

Supplementary Table 3. Full-texts prioritised from the TLR to inform Delphi panel questionnaires

Author (Year)	Title
Amador <i>et al.</i> (2018)	Treatment with chenodeoxycholic acid in cerebrotendinous xanthomatosis: clinical, neurophysiological, and quantitative brain structural outcomes
Batta <i>et al.</i> (2004)	Hydrophilic 7 β -hydroxy bile acids, lovastatin, and cholestyramine are ineffective in the treatment of cerebrotendinous xanthomatosis
Beppu <i>et al.</i> (1982)	Serum bile acid profiles in cerebrotendinous xanthomatosis
Berginer <i>et al.</i> (1982)	EEG in cerebrotendinous xanthomatosis (CTX)
Berginer <i>et al.</i> (1984)	Long-term treatment of cerebrotendinous xanthomatosis with chenodeoxycholic acid
Bleye <i>et al.</i> (2016)	Update on newborn dried bloodspot testing for cerebrotendinous xanthomatosis: an available high-throughput liquid-chromatography tandem mass spectrometry method
DeBarber <i>et al.</i> (2018)	Newborn screening for cerebrotendinous xanthomatosis is the solution for early identification and treatment
DeBarber <i>et al.</i> (2014)	A blood test for cerebrotendinous xanthomatosis with potential for disease detection in newborns
Duell <i>et al.</i> (2018)	Diagnosis, treatment, and clinical outcomes in 43 cases with cerebrotendinous xanthomatosis
Ginanneschi <i>et al.</i> (2013)	Polyneuropathy in cerebrotendinous xanthomatosis and response to treatment with chenodeoxycholic acid
Inglese <i>et al.</i> (2003)	Quantification of brain damage in cerebrotendinous xanthomatosis with magnetization transfer MR imaging
Köhler <i>et al.</i> (2018)	Adulthood leukodystrophies
Mascalchi and Vella. (2018)	Neuroimaging Applications in Chronic Ataxias
Mignarri <i>et al.</i> (2017)	The spectrum of magnetic resonance findings in cerebrotendinous xanthomatosis: redefinition and evidence of new markers of disease progression
Mignarri <i>et al.</i> (2014)	A suspicion index for early diagnosis and treatment of cerebrotendinous xanthomatosis

Author (Year)	Title
Mignarri <i>et al.</i> (2016)	Evaluation of cholesterol metabolism in cerebrotendinous xanthomatosis
Mignarri <i>et al.</i> (2011)	Clinical relevance and neurophysiological correlates of spasticity in cerebrotendinous xanthomatosis
Nie <i>et al.</i> (2014)	Cerebrotendinous xanthomatosis: a comprehensive review of pathogenesis, clinical manifestations, diagnosis, and management
Pilo-de-la-Fuente <i>et al.</i> (2011)	Cerebrotendinous xanthomatosis in Spain: clinical, prognostic, and genetic survey
Pitt (2007)	High-throughput urine screening for Smith–Lemli–Opitz syndrome and cerebrotendinous xanthomatosis using negative electrospray tandem mass spectrometry
Salen <i>et al.</i> (1991)	Biochemical abnormalities in cerebrotendinous xanthomatosis
Salen and Steiner. (2017)	Epidemiology, diagnosis, and treatment of cerebrotendinous xanthomatosis (CTX)
Stelten <i>et al.</i> (2018)	Autism spectrum disorder: an early and frequent feature in cerebrotendinous xanthomatosis
Stelten <i>et al.</i> (2018)	Movement disorders in cerebrotendinous xanthomatosis
Vaz <i>et al.</i> (2017)	A newborn screening method for cerebrotendinous xanthomatosis using bile alcohol glucuronides and metabolite ratios
Verrips <i>et al.</i> (2000)	Presence of diarrhea and absence of tendon xanthomas in patients with cerebrotendinous xanthomatosis
Verrips <i>et al.</i> (1999)	Effect of simvastatin in addition to chenodeoxycholic acid in patients with cerebrotendinous xanthomatosis
Waterreus <i>et al.</i> (1987)	Cerebrotendinous xanthomatosis (CTX): a clinical survey of the patient population in The Netherlands
Yahalom <i>et al.</i> (2013)	Neurological outcome in cerebrotendinous xanthomatosis treated with chenodeoxycholic acid: early versus late diagnosis
Zubarioglu <i>et al.</i> (2017)	Early diagnosed cerebrotendinous xanthomatosis patients: clinical, neuroradiological characteristics and therapy results of a single center from Turkey

CTX: Cerebrotendinous xanthomatosis; EEG: Electroencephalogram; MR: Magnetic resonance.