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ECTRODACTYLY – RARE INDIAN DISORDER : A CASE REPORT

Dr. Tarun Singh, Senior Resident [1]Dr. Deepak Gupta, Senior Consultant [1]Dr. Satish Kumar, Consultant [1] Dr. Patil Tushar Liladhar, Consultant Radiologist [2]

[1] Dept. of Pediatrics, Sanjay Gandhi Memorial Hospital, Mangolpuri, Govt. of NCT of Delhi, Delhi- 110083

[2] Suman Diagnostics, Pune, Maharashtra

Introduction:

Ectrodactyly or Split Hand & Split Foot Malformation (SHFM) is congenital absence of one or more central/median rays or digits creating cone shaped clefts of hands and/or feet.[(1)(2)] Term ectrodactyly was originated from greek words- Ektroma (abortion) and Daktylos (Finger). Incidence reported is 1:90,000 live births with no sex predilection. Ist documented case was in a tribe of Guiana Indians in 1770.[(2)] It was called "Crab claw" deformity for first time by Von Walker in 1829 and "Lobster claw" by Cruveilhier in 1842.[(3)] It is also known as Karsch Neugebauer Syndrome.[(4)]

Basic embryologic abnormality involved is failure to maintain normal functioning apical ectodermal ridge, which leads to failure to differentiate the autopod (hand or foot).[(2)] It occurs in two forms, either isolated or as component of a syndrome. Any of these two forms can be sporadic or familial, of which former being more common.[(3)] In familial form, most common pattern of inheritance is Autosomal dominant with cases of Autosomal recessive and X linked inheritance reported rarely.[(2)]

EEC syndrome is characterised by triad of ectrodactyly, ectodermal dysplasia and facial clefting (lip and/or palate) which was first described by Cockayne in 1936. Incidence of syndrome is very rare and is found to be 1.5 per hundred million live births.[(5)] Term EEC syndrome was coined by Rudiger et al in 1970. Most common mutations were seen that involves TP63 and TP73L genes.[(6)] Prenatal diagnosis was first reported in 1980 and most patients lived normal life with no or minimal intellectual deficiency.[(7)]

There are five different mutations known to be associated with SHFM, of which Type I being the most common, is due to chromosome 7 mutation.[(8)] It involves a region containing 2 homeobox genes DLX 5 and DLX 6.[(4)] Other associated syndromes are Carpenter's syndrome, Delange syndrome, Goltz syndrome & Miller syndrome.[(3)]

Case report:

We report a case of term male child born by normal vaginal delivery at our hospital weighing 2.6 kg. His mother received routine Ante Natal care and all antenatal ultrasounds (2 dimensional) were reported normal. No unknown medications were taken except for iron folic acid and calcium supplementation.

On physical examination, there were only 2 toes present in bilateral foot and 2 fingers in each hand. There was no other congenital abnormality seen.

He was shifted to NICU for further evaluation. Routine treatment was started in form of intravenous fluids and IV antibiotics. Infantogram was done which confirmed the diagnosis. Baby was shifted from NICU on day 2 of life and then discharged and referred to higher center for further evaluation.

Probable pattern of inheritance was Autosomal recessive as there was no positive family history.

Relevant images are shown below.

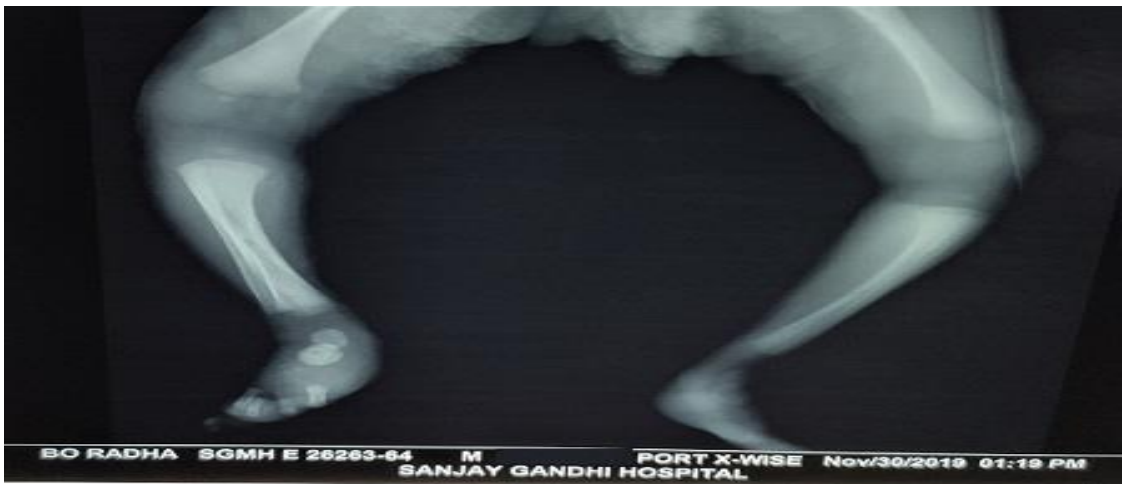
Figures:


RIGHT HAND


RIGHT LEG


SEX:
LEFT HAND


LEFT LEG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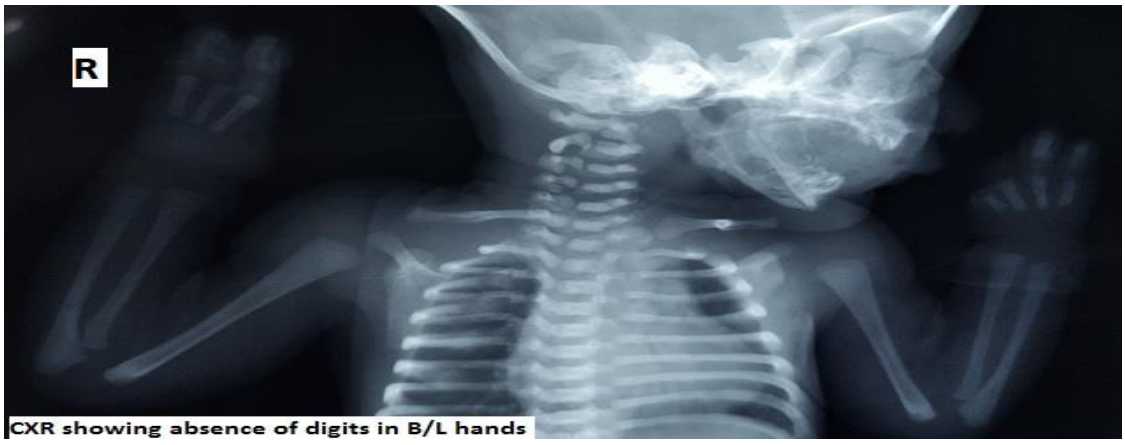
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CXR showing absence of digits in B/L hands

Discussion:

Ectrodactyly, also known as SHFM, involves complete absence of central rays of the autopod due to which each hand and foot divides into 2 parts giving it a "Lobster claw" appearance.[1] Its a rare genetic disorder with incidence of 1: 90,000 live births with no sex predilection.[2]

It is associated with EEC syndrome which is usually inherited as Autosomal dominant disorder. Other associated orofacial abnormalities includes lack of permanent incisors, adontia, enamel hypoplasia, deeply furrowed tongue, photophobia.[9]

Bernstein et al described sudden death of infants due to hypohidrotic ectodermal dysplasia.[9] Birch-Jensen after investigating 30 families found 90% cases of SHFM are sporadic and only 10% are inherited.[10]

Blauth and Borisch proposed a radiographic classification of cleft feet and 6 grades were given.[2] Verma et al reported a family with atypical split hand & foot deformity found in 4 children from 2 related parents.[11]

Hartsfield et al in 1984 described first case of ectrodactyly associated with Holoprosencephaly.[5] In recent studies, chromosome 19 with regions of D19S894 & D19S416 has been postulated as Locus for EEC syndrome.[6] Also two locus model has also been suggested in which dominant mutation leading to SHFM being controlled by a gene at another locus.[12]

Diagnosis could be made using antenatal fetal ultrasonography.[8] The two studies conducted by Budorick N.E. et al and Hull A.D. et al showed better visualization and delineation of complex malformations like ectrodactyly or EEC syndrome when used 3D ultrasonography for fetal assessment during ante natal period instead of 2D ultrasonography.[13] Also in a study by Allen & Maestri, 3D sonography rendered better view for facial anomalies and ectrodactyly of limbs.[14] In our case, 2D ultrasound was used in 1st trimester which was reported normal and no subsequent ultrasound was done.

It can be treated surgically in order to improve function and appearance. Prosthetics can also be useful.[7] Genetic counselling should be offered to parents to explain the prognosis and possibility of recurrence of disease in future siblings.

Conclusion:

Importance of the early diagnosis of ectrodactyly or associated syndrome should be emphasized in order to decrease morbidity and mortality. We reort a case of ectrodactly with the clinical findings associated and infantogram confirming it. In this reported case, there was no positive family history and inheritance pattern suspected was autosomal recessive which is rare inheritance pattern for ectrodactyly. Antenatal ultrasound was reported normal. The attributed reason being use of 2D ultrasonography which is an inferior mode for detection of skeletal deformities as compared to 3D ultrasonography. It may be further emphasized that in suspected skeletal deformities/dysplasias, antenatal screening should be done by 3D ultrasonography, wherever possible, to improve the detection rate. Proper management would require multidisciplinary approach which includes plastic surgeon, dermatologist, ophthalmologist, pediatric endocrinologist and neurologist.

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