

Overview**Useful For**

Establishing a diagnosis of lecithin-cholesterol acyltransferase deficiency

Evaluating the extent of metabolic disturbance by bile stasis or liver disease

Method Name

EnzymaticColorimetric

NY State Available

Yes

Specimen**Specimen Type**

Serum

Specimen Required**Collection Container/Tube:**

Preferred: Serum gel

Acceptable: Red top

Submission Container/Tube: Plastic vial

Specimen Volume: 1 mL

Collection Instructions: Centrifuge and aliquot serum into plastic vial. Send refrigerated.

Forms

If not ordering electronically, complete, print, and send a [Cardiovascular Test Request Form](#) (T724) with the specimen.

Specimen Minimum Volume

0.5 mL

Reject Due To

Gross hemolysis	Reject
Gross lipemia	OK
Gross icterus	Reject

Specimen Stability Information

Specimen Type	Temperature	Time	Special Container
Serum	Refrigerated (preferred)	7 days	

Specimen Type	Temperature	Time	Special Container
	Frozen	60 days	
	Ambient	24 hours	

Clinical and Interpretive

Clinical Information

Cholesterol in the blood serum is normally 60% to 80% esterified with fatty acids, largely as a result of the action of the enzyme lecithin-cholesterol acyltransferase (LCAT), which circulates in the blood in association with the high-density lipoproteins.

Familial deficiency of LCAT is uncommon, usually occurring individuals of northern Europe descent, and is associated with erythrocyte abnormalities (target cells) and decreased (20% or less) esterification of serum cholesterol. This is associated with early atherosclerosis, corneal opacification, hyperlipidemia, and mild hemolytic anemia.

Persons with liver disease may have impaired formation of LCAT and, therefore, an acquired LCAT deficiency and reduced cholesterol ester concentration.

Reference Values

> or =18 years: 60-80% of total cholesterol

Reference values have not been established for patients who are less than 18 years of age.

Interpretation

In patients with lecithin-cholesterol acyltransferase deficiency, the concentration of unesterified cholesterol in serum may increase 2 to 5 times the normal value, resulting in a decrease in esterified serum cholesterol to 20% or less of the total serum cholesterol.

Cautions

In a severe form, cholesteryl ester storage disease can be fatal before the age of 1 year (Wolman's disease) and, in some less severe form cases, may be undetected until adulthood. This defect is caused by a deficiency of a lysosomal enzyme, acid cholesteryl ester hydrolase (also known as acid lipase). It causes accumulation of cholesteryl esters in tissues, but it has no effect on the percentage of cholesterol that circulates in esterified form in the blood serum. Detection of the defect requires careful evaluation of the cholesteryl ester hydrolase activity and cholesteryl ester content of leukocytes, cultured fibroblasts, and liver biopsy.

Result can be falsely decreased in patients with elevated levels of N-acetyl-p-benzoquinone imine (NAPQI)-a metabolite of acetaminophen, N-acetylcysteine (NAC), and metamizole.

Clinical Reference

1. Meikle PJ, Mundra PA, Wong G, et al: Circulating lipids are associated with alcoholic liver cirrhosis and represent potential biomarkers for risk assessment. *PLoS One*. 2015 Jun 24;10(6):e0130346. doi: 10.1371/journal.pone.0130346
2. Leach NV, Dronca E, Vesa SC, et al: Serum homocysteine levels, oxidative stress and cardiovascular risk in non-alcoholic steatohepatitis. *Eur J Intern Med*. 2014 Oct;25(8):762-767. doi: 10.1016/j.ejim.2014.09.007

3. Santamarina-Fojo S, Hoeg JM, Assmann G, Brewer B: Lecithin cholesterol acyltransferase deficiency and fish eye disease. In: Valle DL, Antonarakis S, Ballabio A, Beaudet AL, Mitchell GA. eds. The Online Metabolic and Molecular Bases of Inherited Disease. McGraw-Hill; 2019. Accessed June 8, 2021. Available at <https://ommbid.mhmedical.com/content.aspx?sectionid=225539713&bookid=2709>

Performance

Method Description

Free cholesterol reacts with cholesterol oxidase to generate hydrogen peroxide which reacts with 3,5-dimethoxy-*N*-(2-hydroxy-3-sulfopropyl) aniline sodium (DAOS) and 4-aminoantipyrene to produce a blue pigment. The product is proportional to the serum free cholesterol concentration. (Fujifilm Free Cholesterol E. Fujifilm Wako Pure Chemical Corporation; 04/01/2018)

PDF Report

No

Day(s) Performed

Monday through Friday

Report Available

1 to 3 days

Specimen Retention Time

7 days

Performing Laboratory Location

Rochester

Fees and Codes

Fees

- Authorized users can sign in to [Test Prices](#) for detailed fee information.
- Clients without access to Test Prices can contact [Customer Service](#) 24 hours a day, seven days a week.
- Prospective clients should contact their Regional Manager. For assistance, contact [Customer Service](#).

Test Classification

This test was developed, and its performance characteristics determined by Mayo Clinic in a manner consistent with CLIA requirements. This test has not been cleared or approved by the US Food and Drug Administration.

CPT Code Information

84311

LOINC® Information

Test ID	Test Order Name	Order LOINC Value
CHLE	Cholesteryl Esters, S	21197-9

Result ID	Test Result Name	Result LOINC Value
CHLES	Cholesteryl Esters, S	21197-9