

Opis choroby *

Definicja

A rare ciliopathy characterized by the association of nephronophthisis and liver fibrosis. Renal manifestations include chronic renal failure, polyuria, polydipsia, anemia, as well as increased echogenicity on renal ultrasound and interstitial fibrosis and tubular dilation on biopsy. Hepatic involvement manifests as hepatosplenomegaly with extensive fibrosis, destruction of the bile ducts, and cholestasis. Mild psychomotor retardation and ocular symptoms, such as strabismus, nystagmus, retinal degeneration, and anisocoria, have been reported in some patients.

Dane

Klasyfikacja

Choroba

Synonimy

Boichis disease

Choroba Boichisa

Zespół nefronoftyzy i włóknienia wątroby

Nephronophthisis-hepatic fibrosis syndrome

Kod ORPHA

84081

Kod OMIM

613550

Kod ICD10

Q61.8

Kod ICD11

GB8Y

*Źródło

orphanet