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Case Report

Ectrodactyly ectodermal dysplasia and cleft lip and palate in new born: a rare case report

Lakhan Poswal, Mahima Joshi*, Ritvika Jyani, Akanksha Sharma, Kapil Sharma

Department of Pediatrics, RNT Medical College, Udaipur, Rajasthan, India

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***Correspondence:**
Dr. Mahima Joshi,
E-mail: mahimajoshi97@gmail.com

ABSTRACT

Ectrodactyly, ectodermal dysplasia, and cleft lip/palate syndrome (EEC syndrome) is a rare genetic disorder caused by mutations in the TP63 gene. It is characterized by a combination of limb malformations (split-hand and split-foot), ectodermal dysplasia (involving hair, nails, skin, and teeth), and cleft lip/palate. We report a case of a full-term male newborn with low birth weight and intrauterine growth restriction who presented with multiple congenital anomalies, including bilateral ectrodactyly, wide unilateral cleft lip and palate, sparse scalp hair, dysplastic nails, reduced dental buds, and mild ophthalmic and genitourinary anomalies. Genetic testing identified a pathogenic TP63 gene variant, confirming the diagnosis of EEC syndrome. EEC syndrome follows an autosomal dominant inheritance pattern with variable expression and requires a multidisciplinary approach for management. This includes dermatological, surgical, dental, and ophthalmological interventions, with a focus on improving the quality of life. Genetic counselling is crucial for affected families to guide future pregnancies and provide emotional and medical support. Early diagnosis and supportive therapies are essential for enhancing the child's quality of life.

Keywords: Ectrodactyly, Ectodermal dysplasia, Autosomal dominant, TP63 gene, Split-hand, Split-foot

INTRODUCTION

EEC syndrome, short for ectrodactyly-ectodermal dysplasia-cleft lip/palate syndrome, is a rare genetic condition that occurs in approximately 1 out of every 90,000 individuals. It is also referred to as split hand/split foot ectodermal dysplasia cleft syndrome, or colloquially as lobster claw hand/foot due to its characteristic limb malformations.[1] Ectrodactyly, a key feature, typically impacts the central digits of the hands and feet, though its appearance can vary significantly.

A cleft lip and/or cleft palate-gaps in the upper lip and roof of the mouth-are also common. The ectodermal dysplasia aspect involves abnormalities in structures that originate from the embryonic ectoderm, including the skin, hair, nails, teeth, and sweat glands. Individuals with EEC syndrome may also experience issues with the eyes and genitourinary system. Despite these physical manifestations, cognitive development and intelligence are usually unaffected. Most cases result from mutations in the TP63 gene and can arise spontaneously or be passed down through an autosomal dominant inheritance pattern.[2]

CASE REPORT

A full-term LBW IUGR male newborn (Figure 1), birth weight of 1.8 kg was delivered via normal vaginal delivery at 37 weeks of gestation, presented with multiple congenital anomalies on day 1 of life in outborn NICU. The pregnancy was uncomplicated, G5P2L2A2, with no significant maternal illnesses or teratogenic exposures. Antenatal USG suggestive of enlarged fetal right kidney and multiple anechoic cyst suggestive of multicystic dysplastic kidney. First baby died at the age of 4 year and there was family history of similar congenital anomaly in elder sibling. On physical examination, the newborn had

limb anomalies which includes bilateral ectrodactyly with absence of central digits of both hands and feet, characteristic of split-hand and split-foot malformation (Figure 2,3) along with a wide unilateral cleft lip and cleft palate. Ectodermal dysplasia features were sparse scalp hair, hypoplastic eyebrows, and dry, scaly skin. The nails were dysplastic, and the infant had a reduced number of dental buds on oral examination. Ophthalmic anomalies include mild corneal erosions and sparse eyelashes were noted. And mild hypospadias without other structural abnormalities.

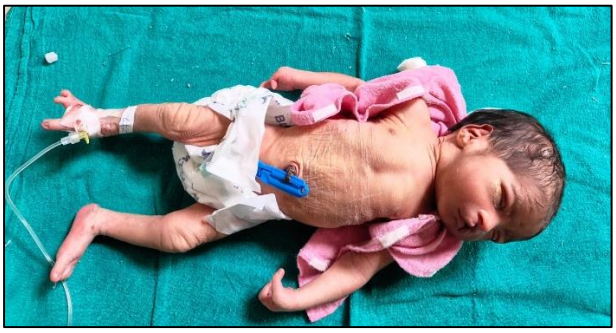

Figure 1: Full-term LBW IUGR male newborn.

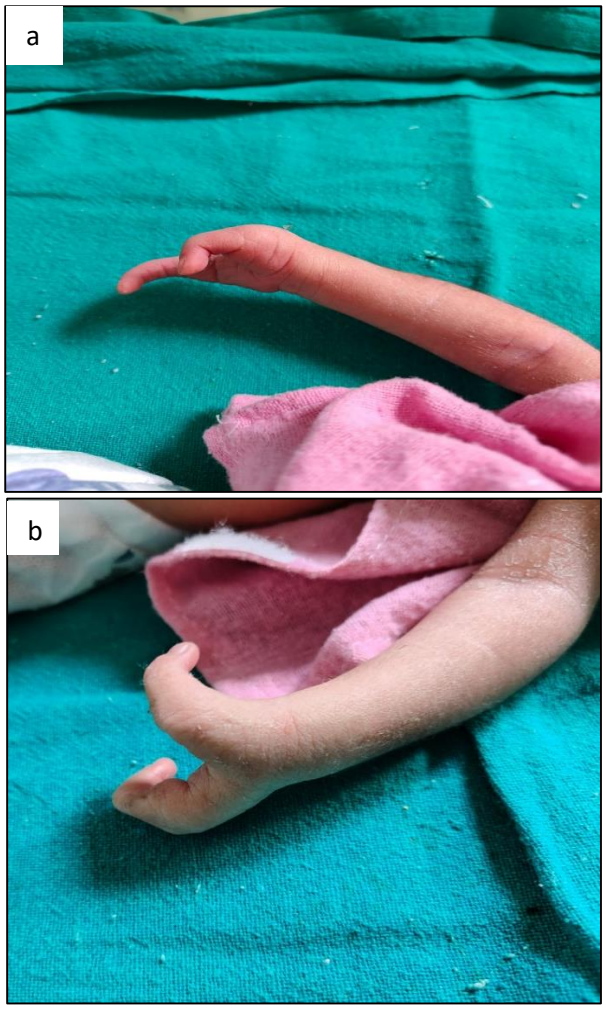

Figure 2 (a and b): Split hand.

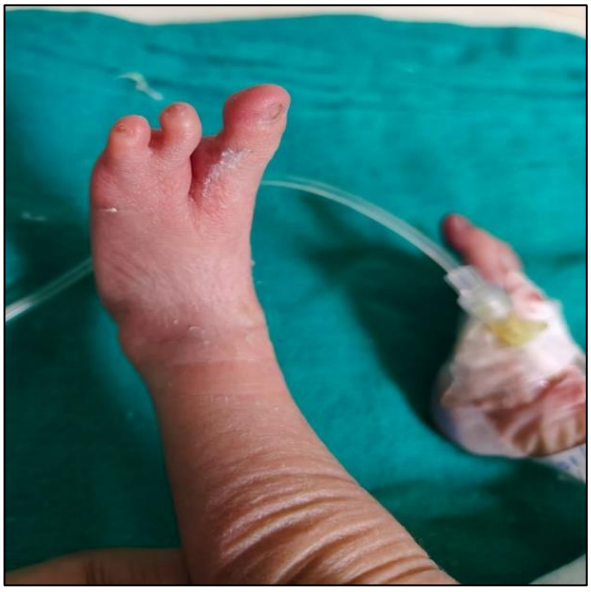

Figure 3: Split foot.

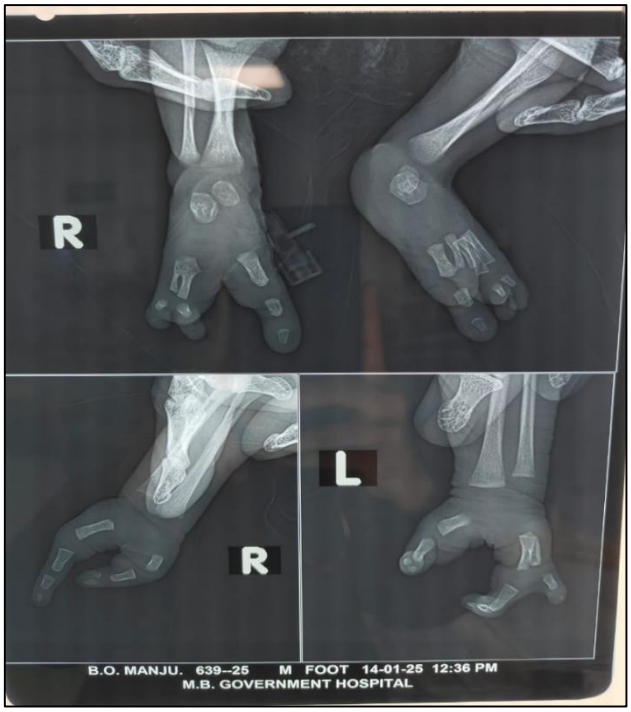

Figure 4: X-ray upper limb and lower limb.

Routine investigations including CBC, RFT, LFT and sepsis screen was done. S.urea (25.2 mg/dl), S. creat (1.5 mg/dl), Hb (22.4 gm/dl), plt (240 k per microlitre), TLC (6.01 k per microlitre), CRPQ (7.61 mg/dl). X-rays of the upper and lower limbs confirmed the absence of central rays, characteristic of ectrodactyly (Figure 4). USG abdomen and pelvis suggestive of multicystic dysplastic kidney. 2D ECHO was done and was found to be normal. OAE was normal. Genetic testing identified a pathogenic variant in the TP63 gene, confirming the diagnosis of EEC syndrome. Further, follow up was not done.

DISCUSSION

EEC syndrome is an autosomal dominant genetic disorder primarily caused by mutations in the p63 gene-a transcription factor related to the p53 gene family and known for its role in tumor suppression. The hallmark features of EEC syndrome include ectrodactyly, which involves missing digits and syndactyly; ectodermal dysplasia, which can present as abnormal secondary dentition, reduced sweating, brittle or misshapen nails, and sparse, dry, or lightly pigmented hair; and cleft lip and/or palate. Although the term "lobster claw deformity" was historically used to describe limb anomalies in this syndrome, it is now considered outdated and inappropriate.[3]

Research indicates that individuals with isolated (sporadic) EEC cases often exhibit more severe symptoms compared to those with familial forms4. Previous studies have also highlighted lacrimal duct anomalies as a significant diagnostic criterion. Ophthalmological manifestations commonly seen in affected individuals include entropion, wide-set eyes (hypertelorism), absent lacrimal puncta, sparse eyelashes (hypotrichiasis), eyelid inflammation (blepharitis), sensitivity to light (photophobia), corneal clouding, and inflammation of the tear sac (dacryocystitis).[5] Some individuals may also experience genitourinary malformations. Conductive hearing loss, frequently linked with cleft lip and palate, has also been reported.

When diagnosing EEC syndrome, it is important to consider other conditions with overlapping features. These include Split-hand/Split-foot Malformation (SHFM), a congenital anomaly marked by a deep cleft in the hands or feet due to missing central digits.[6] Another condition to consider is Acro-cardio-facial syndrome (ACFS), a rare genetic disorder characterized by SHFM, facial abnormalities, cleft lip/palate, congenital heart defects, genital malformations, and intellectual disability.[7]

Hay-Wells syndrome, which shares features such as clefting, ectodermal abnormalities, and ankyloblepharon, filiform adnatum (fusion of eyelids).[8] Additionally, Acro-Dermato-Ungual-Lacrimal-Tooth (ADULT) syndrome presents with features like ectrodactyly, syndactyly, nail dysplasia, underdeveloped nipples and breasts, extensive freckling, blocked tear ducts, frontal baldness, and early tooth loss.[9]

Management of EEC syndrome requires a collaborative, multidisciplinary approach. Specialists such as dermatologists, plastic and dental surgeons, ophthalmologists, and nephrologists may all play a role in providing comprehensive care.

Surgical interventions to address physical anomalies, particularly of the skin, eyes, and oral cavity, can significantly enhance the patient's quality of life. Genetic counselling is recommended for affected individuals and their families.[10] Prenatal diagnosis using cleft lip and palate as a marker can also be done in the case of families with affected children.

CONCLUSION

This case highlights a newborn with typical manifestations of the syndrome, such as split-hand and split-foot malformation, cleft lip and palate, ectodermal dysplasia, and ophthalmological and genitourinary anomalies. Genetic testing confirmed a pathogenic variant in the TP63 gene, solidifying the diagnosis. While the condition is autosomal dominant with variable expression, management requires a comprehensive multidisciplinary approach to address the dermatological, dental, ophthalmological, and surgical needs of the patient. Early recognition, genetic counselling, and supportive therapies are essential for improving quality of life and offering the family appropriate guidance for future pregnancies.

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