

Opis choroby *

Definicja

A rare clinical variant of hereditary nephronophthisis characterized by reduced renal concentrating ability, chronic tubulointerstitial nephritis, cystic renal disease, and progression to end-stage renal disease (ESRD) before 3 years of age.

Dane

Klasyfikacja Podtyp kliniczny

Synonimy
Autosomal recessive infantile NPHP
Autosomal recessive childhood NPHP
Autosomal recessive nephronophthisis childhood
Autosomal recessive infantile nephronophthisis

Kod ORPHA
93591

Kod OMIM
602088

Kod ICD10
Q61.5

Kod ICD11

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*Źródło

orphanet