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IMAGES IN PEDIATRIC SCIENCES

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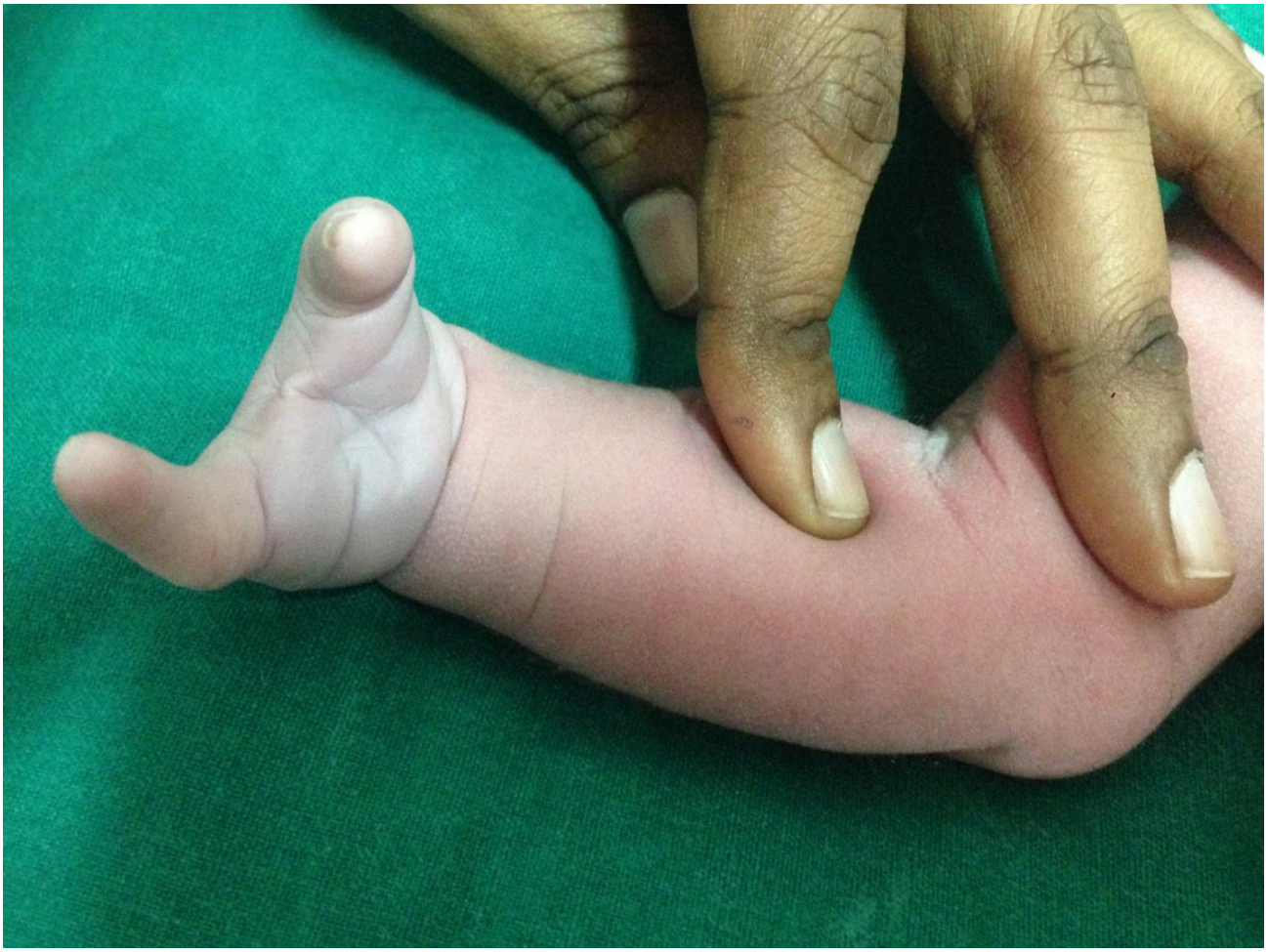

Figure 1. Claw hand or lobster hand

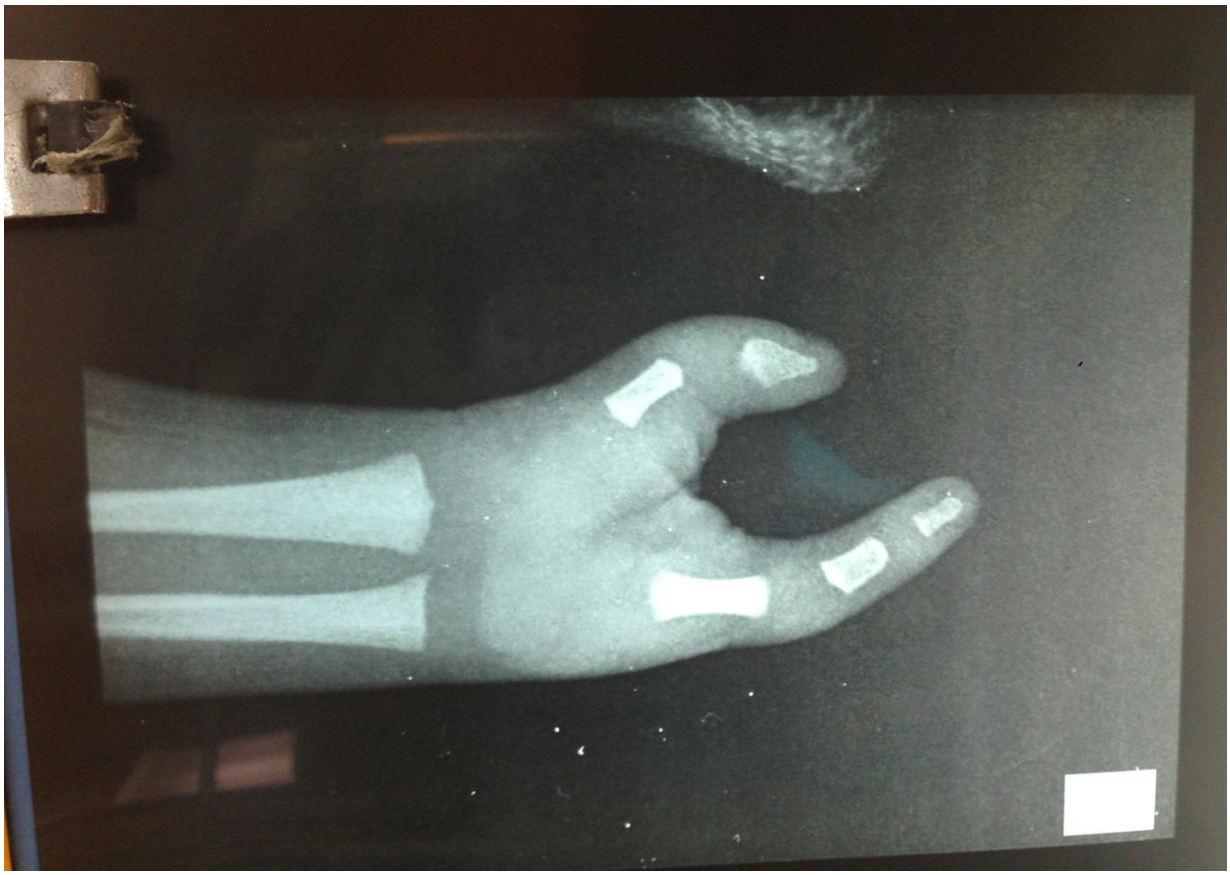

Figure 2. X ray of right wrist joint: a deficiency of three central digits of hand, characterized by a deep U shaped central bony defect.

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).

24 year old female delivered a full term male new born by caesarean section with birth weight 2.32 kg. 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.

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 (3).

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, ectrodermal craniofacial findings and orofacial clefting and deafness (6). 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 (7).

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