

Disease

Cerebrotendinous xanthomatosis¹

Gene

CYP27A1

Inheritance	AR
Current Treatments	chenodeoxycholic acid and Ctexli (chenodiol)
Evidence for Treatment	expert opinion
Non-Molecular Testing Availability	yes
Non-Molecular Test	cholestanol level ²
ClinGen	https://actionability.clinicalgenome.org/ac/ui/Pediatric/ui/stg2SummaryRpt/AC1084
Disease Frequency (per 100,000)	0.48 ³
Genes Included in Disease Frequency	CYP27A1
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=Cerebrotendinous+xanthomatosis&term=&cntry=&state=&city=

References

- ¹ <https://www.ncbi.nlm.nih.gov/books/NBK1409/#ctx.Summary>
- ² <https://www.ncbi.nlm.nih.gov/books/NBK1409/#ctx.Diagnosis>
- ³ <https://www.ncbi.nlm.nih.gov/pubmed/26643207>