DISEASE:
Cerebrotendinous xanthomatosis

NAME:	Cerebrotendinous xanthomatosis
DESCRIPTION:	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.
ORPHACODE:	909
SYNONYMS:	CTX Sterol 27-hydroxylase deficiency
XREF(S):	Orphanet OMIM
CREATED:	23 May 2018 - 17:24
CHANGED:	02 Aug 2018 - 08:54

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