

Opis choroby *

Definicja

A rare metabolic liver disease characterized by progressive liver disease and early cirrhosis due to accumulation of toxic cholesterol metabolites, which are detectable in bile, plasma, and urine, in association with dental abnormalities such as general hypomineralization and enamel hypoplasia, as well as occurrence of supernumerary teeth. There have been no further descriptions in the literature since 1996.

Dane

Klasyfikacja

Choroba

Synonimy

Lyngstadaas syndrome

Zespół Lyngstadaasa

Kod ORPHA

3196

Kod OMIM

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Kod ICD10

K76.8

Kod ICD11

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*Źródło

orphanet