Opis choroby *

Definicja

A rare autosomal dominant tubulointerstitial kidney (ADTKD) disease due to MUC1 mutations characterized clinically by a bland urinalysis (absence of blood or protein in the urine), and chronic kidney disease leading to end-stage kidney disease (ESKD) between 20 and 80 years.

Dane

Klasyfikacja
Podtyp kliniczny

Synonimy
ADTKD-MUC1
Autosomalna dominująca kanalikowo-śródmiąższowa choroba nerek związana z MUC1
MCKD1
MUC1-related medullary cystic kidney disease
MUCI-related ADTKD
Medullary cystic kidney disease type 1

Kod ORPHA
88949

Kod OMIM
174000

Kod ICD10
Q61.5

Kod ICD11
GB82

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*Źródło

orphanet