

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: Plasma acylcarnitines: C4-OH is typically elevated in SCHAD. Urine organic acids: Hydroxy-dicarboxylic acids may be elevated. Plasma insulin may also be elevated. Molecular genetic testing 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.



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

Newborn Genetic Screening, New Mexico Department of Health 877-890-4692, www.nmhealth.org/about/phd/fhb/cms/nbgs/

Genetics/Metabolic Consultants

Michael Marble, MD University of New Mexico Metabolic Clinic 505-272-6631, hsc.unm.edu/directory/marble-michael.html

Information for Clinicians and Families

New Mexico Medical Home Portal (see Newborn Disorders and Parents & Families sections) nm.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

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/

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