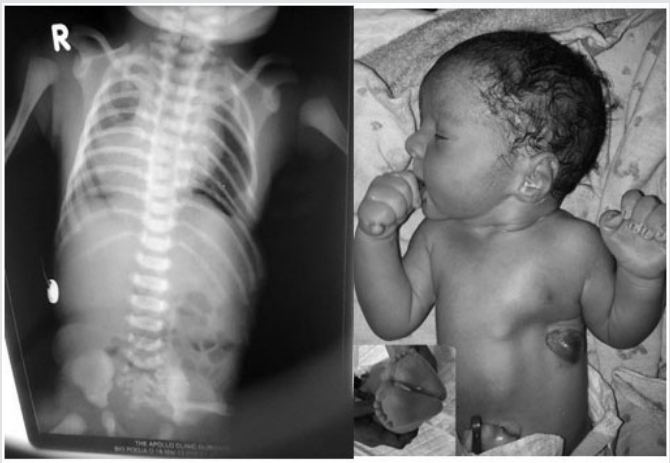
SPOT DIAGNOSIS (IMAGE GALLERY)



SHORT RIB POLYDACTYLY SYNDROME TYPE III

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A one day old male new-born presented with in-drawing of chest, bulged-out skin bereft lesion in left lower chest-wall, polysyndactyly in all for limbs. Chest x-ray showed the presence of hypoplastic ribs 5th – 10th on the left side and dextrocardia. On ultrasound, the bulged-out lesion seemed to be protruded spleen; bilateral polycystic-kidney was also evident. Echocardiography showed dextrocardia with ventricular septal defect. MRI abdomen corroborated USG findings. The chest lesion got cured by topical antibiotic ointment application.

What is the diagnosis?

Short-rib-polydactyly-syndrome type III (SRPS3), which is also called Verma-Naumoff syndrome. It is a very rare syndrome characterised by short ribs, various gastrointestinal, cranial structural abnormality, post-axial polydactyly, cystic renal disease, heart and laterality disorder, ambiguous genitalia. (1) Homozygosity or compound heterozygosity for missense or nonsense mutations in the cytoplasmic dynein 2 heavy chain 1 gene (DYNC2H1) are reported in SRPS3. (2) Differential diagnosis include Ellis-van Creveld syndrome, Acropectoral syndrome, Greig-cephalopolysyndactyly syndrome, Saldino-Noonan syndrome, Acrocallosal syndrome. The Verma-Naumoff type was first published in 1983 as a subtype of Type I, since most of its characteristics are the same as those of Type I. It differs from Type I as the bones of the extremities and the iliac bones show better ossification, and visceral organ anomalies occur less often. (3) Generally this syndrome is diagnosed by pre and post natal ultrasonography, radiography, post mortem examination and genetic analysis. (4) But there is no treatment or prevention available for this syndrome.

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