

## **Amelia with Anorectal and External Genital Atresia**

A term neonate was delivered vaginally with weight 2980 g and Apgar score of 4 at 1 min and 8 at 5 min. Examination revealed scoliosis (thoraco-lumbar) to right, only one (right) lower limb, no cloacal (anal, urinary or genital) opening and syndactyly of right 3rd and 4th fingers (*Fig. 1*). Radiology showed thoraco-lumbar scoliosis to right and hypoplastic sacrum, crowding of ribs of left side with bifurcation of 8th rib, hypoplastic left iliac bone with no hip joint and lower limb. Abdominal ultrasound showed normal organs except non visualization of left kidney, rudimentary urinary bladder, rectum and lower colon. Echocardiography was normal. The neonate died at 5 days of life.

Non syndromic limb defects with a major malformation has a prevalence of 12.9 per 100,000 population. Amelia has a reported incidence of 1.5 per 100,000 newborn infants. Specific limb defects occur with distinct set of malformations. Preaxial limb defects occurred frequently with microtia, esophageal atresia, anorectal or cloacal atresia, heart defects, unilateral kidney dysgenesis and some axial skeleton defects. Post axial defects may present with hypospadias, transverse defects, craniofacial defects, micrognathia and ring constrictions etc. Amelia occurred more frequently with



*Fig. 1. Baby with absent left lower limb and absence of anal and genital orifices.*

anorectal atresia, omphalocele, severe genital defects, unilateral kidney dysgenesis, gastroschisis and ring constrictions.

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