Case Report

Jeune Syndrome - a Case Report

Baljinder Kaur, Professor and Head, **Navpreet Kaur,** Junior Resident
Department of Paediatrics, Government Medical College Patiala (Punjab) India

Corresponding Author:
Navpreet Kaur, Junior Resident
Email: navpreetriar74@gmail.com

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Abstract:

Jeune syndrome is a rare autosomal recessive disease characterized by narrow thoracic cage and short limbed dwarfism. More than half (70%) of affected babies die in their early childhood from pulmonary hypoplasia and respiratory distress due to small size of the thorax. Growth retardation and chronic renal failure may occur in patients who survive respiratory failure. Here in our institute a newborn baby was born with jeune syndrome, diagnosed clinically by short rib cage, presented with respiratory distress on day 1 of life. Baby was started on oxygen by positive pressure ventilation and eventually shifted to mechanical ventilation. Symptomatic treatment continued with iv fluids and iv antibiotics but the baby couldnot survive beyond 7 days of life.

Key Words:

Asphyxiating Thoracic Dystrophy, Continuous Positive Airway Pressure, C - reactive protein, Occipito-frontal Diameter, Total Leukocyte Count

Introduction:

Jeune syndrome was first described in 1955, has a reported incidence of 1 in 100,000–130,000 live births and is popularly recognized as a skeletal dysplasia (asphyxiating thoracic dystrophy). This was first described in two siblings with severely narrow thorax.[1]

Jeune syndrome is a rare autosomal recessive skeletal dysplasia characterized by small thorax, short ribs and polydactyly.

International Working Group on Constitutional Diseases of Bone classified short rib and polydactyly syndrome into 6 types:

- Type I (Saldino- Noonan)
- Type II (Magewski)
- Type III (Verma-Naumoff)
- Type IV (Beemer langer)
- Type V Jeune syndrome (asphyxiating thoracic dystrophy)
- Type VI (Ellis van Creveld) syndrome.

Type I-IV are lethal, however Jeune syndrome and Ellis van Creveld syndrome which have often but not always lethal character.[2]

Jeune syndrome is known to be genetically heterogeneous. A locus has been identified on chromosome 15q13 in which recently mutation is found in the IFT80 gene.[3]

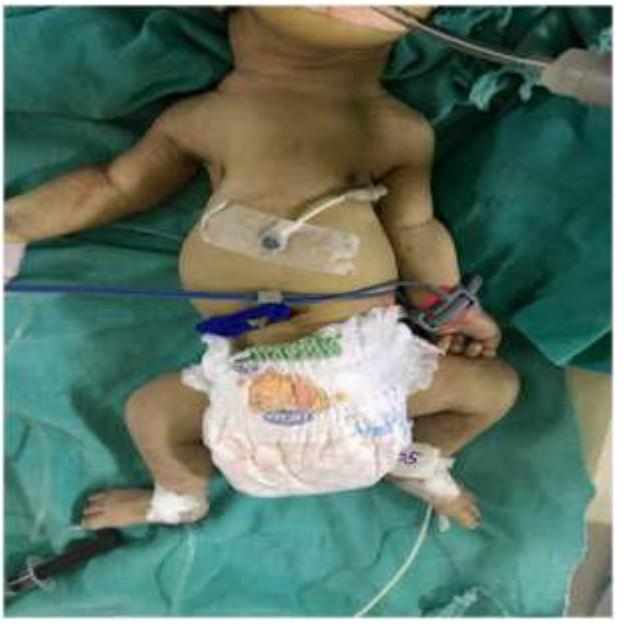
This syndrome is a disorder of bone growth and is characterized by a short sternum narrow and short-rib thorax, and hypoplastic iliac wings. The long bones are either normal or mildly shortened. [4,5] This syndrome is a rare multisystem skeletal dysplasia estimated to occur in 1 per 1,00,000-1,30,000 live births.[6]

In jeunesyndrome , the thorax is small and it results in respiratory distress and recurrent respiratory infection in the neonatal period and infancy. This syndrome has wide spectrum of symptoms beginning from a latent form to lethal condition. In severe cases the progressive respiratory failure may lead to death of neonate. Progressive renal failure is typical for mild form of Jeune syndrome.[5]

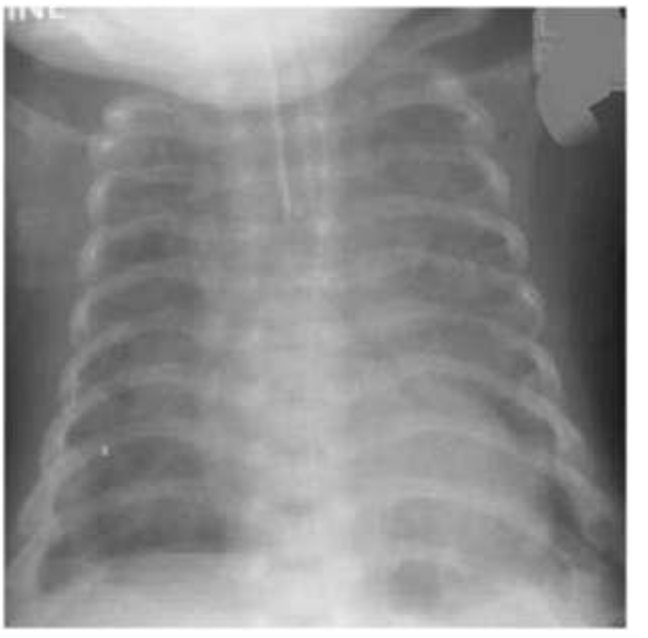
In 60-80% of Jeune syndrome prognosis is poor and often leads to early death of infant.[7]We describe here one such case with the diagnosis based on clinical and radiological findings.

Case report:

A newborn baby was born out of nonconsanguious marriage. Mother was on regular



Pic 1. A newborn baby diagnosed with Jeune syndrome.



Pic 2. X-ray of baby with Jeune syndrome showing small chest, high up clavicle and horizontal ribs.

antenatal check-up and her antenatal health was good. The baby was delivered at term in Rajindra hospital Patiala, by normal vaginal delivery. Weight of the baby was 2630 grams. Occipito frontal circumference (OFC) was 35 cms. Length of the baby was 49 cms. Abdominal examination liver was palpable and spleen was not palpable. Other systems did not reveal any abnormality.

Cry of the baby was immediate with APGAR score of 9 at 1 minute and 10 at 5 minutes. Vitals of the baby were: respiratory rate was 68 / min, heart rate was 136 beats per min and baby was warm to touch with no temperature instability. On examination respiratory distress was present, in the form of nasal flaring, tachypnoea, subcostal, intercostals and suprasternal retractions. On auscultation, grunt was present. Baby was not maintaining oxygen saturation on room air. Baby was started on oxygen by CPAP, but due to continued respiratory distress, baby was intubated on day 1 of life and mechanical ventilation was started.

On investigation haemoglobin was 17 gm/ dl, TLC count was 16500/ cmm and platelet count was within normal limits. CRP of the baby was negative on day 1 of life, when it was repeated after 72 hours of life, it was found to be positive. Serum electrolyte was with in normal limits. Serum calcium was 9.2 mg / dl.

X-Ray of the chest revealed small chest, high up clavicle and horizontal ribs. So on the basis of clinical examination finding and radiological evidence the patient was diagnosed as Asphyxiating thoracic dystrophy (ATD) with late onset neonatal sepsis. Baby was managed with mechanical ventilation, iv antibiotics, iv fluids . Respiratory distress of the baby was not improving with increasing pressures of the ventilator. Baby could not survive beyond 7 days of life.

Discussion:

The clinical features of Jeune syndrome are small, narrow chest and variable limb shortness. Associated congenital abnormalities can be postaxial polydactyly of both hands and/ or feet in 20% of cases.

The characteristic radiological features are narrow thorax, horizontally oriented ribs having expanded anterior ends, horizontally elevated clavicles ("handle-bar" clavicles), and pelvic changes in the form of short iliac wings, trident appearance of acetabulum, relatively short and wide long bones of the extremities and hypoplastic phalanges of both hands and feet with cone shaped epiphyses.[8]

The radiographic findings are so typical that distinction from other skeletal dysplasia is not difficult. In Ellis van Creveld syndrome the appearance of the pelvis is indistinguishable but the

involvement of the thorax is less pronounced.[9]

Nowadays with dynamic development of ultrasound techniques earlier diagnosis is possible; especially in severe cases of this disease. Antenatal diagnosis of this entity has been reported by Mistry *et al.*[8] and few other investigators, who have documented thoracic hypoplasia and short ribs on antenatal ultrasound scan. The antenatal detection of a short rib anomaly warrants counselling of parents of the inevitable, recurrent respiratory complications and poor prognosis despite recent surgical treatment options. The recurrence risk of this autosomal recessive condition in future offspring should be explained to the parents, who should also be advised genetic evaluation.

Jeune syndrome is sometimes compatible with life although respiratory failure and infections are often fatal during infancy. For those patients who survive infancy, the thorax tends to revert to normal with improving respiratory function. This suggests that the lungs have a normal growth potential and the respiratory problems are secondary to restricted rib cage deformity.[7] In milder types of Jeune syndrome it is possible to surgically expand thoracic cavity to prevent respiratory distress.[10]

The differential diagnosis of Jeune syndrome in the antenatal and postnatal period includes other short-rib dysplasias, namely Ellis-van Creveld syndrome , and short rib-polydactyly syndrome. Other differential diagnosis of JATD includes cranio-ectodermal dysplasia and nephronophthisis. Ellis-van Creveld syndrome is characterized by finger nail dysplasia, polydactyly and heart defects. These features were conspicuous by their absence in our patient. Absence of polydactyly further excluded short-rib polydactyly syndrome. Cranio-ectodermal dysplasia is a ciliopathy with skeletal involvement along with the presence of characteristic facial and ectodermal features. There was no cranio-facial involvement in our patient excluding cranio-ectodermal dysplasia. Nephronophthisis was excluded as there was no renal involvement in our patient.[8,11]

Conclusion:

The correct diagnosis of Jeune syndrome should be important as recurrence risk of this autosomal recessive condition in future offspring should be explained to the parents, who should also be advised genetic evaluation.

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