Jeune Syndrome

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Introduction

Jeune syndrome is hereditary hypoplasia of the thorax, associated with pelvic skeletal abnormality. It is a rare autosomal recessive disorder characterized by typical skeletal dysplasias, such as narrow thorax and micromelia, with respiratory and renal manifestations. Respiratory manifestations vary widely from respiratory failure and infantile death to latent phenotype without respiratory symptoms.

Case Report

A term, small for date, male neonate was born to a 26 years old 2nd gravida mother out of second degree consanguineous marriage. At birth, baby had intractable birth asphyxia. Birth weight of the baby was 2000 gms, supine length was 44 cms (ponderal index-2.35) and head circumference was 33 cms. He had micrognathia with bilateral club foot and pedal oedema (bilaterally). Thoracic cage appeared to be small and chest

Fig. 1 : Clinical photograph showing micrognathia, constricted thoracic cage and bilateral club feet

circumference was 30 cms. Antenatal ultrasonography revealed polyhydramnia with bilateral renal cysts.

Baby had respiratory distress after birth, requiring continuous assisted ventilation. Radiological examination of chest revealed small, bell shaped rib cage with diminished lung volume bilaterally, suggestive of lung hypoplasia. The baby died after 72 hrs of birth due to progressive respiratory insufficiency.

Discussion

In 1955, Jeune et al, described familial asphyxiating thoracic dystrophy in a pair of siblings with severe narrowing of thoraxes. Also known as 'Asphyxiating Thoracic Dystrophy' or 'Thoracic Pelvic Phalangeal dystrophy', it is an autosomal recessive disorder, whose gene has not been mapped, but various studies have shown the locus to be on 12p and 15q13 [1]. It is a rare, potentially lethal congenital dwarfism with estimated incidence of 1 per 100,000-130,000 live births.

Classic manifestations in infancy include dwarfism with short ribs, short limbs and characteristic radiographic changes in the ribs (small bell shaped thorax) and pelvis (short iliac bones with acetabular spurs) [2]. Severity of clinical and radiological features is variable. Lung hypoplasia, presumably due to restricted thoracic cage, causes alveolar hypoventilation, and approximately 60-70% patients of Jeune syndrome die from respiratory failure in infancy. Chronic renal failure may ensue in survivors. This is due to associated bilateral microcystic renal disease, which gradually progresses to tubular atrophy and renal failure.

Variability in clinical, radiological and pathological manifestations may be related to genetic heterogeneity. The various manifestations include post axial polydactyly of hands and feet, retinal degeneration [3], intestinal malabsorption, polycystic liver disease, congenital hepatic cirrhosis, cardiac failure due to increased pulmonary vascular resistance, cystic changes of pancreatic ducts

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causing pancreatic exocrine insufficiency and occasional involvement of the teeth, nails and other organs of the body [4].

Changes in the rib cage, pelvis and limbs are rather similar to those of the Ellis-van Creveld syndrome and there is a speculation that Jeune syndrome represents the severe end of the Ellis-van Creveld syndrome spectrum [5]. Other conditions which have similar presentation are achondrogenesis, achondroplasia, osteogenesis imperfecta, thanatophoric dwarfism and hypophosphatasia [6].

Prenatal ultrasonography may show narrow thorax, short hypoplastic ribs and short tubular bones [7,8]. Other ultrasonographic findings include polyhydramnios and absent or feeble respiratory movements.

Conflicts of Interest

None identified

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