

Images in Clinical Practice

Jeune Syndrome

A 3-month-old male patient presented with respiratory distress since birth. There was rhizomelic shortening of upper limbs, simian crease and clinodactyly on both sides. Respiratory rate 80/minute, with sub costal recession, chest was elongated with short ribs (*Fig. 1*), fine crepts on deep inspiration, no cardiac murmur, abdomen soft, liver just palpable.

Anthropometric measurement were weight 3.350 kg, Head circumference 38 cms, chest circumference 27.5 cm, length 56



Fig. 1. Jeune syndrome. Note the small, narrow thoracic cage, subcostal recession, rhizomelic shortening of upper limb.



Fig. 2. X-ray chest-Short rib, high position of clavicle.

cms, (US: LS ratio was 1.6). X-ray chest revealed short horizontal ribs, elongated chest with high placed clavicles (*Fig. 2*). X-ray pelvis depicted hypoplastic iliac wings, horizontal acetabular roof, and spur like projection at lower margin of sciatic notches. Head and abdominal sonography were normal. A diagnosis of Jeune syndrome was made.

Jeune syndrome or asphyxiating thoracic dystrophy (ATD) is a rare autosomal recessive skeletal dysplasia characterized by a small thorax, predominantly rhizomelic brachemelia, renal and hepatic anomalies. Half of the cases have polydactyly. Radiological confirmation of diagnosis is essential. The ribs are short, and the ilia of the pelvis are small, horizontal acetabular roof medial bony projection is visible to give a

trident appearance. Premature ossification of capital femoral epiphysis is seen. New born with thoracic dysplasia associated with lung dysplasia usually die during the neonatal period due to respiratory failure. In those who survive, chronic renal failure is a common cause of death. Renal histopathology reveals cystic changes later peri-glomerular fibrosis. Cirrhosis liver is also one of the cause of early morbidity. At present there is no biochemical or genetic marker, which could be used for prenatal diagnosis of ATD.

However, prenatal ultranographic measurements like TC/ AC (Thoracic circumference/ Abdominal circumference) and RCP (Rib Cage Perimeter)/TC helps in diagnosing skeletal dysplasia associated with small thorax.

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Aplasia Cutis Congenita

A 2-day-old female neonate was with a small skin lesion on the scalp. The lesion was 2 × 1 cm area on the right parietal region covered with a thin, partial thickness membrane like covering, with slight fluid accumulation underneath, giving the appearance of a blister (*Fig. 1*). There was a definite underlying bony defect 1 × 0.5 cm. The child had no neurological deficit and the ultrasound examination of the abdomen and echo-cardiographic evaluation were normal. A clinical diagnosis of Aplasia cutis congenital was made and the child was managed conservatively with paraffin gauze dressings (to avoid drying and eschar formation). The child had complete epithelization at two months of age.

Aplasia cutis congenita is a rare disorder associated with a complete or partial absence of an area of skin. Although this may occur anywhere on the body, about 80% of these lesions involve the scalp, usually the vertex. The size of the lesion may vary from a barely perceptible lesion to lesions larger than 10



Fig. 1. Head demonstrating a small area with absence of skin, which was covered by a thin parchment like membrane.

cm. The smaller lesions are usually partial thickness defects whereas larger defects are likely to be full-thickness with underlying bony defects in 15-30%. They are known to be associated with cardiovascular, neurological, spinal or chromosomal abnormalities and may be familial.