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Case Report

Ectrodadactyly: a rare case report

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ABSTRACT

Ectrodactyly also called as split hand or foot malformation (SHFM).Congenital absence of all or part of one or more fingers or toes is defined as Ectrodactyly. Irregular pattern of inheritance exists for ectrodactyly, may be autosomal dominant, autosomal recessive or X linked. In this condition, the median cleft of the hand gives the appearance of lobster claw-so called 'Lobster Claw Hand'. Ectrodactyly may be very commonly associated with other anomalies of face, eyes and skeletal system. We report a rare case of unilateral ectrodactyly of right hand.

Keywords: Ectrodactyly, Split hand, Foot malformation, Median Cleft

INTRODUCTION

Ectrodactyly, also known as Lobster claw hand, is a rare limb malformation involving the central rays of autopod and presenting with syndactyly, median clefts of the hands and feet and aplasia or hypoplasia of the phalanges, metacarpals or metatarsals.[1] The absence of central digital rays gives rise to the median cleft, resulting in the lobster appearance of hands and feet.

Ectrodactyly has also been known as split hand or foot malformation (SHFM), or Karsch Neugebauer syndrome or EEC (Ectrodactyly- ectodermal Dysplasia-clefting) syndrome.[2]

We report a rare case with Ectrodactyly in new-born.

CASE REPORT

24 year old female delivered a full term male new born by caesarean section with birth weight 2.32kg, born by non-consanguineous marriage. Antenatal scan was normal.

On physical examination right hand showed claw shaped deformity (Figure 1) with absence of middle three fingers with normal development of other bones of same limb and the other three limbs was normal. No other obvious deformities. Systemic examination was found to be normal. Radiograph was taken which showed there is a deficiency of three central digits of hand, characterized by a deep U shaped central bony defect (Figure 2) and normal development of other bones in the same hand. Ultrasonography of abdomen and Echo showed normal study.

DISCUSSION

Ectrodactyly involves median clefts of the hands and feet with associated syndactly, aplasia/hypoplasia of phalynges, metacarpals and metatarsals.[3] First case of Ectrodactyly was described in 1936. Prenatal diagnosis of ectrodactyly was first reported in 1980.[4] The anamoly of ectrodactyly develops secondary to one of the chromosomal mutation which results in failure of AER (Apical epidermal ridge) to produce molecules that signals nearby cells to differentiate into digital rays, so far only mutations known to underline SHFM in humans have been found in TP63 gene.[5] Five different genetic mutations are known to be associated with SHFM. Type 1, the most frequent variety is due to mutation on chromosome 7 in a region contains 2 homeobox genes DLX5 and DLX6.[6]

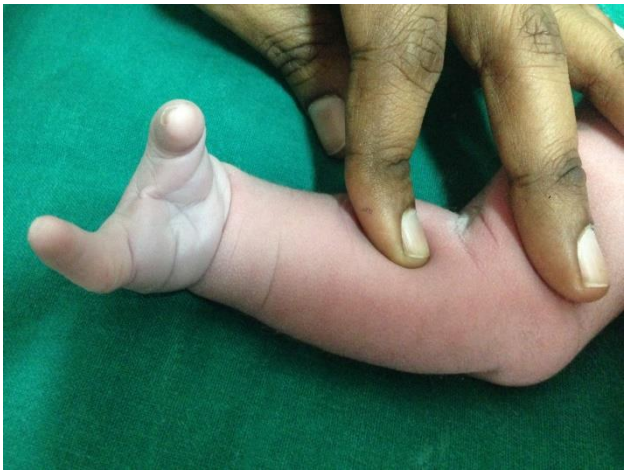

Figure 1: Right hand showed claw shaped deformity with absence of middle three fingers.

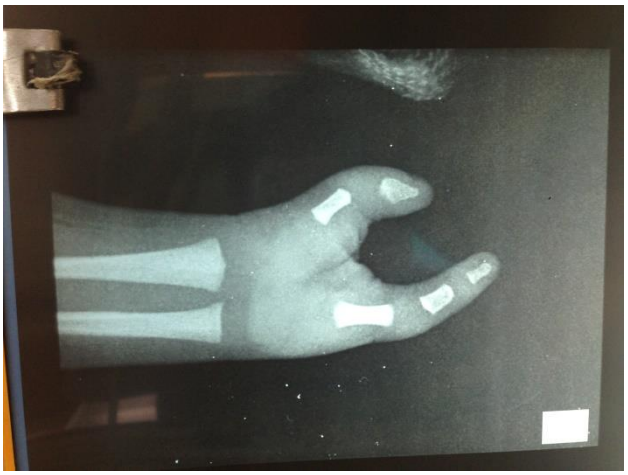

Figure 2: Deep U shaped central bony defect.

Two expressions of SHFM occurs, one with isolated involvement of limbs known as the Non syndromic form and the second, the syndromic form with associated anamolies such as tibal aplasia, mental retardation, ectodermal craniofacial findings and orofacial clefting and deafness.[7] The syndromic form has a variable degree of expression. The non syndromal SHFM limited to the hands and feet usually follows the pattern of inheritance of a regular autosomal dominant gene with high penetrance.[8]

We report a case of non syndromic type of SHFM as there is no associated anamolies and the probable pattern of inheritance is autosomal recessive as no other family member are affected. Ectrodactyly can be treated surgically in order to improve function and appearance prosthetics may also be used.[9]

CONCLUSION

Cerebral sinus-venous thrombosis is a rare but life-threatening disease. Oral contraceptives are one of the leading iatrogenic causes. The true incidence of CSVT secondary to OCP's is not known. Growing knowledge about use of OCP's and its widespread availability has led to its usage without any medical supervision leading to complications. This case enlightens about importance of counselling and educating parents on early clinical signs and insisting on regular follow-up, could lead to early diagnosis and intervention.

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