Opis choroby *

Definicja

A form of autosomal dominant tubulointerstitial kidney disease (ADTKD) due to UMOD mutations that is clinically characterized by bland urinalysis (absence of blood or protein in the urine), chronic kidney disease (CKD) leading to end-stage kidney disease (ESKD) between 20 and 80 years, and gout occurring in 50% of affected individuals.

Dane

Klasyfikacja
Podtyp kliniczny

Synonimy
ADTKD-UMOD
Autosomalna dominująca kanalikowo-śródmiąższowa choroba nerek związana z UMOD
Familial juvenile hyperuricemic nephropathy type 1
MCKD2
Medullary cystic kidney disease type 2
UMOD-related ADTKD
Uromodulin-associated kidney disease

Kod ORPHA
88950

Kod OMIM
162000

Kod ICD10
Q61.5

Kod ICD11
GB82

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*Źródło

orphanet