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Minnesota Department of Health

Laboratory Evaluation of the Infant with a Positive Newborn Screen for a Metabolic Disorder

What is a positive newborn screen for a metabolic disorder?

A positive newborn screen indicates that tandem mass spectrometry has detected an abnormal chemical pattern suggestive of a metabolic disorder. However, other contributing factors may cause a false positive result. Because the newborn screen is not diagnostic, follow-up evaluation is required.

What should providers do?

1. **Speak with a pediatric metabolic specialist.** These specialists are your best source of information and guidance specific to your patient's screening result and both the laboratory and clinical follow-up that is required. Metabolic specialists are available on call at the University of Minnesota (888-543-7866) and the Mayo Clinic (800-533-1710).
2. Collection of laboratory specimens may be requested by the pediatric metabolic specialist. Use the information on this sheet to work with your lab to arrange for appropriate collection of samples and submission to a reference laboratory as soon as possible.
3. If the infant's condition worsens; you have difficulty collecting the specimens; or you have questions about the process or the laboratory results sent to you by the reference lab, call the metabolic specialist.

Important notes

- Newborn screening is done on whole blood. Follow-up plasma/urine testing separates true positives - which require prompt treatment - from false positives.
- Because the disorders tested for in newborn screening are rare, your lab staff and even your reference lab may be unfamiliar with the tests you are requesting. You may need to speak directly with the specialty lab involved.

Laboratory tests and specimen requirements

Frequently, the metabolic specialist will direct you to collect specimens for **one or more** of these tests:

<i>Test Name</i>	<i>Specimen Requirements</i>
<u>Plasma acylcarnitines</u>	0.1 mL frozen plasma in a Sodium heparin green top tube
<u>Plasma carnitines</u> (Free, Total, Esterified)	0.5 mL frozen plasma in a Sodium heparin green top tube
<u>Plasma amino acids</u>	0.5 mL frozen plasma in a Sodium heparin green top tube
<u>Urine acylglycines</u>	5.0 mL random urine, frozen
<u>Urine organic acids</u>	4.0 mL random urine, frozen

Reference laboratories

- These specialized tests are available in only a few laboratories in the country. Use your facility's send-out laboratory as a resource. Your send-out lab will likely be able to help you determine the logistics of collecting the sample.
- Using one reference laboratory for all the ordered tests often allows for collection of smaller specimen volumes and shorter turn around times.
- For additional laboratory resources, you can call the metabolic specialists or one of the reference labs serving Minnesota or look online at www.health.state.mn.us/newbornscreening.

