# Newborn Screening ACT Sheet
# [Elevated C4-OH Acylcarnitine]
## 3-Hydroxyacyl-Coenzyme A Dehydrogenase Deficiency
## Medium/Short Chain Acyl-CoA Dehydrogenase (M/SCHAD) Deficiency

**Differential Diagnosis:** Medium/Short-chain hydroxyacyl-CoA dehydrogenase (M/SCHAD) deficiency.

**Condition Description:** M/SCHAD is a fatty acid oxidation (FAO) disorder. Fatty acid oxidation occurs during prolonged fasting and/or periods of increased energy demands (fever, stress) when energy production relies increasingly on fat metabolism. In an FAO disorder, fatty acids and potentially toxic derivatives accumulate because of a deficiency in one of the FAO enzymes.

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*YOU SHOULD TAKE THE FOLLOWING ACTIONS:*

- Contact family **IMMEDIATELY** 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. If infant becomes even mildly ill (poor feeding, vomiting, or lethargy), immediate treatment with IV glucose is needed.
- Report finding to newborn screening program.

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**Diagnostic Evaluation:** Plasma acylcarnitines will show increased C4-OH acylcarnitine. Urine organic acids will show increased hydroxy-dicarboxylic acids. Plasma insulin may be elevated. M/SCHAD gene sequencing can confirm the diagnosis.

**Clinical Considerations:** The neonate is usually asymptomatic, although hypoglycemia and hyperinsulinism may be present. Severe hypoglycemia and severe hyperinsulinism may appear later. Sudden death in infancy has been reported.

**Additional Information:**

Gene Reviews
OMIM

**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 Reviews**
http://ghr.nlm.nih.gov/condition=3hydroxyacylcoenzymeadehydrogenasedeficiency

**OMIM**
http://www.ncbi.nlm.nih.gov/entrez/dispomim.cgi?id=231530

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

**Testing**
http://www.ncbi.nlm.nih.gov/sites/GeneTests/lab/clinical_disease_id/26811?db=genetests

**Clinical Services**
http://www.ncbi.nlm.nih.gov/sites/genetests/clinic?db=genetests

**Find Genetic Services**
http://www.acmg.net/GIS/Disclaimer.aspx