

Spondylocostal Dysostosis (Jarcho Levin Syndrome)

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A term female neonate with a birth weight of 3.25 kg was tachypneic at birth. The neonate had a short neck, scoliosis to the right side of the chest and asymmetric chest wall with heart sounds heard on the right side of the chest (Fig. 1). The neonate's rib cage was deficient on the left side with a soft tissue swelling moving with respiration. It was suggestive of lung herniation. Chest skiagram showed congenital absence of left 6th and 7th ribs, partial fusion of left 8th and 9th ribs, partial fusion of right 5th, 6th and 7th ribs, scoliosis of the spine to the right side of chest and hemivertebra (Fig. 2). The echocardiography was



Fig. 1 The neonate with asymmetric chest wall

normal. These features were consistent with the diagnosis of Spondylocostal dysostosis or Jarcho-Levin syndrome.

This syndrome is characterized by fused or malformed vertebrae and small, fused or missing ribs. Associated features include short neck and short stature [1]. It is caused by mutation in any of the 4 genes—*DLL3*, *MESP2*, *LFNG* and *HES7* [2]. Treatment is mainly supportive.

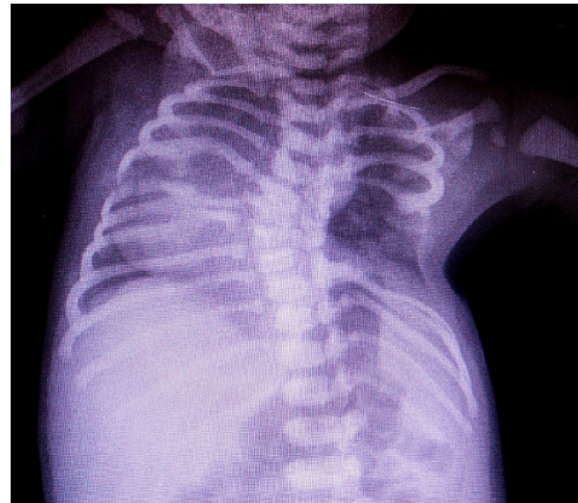


Fig. 2 Skiagram showing absence of left 6th and 7th ribs, partial fusion of left 8th and 9th ribs and partial fusion of right 5th, 6th and 7th ribs with scoliosis of the spine to the right side of chest and hemivertebra

Compliance with Ethical Standards

Conflict of Interest None.

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References

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The parent's consent was taken to use photographs that may reveal the identity of the patient.

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