

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.

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.

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](#)

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://ghr.nlm.nih.gov/condition=3hydroxyacylcoenzymeadehydrogenasedeficiency>

OMIM

<http://www.ncbi.nlm.nih.gov/entrez/dispmim.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>

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