

## Opis choroby \*

### Definicja

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).

### Dane

Klasyfikacja	Synonimy
Choroba	PKDTS
	Zespół przyległych genów twardzina układowa/wielotorbielowość nerek
	TSC2/PKD1 contiguous gene syndrome
	Tuberous sclerosis/polycystic kidney disease
	contiguous gene syndrome

Kod ORPHA	Kod OMIM	Kod ICD10
88924	600273	Q61.2

**Kod ICD11**  
LD2F.1Y

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### \*Źródło

orphanet