

Overview

Useful For

Confirmation of diagnosis of carnitine palmitoyltransferase II deficiency

Carrier screening in cases where there is a family history of carnitine palmitoyltransferase II deficiency, but disease-causing mutations have not been identified in an affected individual

Reflex Tests

Test ID	Reporting Name	Available Separately	Always Performed
CULFB	Fibroblast Culture for Genetic Test	Yes	No

Testing Algorithm

If skin biopsy is received, fibroblast culture will be added and charged separately.

Special Instructions

- [Molecular Genetics: Biochemical Disorders Patient Information](#)
- [Informed Consent for Genetic Testing](#)
- [Blood Spot Collection Card-Spanish Instructions](#)
- [Blood Spot Collection Card-Chinese Instructions](#)
- [Informed Consent for Genetic Testing \(Spanish\)](#)
- [Blood Spot Collection Instructions](#)

Method Name

Polymerase Chain Reaction (PCR) Followed by DNA Sequence Analysis

NY State Available

Yes

Specimen

Specimen Type

Varies

Shipping Instructions

Specimen preferred to arrive within 96 hours of draw.

Specimen Required

Patient Preparation: A previous bone marrow transplant from an allogenic donor will interfere with testing. Call 800-533-1710 for instructions for testing patients who have received a bone marrow transplant.

Submit only 1 of the following specimens:

Preferred:

Specimen Type: Whole blood

Container/Tube:

Preferred: Lavender top (EDTA) or yellow top (ACD)

Acceptable: Any anticoagulant

Specimen Volume: 3 mL

Collection Instructions:

1. Invert several times to mix blood.
2. Send specimen in original tube.

Specimen Stability Information: Ambient (preferred)/Refrigerated

Acceptable:

Specimen Type: Cultured fibroblasts

Container/Tube: T-75 or T-25 flask

Specimen Volume: 1 Full T-75 flask or 2 full T-25 flasks

Specimen Stability Information: Ambient (preferred)/Refrigerated <24 hours

Specimen Type: Skin biopsy

Supplies: Fibroblast Biopsy Transport Media (T115)

Container/Tube: Sterile container with any standard cell culture media (eg, minimal essential media, RPMI 1640). The solution should be supplemented with 1% penicillin and streptomycin. Tubes can be supplied upon request (Eagle's minimum essential medium with 1% penicillin and streptomycin, T115).

Specimen Volume: 4-mm punch

Specimen Stability Information: Refrigerated (preferred)/Ambient

Specimen Type: Blood spot

Supplies: Card - Blood Spot Collection (Filter Paper) (T493)

Container/Tube:

Preferred: Collection card (Whatman Protein Saver 903 Paper)

Acceptable: Ahlstrom 226 filter paper, or Blood Spot Collection Card (T493)

Specimen Volume: 2 to 5 Blood spots on collection card (Whatman Protein Saver 903 Paper; Ahlstrom 226 filter

paper; or Blood Spot Collection Card, T493)

Collection Instructions:

1. An alternative blood collection option for a patient >1 year of age is finger stick.
2. Let blood dry on the filter paper at ambient temperature in a horizontal position for 3 hours.
3. Do not expose specimen to heat or direct sunlight.
4. Do not stack wet specimens.
5. Keep specimen dry.

Specimen Stability Information: Ambient (preferred)/Refrigerated

Additional Information:

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

Forms

1. New York Clients-Informed consent is required. Document on the request form or electronic order that a copy is on file. The following documents are available in Special Instructions:

-[Informed Consent for Genetic Testing](#) (T576)

-[Informed Consent for Genetic Testing-Spanish](#) (T826)

2. [Molecular Genetics: Biochemical Disorders Patient Information](#) (T527) in Special Instructions

3. If not ordering electronically, complete, print, and send an [Inborn Errors of Metabolism Test Request](#) (T798) with the specimen.

Specimen Minimum Volume

Blood: 1 mL

Blood Spots: 5 punches-3 mm diameter

Reject Due To

All specimens will be evaluated at Mayo Clinic Laboratories for test suitability.

Specimen Stability Information

Specimen Type	Temperature	Time	Special Container
Varies	Varies		

Clinical and Interpretive

Clinical Information

Carnitine palmitoyltransferase II (CPT II) deficiency is an autosomal recessive disorder of long-chain fatty-acid oxidation. There are 3 distinct clinical phenotypes: a lethal neonatal form, an early-onset infantile form, and a late-onset adult myopathic form. The lethal neonatal and early-onset infantile forms are characterized by liver failure, cardiomyopathy, seizures, hypoketotic hypoglycemia, peripheral myopathy and early death. The adult-onset myopathic form is the most common type and is characterized by exercise-induced muscle pain and weakness and may be associated with myoglobinuria. Males are more likely to be affected with the myopathic form than females.

Initial screening can be done with plasma acylcarnitines. Definitive diagnosis can be made by detection of reduced CPT enzyme activity. Mutations in the *CPT2* gene are responsible for CPT II deficiency and sequencing of this gene is recommended after positive biochemical analysis.

Reference Values

An interpretive report will be provided.

Interpretation

All detected alterations are evaluated according to American College of Medical Genetics recommendations.⁽¹⁾ Variants are classified based on known, predicted, or possible pathogenicity and reported with interpretive comments detailing their potential or known significance.

Cautions

[A small percentage of individuals who are carriers or have a diagnosis carnitine palmitoyltransferase II \(CPT II\) deficiency may have a mutation that is not identified by this method \(eg, promoter and deep intronic mutations\). The absence of a mutation, therefore, does not eliminate the possibility of positive carrier status or the diagnosis of CPT II deficiency. For carrier testing, it is important to first document the presence of a CPT2 gene mutation in an affected family member.](#)

In some cases, DNA alterations of undetermined significance may be identified.

Rare polymorphisms exist that could lead to false-negative or false-positive results. If results obtained do not match the clinical and biochemical findings, additional testing should be considered.

Test results should be interpreted in the context of clinical findings, family history, and other laboratory data. Errors in our interpretation of results may occur if information given is inaccurate or incomplete.

Clinical Reference

1. Richards S, Aziz N, Bale S, et al: Standards and guidelines for the interpretation of sequence variants: a joint consensus recommendation of the American College of Medical Genetics and Genomics and the Association for Molecular Pathology. *Genet Med* 2015 May;17(5):405-424
2. Bonnefont JP, Djouadi F, Prip-Buus C, et al: Carnitine palmitoyltransferases 1 and 2: biochemical, molecular and medical aspects. *Mol Aspects Med* 2004 Oct-Dec;25(5-6):495-520
3. Siguake E, Rakheja D, Kitson K, Bennet M: Carnitine palmitoyltransferase II deficiency: a clinical, biochemical, and molecular review. *Lab Invest* 2003 Nov;83(11):1543-1554

Performance

Method Description

Bidirectional sequence analysis is performed to test for the presence of a mutation in all coding regions and intron/exon boundaries of the *CPT2* gene. (Unpublished Mayo method)

PDF Report

No

Day(s) and Time(s) Test Performed

Performed weekly, Varies

Analytic Time

14 days

Maximum Laboratory Time

20 days

Specimen Retention Time

Whole Blood: 2 weeks (if available) Extracted DNA: 3 months

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 U.S. Food and Drug Administration.

CPT Code Information

81404-CPT2 (carnitine palmitoyltransferase 2) (eg, carnitine palmitoyltransferase II deficiency), full gene sequence

Fibroblast Culture for Genetic Test

88233-Tissue culture, skin or solid tissue biopsy (if appropriate)

88240-Cryopreservation (if appropriate)

LOINC® Information

Test ID	Test Order Name	Order LOINC Value
CPT2Z	CPT2 Gene, Full Gene Analysis	94195-5

Result ID	Test Result Name	Result LOINC Value
53887	Result Summary	50397-9
53888	Result	82939-0
53889	Interpretation	69047-9
53890	Additional Information	48767-8
53891	Specimen	31208-2
53892	Source	31208-2
53893	Released by	18771-6