DISEASE:
Short rib-polydactyly syndrome, Verma-Naumoff type

NAME:	Short rib-polydactyly syndrome, Verma-Naumoff type
DESCRIPTION:	A rare ciliopathy with major skeletal involvement characterized by short ribs and extremely narrow thorax, severely shortened tubular bones with round metaphyseal ends and lateral spikes, and anomalies of multiple organs such as the heart, kidneys, liver, pancreas, intestine, and genitalia, with occasional occurrence of situs inversus totalis. Cleft lip/palate and polydactyly may also be present. The syndrome is fatal prenatally or in the perinatal period.
ORPHACODE:	93271
SYNONYMS:	Short rib-polydactyly syndrome type 3
XREF(S):	Orphanet MeSH OMIM OMIM OMIM OMIM ICD-10

ANALYTE(S):	IFT80 DYNC2H1 WDR35 DYNC2I1 DYNC2I2
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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
- dynein 2 intermediate chain 1
- dynein 2 intermediate chain 2
- intraflagellar transport 80
- WD repeat domain 35

Related Gene Panels

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

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