

Opis choroby *

Definicja

Cerebrotendinous xanthomatosis (CTX) is an anomaly of bile acid synthesis (see this term) characterized by neonatal cholestasis, childhood-onset cataract, adolescent to young adult-onset tendon xanthomata, and brain xanthomata with adult-onset neurologic dysfunction.

Dane

Klasyfikacja

Choroba

Synonimy

CTX

CTX

Niedobór 27-hydroksylazy sterolowej

Sterol 27-hydroxylase deficiency

Kod ORPHA

909

Kod OMIM

213700

Kod ICD10

E75.5

Kod ICD11

5C52.11

*Źródło

orphanet