Hamdan Medical Journal 2012; 5:375–378 (http://dx.doi.org/10.7707/hmj.v5i3.151)

CASE REPORT

Antenatal diagnosis of ectrodactyly with fibular aplasia – a case report

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Abstract

Introduction. Split-hand or split-foot malformation associated with a fibula aplasia is a rare limb malformation; it results from failure of formation of parts of hands, feet or both due to a variable deficiency of central rays of the autopad. This anomaly can be isolated or associated with genetic or non-genetic syndromes. Routine examination of fetal hands and feet during second-trimester ultrasonography should make the detection of hand and foot malformations more frequent.
Method. We report a case of familial non-syndromic form of ectrodactyly with aplasia of fibula because of its rarity, detected by ultrasonography. The anomaly was confirmed after birth and conservative orthopaedic management with further surgical reparation later was decided.
Conclusion. Although it is rarely seen, limbs defects may be detected by detailed prenatal ultrasonography; however, the impact of these anomalies is assessed after birth, to choose the best option for the correction of the defect.

Introduction

Split-hand or split-foot malformation (SHFM), also known as ectrodactyly, is a congenital limb malformation characterized by a deep median cleft of the hand and/or foot due to the absence of the central rays, the third digit, giving a characteristic appearance of 'lobster claw' hands. SHFM associated with fibular aplasia is considered to be a rare disorder and is thought to be inherited in an autosomal dominant fashion with reduced penetrance and variable expression.[1–3]

Split-hand or split-foot malformation 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.[4,5]

Split-hand or split-foot malformation occurs in 1 per 90 000 live births and was first described in 1936[6,7] with no sex predilection. Its association with other bone anomalies is rare.

Two modes of expression exist for SHFM: an isolated non-syndromic form limited to the limbs and syndromic expression involving other anomalies. Most cases of isolated ectrodactyly do not require surgical intervention and most individuals with isolated ectrodactyly live normal lives with modest functional impairment of the hands.[4]

Ectrodactyly can be prenatally diagnosed using ultrasound in the first or second trimester using two- or three-dimensional (3D) ultrasonography. Aplasia of other bones including the tibia and ulna may also occur.[4]

We report a case of a patient with previous family history of limb defects who was pregnant with a fetus which had ectrodactyly involving both hands and feet with other bone and limb abnormalities.

Case report

A 28-year-old healthy pregnant woman, Gravida 4 Para 3 (G3P2), non-consanguinity, was referred to our hospital after the detection of limb anomalies in the fetus at 29 weeks.

The patient's medical history revealed several cases of limb defects in her husband's family. Previous obstetric history showed that in 2007 she had delivered a healthy newborn, followed in 2009 by another delivery of a child with an isolated limb defect: oligodactyly (right hand with three fingers, left hand with four fingers and right foot with three toes). The couple had a genetic consultation. The chromosomal study results were normal and no mutation was found.

During her current pregnancy, the patient was followed in a private structure. She was referred to the hospital at 29 weeks of gestation. A detailed ultrasonography was carried out, revealing a singleton fetus consistent with gestational age. Both upper hands had deformities: oligodactyly with syndactyly suggestive of lobster claw, presence of both radius and ulna. Both tibias appeared shorter, with an absence of fibula and bilateral talipes (Figure 1). Amniotic fluid was increased.

The findings were explained to the patient in detail; however, as she was referred to us late in her pregnancy, and in view of her previous history, it was decided she would continue the pregnancy; the chromosomal study had not yet been done. She was given anti-D (Rho) immunoglobulin as she was rhesus negative.

During her follow-up, repeated ultrasonography was carried out. At ≥ 34 weeks, the findings were consistent with a fetus corresponding to gestational age with severe oligohydramnios (1 pocket = 26 mm) and increased umbilical artery Doppler, but normal middle cerebral artery Doppler. She was induced at 36 + 1 weeks with 3-mg tablet of Prostin E_2 (Pfizer Inc.).

She delivered a female newborn, Apgar score (a measure of newborn well-being) = 9 (at 1 minute); 10 (at 5 minutes), weight = 2430 g. Neonatal examination confirmed the anomalies, with the right hand having three fingers, the left hand having four fingers with syndactyly, including thumbs, and short and deformed legs and club feet, with two toes on each foot (Figure 2).

X-ray skeletal survey confirmed that there were three digits on the right hand and four on the left with cleft noted between the second and third metacarpals.

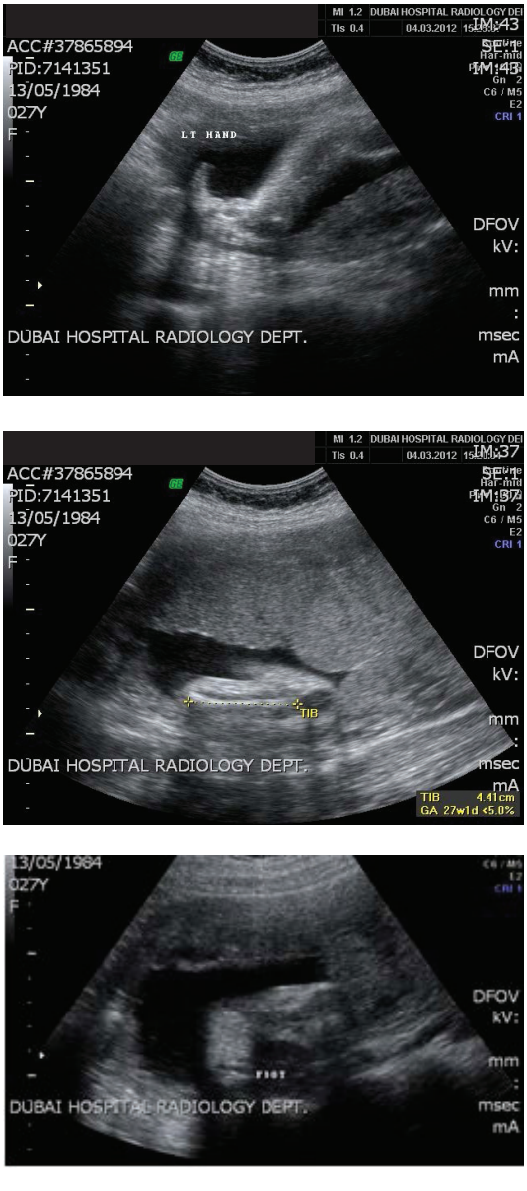

FIGURE 1 Showing hands deformities, oligodactyly and absence of fibula.

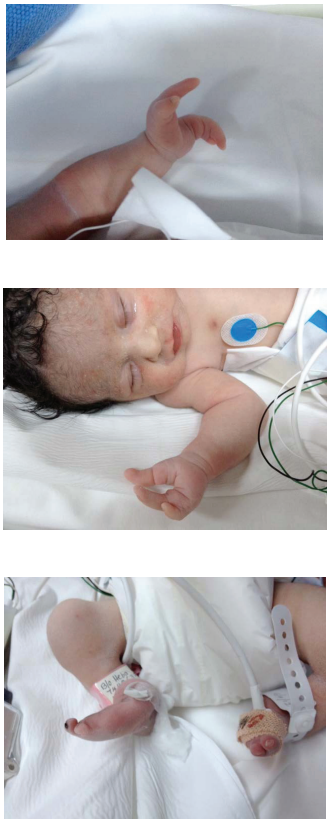

FIGURE 2 Limbs defects at birth.

The absence of fibula was also noted with deviation deformity of the lower end of the left tibia (Figure 3).

The newborn was discharged on the second day after birth in good condition with an appointment to attend the orthopaedic paediatric clinic.

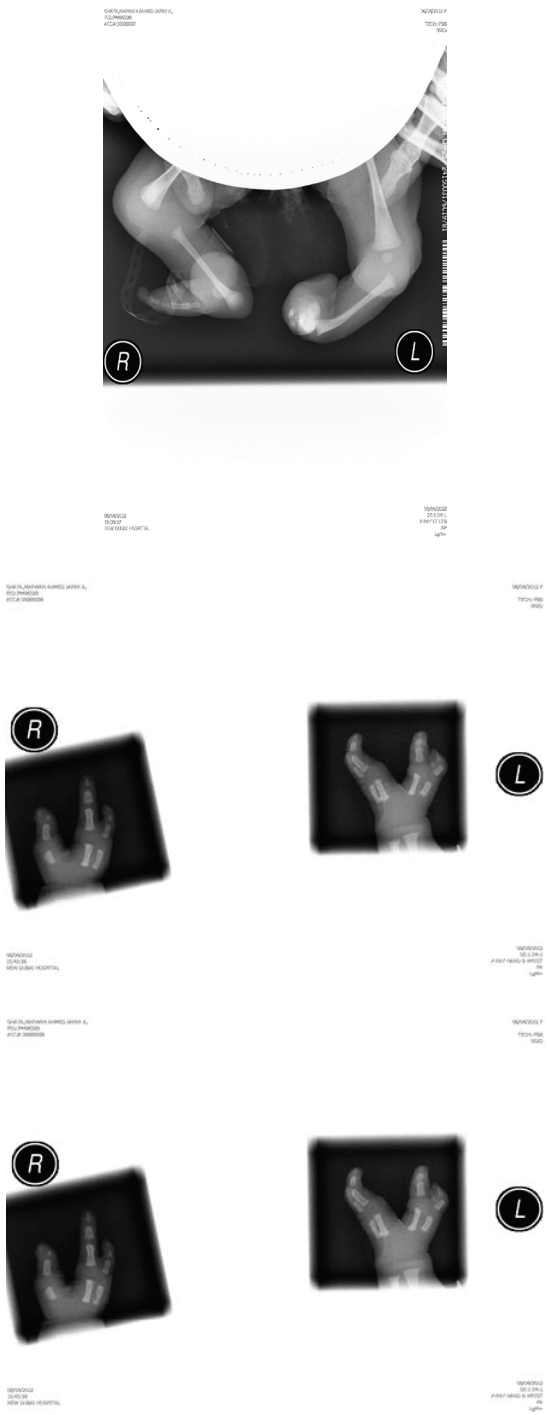

FIGURE 3 X-ray findings.

A cytogenic study showed a normal female (46 XX).

Discussion

Split or cleft hand results from a longitudinal deficiency of the central digits. The main pathogenic mechanism is probably a failure of the median apical ectodermal ridge in the developing limb bud.[5] Examination of the extremities should be carried out during a detailed second trimester ultrasonography. It may help to diagnose many syndromes. Anomalies of the hands and feet may involve all limbs or a single distal ray. The assessment of extremities is important as it allows time for counselling of the parents and the institution of proper care after birth.

A 3-dimensional sonographic image of the anomaly confirms the diagnosis. Most cases of isolated ectrodactyly do not require surgical intervention, and most individuals with isolated ectrodactyly live normal lives with modest functional impairment of the hands.[3]

Currently, if needed, there are several treatments that can normalize the appearance of the hands, but the anomalous hands work exactly the same way as regular-shaped hands. The prognosis for most people with ectrodactyly syndrome is very good. Some people with ectrodactyly use prosthetic hands, minimizing the chances that others will notice their condition. The condition can also be treated surgically. Early physical and occupational therapy can help those with ectrodactyly to adapt to learn, write, pick things up and become fully functional. Genetic findings could have major implications on the clinical diagnosis and treatment of not only ectrodactyly, but also many other related syndromes.

Conclusion

Ectrodactyly is a rarely seen limb defect which may be detected by detailed prenatal ultrasonography. This case would give us a better understanding of this genetic limb defect and enable us to provide appropriate and timely care to these infants after birth. The real impact of these anomalies is usually assessed after birth, helping to choose the best option for the defect's correction.

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