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Short rib-polydactyly syndrome, Verma-Naumoff type

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. <u>Short rib-</u> polydactyly syndrome, Verma-Naumoff type. ORPHA:93271

Short rib-polydactyly syndrome, Verma-Naumoff type is a short rib-polydactyly syndrome characterized by short limb dwarfism, short ribs with thoracic dysplasia, postaxial polydactyly and protuberant abdomen. Associated multiple malformations include cardiovascular defects, renal agenesis /hypoplasia, abnormal cloacal development (ambiguous genitalia, anal atresia) and cerebellar hypoplasia. Short rib-polydactyly syndrome, Verma-Naumoff type follows an autosomal recessive mode of transmission. The disease is usually fatal in the perinatal period.