

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

*Źródło

orphanet