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Case Report

MULTIPLE CONGENITAL ANOMALIES WITH ABSENCE OF DISTAL LOWER LIMB DEFECTS

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ABSTRACT
In early 1960s, high rates of limb reduction were associated with expectant mothers taking the drug thalidomide. As a result of this, congenital anomaly surveillance systems were established to try and prevent further such episodes. Consequent to improved technologies limb reductions were found to be associated with chorionic villus sampling in early pregnancies. The present case is a still born foetus of a 28 year old multiparous mother with G II P I Congenital anomalies in the thorax revealed hypoplastic lungs and cystic kidneys. Other causes of focal limb loss include the amniotic band syndrome, thalidomide exposure and caudal regression syndrome. Hence, the present case is being reported.

Key Words: *Distal Limb, Club Foot, Hypoplasia, Cystic Kidney*

INTRODUCTION
A distruption of limb vasculature after theperiod of embryonic development can result instructural anomalies. This disruption can be due totangled strands of amnion that encircle embryonicstructures, placental emboli or death of amonozygotic twin *(Torpin 1965, Van Allen 1982, Hoyme 1981)*. In 1991 a report from Oxford (Firth et al, (1991)first suggested the link between chorionic villus sampling (CVS) and severe limb deficiencies. Further studies confirmed this association with defects where the distal part of one or more limbs or digits is missing across its entire width (transverse terminal deficiency) The earlier in pregnancy that CVS took place the more extensive the defect (Firth et al, (1991) .Very few reported cases on absence of foot are available in literature. Hence, the present case is being reported.

Case
The present case is a still born foetus of a 28 year old multiparous mother with G II P I. There is no history of consanguineous marriage. There is no abnormal drug or alcohol intake. Ultrasonography revealed a male live foetus with limb reduction defects and renal defects. Pregnancy was terminated during 28 weeks of gestation. At necropsy, the foetal weight recorded 2,000 grams. Macroscopic observation revealed polydactyly of hands on both sides with lower limb reduction and achieria of foot on right side and club foot on the left side (fig.1).
Postmortem radiography revealed normal vertebral bodies that appeared square, with extremely short ribs. There is limb reduction on the right side with shorter femur comparatively; lower end of the tibia pointed and absence of entire foot bones (fig. 2). On the left side, the remaining appendicular and axial bones appeared normal. Congenital anomalies in the thorax revealed hypoplastic lungs (fig.3) and heart with patent foramen oval. Abdominal visceral anomalies revealed cystic kidneys (fig.4). Chromosomal analysis revealed 46, XY.

DISCUSSION
The prevalence of limb reduction deformities is about 1 per 20 000 births. In general, limb deficiency of the upper extremity is an isolated anomaly, whereas congenital amputation of the leg or bilateral amputations or reductions of all limbs are usually part of a genetic syndrome .Isolated amputation of an extremity can be due to amniotic band syndrome, exposure to a teratogen or a vascular accident (Gianluigi Pilu et al. 2002). Limb reduction defects (especially those involving the radius or thumb) have

been associated with chromosomal anomalies. It has been calculated that, when limb reduction defects are present, the chance of having trisomy 18 is 1 in 17(Twining P (1995).

Figure 1. Absence of foot on right side and club foot on the left side

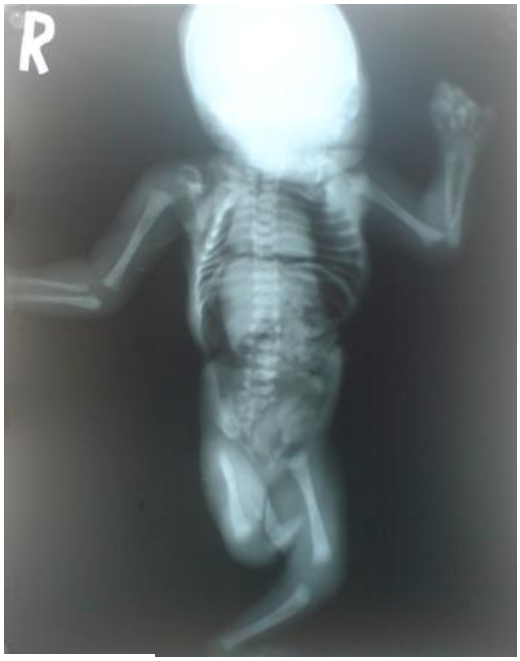

Figure 2. Tibia pointed and absence of entire foot bones

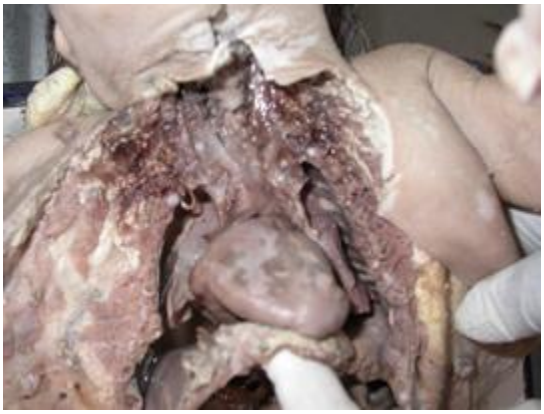

Figure 3. Hypoplastic lungs

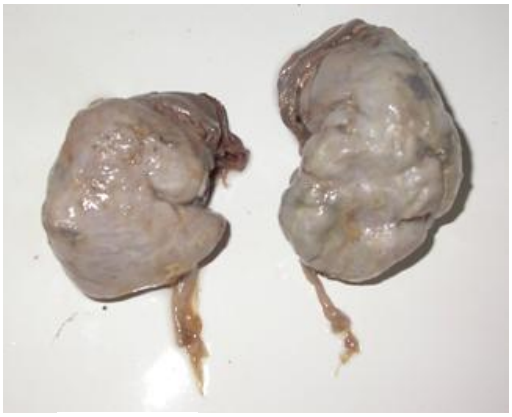

Figure 4. Cystic kidneys

Absence of left hand was detected at 18 weeks of pregnancy of a 32-year-old woman in a case reported by Andrey Volkov (2007). Cuillier F et al (2004) in a case report found unilateral agenesis of the fibula, hypoplastic tibia and unilateral club-foot with three metatarsal bones .

Absence of foot is very rare and isolated limb reduction deformities, such as complete absence of extremities, absence of the hand or foot, phocomelia (seal limb) or aplasia–hypoplasia of the radius or ulna, are often inherited as part of a genetic syndrome. Other causes of focal limb loss include the amniotic band syndrome, thalidomide exposure and caudal regression syndrome. Absence of an extremity or a segment of an extremity is referred to as 'limb deficiency' or 'congenital amputation'(Gianluigi Pilu et al. 2002).

Severe shortening of the limbs, absence of the right fibula was observed by (Maruotti et al. 2009) in a case report of prenatal diagnosis of Ulbright Holde's syndrome. In addition, radiohumeral fusion, a Potter-like face with a beaked nose, a high palate and small mouth, a short neck, hypoplastic lungs, and cerebellar hypoplasia were also recorded (Maruotti et al. 2009). Absence of hand or foot with multiple congenital anomalies has a fatal outcome due to severe respiratory insufficiency. Though, it is not inherited, the primary cause may have a genetic reason like autosomal recessive polycystic kidney.

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