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RESEARCH ARTICLE

FAMILIAL SPLIT-HAND/FOOT MALFORMATION: (LOBSTER CLAW ANOMALY)

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ABSTRACT

Split Hand/split-Foot Malformation (SHFM) is a congenital anomaly with failure of development of the central digital rays of hand or foot to a variable extent. It is characterized by hypoplasia/aplasia of the phalanges, toes, metacarpals and metatarsals. The presentation may be an isolated anomaly or may be associated with syndrome and thus have variable pattern of inheritance. SHFM may occur as an isolated entity or as part of a syndrome. Both forms are frequently found in association with chromosomal rearrangements such as deletions or translocations.

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INTRODUCTION

Case History

This 28yr old male patient hospitalised for treatment of acute viral hepatitis A. On general physical examination apart from icterus there was a peculiar absence of central arrays of both hands and feet (Figure 1 & 2). Systemic examination was unremarkable. Patient gave history of similar absence of central rays of both hands and feet's in his family members- two brothers, one sister, his son, nephews, father, uncle and grandfather (Total Males-9 and female-1) with variable severity. On making pedigree the affection was autosomal dominant with variable penetrance.There were no associated anomalies, such as ectodermal dysplasia, cleft lip/palate, malformations of the long bones or internal organs, and overt mental retardation. Split-hand foot malformation (SHFM) is also known as ectrodactyly. This is a rare genetic disorder with an incidence of 1:90000 (Moerman and Fryns, 1998). It is acongenital limb defect affecting predominantly the central rays of hands and/or feet. SHFM is a clinically heterogeneous abnormality, which ranges from a relatively mild defect, such as hypoplasia of a single phalanx or syndactyly, to the aplasia of one or more central digits (i.e., classical cleft also known as lobster-claw anomaly) (Köhler *et al*., 1989). In utero valproic acid exposure can cause ectrodactyly rarely (Köhler *et al*., 1989). The condition is mostly sporadic, familial forms are uncommon (Ahmad *et al*., 1987).

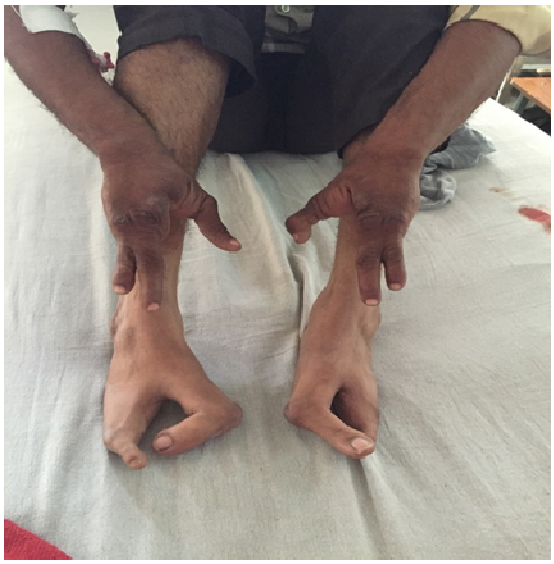

Figure 1. Patient with SHFM phenotype -Lobster Claw Anomaly (Ectrodactyly)

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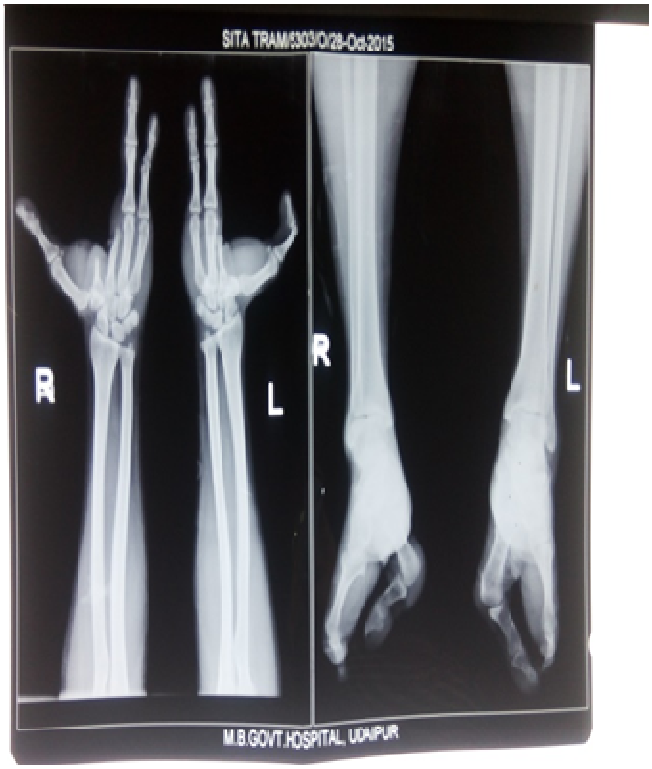

Figure 2. X-ray Of both upper and lower limb showing absence of central rays of extremities

In the majority of cases SHFM undergoes autosomal dominant mode of inheritance with reduced penetrance, while X linked and autosomal recessive forms occur more rarely (Ahmad *et al*., 1987; Pascal *et al*., 2003). Common anomalies associated with ectrodactyly include tibial aplasia, craniofacial defects, and genitourinary abnormalities (Pinette *et al*., 2006).

Table 1: Types of SHFM syndromes

Type	Description
I	Web space not narrowed
IIA	Web space mildly narrowed
IIB	Web space severely narrowed
III	Syndactylized fingers and web space obliterated
IV	Thumb web space is merged with the cleft
V	Thumb web space no longer present.

(Sunil K Mahavar and Ashutosh Chaturved, 2016)

REFERENCES

Ahmad M, Abbas H, Haque S. and Flatz G. 1987. X-chromosomally inherited split-hand/split-foot anomaly in a Pakistani kindred. *Hum Genet*., 75(2):169–173

Moerman, P. and Fryns, J.P. 1998. "Ectodermal dysplasia, Rapp-Hodgkin type in a mother and severe ectrodactyly-ectodermal dysplasia-clefting syndrome (EEC) in her child". *American Journal of Medical Genetics Part A*. 63 (3): 479–81

Pascal. H.G. *et al*. 2003. Pathogenesis of split-hand/split-foot malformation. Human Molecular Genetics 12. 1. R51–R60. 10.1093/hmg/ddg090.

Pinette M, Garcia L, Wax JR, Cartin A. and Blackstone J. 2006. Familial ectrodactyly. *J Ultrasound Med*., 25:1465–7.

Prenatal diagnosis of the ectrodactyly, ectodermal dysplasia, cleft palate (EEC) syndrome.Köhler R, Sousa P, Jorge CS *J Ultrasound Med*., 1989 Jun; 8(6):337-9

Sunil K Mahavar and Ashutosh Chaturved, 2016. Split Hand / Foot Malformation Syndrome with Cerebral Degeneration. *Journal of The Association of Physicians of India (JAPI)*, Vol. 64.

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