DISEASE:
Jeune syndrome

NAME:	Jeune syndrome
DESCRIPTION:	Jeune syndrome, also called asphyxiating thoracic dystrophy, is a short-rib dysplasia characterized by a narrow thorax, short limbs and radiological skeletal abnormalities including 'trident' aspect of the acetabula and metaphyseal changes.
ORPHACODE:	474
SYNONYMS:	Asphyxiating thoracic dystrophy of the newborn JATD Jeune asphyxiating thoracic dystrophy

XREF(S):	Orphanet OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM MeSH MedDRA ICD-10
ANALYTE(S):	KIAA0753 DYNC2LI1 IFT80 DYNC2H1 TTC21B WDR19 IFT140 DYNC2I1 IFT172 DYNC2I2 CEP120
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RELATED CONTENT

Related Genetic Tests

- Ciliopathy (gene panel)
- Nephrotic syndrome, Focal Segmental Glomerulosclerosis (FSGS) , Alport syndrome and podocytopathy (gene panel)
- cleft lip with/whitout cleft palate (virtual gene panel)

Related Laboratories

- Centre de Génétique Médicale UCL
- Centre de Génétique-Institut de Pathologie et de Génétique (IPG)
- Centrum Medische Genetica - UZ Gent

Related Analytes

- centrosomal protein 120
- dynein cytoplasmic 2 heavy chain 1
- dynein 2 intermediate chain 1
- dynein 2 intermediate chain 2
- dynein cytoplasmic 2 light intermediate chain 1
- intraflagellar transport 140
- intraflagellar transport 172
- intraflagellar transport 80
- KIAA0753
- tetratricopeptide repeat domain 21B
- WD repeat domain 19

Related Gene Panels

- Ciliopathy (120 genes) - UGent
- Cleft lip and palate / dysmorphic facial features / craniofacial anomalies (255 genes)) - UCL
- Nephrotic syndrome, FSGS, Alport syndrome (76 genes) - IPG
