

Test Definition: CTXWB

Cerebrotendinous Xanthomatosis, Blood

Reporting Title: Cerebrotendinous Xanthomatosis, B

Performing Location: Rochester

Ordering Guidance:

For assessment of bile acid malabsorption in patients with irritable bowel syndrome-diarrhea, order 7AC4 / 7AC4, Bile Acid Synthesis, Serum.

This test is also available as a part of a panel; see HSMWB / Hepatosplenomegaly Panel, Blood. If this test (CTXWB) is ordered with either GPSYW / Glucopsychosine, Blood or OXYWB / Oxysterols, Blood, the individual tests will be canceled and HSMWB ordered.

Specimen Requirements:

Container/Tube:

Preferred: Lavender top (EDTA)

Acceptable: Green top (sodium heparin, lithium heparin), yellow top (ACD B)

Specimen Volume: 1 mL

Collection Instructions: Send whole blood in original vial. Do not aliquot.

Forms:

- 1. Biochemical Genetics Patient Information (T602)
- 2. If not ordering electronically, complete, print, and send a <u>Biochemical Genetics Test Request</u> (T798) with the specimen.

Specimen Type	Temperature	Time	Special Container
Whole blood	Refrigerated (preferred)	72 hours	
	Ambient	48 hours	

Result Codes:

Result ID	Reporting Name	Туре	Unit	LOINC®
BA4365	Interpretation (CTXWB)	Alphanumeric		59462-2
BA4363	7a-hydroxy-4-cholesten-3-one	Numeric	nmol/mL	92762-4
BA4364	7a,12a-dihydroxycholest-4-en-3-one	Numeric	nmol/mL	92759-0
BA4366	Reviewed By	Alphanumeric		18771-6

LOINC® and CPT codes are provided by the performing laboratory.

Supplemental Report:

No

CPT Code Information:

82542

Reference Values:



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7-ALPHA-HYDROXY-4-CHOLESTEN-3-ONE (7a-C4)

Cutoff: < or =0.750 nmol/mL

7-ALPHA,12-ALPHA-DIHYDROXYCHOLEST-4-en-3-ONE (7a12aC4)

Cutoff: < or =0.250 nmol/mL