Spondylocostal Dysostoses

A 22-year-old gravida 2 mother delivered a female baby at 35 weeks of gestation. The parents were first cousins and there was no family history of short stature or spinal abnormality. The mother’s antenatal period was uneventful. The baby’s birth weight was 2010 g, length was 39 cm and head circumference 31 cm. Physical examination revealed a short neck and trunk with apparently long limbs and a protuberant abdomen (Fig. 1). Systemic examination was otherwise normal. Radiographic examination showed multiple vertebral anomalies (hemivertebrae, absent vertebrae and fused vertebrae) and rib deformities (fused and absent ribs) (Fig. 2). Abdominal ultrasonography and echo-cardiography were normal.

The clinical and radiological features were consistent with the diagnosis of spondylocostal dysostosis (SCD). She did not develop respiratory distress and was discharged on exclusive breast feeds.

Spondylocostal dysostoses are a heterogeneous group of disorders characterized by multiple vertebral and rib anomalies. Affected individuals have short trunked dwarfism of prenatal onset associated with non-progressive kyphoscoliosis, and restrictive lung disease that is usually the cause of early death. Radiologically, the disease is characterized by vertebral malformations including hemivertebrae, block vertebrae, fused vertebrae and spina bifida and deformities of the ribs that include absent ribs and bifid or fused ribs, which give the typical “crab like”, or “fan like” appearance. Associated malformations include those of the

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**Fig. 1. Clinical photograph of the baby showing a short trunk and neck**

**Fig. 2. X-ray of the spine showing multiple vertebral and rib anomalies.**
Images in Clinical Practices

SCD families that the defective locus is on chromosome 19q13. Subsequent mutation analysis has determined that mutations in the human somitogenesis gene, delta-like 3 (DLL 3), which encodes a ligand for the Notch signalling pathway causes autosomal recessive SCD.

Prenatal ultrasound diagnosis is possible and characteristically shows the presence of fanned out ribs from fused thoracic vertebral bodies. Reconstructive surgery of the chest including titanium rib implants has been described in the treatment of this disorder.

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Tinea Faciei

A two-week-old female infant presented with a rash first noted for two days (Fig. 1). The rash was on the face and scalp. There were multiple lesions with an annular, raised, scaly margin that was erythematous and pustular in part, central depigmentation was also noted. The mother had a similar rash two years earlier. In the subsequent two years several members of the extended family had developed a similar rash. The differential diagnosis considered were Neonatal Lupus or Tinea Faciei. Fungal scrapings taken from the baby were negative on direct examination.