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
Choroba

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

Kod ORPHA
88924

Kod OMIM
600273

Kod ICD10
Q61.2

Kod ICD11
LD2F.1Y

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*Źródło

orphanet