

**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)

**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
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