

Opis choroby *

Definicja

Cholestasis-lymphedema syndrome is a rare genetic disorder characterized by neonatal intrahepatic cholestasis, often lessening and becoming intermittent with age, and severe chronic lymphedema which mainly affects the lower limbs. Patients often present with fat malabsorption leading to failure to thrive, fat soluble vitamin deficiency with bleeding, rickets, and neuropathy. In 25% of cases, cirrhosis occurs during childhood or later in life.

Dane

Klasyfikacja

Choroba

Synonimy

Agenaes syndrome

Zespół Aagenaesa

Kod ORPHA

1414

Kod OMIM

214900

Kod ICD10

Q82.0

Kod ICD11

DB99.6Y

*Źródło

orphanet