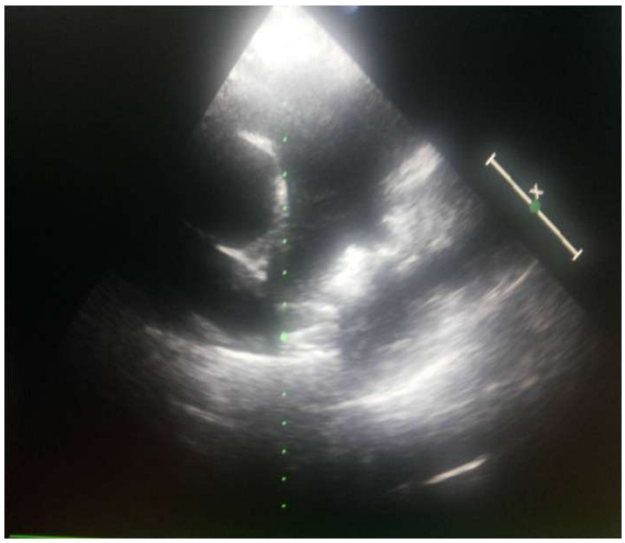

and recurrent respiratory infections (Roelfsema *et al*., 1996; Obel *et al*., 1993; Tucker *et al*., 1990; Christodoulou *et al*., 1989). The three cardinal symptoms of the EEC syndrome are ectrodactyly, ectodermal dysplasia, and clefting. The term "incomplete EEC syndrome" was suggested by Pries *et al*. (1974) for cases in which one or more of the cardinal symptoms was missing. Diagnosis of isolated cases of EEC can be established when two of the three cardinal symptoms are present, while familial cases are diagnosed with only one cardinal symptom in addition to the presence of a first degree relative with definitive EEC syndrome. Ectrodactyly-Ectodermal dysplasia-Clefting (EEC) Syndrome is inherited as autosomal dominant disorder of ectodermal and/or mesodermal interaction with highly variable expression and reduced penetrance (Roelfsema *et al*., 1996; McKusick *et al*., 1992; Fryns *et al*., 1990; Gorlin *et al*., 1990). Autosomal recessive and X linked inheritance occur more rarely. In EEC Syndrome mutations of the p63-[alpha] gene, a homolog of the p53 oncogene, have been described as the causative defect resulting in amino acid substitutions eliminating the DNA binding capability of the p63 protein. Three EEC loci have been identified. Fukushima *et al*. (1993) identified the break points of the EEC1 balanced translocation to 7q11.21 and 9p12. EEC2 has been defined by D19S894 and D19S416 on chromosome 19 (O'Quinn *et al*., 1998). It is noted that a locus, representing the proto-oncogene BCL3, resides on at 19q13. The recent locus at 3q27 described by Celi *et al*. (1999) represents EEC3.

In a retrospective analysis of 230 cases of the EEC syndrome, Roelfsema *et al*. (1996) determined that ectrodactyly was present in 84%, ectodermal dysplasia in 77%, and clefting in 68% of patients. Roelfsema *et al*. (1996) concluded that isolated cases of EEC syndrome are more severely affected than familial cases. More analyses of the gene(s) involved in EEC as well as the functional interactions of genes are required in order to better understand why some symptoms are more common than others. In another analysis of 31 cases of EEC syndrome, cleft lip/palate was present in 40% of patients, abnormal ears in 70%, sparse, short hair , scanty eyebrows, plantar/solar hyperkeratosis in 100% each ,abnormal teeth in 70%, abnormal nails in 60% and syndactyly in 65% of patients.

Ectrodactyly is also known as Split hand/split foot malformation (SHFM) or lobster claw syndrome/ cleft hand. Incidence is at ~ 1 in 90,000 - 150,000 births.Ectrodactyly can be caused by deletions, translocations, and inversions in chromosome 7 (7q). Typical hand manifest in the complete or incomplete absence of the middle finger with "V"-shaped cleft (Simon *et al*.,). The digits at the borders of the cleft might be syndactilyzed, and one or more digits can be absent and One to four limbs involved.Syndromes associated with ectrodactyly include-EEC syndrome, Ectrodactyly – Ectodermal Dysplasia - Macular Dystrophy(EEM) syndrome, Ectrodactyly - Cleft Palate (ECP)syndrome, SHFM with sensorineural hearing loss, SHFM with microphthalmia, colobomas and Microcephaly - microphthalmia -ectrodactyly – prognathism syndrome. Ectodermal dysplasias include- Maxillary hypoplasia; Hearing loss, small ears, low set ears and malformed auricles; Blue irides, blepharitis, dacryocystitis and lacrimal duct abnormalities; Broad & flat nasal tip ; Cleft lip, cleft palate and xerostomia; Tooth agenesis, microdontia and caries; Choanal atresia; Hypoplastic nipples of breast; skin changes including hypohydrosis and hyperkeratosis; Nail abnormalities including dystrophic and pitted nails; Hair changes namely light colored hair, sparse & thin scalp hair, pubic hair and axillary hair, sparse eyebrows and eyelashes; Genital anomalies including micropenis & cryptorchidism in males, and transverse vaginal septum in females; Renal anomalies including Renal agenesis,

renal dysplasia and hydronephrosis; Ureteral malformations including vesicoureteral reflux and ureterocele.

There are a number of syndromes that must be considered within the differential diagnosis of clinical symptoms described in EEC Syndrome, including Rapp-Hodgkin syndrome, Rosselli-Gulienetti syndrome, Hay-Wells syndrome, Bowen-Armstrong syndrome, Goltz-Gorlin syndrome, Odontotrichomelic hypohidrotic dysplasia, EEM syndrome, ODD syndrome, ECP syndrome, Roberts syndrome, Herrmann syndrome, syndrome of Fontaine, ADULT syndrome and Limb-mammary (LMS) syndrome, Ectrodactyly –Cleft Palate (ECP) syndrome, Acro-cardio-facial syndrome (ACFS), and Ectrodactyly- Polydactlyly.

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