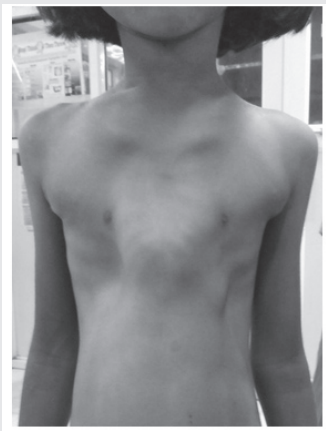
SPOT DIAGNOSIS (IMAGE GALLERY)



SMALL AND NARROW CHEST

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A 7 year old girl presented in emergency with severe respiratory distress. She had frequent episodes of respiratory difficulty since birth. On examination, her chest was small and narrow with abnormal protrusion. Upon auscultation, bilateral rhonchi were present. Her X-ray chest was suggestive of narrow chest with short and horizontal ribs.

What is the diagnosis?

Asphyxiating thoracic dystrophy (Jeune syndrome) is a rare disease characterized by small thorax and short limb dwarfism with multi-organ involvement. (1,2) The inheritance of asphyxiating thoracic dystrophy is autosomal recessive. A locus has been identified on chromosome 15q13. (3) Diagnosis is based on clinical and radiographic findings. The radiological appearance include narrow chest cage, especially in its upper part, with the relative flaring of lower ribs, which accounts for the bell shape. The ribs are short and horizontal in position, their distal metaphyses widened and the costochondral junctions expanded and irregular. Clavicles are high. (4) The small, narrow thorax often results in respiratory distress and recurrent respiratory infections in the neonatal period and infancy. (2) The severity of thoracic constriction varies. Treatment is primarily symptomatic support. Patients surviving infancy may develop progressive renal failure later in life. (5) Liver, pancreas and retinal abnormalities have also been described. The thoracic malformation tends to become less severe with age, resulting in less respiratory problems. (2)

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