

Disease

Congenita bile acid synthesis defect type 6^{1, 2}

Gene

ACOX2

Inheritance	AR
Current Treatments	ursodeoxycholic acid
Evidence for Treatment	expert opinion
Non-Molecular Testing Availability	yes
Non-Molecular Test	urine fast atom bombardment ionization mass spectrometry (FAB-MS) analysis ²
Experimental / Investigational Therapies	https://clinicaltrials.gov/search?cond=Bile%20Acid%20Synthesis%20Disorder&city=

References

¹ <https://omim.org/entry/617308>

² <https://pubmed.ncbi.nlm.nih.gov/35395098/>