

Reporting Title: Cerebrotendinous Xanthomatosis, BS**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 HSMBS / Hepatosplenomegaly Panel, Blood Spot. If this test (CTXBS) is ordered with either GPSY / Glucopsychosine, Blood Spot or OXYBS / Oxysterols, Blood Spot, the individual tests will be canceled and HSMBS ordered.

Specimen Requirements:

Supplies:

- Card-Blood Spot Collection (Filter Paper) (T493)
- Card-Postmortem Screening (Filter Paper) (T525)

Container/Tube:

Preferred: Blood Spot Collection Card (Filter Paper)

Acceptable: Whatman Protein Saver 903 filter paper, PerkinElmer 226 filter paper, Munktell filter paper, Postmortem Screening Card or collected with EDTA, sodium heparin, lithium heparin, or ACD B-containing devices

Specimen Volume: 2 Blood spots

Collection Instructions:

1. Let blood dry completely on filter paper at ambient temperature in a horizontal position for a minimum of 3 hours.
2. At least 1 spot should be complete (ie, unpunched).
3. Do not expose specimen to heat or direct sunlight.
4. Do not stack wet specimens.
5. Keep specimen dry.

Additional Information:

1. For collection instructions, see Blood Spot Collection Instructions
2. For collection instructions in Spanish, see Blood Spot Collection Card-Spanish Instructions (T777)
3. For collection instructions in Chinese, see Blood Spot Collection Card-Chinese Instructions (T800)

Specimen Minimum Volume:

1 Blood spot

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
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Whole blood	Refrigerated (preferred)	10 days	FILTER PAPER
	Frozen	59 days	FILTER PAPER
	Ambient	10 days	FILTER PAPER

Result Codes:

Result ID	Reporting Name	Type	Unit	LOINC®
BA4361	Interpretation (CTXBS)	Alphanumeric		59462-2
BA4359	7a-hydroxy-4-cholesten-3-one	Numeric	nmol/mL	92763-2
BA4360	7a,12a-dihydroxycholest-4-en-3-one	Numeric	nmol/mL	92760-8
BA4362	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 =0.750 nmol/mL

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

Cutoff: < or =0.250 nmol/mL