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Short rib-polydactyly syndrome, Beemer-Langer type

INSERM

Source

INSERM. (1999). *Orphanet: an online rare disease and orphan drug data base. Short rib-polydactyly syndrome, Beemer-Langer type. ORPHA:93268*

Short rib-polydactyly syndrome (SRPS), Beemer-Langer type is an extremely rare type of SRPS developing prenatally or immediately after birth and characterized by short and narrow thorax with horizontally oriented ribs. Other bone features include small iliac bones, short tubular bones, bowing of long bones and rarely pre- and post-axial polydactyly. Brain defects are common and some cases of cleft lip, absent internal genitalia and renal, biliary and pancreatic cysts have been reported. The course is rapidly fatal.