

Duplicate Tail Like Right Lower Limb: A Rare Congenital Malformation

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ABSTRACT

Introduction: Polymelia (supernumerary limbs) or congenital duplication of limb is an extremely rare condition in humans, with only few cases reported in the literature.

Case Report: This is a case of newborn female born with a tail-like extra right leg, which unfortunately was not detected during the routine antenatal ultrasonography.

Conclusion: Early diagnosis can facilitate the management planning and psychological preparation of the parents. Surgical resection of the accessory limb at an early age is recommended in patients with polymelia.

Keywords

CT (computed tomography), AER (Apical Ectodermal Ridge), ZPA (Zone of proliferation activity), USG (ultrasound), Polymelia.

Introduction

Congenital duplication of the lower limb is a rare condition that can be complete or incomplete. Polymelia is defined as the presence of accessory limbs attached to various body parts and may be classified as cephalomelia (if attached to the head), notomelia (to the backbone), thoracomelia (to the thorax), and pyromelia (to the pelvis).

Case Report

This is very rare case of newborn with supernumerary tail-like right leg.

Family history revealed that the mother is a 20-year-old primigravida of Syrian nationality married to 31-year-old husband with no history of consanguinity. No history of hereditary diseases or major Dysmorphology in the family. No history of accidental or work-related exposure to genotoxic agents.

She had regular antenatal care and was found to have low Hb and vitamin B 12, which was corrected. She received methyl dopa Low 250 mg twice daily for hypertension detected during the last month of her pregnancy. Although she had five antenatal USG examinations (2 during the second trimester and 3 during the third trimester of pregnancy), the extra limb was not detected. At 39+3 weeks labor was induced with prostaglandins due to Oligohydramnios detected in the last USG. A baby girl was born weighing 2990 grams with APGAR scores of 9 and 10 at one and five minutes, respectively. The extra limb-like structure was noted at birth with no other external anomalies elsewhere (Figure 1A+B). The limb was attached inferomedial to the right gluteal area, lateral to the anal orifice, with a thick fold of skin surrounding the origin. It hung loosely with no spontaneous movements and extending up to the level of mid-leg. The proximal portion was swollen with localized erythema (Figure 2). Distally, the foot had a deep first interdigital cleft, giving the appearance of two separate (Figure 3). The baby was transferred to NICU for further evaluation and observation. She remained stable with normal observations. Complete blood counts, renal and liver function tests were normal. The routine metabolic screening was also found to be normal.

Skeletal survey showed the presence of femur, metatarsal and phalangeal bones in the accessory limb, along with tibial and fibular malformations (Figure 4). Hip bones and bones in the other lower limbs appeared normal. Neurosonogram and USG abdomen were unremarkable except for a suspicious small hemangioma in the right lobe of the liver (Figure 5).

CT scan (Figure 6) images showed poorly formed accessory acetabular and ilial bones connected to a well-formed femur. Distal to the knee, bones were poorly formed. The tarsal bones were not seen but metatarsal and phalangeal bones were noted. Small muscular tissue was noted adjacent to the acetabulum. Adipose tissue was seen adjacent to the femur and knee joint. A small cystic area (1.5x1.3cm) was noted posterior to the rectum.

Molecular study of whole-genome array yielded a normal hybridization pattern. The baby remained clinically stable and was discharged home on the fourth day with follow up in the pediatric orthopedics department.



Picture 4 Skeletal survey.



Figure 1A and 1B: Extra limb-like structure AP; B: extra limb-like structure LATERAL



Picture 5 hemangioma at liver



Figure 2: Proximal portion was swollen & erythema



Figure 3: Two separate feet



Picture 6: CT scan.

Discussion

Isolated limb duplication is a rare congenital condition with only few cases documented.

Limb development involves a very large number of genes [1]. One gene widely associated with the development of supernumerary limbs is the mouse mutant disorganization Ds gene (OMIM:223200) [2,3] which is a semi dominant gene with variable penetrance in heterozygotes and lethal in homozygotes. In 67% of heterozygotes, multiple defects are observed while the rest have a single defect in which polydactyly is predominant [4].

Human limbs start developing from the fourth week after fertilization and follow proximal to distal and dorsal to the ventral pattern [5]. Each limb bud consists of a mass of mesenchyme covered by a layer of ectoderm. The limb growth is promoted from the apical ectodermal ridge (AER) which is a condensation of the ectodermal cells at the tip of the developing limb bud. The mesodermal cells subjacent to the AER, referred to as the zone of proliferation activity (ZPA), stimulate the AER formation [6]. The endogenous Sonic hedgehog signal in the ZPA and fibrous growth factor 4 in the AER stimulate and maintain mutually [7,8].

After reviewing many literatures, several etiological theories of limb duplication have been suggested.

Hanley suggested that bifurcation or splitting of the limb bud may result in limb duplication [9]. McCredie suggested that with thalidomide exposure, the embryo may suffer damage to the neural crest or peripheral nerves, which leads to dysmelia and associated visceral defects [10].

Griffet noted the association of congenital anomalies of the limbs and the kidney (acro-renal). This may be explained by the fact that at the 4th week of gestation, the lower limb bud and mesonephric structures (from which the ureteric buds give rise to the renal system) are in close locations [11].

The possible etiological factors could be genetic (trans-gene, chromosomes) or environmental (infectious agent, toxin). Over 80% of children born to mothers who took thalidomide had limb defects. Abnormalities like polydactyly, polydactyly, and other congenital defects have been associated with antenatal exposure to progesterone preparations.

Our patient has characteristics very similar to a previous case reported by Mekonnen et al. [12].

Conclusion

This rare presentation of tail like duplication of the lower limb can happen due to many factors and should be detected by antenatal ultrasonography. Early diagnosis can facilitate the management, planning and psychological preparation of the parents.

Consent

We obtained written informed consent from the patient's parents to publish this case report and any accompanying images. A copy of written consent is available for review by the editor in chief of this journal.

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