DISEASE:
Autosomal dominant polycystic kidney disease type 1 with tuberous sclerosis

NAME:	Autosomal dominant polycystic kidney disease type 1 with tuberous sclerosis
DESCRIPTION:	A rare contiguous gene syndrome involving a partial deletion of chromosome 16 and characterized by early-onset and severe polycystic kidney disease with various manifestations of tuberous sclerosis (multiple angiomyolipomas, lymphangioleiomyomatosis and periventricular calcifications of the central nervous system).
ORPHACODE:	88924
SYNONYMS:	PKDTS TSC2/PKD1 contiguous gene syndrome Tuberous sclerosis/polycystic kidney disease contiguous gene syndrome
XREF(S):	Orphanet OMIM ICD-10
ANALYTE(S):	TSC2 PKD1
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RELATED CONTENT

Related Genetic Tests

- Ciliopathy / polycystic kidney and liver diseases / ADTKD/ nephronophtisis / Bardet-Biedl syndromes and kidney cancers (gene panel)
- Polycystic kidney disease type 1 and 2

Related Laboratories

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

Related Analytes

- polycystin 1, transient receptor potential channel interacting
- TSC complex subunit 2

Related Gene Panels

- Ciliopathy, polycystic kidney and liver diseases, ADTKD, nephronophtisis, Bardet-Biedl syndromes and kidney cancers (146 genes) - IPG