

Reporting Title: Cerebrotendinous Xanthomatosis, P

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 HSMP / Hepatosplenomegaly Panel, Plasma. If this test (CTXP) is ordered with either GPSYP / Glucopsychosine, Plasma or OXNP / Oxysterols, Plasma, the individual tests will be canceled and HSMP ordered.

Specimen Requirements:

Collection Container/Tube:

Preferred: Lavender top (EDTA)

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

Submission Container/Tube: Plastic vial

Specimen Volume: 0.3 mL

Collection Instructions:

1. Centrifuge at 4 degrees C if possible
2. Aliquot plasma into a plastic vial. **Do not disturb or transfer the buffy coat layer.**
3. Send frozen.

Forms:

1. [Biochemical Genetics Patient Information](#) (T602)
2. If not ordering electronically, complete, print, and send a [Biochemical Genetics Test Request](#) (T798) with the specimen.

Specimen Type	Temperature	Time	Special Container
Plasma	Frozen	65 days	

Result Codes:

Result ID	Reporting Name	Type	Unit	LOINC®
BA4379	Interpretation (CTXP)	Alphanumeric		59462-2
BA4376	7a-hydroxy-4-cholesten-3-one	Numeric	nmol/mL	92761-6
BA4377	7a,12a-dihydroxycholest-4-en-3-one	Numeric	nmol/mL	92758-2
BA4378	Reviewed By	Alphanumeric		18771-6

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

Supplemental Report:

No

CPT Code Information:

82542

Reference Values:

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

Cutoff: < or =0.300 nmol/mL

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

Cutoff: < or =0.100 nmol/mL