DISEASE:
Short rib-polydactyly syndrome, Majewski type

NAME:	Short rib-polydactyly syndrome, Majewski type
DESCRIPTION:	A rare ciliopathy with major skeletal involvement characterized by a hypoplastic thorax with short ribs and protuberant abdomen, micromelia with particularly short tibiae with ovoid configuration, pre- and postaxial polydactyly, brachydactyly, hypoplasia or aplasia of nails, and dysmorphic craniofacial features (such as prominent forehead, low-set and malformed ears, short and flat nose, lobulated tongue, micrognathia, and cleft lip/palate). Additional reported manifestations include urogenital, gastrointestinal, cardiovascular, and cerebral malformations, among others. The condition is fatal in the neonatal period.
ORPHACODE:	93269
SYNONYMS:	Short rib-polydactyly syndrome type 2
XREF(S):	Orphanet ICD-10 OMIM OMIM
ANALYTE(S):	TRAF3IP1 DYNC2H1 NEK1
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RELATED CONTENT

Related Genetic Tests

- Ciliopathy (gene panel)
- cleft lip with/whitout cleft palate (virtual gene panel)

Related Laboratories

- Centre de Génétique Médicale UCL
- Centrum Medische Genetica - UZ Gent

Related Analytes

- dynein cytoplasmic 2 heavy chain 1
- NIMA related kinase 1
- TRAF3 interacting protein 1

Related Gene Panels

- Ciliopathy (120 genes) - UGent
- Cleft lip and palate / dysmorphic facial features / craniofacial anomalies (255 genes)) - UCL

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