

Newborn Screening ACT Sheet [Elevated C0/C16+C18] Carnitine Palmitoyl Transferase 1 Deficiency (CPT1)

Differential Diagnosis: Carnitine palmitoyl transferase 1 deficiency (CPT1).

Condition Description: This disorder is caused by a deficiency of the enzyme CPT1, preventing the fatty acid carnitine-acylcarnitine linkage required to transport fatty acids into the mitochondria. This results in accumulation of free carnitine (C0) and prevents the fatty acid oxidation response necessary to generate energy during fasting and increased energy needs (fever, stress).

YOU SHOULD TAKE THE FOLLOWING ACTIONS:

- Contact family to inform them of the newborn screening result and ascertain clinical status (lethargy, seizures).
- Consult with pediatric metabolic specialist.
- Evaluate the newborn (lethargy, hepatomegaly, seizures); initiate emergency treatment as indicated by metabolic specialist.
- Initiate timely confirmatory/diagnostic testing as recommended by specialist.
- Educate family about signs, symptoms and need for urgent treatment of hypoglycemia (lethargy, seizures).
- Report findings to newborn screening program.

Diagnostic Evaluation: Plasma acylcarnitine showing elevated free carnitine C0 with low or normal long-chain acylcarnitines. CPT1 enzyme assays and CPT1A gene sequencing establish the diagnosis.

Clinical Considerations: CPT1 can have a variable presentation. Critical hypoketotic hypoglycemia is a common presenting feature. Newborns may appear asymptomatic but can progress to fasting hypoketotic hypoglycemia, lethargy, hepatomegaly, and seizures, usually precipitated by fasting or acute illness.

Additional Information:

[Gene Reviews](#)

[Genetics Home Reference](#)

Referral (local, state, regional and national):

[Testing](#)

[Clinical Services](#)

[Find Genetic Services](#)

Disclaimer: This guideline is designed primarily as an educational resource for clinicians to help them provide quality medical care. It 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. Adherence to this guideline does not necessarily ensure a successful medical outcome. 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 guideline. Clinicians also are advised to take notice of the date this guideline was adopted, and to consider other medical and scientific information that become available after that date.

LOCAL RESOURCES: Insert State newborn screening program web site links

State Resource site (insert state newborn screening program website information)

Name	<input type="text"/>
URL	<input type="text"/>
Comments	<input type="text"/>

Local Resource Site (insert local and regional newborn screening website information)

Name	<input type="text"/>
URL	<input type="text"/>
Comments	<input type="text"/>

APPENDIX: Resources with Full URL Addresses

Additional Information:

Gene Reviews

<http://www.ncbi.nlm.nih.gov/bookshelf/br.fcgi?book=gene&part=cpt1a>

Genetics Home Reference

<http://ghr.nlm.nih.gov/gene=cpt1a>

Referral (local, state, regional and national):

Testing

http://www.ncbi.nlm.nih.gov/sites/GeneTests/lab/clinical_disease_id/3164?db=genetests&country=United%20States

Clinical Services

<http://www.ncbi.nlm.nih.gov/sites/genetests/clinic?db=genetests>

Find Genetic Services

<http://www.acmg.net/GIS/Disclaimer.aspx>

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