

DISEASE:
Short rib-polydactyly syndrome, Beemer-Langer type

NAME:	Short rib-polydactyly syndrome, Beemer-Langer type
DESCRIPTION:	A rare ciliopathy with major skeletal involvement characterized by short ribs and hypoplastic thorax, small iliac bones, short tubular bones with smooth metaphyseal margins, and bowed radii and ulnae. The tibiae are relatively well tubulated and longer than the fibulae. There is a high frequency of brain defects, while post-axial polydactyly is rare. Additional features may include cleft lip, absence of internal genitalia, and renal, biliary, and pancreatic cysts, among others.
ORPHACODE:	93268
SYNONYMS:	Short rib-polydactyly syndrome type 4
XREF(S):	<u>Orphanet</u> <u>ICD-10</u> <u>OMIM</u>
ANALYTE(S):	<u>IFT80</u> <u>IFT122</u>
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