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Cerebrotendinous xanthomatosis

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base.

Cerebrotendinous xanthomatosis. ORPHA:909

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.