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Case Report

Split hand/split-foot malformations: a report of four cases in a family with variable presentations

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ABSTRACT

Split hand/split-foot malformation 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. We report a family of 10 members; four of which are affected with autosomal recessive pattern of inheritance. We discuss here the clinical presentation, genetic inheritance, prenatal diagnosis and treatment for the malformation.

Keywords: Split hand, Split foot, Malformation

INTRODUCTION

Split-Hand/split-Foot Malformation (SHFM) is also known as ectrodactyly or cleft hand/foot, is rare form of congenital disorder. The word ectrodactyly is derived from Greek ektroma (abortion) and daktylos (finger). There is a median cleft in the hand and/or feet hence often called as "lobster claw". The incidence varies from 1/90000 to 1/10000 depending on genetic heterogenecity (Table 1). The condition may be unilateral or bilateral and may occur as an isolated anomaly or may be associated with other various malformation in the affected extremity (syndromic association) and systemic features. Majority of the cases live a normal life with very little effect on limb function.

Isolated variety (SHFM-1) is a type I failure of formation - longitudinal arrest with transverse terminal deficiency affecting the central digital rays of hand. In contrast to radial or ulnar deficiencies; there is no proximal deficiencies of the bones muscles, tendons, vessels and nerves and has autosomal dominant inheritance and is the only form with sensorineural hearing loss. However other varieties may be associated with other anomalies of the limb or systemic features.

Table 1: Genetic heterogenicity of SHFM.

Type	Chromosomal abnormality
SHFM1	7q21-q22
SHFM2	Xq26
SHFM3	FBXW4/DACTYLIN at 10q24
SHFM4	TP63 at 3q27
SHFM5	DLX1 and DLX 2 at 2q31

Syndromic association of SHFM

- Split-Hand-Foot Malformation Syndrome
- Ectrodactyly–ectodermal dysplasia–cleft syndrome
- Silver-Russell syndrome

- Cornelia de Lange syndrome
- Acrorenal syndrome
- Focal dermal hypoplasia
- Ectrodactyly and cleft palate syndrome
- Ectrodactyly/mandibulofacial dysostosis
- Ectrodactyly and macular dystrophy

CASE REPORT

We report a family of ten blood related members out of which four members were affected. The pedigree chart is depicted (Figure 5) which shows the autosomal recessive pattern of inheritance with variable degree of penetrance involving both male and female. No consanguineous marriage were reported. There was no significant prenatal or perinatal history. The members of the family were having variable complaints. Four cases including two males and two females were examined. One case (mother) had unilateral involvement with isolated absence of third digital ray (case-1) with a V-shaped cleft. She was able to hold the objects and able to write down, no other features were found. The son of the affected mother had bilateral involvement (case-2).

On right side he had a cleft hand with absence of central digital ray and also had bony fusion of first and second metacarpal (syndactyly) along with variable fusion of carpals, had a good grip with right hand. On left side he had only first digital ray with scaphoid and trapezium and absence of rest of the elements. The daughter of the affected mother (case-3) had affected right hand with absence of phalanges of third digit and bony fusion of first and second ray. The cleft deformity was U shaped, she had good grip. The grandson of the affected mother (case-4) had right lower limb involvement instead as cleft foot with absence of second and third digit and absence of some corresponding tarsal. He also had tibial hemimelia on same side and having difficulty in walking.

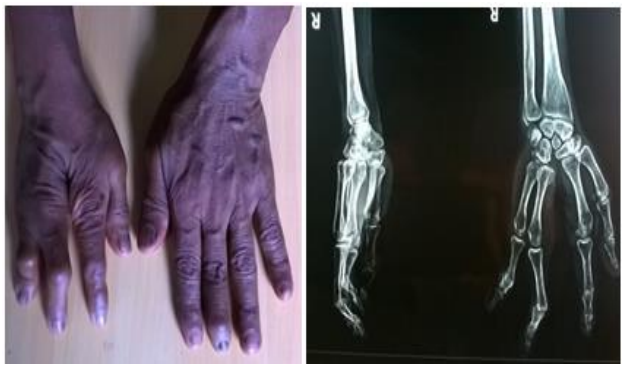

Figure 1: Unilateral involvement with isolated absence of third digital ray (case-1) with a V-shaped cleft.

Figure 2: The son of the affected mother had bilateral involvement (case-2).

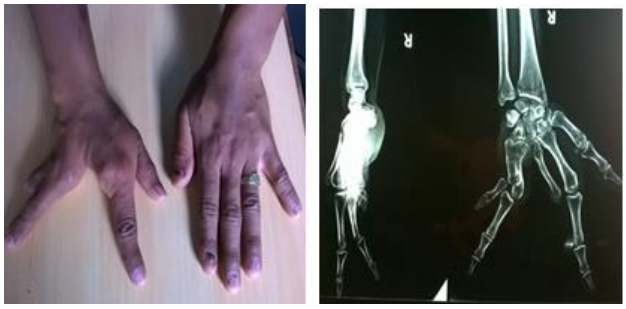

Figure 3: The daughter of the affected mother (case-3) had affected right hand with absence of phalanges of third digit and bony fusion of first and second ray.

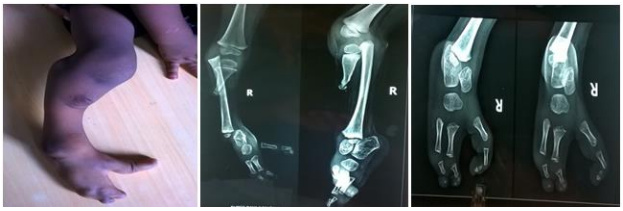

Figure 4: The grandson of the affected mother (case-4) had right lower limb involvement instead as cleft foot with absence of second and third digit and absence of some corresponding tarsal.

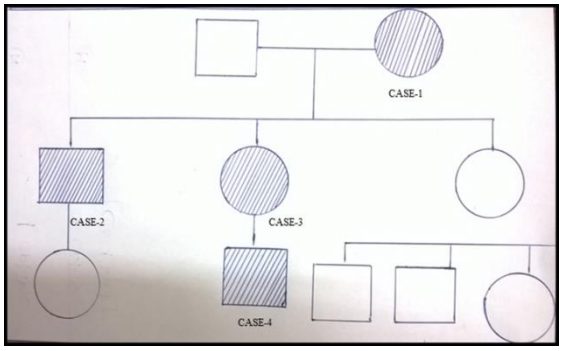

Figure 5: Pedigree chart.

DISCUSSION

Split hand foot syndrome (SHFM) is a syndrome of limb malformation which affects central rays of hand and foot. The term ectrodactyly is used instead of SHFM by some authors. The presentation of SHFM may vary from syndactyly, aplasia and/or hypoplasia of the phalanges, metacarpals and metatarsals and, median clefts of the

hands and feet. Severity may differ not only between patients, but also between limbs of a single individual. SHFM caused by many gene defect. The syndrome may be inherited as autosomal-dominant with reduced penetrance,[1] autosomal-recessive[2] and X-linked forms[3] depending upon chromosomal mutation.

Depending on clinical presentation, SHFM has two types: syndromic and non syndromic. While non-syndromic form affect the limbs only, the syndromic form, with associated with tibial aplasia, mental retardation, ectodermal and craniofacial findings, cleft lip/palate and deafness.[5] The syndromic form is variable in inheritance. The non-syndromal SHFM mostly inherited as a autosomal dominant gene with a high penetrance.[6] But in some cases of non syndromic SHFMs recessive inheritance pattern has been observed.[7-10] Non-syndromic SHFM has five types depending on the involvement of chromosome. SHFM1 has been associated with mutation of chromosome 7. Similarly SHFM2, SHFM3, SHFM4and SHFM5 has been associated with X chromosome, chromosome 10, chromosome 3 and chromosome 2 respectively. Split-hand deformity presents in two forms clinically. In the lobster claw variety the third digit is usually absent. The cone-shaped cleft that tapers divides the hand into two parts and tappers towards wrist. The deformed hand has an appearance of lobster claw. The remaining fingers or parts of fingers on each side of the cleft are often joined or webbed together. The cleft mostly involves both hand which might differ in severity and may involve feet. In the second type of split-hand deformity, there is no cleft but fifth digit is only present (monodactyly).

The diagnosis of SHFM is based on clinical examination of limbs at birth. X-rays is required to identify the skeletal anomalies by DNA analysis for. Molecular genetic testing is available only for SHFM4. Ultrasonography in antennal period is helpful for early diagnosis.[5,12] For treatment, prosthetics[4] are useful to achieve normal functioning. Reconstructive surgery for deformed limb is done when necessary. Other treatment is symptomatic and supportive. Genetic counseling of parents is helpful.

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