

## 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

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#### \*Źródło

orphanet