Newborn Screening ACT Sheet
[Elevated C14:1 +/- other long-chain acylcarnitines]
Very Long-Chain Acyl-CoA Dehydrogenase (VLCAD) Deficiency

***Differential Diagnosis*:** Very long-chain acyl-CoA dehydrogenase (VLCAD) def.

Condition Description- VLCAD deficiency is a fatty acid oxidation (FAO) disorders. 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 a FAO disorder, fatty acids and potentially toxic derivatives accumulate because of a deficiency in one of the mitochondrial FAO enzymes.

MEDICAL EMERGENCY - TAKE THE FOLLOWING IMMEDIATE ACTIONS:

- Contact family to inform them of the newborn screening result and ascertain clinical status (poor feeding, vomiting, lethargy).
- Consult with pediatric metabolic specialist.
- Evaluate the newborn (poor feeding, lethargy, hypotonia, hepatomegaly, arrhythmia, evidence of cardiac decompensation). If signs are present or infant is ill, initiate emergency treatment with IV glucose and oxygen. Transport to hospital for further treatment in consultation with metabolic specialist. If infant is normal initiate timely confirmatory/diagnostic testing, as recommended by specialist.
- Educate family about need for infant to avoid fasting. Even if mildly ill, immediate treatment with IV glucose is needed.
- Report findings to newborn screening program.

Diagnostic Evaluation: Plasma acylcarnitine profile may show increased **C14:1 acylcarnitine** (and lesser elevations of other long chain acylcarnitines). Diagnosis is confirmed in consultation with the metabolic specialist by mutation analysis of the **VLCAD gene** and additional biochemical genetic tests.

Clinical Expectations: VLCAD deficiency may present acutely in the neonate and is associated with high mortality unless treated promptly; milder variants exist. Features of severe VLCAD deficiency in infancy include hepatomegaly, cardiomyopathy and arrhythmias, lethargy, hypoketotic hypoglycemia, and failure to thrive. Treatment is available.

Additional Information:
(Click on the name to take you to the website. Complete URLs are listed in the Appendix)

New England Consortium of Metabolic Programs

VLCAD Emergency Protocol

Genetics Home Reference

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

Testing

Gene Tests

Clinical

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APPENDIX: Resources with Full URL Addresses

Additional Information:

New England Consortium of Metabolic Programs
http://www.childrenshospital.org/newenglandconsortium/NBS/VLCADD/vlcadd_protocol.htm

VLCAD Emergency Protocol http://www.childrenshospital.org/newenglandconsortium/NBS/VLCADD.html

Genetics Home Reference
http://ghr.nlm.nih.gov/condition=verylongchainacylcoenzymeadehydrogenasedeficiency

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

Testing http://biochemgen.ucsd.edu/UCSDW3BG/Labchoose.asp

Gene Tests
http://www.genetests.org/servlet/access?prg=j&db=genetests&site=gt&id=8888891&fcn=c&qry=2927&res=nous&res=nointl&key=jXHXXy8dPUhIt&show_flag=c

Clinical
http://www.genetests.org/servlet/access?id=8888891&key=RRqZcXXEUiAx9&fcn=y&fw=zLsf&filename=/clinicsearch/searchclinic.html

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