Lumbo-costo-vertebral syndrome with inguinal hernia and other anomalies

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DESCRIPTION

A 20 day-old full-term vaginally delivered baby boy, weighing 3.25 kg, presented to the surgical department with deformity of spine and lower limbs, swelling in left groin and left lumbar region since birth. Antenatal ultrasound was not available. On examination, the child was active, crying well, lips were cyanosed and there was no pedal oedema. Cardiovascular examination revealed the presence of systolic murmur. Abdominal examination showed the presence of left lumbar and inguinal hernia (figure 1). Telipus equinovarus deformity was present bilaterally in lower limbs. Scoliosis of spine with concavity towards left side was present. Echocardiography showed tetralogy of Fallot. Ultrasound scan of the abdomen revealed the absence of left kidney, left lumbar and inguinal hernia. X-ray showed scoliosis, hemivertebrae of thoracic region, left rib anomalies, herniation of bowel loops in left lumbar region and left scrotum (figure 2).

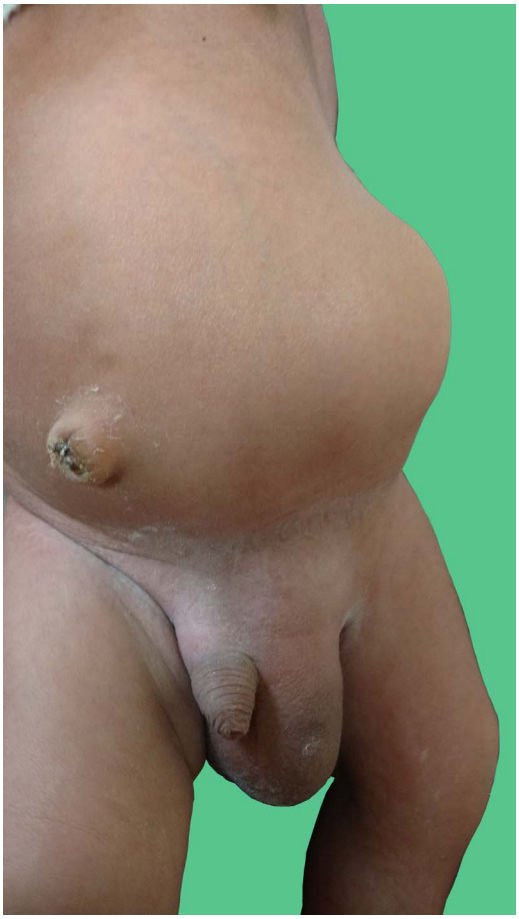

Figure 1 Left lumbar and inguinal hernia.

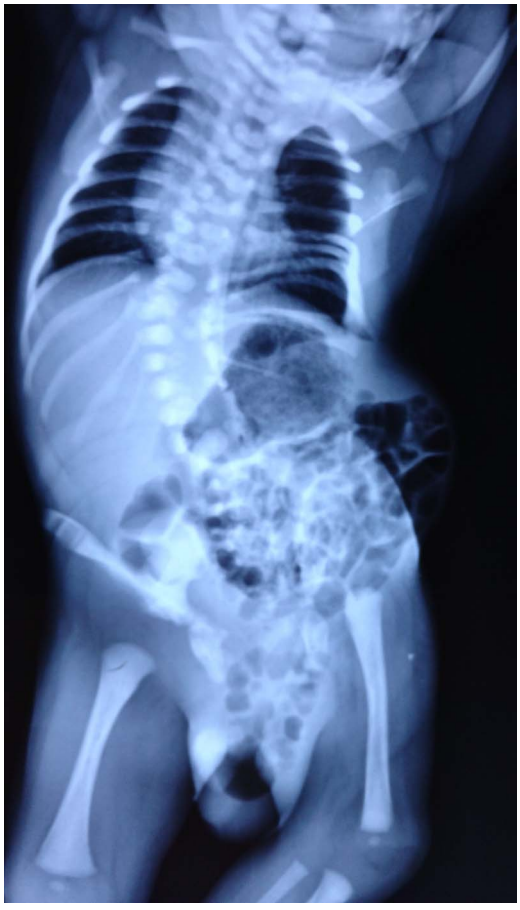

Figure 2 Costo-vertebral anomalies, scoliosis, bowel loops in lumbar and inguinal hernia sac.

Learning points

- Lumbo-costo-vertebral syndrome includes hemivertebra, congenital anomalies of ribs, anterior myelomeningocele and abdominal wall muscular hypoplasia.
- Identification of anomalies associated with lumbo-costo-vertebral syndrome should therefore direct prompt thorough evaluation for other anomalies.
- Evaluation should include radiographic studies and appropriate consultation of specialists.

Lumbo-costo-vertebral syndrome is a rare disorder comprising of vertebral, ribs and abdominal musculature anomalies along with other associated anomalies such as inguinal hernia, telipus

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equinovarus, absence of kidney, congenital heart disease and many others.[1–3] It has been postulated that single somatic mutation during the third to fifth week of gestation lead to the development of this constellation of disorder.[1] Lumbo-costo-vertebral syndrome with bony, cardiac and renal anomalies was found to be an associated anomaly to VACTERL syndrome.[3] This case represents rare association of congenital anomalies and highlights the requirement of multidisciplinary subspecialty in management.

Contributors RSS, SSP and SKR have seen the case, advised necessary investigation, prepared the manuscript and edited. RSS will be guarantor for this manuscript.

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Patient consent Obtained.

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