Opis choroby *

Definicja

A form of autosomal dominant tubulointerstitial kidney disease (ADTKD) due to variants in or whole gene deletions of HNF1B, which is characterized by chronic tubulo-interstitial nephritis, that manifests with nonsignificant urinalysis and slowly progressive renal failure. It can be associated with cystic kidney dysplasia, early onset diabetes and extrarenal manifestations.

Dane

Klasyfikacja
Podtyp kliniczny

Synonimy
ADTKD-HNF1B
Dysfunkcja nerek - cukrzyca o wczesnym poczatku
MODY5
Torbiele nerek - MODY
Zespół RCAD
Autosomalna dominująca kanalikowo-śródmiąższowa choroba nerek związana z HNF1B
HNF1B-MODY
HNF1B-related nephropathy
MODY5
Maturity-onset diabetes of the young type 5
RCAD syndrome
Renal cysts and diabetes syndrome
Renal dysfunction-early-onset diabetes syndrome

Kod ORPHA
93111

Kod OMIM
137920

Kod ICD10
N11.8

Kod ICD11
5A13.6

---

*Źródło

orphanet