

Disease

Congenital bile acid synthesis defect type 4^{1, 2, 3}

Gene

AMACR

Inheritance	AR
Current Treatments	cholic acid, restriction of phytanic and pristanic acids
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/ct2/results?cond=Bile+Acid+Synthesis+Disorder&term=&cntry=&state=&city=&di

References

- ¹ <https://omim.org/entry/214950>
- ² <https://pubmed.ncbi.nlm.nih.gov/12512044/>
- ³ <https://pubmed.ncbi.nlm.nih.gov/34267495/>