
DISEASE:
Short rib-polydactyly syndrome type 5

NAME:	Short rib-polydactyly syndrome type 5
DESCRIPTION:	A rare ciliopathy with major skeletal involvement characterized by short ribs, micromelia, limb bowing, polysyndactyly, absent ossification of the radii, tibiae and fibulae, as well as the bony elements of the hands and feet, and hypoplastic scapulae. Additional hallmarks of ciliopathic disease, such as laterality defects and cystic kidneys, have also been observed.
ORPHACODE:	498497
XREF(S):	<u>Orphanet</u> <u>ICD-10</u> <u>OMIM</u>
ANALYTE(S):	<u>WDR35</u>
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