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3 GENERATIONS OF LOBSTER CLAW SYNDROME: A RARITY

Paediatrics

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ABSTRACT

Ectrodactyly also known as Split hand/foot malformation is a rare limb deformity with majorly expressing as an autosomal dominant inheritance with variable penetrance, commonly known as "lobster claw hand". Usually, it involves midline clefts of the hands and feet with syndactyly. Here we are reporting an autosomal dominant non-syndromic ectrodactyly with syndactyly in an Indian family expressing three generations.

KEYWORDS

Ectrodactyly, Split Hand Deformity, Three Generations

INTRODUCTION:

Ectrodactyly, split hand, or cleft hand (derived from Greek *ektroma* 'abortion' and *daktylos* 'finger') involves the deficiency or absence of one or more central digits of the hand or foot and is also known as split hand/split foot malformation (SHFM). The rare combination of ectrodactyly (lobster claw deformity), ectodermal dysplasia and cleft lip with or without cleft palate has been described as EEC syndrome.

It was first described by Cockayne in 1936 but the acronym was coined by Rudiger et al. Ectrodactyly may be associated with syndactyly, aplasia or hypoplasia of phalange, metacarpals and metatarsals. The incidence is rare, generally 1 per 90000 live births. It may present either as an isolated case or in association with a syndrome. In syndromic form, ectrodactyly occurs with other anomalies such as ectodermal dysplasia, cleft lip and palate, genitourinary and other anomalies of ectodermal origin. Often in genetic inheritance, it can present as autosomal dominant pattern with reduced penetrance. Infrequently it exhibits as autosomal recessive and X linked forms.

Here we describe a case of ectrodactyly in isolated non syndromic form claw in three living generation of an Indian family which presented as autosomal pattern of inheritance with sporadic occurrence.

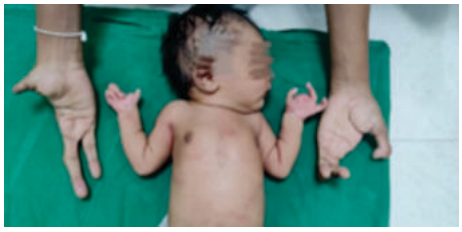
Case Report:

A 25 year old primigravida mother, with malformation involving upper and lower limb, registered and immunized at a primary health center was brought to a tertiary care center at 9 months of amenorrhea in view of pain in abdomen. She was on her regular antenatal check-ups and received regular course of medicines. She had undergone 2 ultrasonography, but no abnormality was detected. History of consanguinity, radiation exposure and drug exposure was ruled out.

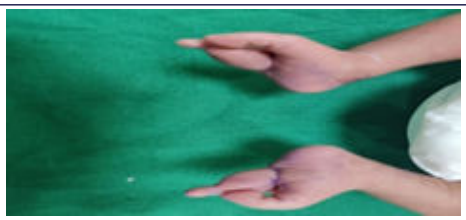
A female baby was born by spontaneous vaginal delivery, cried immediately after birth, with a birth weight of 2.5 kgs with evidence of malformation of upper and lower limb noted at birth. No resuscitation was required. Breast feeding was initiated within 30 minutes after birth.

On Examination:

1. Patient:



a. Hand: Medial cleft in the metacarpals with aplasia of phalanges of 2nd and 3rd finger with classical split hand deformity

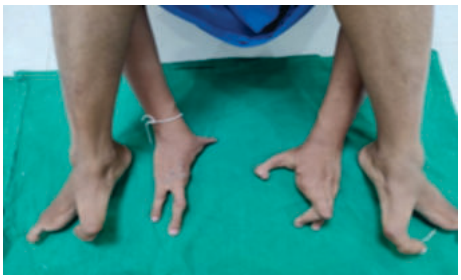


b. Legs: There is a medial cleft in the metatarsals with aplasia of phalanges of 2nd, 3rd and 4th toes of both feet with classical cleft foot or split foot appearance.

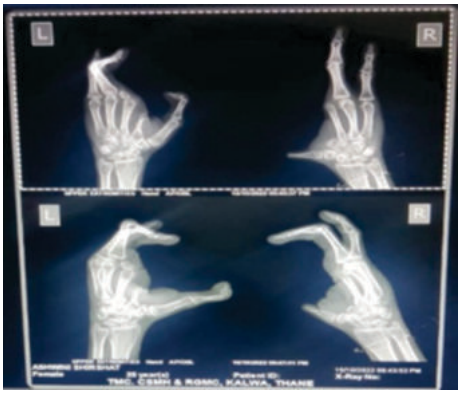
Her echocardiogram revealed double outlet right ventricle. There was no visible cleft lip or palate. Hair and nails were normal. There was no auditory or neurological deficit.

There was no facial cleft or ocular abnormality. Abdominal ultrasonography did not show any abnormality

2. Mother:

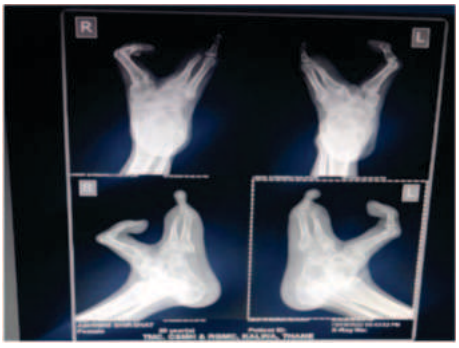


a. Ectrodactyly of both hand and feet



b. Hands: Both her hands showing lengthening and broadening of the digits on x-ray. There is a medial cleft in the metacarpals,

dividing the hand into two portions with aplasia of 2nd and 3rd phalanges of both hands. Syndactyly of 4th and 5th finger of left hand. The growth of the digits is more as compared to other body parts.



c. Lower limb: absent 3rd and 4th metatarsal with absent phalanges of mid 2 digits on x-ray

Flexion deformity of 1st digit
Systemic examination of the mother did not revela any other significant abnormality. Intelligence was normal.

3. Patient's Grandfather (Mother's father):

a. Hand: There is a medial cleft in the metacarpals dividing the hand into 2 portions. Syndactyly of 3rd and 4th finger noted in right hand with polydactyly of fingers in both the hands with clinodactyly of 5th finger in both hands.

b. Lower limb: There is lengthening and broadening of digits with a medial cleft at metatarsals with absent phalanges of mid 3 digits.

Systemic examination of the grandfather did not reveal any other significant abnormality. Intelligence was normal.

DISCUSSION:

Ectrodactyly was first documented in 1770 among a tribe of some Guiana in India. Other names for ectrodactyly include split-hand/foot malformation (SHFM), crab-claw deformity - first used by VonWalter in 1829, and lobster claw - first used by Cruveilhier in 1842.

The basic embryologic abnormality in ectrodactyly is failure to maintain a normal functioning apical ectodermal ridge (AER), leading to failure of differentiation of the autopod (hand or foot). In ectrodactyly, the hand and/or foot appear split into two halves with failure of development of the phalanx, metacarpal and/or metatarsal bones of one or more fingers and or toes. Along with facial anomalies as mentioned earlier various dermatological manifestations include hyper keratosis, thickened scaly skin to hypo pigmented dry skin with poor hair growth. Others include dysplastic nails and peg-shaped teeth. Tooth decay (dental caries) is very common. There are other associated abnormalities with sweat, salivary, lacrimal, and sebaceous glands. There have been few rare case reports of associated anomalies of the genitourinary system with various spectrum of malformations ranging from renal agenesis, renal stone to hydronephrosis.

Intelligence is usually preserved, however there may delay in speech development, which is due to associated hearing loss. Few long-term case report has shown their progression to Hodgkin lymphoma.

There are 8 main types of EEC syndrome and among which type 1 is the most common variety and TP63 gene mutation being the most common cause.

Other syndromes associated with ectrodactyly include Carpenter's syndrome, cornea DeLange syndrome, Goltz syndrome, and Miller syndrome.

Types:

TYPES	GENES	INHERITENCE	ASSOCIATED CONDITION
Type 1	DLX5, DLX6	Autosomal Dominant	Ectrodactyly and sensineural hearing loss
Type 2	FGF13 and TONDU	X linked Recessive	Along with consanguinity
Type 3	DACTYLIN, BTRC FGF8, POLL	Autosomal Dominant	-
Type 4	TP63	Autosomal Dominant	EEC Syndrome
Type 5	HOXD13	Autosomal Dominant	Ectrodactyly, Monodactyly
Type 6	WNT108	Autosomal Dominant	Syndactyly, Polydactyly
Type 7	ZAK gene	-	Mesoaxial polydactyly
Type 8	EPS15L1 gene	-	Affects embryonic development and neurogenesis

Management of this condition involves multidisciplinary involvement. The confirmation of EEC syndrome can be done by molecular genetic testing for TP63 gene mutation. Antenatal diagnosis is feasible using the molecular genetic testing and samples are obtained using chorionic villus sampling. The prognosis for most people with ectrodactyly syndrome is very good. Various cosmetic prosthesis, amputation or reconstructive surgery might help, depending on the type of deformity and as per case basis. Multidisciplinary care contributes significantly to improve the survival and the quality of life of a child with ectrodactyly. Genetic counseling is recommended for affected individuals and their families.

CONCLUSION:

Ectrodactyly or split hand/foot malformation is a rare congenital malformation of limb with median cleft and hypoplasia of phalanges. Most commonly it follows an autosomal dominant pattern. Surgical correction can be considered. Proper counselling regarding recurrence in future siblings should be done with advise to go for antenatal ultrasound.

Conflict Of Interest: Nil

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