Jarcho-Levin syndrome: a rare syndrome presented with asymptomatic cardiac murmur

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DESCRIPTION

A 2-year-old female presented with asymptomatic continuous murmur. Echocardiogram revealed patent ductus arteriosus (PDA). Chest roentgenogram (figure 1) incidentally showed scoliosis, hemivertebrae (seventh thoracic vertebrae), failure of fusion of the left lower ribs, bifid rib and spina bifida suggestive of Jarcho-Levin syndrome (JLS). The patient underwent successful percutaneous PDA coil (Flipper) closure.

JLS or spondylocostal dysostosis (SCD) is a rare congenital anomaly of the thoracic cage and vertebrae with an estimated incidence of 1 per 40 000 births.[1] For years, the terms JLS or SCD and spondylothoracic dysostosis (STD) have been used inaccurately interchangeably in the literature. The differences between both the syndromes have been described in the literature.[1] Though JLS is considered as less severe, the respiratory compromise and infection pose the most serious threat to affected individuals. Spina bifida is rarely reported with JLS and is a commoner with STD. There are scattered case reports of the association of JLS with congenital heart diseases, like shunt lesions or heterotaxy syndromes.[2] These patients invariably have some degree of respiratory compromise and are prone to respiratory complications. We want to stress on the fact that the patients with JLS should be screened for cardiac defects, and the defects should be treated whenever possible. This is the first case of successful percutaneous PDA device closure in a patient with JLS, to the best of our knowledge. Also the percutaneous approach appears more prudent as it lessens the perioperative, particularly pulmonary, complications.

Learning points

- Jarcho-Levin syndrome is a rare congenital anomaly of the thoracic cage and vertebrae.
- It may be associated with congenital heart disease.
- Percutaneous device closure is a better treatment option, whenever possible.

Competing interests None.

Patient consent Obtained.

Provenance and peer review Not commissioned; externally peer reviewed.

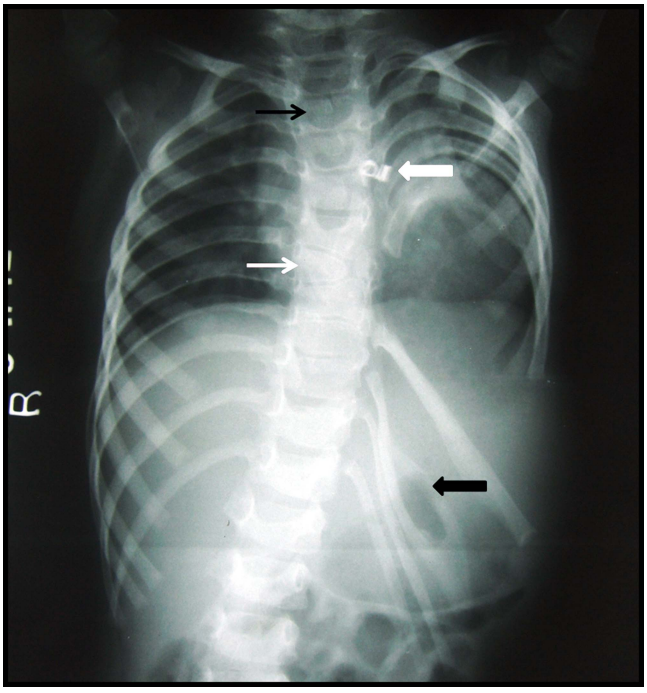

Figure 1 Chest X-ray in posteroanterior view demonstrating scoliosis, hemi-vertebrae (thin white arrow), failure of fusion of the left lower ribs, bifid rib (thick black arrow), spina bifida (thin black arrow) and patent ductus arteriosus coil (thick white arrow) in situ.

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