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CASE REPORT

Ectrodactyly and Prenatal Diagnosis

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Introduction

Ectrodactyly is an autosomal dominant ectodermal dysplasia presenting as bilateral congenital malformed hands and feet [1]. It affects about 1 in 90,000 births with males and females equally as likely to be affected. It is characterized by transverse terminal aphalangia or partial to total absence of the distal segments of fingers. It may involve one or more digits or the full hand and even part of the upper arm. More severe manifestations are hemimelia or amelia. All these abnormalities are considered to represent various degrees of severity of the same anomaly and may be due to an intrauterine vascular occlusion or insufficiency [2]. These different forms are connected with a different genetic mutation. Type I, the most frequent form has been found to be a mutation on chromosome 7 in a region that contains two homeobox genes, DLX5 and DLX6. Usually this is characterized as the split hand/foot deformity due to the absence of the third digit, with clefting into the proximal portion of the hand or foot and syndactyly of remaining digits on each side of the cleft. The hand resembles a lobster claw [3]. The association of ectrodactyly with cleft lip and palate was originally described by Cockayne [4]. It was known as Ectrodactyly-Ectodermal Dysplasia-Cleft lip/palate syndrome (EEC syndrome) [5].

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Case History

A 28-year-old woman with features suggestive of ectrodactyly was referred to our center for genetic counseling in the Department of Fetomaternal Medicine at Centre for Infertility Management and Assisted Reproduction (CIMAR), Edappal Hospitals, Edappal, Malappuram, Kerala. Both her hands were showing lengthening and broadening of the digits. There was a medial cleft in the metacarpals, dividing the hand into two portions. Syndactyly of the remaining fingers was seen. The growth of the digits was more as compared to other body parts. The nails of the affected fingers were maldeveloped (Fig. 1). Her legs also showed ectrodactyly with medial cleft in the metatarsals and syndactyly of the toe fingers (Fig. 2). She could perform all her routine activities and she was a postgraduate schoolteacher. Systemic examination of the patient did not reveal any other anomaly. Abdominal ultrasonography did not show any abnormality. X-ray of both hands and feet confirmed ectrodactyly. Hair and teeth were normal and there were no other congenital malformations. There was no history of consanguinity or any other relevant family history. She had previously undergone two medical terminations of pregnancies as ultrasonographical examination of the fetuses showed splitting of hand and fused fingers suggesting ectrodactyly in the fetus. She was gravida 3 with gestational age corresponding to 11 weeks when she visited our clinic. The fetus was continuously monitored by Voluson 730 pro. The level three targeted scan at 14 weeks showed that the fetus was having

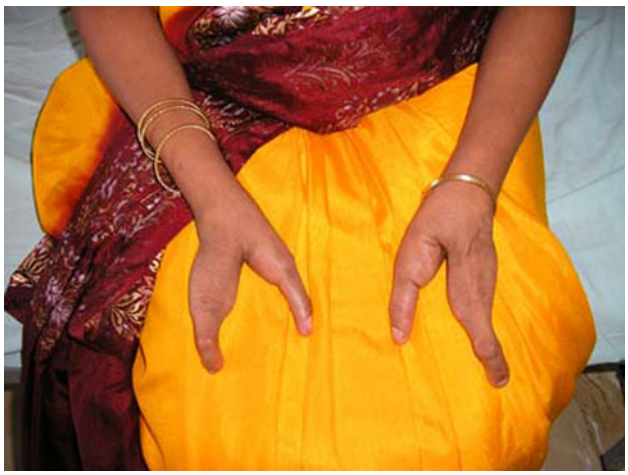

Fig. 1 Showing bilateral ectrodactyly (lobster claw) of the hand. Typical medial cleft with syndactyly of the remaining fingers

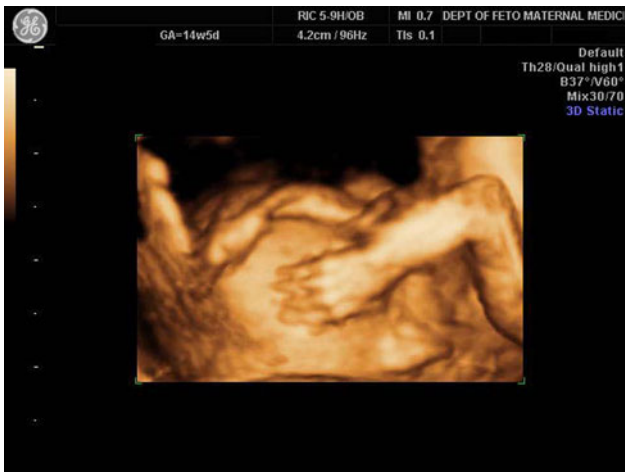

Fig. 3 3D image of the hand kept by the side of fetal head showing normal fingers

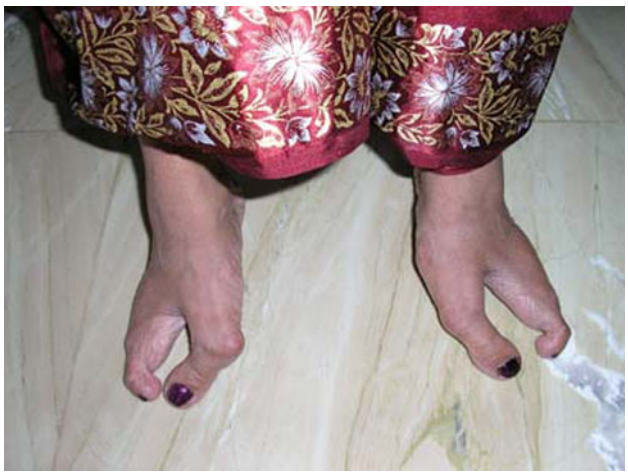

Fig. 2 The same patient as in Fig. 1. Showing bilateral ectrodactyly (lobster claw) of the foot

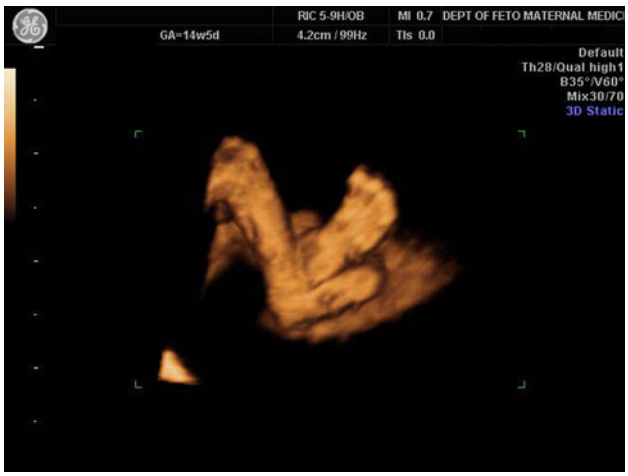

Fig. 4 3D picture of sole of feet of fetus

all the metacarpals and phalanges normal (Figs. 3, 4). The metatarsals and toe phalanges also appeared normal. Ultrasonography was repeated at 19 weeks and the fetus was found to be not affected. She continued her pregnancy till term by continuous ultrasound monitoring and gave birth to a normal male child.

Discussion

Ectrodactyly is a rare autosomal dominant ectodermal dysplasia. It sometimes may be associated with other ectodermal defects. The most common clinical manifestations of EEC syndrome are ectodermal dysplasia, ectrodactyly, cleft lip/palate and tear duct anomalies. The expression of this may be quite variable with reduced penetrance also. In a review of 230 published cases by Roelfsema et al. [6], ectrodactyly was found in 84%, ectodermal dysplasia in 77%, clefting in 68% and anomalies of lacrimal ducts in 59%. Urogenital defects were reported in 52%. Isolated cases were more severely affected than familial cases [6]. In the present case she only showed lobster claw with no other abnormality. The patient was offered genetic counseling and the mode of inheritance explained. Since ectrodactyly is an autosomal dominant disorder, there are 50% chances of recurrence for the future pregnancies. Genetic studies using mutation analysis was explained to the patient but the patient opted out, as it was very expensive and not available in India. In the present case the fetus was continuously monitored by ultrasonography and she delivered a term normal male child. Thus, for certain ectodermal dysplasias like ectrodactyly monitoring by ultrasonography in high risk pregnancy will be

more appropriate rather than expensive gene testing which at times are not available to the patients.

References

1. Kalla G, Garg A. Ectrodactyly. Indian J Dermatol Venereol Leprol. 2002;68:152–3.
2. Deborah K. The dysostoses. In: Rimoin DL, Connor JM, Pyeritz RE and Korf BR, editors. Principles and practice of medical genetics. 4th ed. London: Churchill Livingstone. 2002; pp. 4170–4171.
3. Jung KR, Jeong C, Jong SC. Ectrodactyly. Korean J Radiol. 2003;4(4):243–51.
4. Cockayne EA. Cleft palate-lip, hair lip, docrocystitis and cleft hand and foot. Biometrika. 1936;26:60–3.
5. Pries C, Mittleman D, Miller M, et al. The EEC syndrome. Am J Dis Child. 1974;127:840–4.
6. Roelfsema NM, Cobben JM. The EEC syndrome: a literature study. Clin Dysmorphol. 1996;2:115–27.