

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

*Źródło

orphanet