

Opis choroby *

Definicja

A rare genetic hepatic disease characterized by multiple segmental cystic dilatations of both central and smaller peripheral bile ducts associated with congenital hepatic fibrosis. Age of symptom onset is variable, as is disease progression. Patients present recurrent cholangitis, hepatolithiasis, and cholecystolithiasis. Portal hypertension may appear later in the disease course, and the risk of developing cholangiocarcinoma is increased significantly. The syndrome is often associated with autosomal recessive polycystic kidney disease.

Dane

Klasyfikacja

Zespół wad wrodzonych

Kod ORPHA

480520

Kod OMIM

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Kod ICD10

Q44.6

Kod ICD11

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*Źródło

orphanet