

ACT Sheet

Newborn Screening ACT Sheet [Elevated CO/C16+C18 Acylcarnitine] Carnitine Palmitoyltransferase I Deficiency (CPT I)

Differential Diagnosis: Carnitine palmitoyltransferase I (CPT I); carnitine supplementation.

Condition Description: Carnitine palmitoyltransferase I (CPT I) deficiency disrupts an early component of the carnitine shuttle, the system used to bring long-chain fatty acids into mitochondria for energy production. A deficiency of CPT I activity results in the accumulation of free carnitine and the inhibition of the fatty acid oxidation response necessary to generate energy during fasting and periods of increased energy needs, such as fever or stress. Presentation in the neonatal period is rare.

You Should Take the Following <u>IMMEDIATE</u> Actions:

- Inform family of the newborn screening result.
- Ascertain clinical status (lethargy, seizures).
- Consult with pediatric metabolic specialist the same day.
- Evaluate the newborn (lethargy, hepatosplenomegaly, or seizures). If any of these findings are present or if the newborn is ill, transport to a hospital for further treatment in consultation with the metabolic specialist.
- Initiate confirmatory/diagnostic testing and management, as recommended by specialist.
- Provide the family with basic information about CPT I and its management.
- Report final diagnostic outcome to newborn screening program.

Diagnostic Evaluation: <u>Plasma carnitine (free and total)</u>: Free carnitine is elevated in CPT I deficiency. <u>Dried blood</u> <u>spot or plasma acylcarnitines</u>: Free carnitine is elevated and long-chain acylcarnitines are decreased or are normal. <u>Molecular genetic testing</u> may be required to establish the diagnosis.

Clinical Considerations: CPT I deficiency can have a variable presentation. Critical hypoketotic hypoglycemia leading to lethargy, hepatomegaly, and seizures is a common presenting feature usually precipitated by fasting or acute illness; this presentation is rare in newborns. The Arctic *CPT1* variant is common among the Inuit population of Alaska, Canada, and Greenland. Most infants with this variant remain asymptomatic; those who do develop symptoms usually do so only early in life during periods of fasting or acute illness.

Additional Information:

How to Communicate Newborn Screening Results Emergency Protocols (New England Consortium of Metabolic Programs) Gene Reviews Medline Plus Condition Information for Families- HRSA Newborn Screening Clearinghouse

Referral (local, state, regional, and national):

Find a Genetics Clinic Directory Genetic Testing Registry

This practice resource is designed primarily as an educational resource for medical geneticists and other clinicians to help them provide quality medical services. Adherence to this practice resource is completely voluntary and does not necessarily assure a successful medical outcome. This practice resource should not be considered inclusive of all proper procedures and tests or exclusive of other procedures and tests that are reasonably directed to obtaining the same results. In determining the propriety of any specific procedure or test, the clinician should apply his or her own professional judgment to the specific clinical circumstances presented by the individual patient or specimen. Clinicians are encouraged to document the reasons for the use of a particular procedure or test, whether or not it is in conformance with this practice resource. Clinicians also are advised to take notice of the date this practice resource was adopted, and to consider other medical and scientific information that becomes available after that date. It also would be prudent to consider whether intellectual property interests may restrict the performance of certain tests and other procedures.

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Local Resources (Insert Local Website Links) State Resource Site (Insert Website Information)

Name	
URL	
Comments	

Local Resource Site (Insert Website Information)

Name	
URL	
Comments	

Appendix (Resources with Full URL Addresses)

Additional Information

How to Communicate Newborn Screening Results

• https://www.hrsa.gov/sites/default/files/hrsa/advisory-committees/heritable-disorders/Resources/achdnccommunication-guide-newborn.pdf

Emergency Protocols (New England Consortium of Metabolic Programs)

• https://www.newenglandconsortium.org/cpt-i-deficiency

Gene Reviews

<u>https://www.ncbi.nlm.nih.gov/books/NBK1527/</u>

Medline Plus

<u>https://medlineplus.gov/genetics/condition/carnitine-palmitoyltransferase-i-deficiency/</u>

Condition Information for Families-HRSA Newborn Screening Clearinghouse

<u>https://newbornscreening.hrsa.gov/conditions/carnitine-palmitoyltransferase-i-deficiency</u>

Referral (local, state, regional and national)

Find a Genetics Clinic Directory

https://clinics.acmg.net

Genetic Testing Registry

• <u>https://www.ncbi.nlm.nih.gov/gtr/</u>

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