

Jeune syndrome: Asphyxiating thoracic dystrophy

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Section: Paediatric radiology

Area of Interest: Paediatric Respiratory system

Imaging Technique: Conventional radiography

Case Type: Clinical Cases

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Patient: 17 months, female

Clinical History:

One year five-month-old female child of nonconsanguineous parents presented with complaints of fast breathing since 6 months of age. She had recurrent episodes of lower respiratory tract infections in the past, two of which required mechanical ventilation. On physical examination, the chest was deformed and narrow with central depression. The anthropometric assessment revealed a height below the 25th percentile.

Imaging Findings:

Imaging revealed a narrow elongated "bell-shaped" thorax, short, horizontally oriented ribs with expanded costochondral junctions and high-riding clavicle "handlebar clavicles". Additionally, there was mild squaring of iliac bones with a flattened acetabular roof. Spine radiography showed scalloping of a few dorsal-lumbar vertebral bodies. There was no polydactyly.

Discussion:

Background

Jeune syndrome or asphyxiating thoracic dystrophy is a rare autosomal recessive ciliopathy estimated to occur in 1 per 100,000-130,000 live births. (1) This is a skeletal dysplasia affecting the endochondral bone formation, characterized by a small narrow chest and short-limbed dwarfism along renal and hepatic anomalies. Clinical and radiological findings are used to make the diagnosis. Small thoracic cage and varying limb shortness are traits of ATD. The clinical spectrum can vary significantly. Most patients experienced respiratory problems in the neonatal period of their life. The thoracic malformation becomes less noticeable and respiratory problems diminish with increasing age.

Imaging Perspective

Typical radiographic findings include thoracic cage deformity along with other skeletal deformities. The thorax is narrow and bell-shaped, with short, horizontally oriented ribs, irregular costochondral junctions, and elevated clavicles. (2,3) The clavicles are abnormally elevated, which is known as the handlebar clavicle. Short flared iliac bones with trident acetabula, relatively short and wide long bones of the extremities, short broad phalanges with cone-shaped epiphyses, and polydactyly are other skeletal abnormalities. There is a considerable variation in skeletal deformities.

Outcome

There is a broad spectrum of symptoms, ranging from a latent form to a fatal condition. Although respiratory failure and infections are frequently fatal in infancy, Jeune syndrome is sometimes compatible with life. In severe cases, the neonate may die because of progressive respiratory failure. The overall mortality is up to 70% due to respiratory failure during infancy. (2) When patients survive infancy, the thorax usually returns to normal as their respiratory function improves. Patients who survive childhood are at risk of developing renal tubular acidosis later in life. (1) Renal and hepatic dysfunctions development occur insidiously in patients with Jeune syndrome and lead to complications like renal failure and hepatic cirrhosis later in life. These late complications are other causes leading to the early demise of these children. Our patient had classical thoracic cage abnormalities and further evaluation revealed underlying renal tubular acidosis implying early signs of renal disease.

Written informed patient and parents' consent for publication has been obtained.

Differential Diagnosis List: Jeune syndrome, Thoracolumbar dysplasia, Ellis-van Creveld syndrome, Sensenbrenner syndrome, Paternal uniparental disomy of chromosome 14

Final Diagnosis: Jeune syndrome

References:

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Figure 1

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Description: Chest radiography shows a small narrow chest, high up clavicle, horizontal and short ribs

Origin: © Department of Radiology, St. John's Medical College, Bangalore, India, 2023

Figure 2

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Description: Lateral spine radiograph demonstrates anterior scalloping of a few dorsal-lumbar vertebral bodies **Origin:** © Department of Radiology, St. John's Medical College, Bangalore, India, 2023

Figure 3

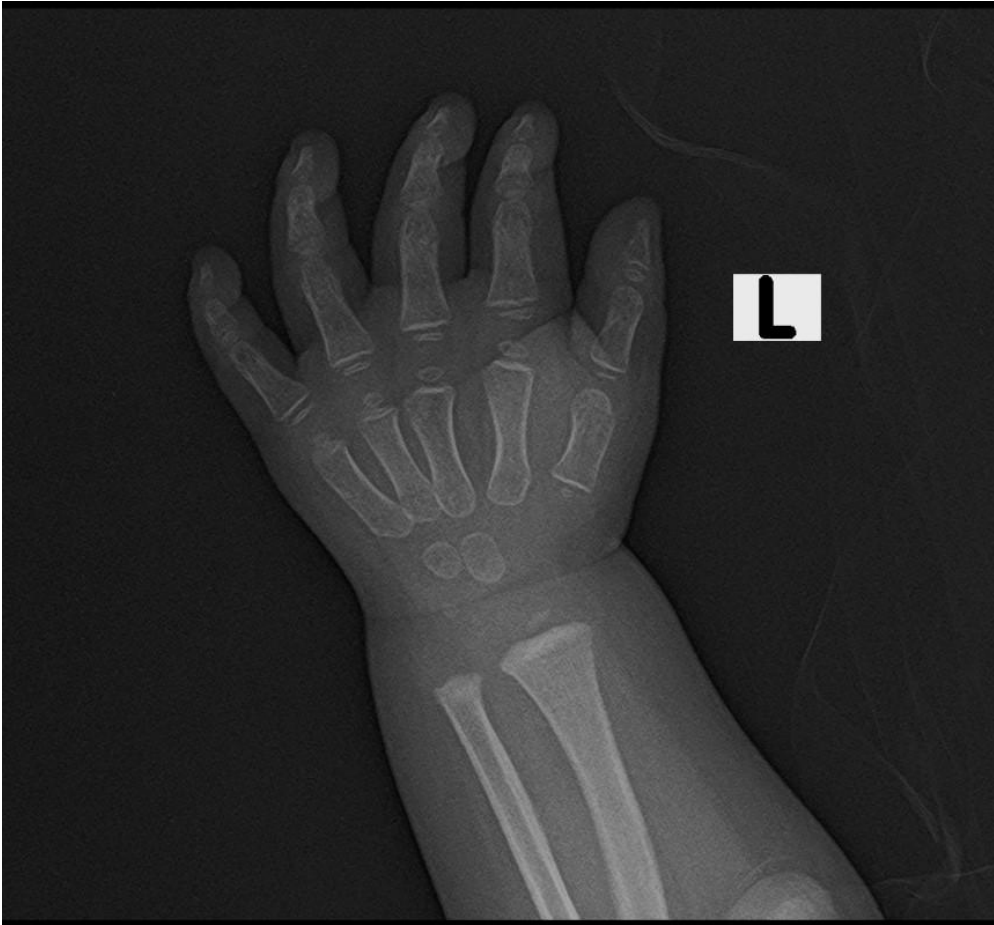
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Description: Pelvis radiograph shows mild squaring of iliac bones with flattened acetabular roof along with metaphyseal bands involving the distal femur **Origin:** © Department of Radiology, St. John's Medical College, Bangalore, India, 2023

Figure 4

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Description: Normal hand radiograph with no evidence of polydactyly **Origin:** © Department of Radiology, St. John's Medical College, Bangalore, India, 2023