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Short rib-polydactyly syndrome, Saldino-Noonan type

INSERM

Source

INSERM. (1999). *Orphanet: an online rare disease and orphan drug data base. Short rib-polydactyly syndrome, Saldino-Noonan type. ORPHA:93270*

Short rib-polydactyly syndrome (SRPS), Saldino-Noonan type is an extremely rare type of SRPS with neonatal onset characterized by polydactyly, hydropic appearance, and small thorax with short horizontal ribs causing fatal cardiorespiratory distress. Affected patients also have extreme micromelia, pointed metaphyses, and a range of other ossification defects (vertebrae, calvaria, pelvis, hand and foot bones). Extrasketal manifestations may include polycystic kidneys, transposition of the great vessels, and atresia of the gastrointestinal and genitourinary systems.