

# Newborn Screening ACT Sheet

## [Elevated C4 Acylcarnitine]

### Short-Chain Acyl-CoA Dehydrogenase (SCAD) Deficiency

**Differential Diagnosis:** Short-chain acyl CoA dehydrogenase (SCAD) deficiency; Isobutyryl-CoA dehydrogenase (IBDH) deficiency (also referred to as isobutyrylglycinuria (IBG)); Ethylmalonic encephalopathy (EE).

**Condition Description:** SCAD deficiency is a fatty acid oxidation (FAO) disorder. FAO occurs during prolonged fasting and/or periods of increased energy demands (fever, stress) when energy production relies increasingly on fat metabolism. IBG results from IBDH deficiency. IBDH is an enzyme involved in the degradation of the branched chain amino acid valine. EE is a related disorder that seems to be due to a defective mitochondrial matrix protein, the precise function of which is yet unknown. In all three conditions, potentially toxic metabolites accumulate.

#### **YOU SHOULD TAKE THE FOLLOWING ACTIONS:**

- Contact family to inform them of the newborn screening result and ascertain clinical status (poor feeding, vomiting, lethargy).
- Consultation with pediatric metabolic specialist.
- Evaluate infant (hypoglycemia, lethargy, metabolic acidosis).
- Emergency treatment if symptomatic.
- Initiate timely confirmatory/diagnostic testing as recommended by specialist.
- Educate family about need for infant to avoid fasting and explain signs, symptoms and need for urgent treatment if infant becomes ill.
- Report finding to newborn screening program.

**Diagnostic Evaluation:** Plasma amino acid analysis which shows increased phenylalanine without increased tyrosine (increased phenylalanine:tyrosine ratio). Urine pterin analysis and red blood cell DHPR assay will identify pterin defects. Consider PAH mutation testing.

**Clinical Considerations:** Asymptomatic in the neonate. If untreated PKU will cause irreversible mental retardation, hyperactivity, autistic-like features, and seizures. Treatment will usually prevent these symptoms. Pterin defects cause early severe neurologic disease (developmental delay/seizures) and require specific therapy.

#### **Additional Information:**

[New England Consortium of Metabolic Programs](#)  
[OMIM EE](#)

#### **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.

# American College of Medical Genetics **ACT SHEET**

## 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:*

#### **New England Consortium of Metabolic Programs**

<http://newenglandconsortium.org/for-professionals/acute-illness-protocols/fatty-acid-oxidation-disorders/short-chain-acyl-coa-dehydrogenase-deficiency-scadd/>

#### OMIM EE

<http://www.ncbi.nlm.nih.gov/entrez/dispmim.cgi?id=602473>

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

#### Testing

[http://www.ncbi.nlm.nih.gov/sites/GeneTests/lab/clinical\\_disease\\_id/3176?db=genetests&country=United%20States](http://www.ncbi.nlm.nih.gov/sites/GeneTests/lab/clinical_disease_id/3176?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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