Newborn Screening ACT Sheet
[Increased Leucine]
Maple Syrup (Urine) Disease

Differential Diagnosis: Maple syrup urine disease (MSUD); hydroxyprolinemia (probably benign).

Condition Description: In MSUD, leucine, isoleucine, and valine (branched chain amino acids) cannot be metabolized further than their α-ketoacid derivatives. The amino acids and organic acids accumulate and produce severe toxicity.

YOU SHOULD TAKE THE FOLLOWING ACTIONS IMMEDIATELY:

- Contact family to inform them of the newborn screening result and ascertain clinical status (poor feeding, vomiting, lethargy, tachypnea).
- Consult with pediatric metabolic specialist.
- Evaluate the newborn (poor feeding, lethargy, tachypnea, alternating hypertonia/hypotonia, seizures). If any sign is present or infant is ill, transport to hospital for further treatment in consultation with metabolic specialist.
- Initiate timely confirmatory/diagnostic testing and management, as recommended by specialist.
- Provide the family with basic information about MSUD and dietary management.
- Report findings to newborn screening program.

Diagnostic Evaluation: In MSUD, plasma amino acid analysis reveals elevations of leucine, isoleucine, alloleucine, and valine (the branched chain amino acids); and urine organic acid analysis reveals abnormal branched-chain hydroxy- and ketoacids. In expanded screening, leucine/isoleucine and hydroxyproline cannot be differentiated, so if the baby has hydroxyprolinemia confirmatory amino acid analysis will show only increased hydroxyproline (a rare and likely benign entity).

Clinical Considerations: MSUD presents in the neonate with feeding intolerance, failure to thrive, vomiting, lethargy and maple syrup odor to urine and cerumen. If untreated, it will progress to irreversible mental retardation, hyperactivity, failure to thrive, seizures, coma, cerebral edema, and possibly death. Hydroxyprolinemia is probably benign.

Additional Information:

Emergency Protocols (New England Consortium of Metabolic Programs)
Gene Reviews
Genetics Home Reference

Referral (local, state, regional and national):

Testing
Clinical Services
Find Genetic Services

ACMG
American College of Medical Genetics and Genomics
Translating Genes Into Health®

LOCAL RESOURCES: Insert State newborn screening program web site links

State Resource site *(insert state newborn screening program website information)*

Name	
URL	
Comments	

Local Resource Site *(insert local and regional newborn screening website information)*

Name	
URL	
Comments	

APPENDIX: Resources with Full URL Addresses

Additional Information:

Emergency Protocols (New England Consortium of Metabolic Programs)
http://newenglandconsortium.org/for-professionals/acute-illness-protocols/organic-acid-disorders/maple-syrup-urine-disease-msud/

Gene Reviews
http://www.ncbi.nlm.nih.gov/bookshelf/br.fcgi?book=gene&part=msud

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

Referral (local, state, regional and national):

Testing
http://www.ncbi.nlm.nih.gov/sites/GeneTests/lab/clinical_disease_id/22186?db=genetests&country=United%20States

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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