

Disease

Bile acid conjugation defect 1¹

Gene

BAAT

Inheritance	AR
Current Treatments	ursodeoxycholic acid or liver transplant
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%20conjugation%20Disorder&viewType=Table

References

¹ <https://omim.org/entry/619232>

² <https://pubmed.ncbi.nlm.nih.gov/23415802/>