CASE REPORT

Jeune's Syndrome: Asphyxiating Thoracic Dysthrophy and Its Late Presentation

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Pak Pediatr J 2025; **49(1):** 102-04

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Received 30th November 2024;
Accepted for publication
3rd February 2025

ABSTRACT

Jeune,s syndrome is an autosomal recessive disease in which there is narrow rib cage and short limbs. Majority of babies (almost 70%) suffering from the disease die in neonatal life or early infancy due to pulmonary hypoplasia and respiratory distress. Those who survive through respiratory failure suffer from chronic renal failure and growth retardation. Here in our institute a 2-year-old male baby was brought with history of recurrent respiratory infections and diagnosed as Jeunes syndrome clinically and radiologically. Child was given oxygen support and symptomatic treatment and discharged to home after 5 days.

Key Words: *Respiratory tract, Pulmonology, Neonate*

INTRODUCTION

Osteochondrodysplasias (skeletal dysplasias) constitute a large, heterogeneous group of mostly genetic disorders with a prevalence of more than 1 in every 5000 newborns. Considering the fact that most of these diseases are life-threatening and underdiagnosed, probably the incidence rate is higher than reported.[1]

The International Working Group on Constitutional Diseases of Bone has classified short ribs and polydactyly syndromes into 6 types:

- Type I (Saldino-Noonan)
- Type II (Magewski)
- Type III (Verma-Naumoff)
- Type IV (Beemer Langer)
- Type V (Jeunes syndrome)
- Type VI (Ellis-van Creveld syndrome)

Except for Types V and VI, which are not always lethal, the rest are invariably lethal.[2]

Jeunes syndrome (JS), also known as asphyxiating thoracic dystrophy, was first described in 1954. It is a congenital multisystem disorder characterized by a constricted and narrow rib cage, polydactyly, and small limbs. Additionally, it is associated with other systemic complications such as impaired renal function, liver fibrosis, pancreatic cysts, and retinal anomalies. This syndrome is extremely rare, with an estimated incidence of 1 per 100,000/130,000 live births. Furthermore, it is an autosomal recessive ciliopathy with a range of clinical presentations.[3]

Genetic heterogeneity may contribute to severe clinical and radiological features. Identified mutations in four genes IFT80, DYNC2H1, TTC21B, and WDR19 are associated with the syndrome. The narrow rib cage results in lung hypoplasia, leading to alveolar hypoventilation, and the disease proves fatal in 90% of neonates due to respiratory failure.[4] In less severe cases, patients may present in late infancy with recurrent pneumonia and/or atelectasis resulting from a rigid chest wall.

Here, we present a case of a 2-year-old exhibiting a narrow rib cage, diagnosed as Jeune syndrome based on clinical and radiological findings.

CASE REPORT

A 2 – year - old boy was born from a non-

consanguineous marriage. The mother had regular prenatal check-ups and was in good health. The baby was born in a private hospital through an emergency lower segment caesarean section due to fetal distress at 36 weeks of gestation and was sent home after 48 hours. The child was kept under observation for 48 hours in the hospital due to late preterm delivery. The baby was doing well until the 5th month of life. After that, the child developed respiratory distress, for which he was admitted to a private hospital and discharged in 7 days. Afterward, the child had multiple episodes of respiratory distress intermittently, with a 3-4 month symptom-free interval. The child was admitted three times for similar complaints of respiratory distress.

Currently, at the age of 2 years, the child presented with dry cough and respiratory distress for the past 5 days, with no significant history of fever, allergen exposure, or relevant family history. On examination, the child had a respiratory rate of 50 breaths per minute, a heart rate of 110 beats per minute, and was not maintaining saturation on room air. On systemic examination, the child had an obvious small-sized chest with a chest circumference of 31 cm and respiratory distress in the form of nasal flaring, intercostal, and subcostal recessions. His upper limb to lower limb ratio was 1.5:1. The rest of the examination was unremarkable. The child was put on a nasal cannula for oxygen support.

On investigation, CBC showed normal hemoglobin of 13 g/dl with normal lymphocyte and platelet count. Biochemistry was also in the normal range. Chest x-ray revealed a small-sized chest. There were short, horizontally oriented ribs and irregular costo-chondral junction. The clavicles were abnormally elevated with a typical handlebar appearance. Echocardiography was normal. So, on the basis of history, examination, and radiological evidence, the patient was diagnosed with asphyxiating thoracic dystrophy i.e., Jeunes syndrome. The child was managed on oxygen support, IV antibiotics, and IV fluids. Dietary counselling was done. The child was discharged after 7 days of treatment and advised follow-up; parents were counselled regarding the disease course.

Fig 1: Patient diagnosed with jeunes syndrome

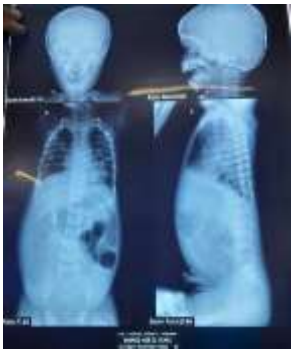

Fig 2: X-ray of baby with Jeune syndrome showing small chest, high up clavicle and horizontal ribs

DISCUSSION

Jeune syndrome (Asphyxiating Thoracic Dystrophy) belongs to a group of skeletal dysplasia known as short-rib thoracic dysplasia. Jeune syndrome affects an estimated 1 in 100,000 to 130,000 live births.[1] It is a rare autosomal recessive ciliopathy characterized by a small, narrow rib cage, variable limb deformity, and additional multiple organ involvement.[5] Polydactyly is reported in 50% of cases.[6] This syndrome is mainly diagnosed by clinical and radiological findings. Clinically, there are short and wide ribs, a narrow bell-shaped chest, and shortened long bones leading to dwarfism. If the chest is extremely narrow, it can restrict lung growth and expansion, leading to lung hypoplasia.[1]

Radiologically, it is characterized by a narrow thorax, handle-bar clavicles, hypoplastic iliac wings, trident appearance of acetabulum, cone-

shaped end of long bones in arms and legs, short hand and feet, and spinal abnormalities.[1] Radiological findings are so typical that they are enough to distinguish from other skeletal dysplasia.

The main concern of Jeune syndrome is respiratory problems and is one of the main concerns for neonatal mortality, reaching up to 80% as mentioned in the literature. Another major complication is renal failure, usually present after 2 years of age. Renal histopathology reveals cystic changes and subsequent periglomerular fibrosis.[6] Some cases with ocular abnormalities have also been reported, usually retinitis pigmentosa. It presents with night blindness initially and can be seen as early as 5 years of age.[5]

At present, there is no biochemical or genetic marker that could be used for prenatal diagnosis of ATD. However, prenatal ultrasonographic measurements like TC/AC (Thoracic circumference/Abdominal circumference) and RCP (Rib Cage Perimeter)/TC help in diagnosing skeletal dysplasia associated with a small thorax.[6] Antenatal detection warrants counselling of parents about the disease and the risk of recurrent chest infections and poor prognosis. The risk of recurrence in future offspring is due to autosomal recessive inheritance. Parents also should be advised for genetic testing.

Jeune syndrome is sometimes compatible with life; the majority of patients with the disease die during the neonatal period due to respiratory failure and recurrent infections. Children often need ventilatory support during the first year of life.[5]

Jeune syndrome, both in the antenatal and postnatal periods, shares similarities with other short-rib dysplasia, specifically Ellis-van Creveld syndrome and short-rib polydactyly syndrome. Additional differential diagnoses for Jeune asphyxiating thoracic dystrophy include cranio-ectodermal dysplasia and nephronophthisis. Ellis-van Creveld syndrome is characterized by finger and nail dysplasia, polydactyly, and heart defects. Polydactyly is specific for short-rib polydactyly syndrome. Cranio-ectodermal dysplasia, a ciliopathy with skeletal involvement, typically presents with characteristic facial and ectodermal features. Nephronophthisis has renal abnormalities.[2]

With the advent of surgical techniques, multiple procedures have been described for the treatment of Jeune syndrome with the primary objective of expanding the thoracic cage, which will lead to lung expansion and improved ventilation. Most of these approaches include median sternotomy with graft interposition.[7]

Conflict of interest: None

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