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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 IMMEDIATE 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

 Content Updated: May 2022 *(Funded in part through MCHB/HRSA/HHS grant #U22MC03957;#UH9MC30770; National Coordinating Center for the Regional Genetics Networks)*

[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/achdnc-communication-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/