# Newborn Screening ACT Sheet
# [Elevated C16 and/or C18:1 Acylcarnitine]
## Carnitine Palmitoyltransferase 2 (CPT2) Deficiency

**Differential Diagnosis:** Carnitine palmitoyltransferase (CPT2) deficiency Carnitine/acylcarnitine translocase (CACT) deficiency;

**Condition Description:** In both the translocase and CPT2 deficiencies, the acylcarnitines cannot be transported into the mitochondria for fatty acid oxidation. Thus, the need for generation of energy from fatty acids during fasting or increased demand (fever, stress) cannot be met. In addition, the neonatal form of CPT2 deficiency is associated with multiple congenital anomalies.

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*YOU SHOULD TAKE THE FOLLOWING ACTIONS:*

- Contact family to inform them of the newborn screening result and ascertain clinical status (poor feeding, vomiting, lethargy).
- Evaluate infant (hepatomegaly, cardiac insufficiency; history of sudden unexpected death in a sibling; dysmorphic facies).
- Consult/refer to a metabolic specialist to determine appropriate follow-up.
- Emergency treatment if symptomatic and/or hypoglycemia is present.
- Report findings to newborn screening program.

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**Diagnostic Evaluation:** Plasma acylcarnitine analysis reveals increased C16 and/or C18:1. Urine organic acid analysis reveals increased lactic acid and dicarboxylic acids.

**Clinical Considerations:** In the neonatal form of CPT2 deficiency, the neonate is profoundly ill with marked hypoglycemia, metabolic acidosis, cardiac arrhythmias, and facial dysmorphism. Only rarely will these infants survive. In the later-onset muscular form of CPT2 deficiency, the neonate is asymptomatic but muscle disease develops in the adolescent or adult years. Translocase deficiency presents similarly to the neonatal form of CPT2 deficiency.

**Additional Information:**

Gene Tests

**Genetics Home Reference**

CPT2

CACT

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

Testing

Clinical Services

Find Genetic Services

*LOCAL RESOURCES:* Insert State newborn screening program web site links

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

| | |
|---|---|
| Name | |
| URL | |
| Comments | |

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

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|---|---|
| Name | |
| URL | |
| Comments | |

APPENDIX: Resources with Full URL Addresses

*Additional Information:*

**Gene Tests**
http://www.ncbi.nlm.nih.gov/bookshelf/br.fcgi?book=gene&part=cpt2

**Genetics Home Reference**
**CPT2** http://ghr.nlm.nih.gov/condition=carnitinepalmitoyltransferaseiideficiency

**CACT** http://ghr.nlm.nih.gov/condition=carnitineacylcarnitinetranslocasedeficiency

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

**Testing**
http://www.ncbi.nlm.nih.gov/sites/GeneTests/lab/clinical_disease_id/3064?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