

# **ACT Sheet**

### **Newborn Screening ACT Sheet**

### [Elevated C4-OH Acylcarnitine] Short Chain Hydroxyacyl-CoA Dehydrogenase (SCHAD) Deficiency

## (also known as 3-Hydroxyacyl-Coenzyme A Dehydrogenase Deficiency, HADH; and Medium/Short Chain Hydroxyacyl-CoA Deficiency, M/SCHAD).

#### Differential Diagnosis: none.

**Condition Description:** SCHAD is a fatty acid oxidation (FAO) disorder. 3-Hydroxyacyl-CoA dehydrogenase deficiency disrupts fatty acid breakdown at the level of short and medium-chain 3-hydroxy-fatty acids. It is associated with elevated C4-hydroxy-acylcarnitine (C4-OH) and a decreased production of energy from fat. 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. Presentation in the neonatal period is rare.

#### You Should Take the Following <u>IMMEDIATE</u> Actions:

- Inform family of the newborn screening result.
- Ascertain clinical status (poor feeding, vomiting, lethargy).
- Consult with pediatric metabolic specialist the same day.
- Evaluate infant (poor feeding, vomiting, lethargy, hypoglycemia, metabolic acidosis). If any of these findings are present or if the neonate is ill, immediately treat with IV glucose and transport to the hospital in consultation with metabolic specialist.
- Initiate confirmatory/diagnostic testing and management, as recommended by the specialist.
- Provide family with basic information about SCHAD and its management, including information about the avoidance of fasting in the newborn.
- Report final diagnostic outcome to newborn screening program.

**Diagnostic Evaluation:** <u>Plasma acylcarnitines:</u> C4-OH is typically elevated in SCHAD. <u>Urine organic acids</u>: Hydroxy-dicarboxylic acids may be elevated. <u>Plasma insulin</u> may also be elevated. <u>Molecular genetic testing</u> may be required to establish the diagnosis.

Clinical Considerations: Neonates with SCHAD deficiency are usually asymptomatic, although hypoglycemia and hyperinsulinism may be present. Severe hypoglycemia and severe hyperinsulinism may appear later. Sudden death in infancy has been reported. Note: Given the limited information available on this specific condition, some links for the similar and more common MCAD (Medium chain Acyl-CoA-Dehydrogenase) deficiency are included.

#### Additional Information:

How to Communicate Newborn Screening Results

Emergency Protocols (New England Consortium of Metabolic Programs) GARD Inform Gene Reviews Medline Plus Condition Information for Families- HRSA Newborn Screening Clearinghouse

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.

© American College of Medical Genetics and Genomics, 2022 Content Updated: May 2022 (Funded in part through MCHB/HRSA/HHS grant #U22MC03957;#UH9MC30770; National Coordinating Center for the Regional Genetics Networks)



# **ACT Sheet**

[Elevated C14:1 +/- other long-chain Acylcarnitines] Very Long-Chain Acyl-CoA Dehydrogenase (VLCAD) Deficiency

### **State and Other Resources**

#### State Newborn Screening Program

Nevada Newborn Screening Program, Nevada State Public Health Laboratory 775-682-6238, med.unr.edu/nsphl/newborn-screening

#### Genetics/Metabolic Consultants

Nicola Longo, MD, PhD, University of Utah Genetics/Pediatrics 801-585-2457, healthcare.utah.edu/fad/mddetail.php?physicianID=u0305101

#### Information for Clinicians and Families

Nevada Medical Home Portal (see Newborn Disorders and Parents & Families sections) nv.medicalhomeportal.org/newborn/very-long-chain-acyl-coa-dehydrogenase-deficiency

#### Parent/Family Support

FOD Family Support Group fodsupport.org/

MitoAction www.mitoaction.org/conditions/vlcad-very-long-chain-acyl-coa-dehydrogenase-deficiency/

#### 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/achdnccommunication-guide-newborn.pdf

#### GARD

rarediseases.info.nih.gov/diseases/5508/vlcad-deficiency

Inform informnetwork.org/?s=vlcad

Gene Reviews www.ncbi.nlm.nih.gov/books/NBK6816/

Medline Plus medlineplus.gov/genetics/condition/very-long-chain-acyl-coa-dehydrogenase-deficiency/

Condition Information for Families-HRSA Newborn Screening Clearinghouse newbornscreening.hrsa.gov/conditions/very-long-chain-acyl-coa-dehydrogenase-deficiency

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

Find a Genetics Clinic Directory clinics.acmg.net

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

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 of the onsidered not 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.

© American College of Medical Genetics and Genomics, 2022 Content Updated: May 2022 (Funded in part through MCHB/HRSA/HHS grant #U22MC03957;#UH9MC30770; National Coordinating Center for the Regional Genetics Networks)