DISEASE:
Joubert syndrome with Jeune asphyxiating thoracic dystrophy

NAME:	Joubert syndrome with Jeune asphyxiating thoracic dystrophy
DESCRIPTION:	A rare genetic developmental defect during embryogenesis characterized by the association of the classic features of Joubert syndrome (congenital midbrain-hindbrain malformations causing hypotonia, abnormal breathing and eye movements, ataxia and cognitive impairment) together with the skeletal anomalies of Jeune asphyxiating thoracic dystrophy (short ribs, long and narrow thorax causing respiratory failure, short-limbs, short stature, and polydactyly). Additional variable manifestations include cystic kidneys, liver fibrosis, and retinal dystrophy.
ORPHACODE:	397715
SYNONYMS:	JBTS with JATD Joubert syndrome with JATD
XREF(S):	Orphanet ICD-10 OMIM OMIM
ANALYTE(S):	CSPP1 KIAA0586
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