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LETTER TO EDITOR (VIEWERS CHOICE)

# ECTRODACTYLY-A RARE LIMB MALFORMATION

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A one year old male child born by non-consanguineous marriage presented with right hand anomaly from birth. On physical examination right hand showed claw shaped deformity (Figure 1) with absence of middle finger, symbrachydactyly of ring and little finger, V shaped cleft in the middle of the hand with normal development of other bones of same limb. The other three limbs was normal. No other obvious deformities were present. Systemic examination was unremarkable. Radiograph of right hand showed a deep V shaped central bony defect (Figure 2) and normal development of other bones in the same hand. Ultrasonography of abdomen and echocardiography were normal. There was no history of similar malformation in other family members.

**Figure 1.** Ectrodactyly

Ectrodactyly is one of the rare limb malformation involving the central rays of autopod clinically 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 results in median cleft with the lobster appearance of hands and feet and hence the name lobster claw

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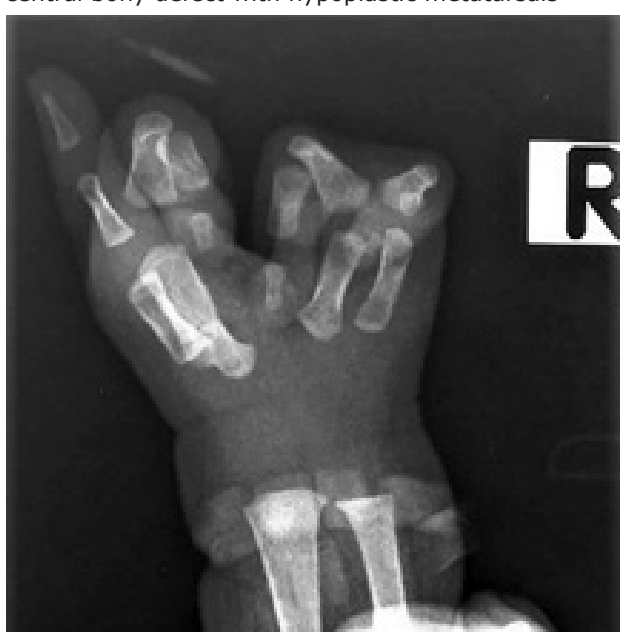
**Figure 2.** X-ray of right hand showed a deep V shaped central bony defect with hypoplastic metatarsals



hand. Ectrodactyly is also known as split hand or foot malformation (SHFM), or Karsch Neugebauer syndrome or EEC (Ectrodactyly- ectodermal Dysplasia-clefting) syndrome.[2] First case of ectrodactyly was reported in 1936[1] and first antenatal diagnosis of ectrodactyly was reported in 1980.[3] Ectrodactyly develops due to failure of apical epidermal ridge to produce molecules that signals nearby cells to differentiate into digital rays. Pattern of inheritance for ectrodactyly may be autosomal dominant, autosomal recessive or X-linked.[4] Seven different genetic mutations are known to be associated with SHFM namely SHFM1 to 6 and SHFM/SHFLD.[4] Two forms of ectrodactyly occur, one with isolated involvement of limbs. It is known as the non-syndromic form, which follows pattern of inheritance of a regular autosomal dominant gene with high penetrance. The second form is the syndromic form with associated anomalies such as tibial aplasia, mental retardation, ectodermal craniofacial findings and orofacial clefting and deafness.[5] Blauth and Borisch classified ectrodactyly into six groups based on the number of metatarsal bones: Type I have five normal

metatarsals; Type II have five metatarsals, which were partially hypoplastic; Type III have four metatarsals; Type IV haave three metatarsals; and Type V haave two metatarsals. Type VI represents the monodactylous cleft foot. Two additional groups include: cleft feet with central polydactyly (polydactylous type) and monodactylous feet with lower-leg diastasis or tibial aplasia or both (the diastatic type).[6] Our patient had non-syndromic type of isolated right hand ectrodactyly, type II according to Blauth and Borisch classification. Ectrodactyly can be treated surgically in order to improve function and appearance. Prosthetics may also be used and requires a multidisciplinary approach for optimal functional, cosmetic, and psychological result.[1]

**Compliance with Ethical Standards**

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**Conflict of Interest:** None

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