

Newborn Screening ACTSheet [Increased C16 and/or C18:1 Acylcarnitine]

Carnitine Palmitoyltransferase II (CPT II) Deficiency and Carnitine Acylcarnitine Translocase (CACT) Deficiency

Differential Diagnosis: Carnitine palmitoyltransferase II (CPT II) deficiency; carnitine acylcarnitine translocase (CACT) deficiency.

Condition Description: Carnitine palmitoyltransferase II (CPT II) deficiency and carnitine acylcarnitine translocase (CACT) deficiency result from defects in the transport of long-chain acylcarnitines into the mitochondria for fatty acid oxidation. As a result, the energy needs derived from fatty acids during fasting or periods of increased demand (e.g., fever, stress) cannot be met. In addition, the neonatal form of CPT II deficiency is associated with multiple congenital anomalies and other organ system involvement, including heart and liver.

You Should Take the Following *IMMEDIATE* Actions:

- Inform family of newborn screening result.
- Ascertain clinical status (poor feeding, vomiting, lethargy); elicit history of sudden unexpected death in a sibling.
- Consult with pediatric metabolic specialist the same day.
- Evaluate the newborn for signs of acute illness such as hypoglycemia, hepatic dysfunction, cardiac insufficiency, 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 a metabolic specialist.
- Initiate confirmatory/diagnostic testing and management
- Provide the family with basic information about CPT II and/or CACT including possible early signs and symptoms.
- Report final diagnostic outcome to newborn screening program.

Diagnostic Evaluation: **Plasma acylcarnitines:** C16 and/or C18:1 are elevated in both CPT II and CACT deficiencies. **Molecular genetic testing** is required to distinguish between the two conditions.

Clinical Considerations: In the neonatal form of CPT II deficiency, the neonate is profoundly ill with marked hypoglycemia, metabolic acidosis, cardiac arrhythmias, cardiomyopathy, liver disease, and facial dysmorphism with renal and brain anomalies. These infants rarely survive. Patients with the more common, later-onset muscular form of CPT II deficiency present in adolescence or adulthood with muscle weakness, exercise intolerance, and rhabdomyolysis. CACT deficiency presents similarly to the neonatal form of CPT II deficiency.

Additional Information:

[How to Communicate Newborn Screening Results](#)
[Emergency Protocols \(New England Consortium of Metabolic Programs\)](#)
[Gene Reviews](#)
[Medline Plus \(CPT II | CACT\)](#)
[Condition Information for Families-HRSA Newborn Screening Clearinghouse](#)
[ClinGen Actionability Report](#)

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.

State and Other Resources

State Newborn Screening Program

Newborn Screening, State of Rhode Island Department of Health
401-921-7619, health.ri.gov/newbornscreening/

Genetics/Metabolic Consultants

Division of Genetics, Lifespan Physician's Group, Hasbro Children's Hospital
401-444-4000, www.lifespan.org/centers-services/childrens-neurodevelopment-center/genetics-and-dysmorphology

Information for Clinicians and Families

Rhode Island Medical Home Portal (see Newborn Disorders and Parents & Families sections)
ri.medicalhomeportal.org/newborn/carnitine-acylcarnitine-translocase-deficiency

Parent/Family Support

FOD Family Support Group
fodsupport.org/

National Resources (with web addresses)

Additional Information

How to Communicate Newborn Screening Results
www.hrsa.gov/sites/default/files/hrsa/advisory-committees/heritable-disorders/Resources/achdnc-communication-guide-newborn.pdf

Emergency Protocols (New England Consortium of Metabolic Programs)
www.newenglandconsortium.org/cpt-ii-deficiency

Gene Reviews
www.ncbi.nlm.nih.gov/books/NBK1253/

Medline Plus

CPT II - <https://medlineplus.gov/genetics/condition/carnitine-palmitoyltransferase-ii-deficiency/>
CACT - <https://medlineplus.gov/genetics/condition/carnitine-acylcarnitine-translocase-deficiency/>

Condition Information for Families-HRSA Newborn Screening Clearinghouse
newbornscreening.hrsa.gov/conditions/carnitine-palmitoyltransferase-ii-deficiency

ClinGen Actionability Report
actionability.clinicalgenome.org/ac/Pediatric/ui/stg2SummaryRpt?doc=AC1022

Referral (local, state, regional and national)

Find a Genetics Clinic Directory
clinics.acmg.net

Genetic Testing Registry
www.ncbi.nlm.nih.gov/gtr/