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CASE REPORT

ECTRODACTYLY IN AN ADULT: A RARE CASE REPORT

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ABSTRACT

Ectrodactyly is one of the rarest congenital malformation of the limb also termed as split hand/split-foot malformation (SHFM).Majority of cases are sporadic, as is the present case, but familial types also exist with autosomal dominant inheritance. The causative factor is defect of central elements of the autopod (Hand/Foot). There is deep median cleft of the hand and/or foot. It is also known as "lobster-claw" deformity because of the cone-shaped clefts of the hands and/or feet. A rare case of severe bilateral SHFM in 46 yrs old male patient, who had adjusted with the normal pursuits of his routine life without much disability, is being presented. Both the feet were divided into two parts by a cone shaped cleft proceeding proximally and resembling a "Lobster claw"(bidactyly). Though in both the hands, the claw deformity was not seen (monodactyly) with aplasia of the rest of the phalanges and metacarpals. The present case represents non-syndromic type of SHFM as other as no known associated anomaly was seen in this patient. In the absence of any functional disability, as in the present case, reconstructive procedures were not advocated.

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INTRODUCTION

Ectrodactyly is a rare structural abnormality of the limbs. The basic embryological defect is failure to maintain normal apical ectodermal ridge thereby causing failure to differentiate the autopod. It is congenital absence of central rays of limbs (Durowaye *et al*., 2011). The term ectrodactyly has its origin from the Greek word Ektroma (Abortion) and Dactylor (Finger) (Pinette *et al*., 2006). The condition was first recorded in 1770 in Guinea Indians (Durowaye *et al*., 2011). Crab-claw deformity was described by Von Walter in 1829 and the term "Lobster-claw" was used by Cruvelheir in 1842 (Duijf, 2003).The condition is common in central African communities (Viljoen *et al*., 1985). Syndromic ectrodactyly is rare and non-syndromic is even rarer condition. Other limb abnormalities like monodactyly, syndactyly, aplasia/hypoplasia of the phalanges, metacarpals and metatarsals are common in ectrodactyly, whereas in syndromic ectrdactyly, associated anomalies may include hearing loss, intellectual deficiencies and ectodermal and craniofacial abnormalities.

Case Report

46 years old male patient presented in the OPD with severe SHFM since birth.

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The patient attended the hospital for corns and callosities on his deformed feet and hands and was not concerned for his congenital defects. Physical examination revealed absence of metacarpals and phalanges from both the hands except bilateral monodactyly and the feet showed typical "Crab-claw" or "Lobster-claw" deformity with cone shaped cleft tapering proximally in both the feet (Fig.1). There was no other associated medical condition like craniofacial deformity or hearing loss. The patient exhibited normal intelligence and was well adjusted to his defects except that presently he developed callosities on the bony prominences of the feet for which he was seeking consultation (Fig.1). His parents and siblings were having no family history of limb deformity. Reconstructive surgery / Remedial surgery was neither recommended as the patient did not have any serious functional disability nor the patient sought the advice for the same. The patient was referred to dermatology for corns/ callosities and was advised X-Ray of the hands and feet, but the patient was lost in the follow up.

DISCUSSION

Ectrodactyly is rare genetic disorder having a prevalence of 1:10,000-1:90,000 live births, with no predilection for gender (Durowaye *et al*., 2011; Kohler *et al*.,1989). *VaDoma* tribe of north Zimbabwe and the Eastern shona kalanga of the Kalahari

desert are famous for their inherited ectrodactyly in some of their families, resulting in the tribe being known as the "two toed or "ostrich footed" Tribe (Farrell.,1984). These are defects of central elements of the autopods (hand/foot) leading to split-hand/split-foot malformation. The present case has four limb involvement. There was monodactyly in the hands and bidactyly in feet leading to "Lobster-claw" deformity of the feet. This is a sporadic case but there exists familial forms also which are predominantly autosomal dominant inheritance. Some cases of SHFM may be due to chromosomal defects like deletions and duplications. Autosomal-recessive and X-linked types do exist but are more rare (Duijf, 2003). Genetically SHFM is heterogeneous with mutation identified at different multiple loci (Nirmala *et al*., 2015). Syndromic SHFM, which is associated with other defects, can present when two or multiple genes are affected by chromosomal rearrangement. The well known and commonest SHFM is Ectrodactyly. Ectodermal-dysplasia-cleft (EEC) syndrome, which is associated with p63 mutation (Barsky *et al*., 1964), is characterized by cleft palate/lip and this patient was having none of these anomalies. The severity of the defect may vary from individual to individual patient and between different limbs of the same patient.

Figure 1.

Failure to maintain median “Apical Ectodermal ridge” (AER) signaling is the pathognomonic mechanism for this condition. This AER signaling may be either due to increased cell death or due to reduced cell proliferation. More severe limb malformation do not occur in ectrodactyly as AER activity defect does not take place in the earliest stages of limb development (Duijf, 2003). Ectrodactyly is known to be associated with many other syndromes like Adams-oliver syndrome, Delange syndrome, Goltz syndrome, Limb-memory syndrome, Carpenter syndrome and Miller syndrome (Durowaye *et al*., 2011), but the presenting patient was not having any associated defect, disability or any other congenital abnormality such as scalp defect, syndactyly, polydactyly, genitourinary anomalies or problems like nasolacrimal ductectasia and buphthalmos. A radiographic classification has divided cleft feet in six grades (blauth *et al*., 1990) and the present case seems to be belonging to grade 5th of this classification which envisages "lobster-claw" foot, complete absence of 2nd, 3rd and 4th rays. Syndromic SHFM is associated with orofacial clefting and neurosensory hearing loss (>35%), ectodermal and craniofacial defects (>35%) and intellectual disability (33%) (Lango *et al*., 2014). The present case was having none of the above defects and seems to be a non-syndromic where only isolated limbs involvement is present. As the patient was not having any functional disability, has adapted himself well to his defects and is living normal life with modest dysfunction of hands only, he was neither recommended any surgical reconstructive procedures nor he sought any remedial surgery for his defects.

In such patients only functional training and physiotherapy should be advised as most of these patients accommodate well and live normal life with only modest functional impairment of the hands. The literature is also lacking in describing suitable techniques for their treatment. Prosthetics have also been used. If a child presents with such anomalies, associated orofacial defects must be looked for. Proper counselling for the possibility of occurrence of such defects in the future siblings, and antenatal diagnosis via ultrasonography should be offered.

REFERENCES

Barsky, A.J. Cleft hand: Classification incidence and treatment. Review of the literature and report of nineteen cases. *J Bone Surg Am*, 1964;46:1707-20.

Blauth, W., Borisch, N.C. 1990. Cleft feet. Proposals for a new classification based on roentgenographic morphology. *Clin Orthop Relat Res*., 258:41-8.

Duijf, P.H., van Bockoven, H., Brunner, H.G. 2003. Pathogenesis of split hand/split foot malformation. *Hum Mol Genet,* 12:R51-60.

Durowaye, M., Adeboye, M., Yahaya-Kongoila, S., Adaje, A., Adesiyun, O., Ernest, S.K., *et al*. 2011. Familial ectrodactyly syndrome in a Nigerian child. A case report. *Oman Med J*., 26:275-8.

Farrell HB (1984)"The two toed Wadoma -familial ectrodactly in Zimbabwe" *S.Afr.Med j*., 65(13):531-3 PMID 6710256.

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

Lango Allen, H., Casewell, R., Xie, W., Xu, X. Wragg, C., Turnpenny, P.D., *et al*. 2014. Next generation sequencing of chromosomal rearangements in patients with split-hand/split-foot malformation provides evidence for DYNC111 exonic enhancers of DLX5/6 expression in humans. *J Med Genet.,* 51:264-7.

Nirmala, S.V.S.G., Sandeep chilamakins, Chaitanya Pavuluri, Sivakumar Nuvvula Sindhuri veluru. Ectrodactyly, A rare anomaly of limbs, Journal of Dr. NTR university of health sciences 2015;4:53-55.

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

Viljoen D, Farrell HM, Brossy JJ, McArthur M, Maheswaran M, Beighton P. Ectrodactyly in central Africa. *S Afr Med J* 1985;68:655-8.

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