

## Radiological Quiz – Obstetric

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Ind J Radiol Imag 2006 16:1:145-146

**T**wenty one year old female (G2P0A1) presented with PIH and hydramnios at 35 weeks gestation. Ultrasound revealed single live fetus with cephalic presentation BPD 87mm (35weeks maturity) narrow thoracic cage, dilated bowel loops and a hypoechoic mass (4x3.8x4.5cm) posterior to urinary bladder. Both kidneys

were hydronephrotic and marked shortening of limb bones was seen (FI 19mm). There was no evidence of bowing of femora. The patient delivered a still born female baby-weight 2.8kg CRI.-39.9cm, head circumference 35cm. Chest circumference 27cm. Given below are the antenatal USG images and postnatal infantogram.



Fig 1



Fig 2



Fig 3

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Received on 28 April 2005; Accepted 10 December 2005

## Radiological Diagnosis

### SHORT RIB POLYDACTYLY SYNDROME (TYPE I) - PRENATAL DIAGNOSIS

SRP Syndrome type 1 is one of the rare varieties of lethal short limb dwarfism with autosomal recessive inheritance. The syndrome characteristically shows severe shortening of long bones with ragged ends and metaphyseal spurs projecting laterally short horizontally placed ribs, hypoplasia of iliac bones with flattened acetabulae, postaxial polydactyly and normal skull spine. Multiple internal abnormalities like hypoplastic cystic kidneys, congenital heart disease, anorectal abnormalities and hypoplastic lungs have also been reported (1,2,3). The present case SRP subtype 1 (Saldino Noonan) was detected in the 35th weeks of gestation showing hydramnios, narrow thorax, micromelia polydactyly bilateral hydronephrotic kidneys and a hypoechoic mass posterior to urinary bladder (Fig 1,2,3). Postmortem pathologic examination of the stillborn baby confirmed these findings and also revealed cloacal abnormality with a blind cystic vagina.

Short rib-polydactyly syndrome (SRP) comprise a group of rare, lethal skeletal dysplasias with autosomal recessive inheritance characterized by short ribs and limbs, polydactyly, hypoplastic thorax and visceral anomalies (hypoplastic cystic kidneys, congenital heart disease, anorectal abnormalities and hypoplastic lungs).

Four types of Short Rib Polydactyly syndrome (SRP) have been reported : Type 1 Saldino Noonan. Type II Majewski, type III Warrenoff, and type IV Beemer-Langer which can be differentiated on the basis of radiological examination (1). SRP type 1 shows severe micromelia with ragged ends and metaphyseal spurs projecting laterally (2): Majewski shows shortened long bones with smooth rounded metaphysis and a classical oval configuration of tibia with normal skull and spine and pelvis. More common association of cleft lip and cleft palate has been reported with Majewski type. In Warrenoff syndrome in addition to narrow thorax, micromelia and post axial polydactyly the spine shows small poorly formed vertebral bodies with wide disc spaces, pelvis is normal and skull shows frontal bossing (2) Type IV SRP shows hypoplastic narrow thorax, rhizomelia, hypoplasia of pubic and ischial ramii with shallow acetabulae, postaxial polydactyly cleft palate, hypoplastic epiglottis and ambiguous genitalia (3).

Important differential diagnosis in a case with narrow thorax and severe micromelia on USG include Asphyxiating Thoracic Dystrophy (AID), Thanatophoric

Dwarfism (TD), Chondroectodermal Dysplasia (CED) and Short Rib Polydactyly syndrome (SRP). However the presence of severe micromelia rules out (ATD and CED while presence of normal spine and absence of severe bowing of femora eliminates the possibility of Thanatophoric Dwarfism.

We feel that it is imperative to be familiar with the features of these rare bone dysplasias so that when recognized early by prenatal ultrasound termination of pregnancy can be offered to the mother. In addition parents must be informed about the risk for subsequent pregnancies depending upon the type of bone dysplasia.

### References

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