



NEWBORN SCREENING NEWSLETTER

Change in Time of Collection:

Rule change targets timing of collection of first newborn screen:

- With the new rule change first newborn screening specimens are required to be collected at **24–48 hours**. Previously this time was 48 hours– 5 days.
- Specimens should **not** be collected prior to 24 hours. Samples collected before 24 hours may produce a false positive or false negative result. First specimen test results are validated only on specimens collected between **24–48 hours**.
- First specimens should be sent to the State Laboratory for testing within 24 hours of collection. Specimens should be sent **daily**.
- There are no changes to timing of second screen collection and there are no changes to sick/preterm protocol.

Rule changes effective June 1, 2015

New Disorder Being Added:

With the new rule change Guanidinoacetate methyltransferase (GAMT) deficiency will be added to the newborn screening panel. GAMT is an autosomal recessive disorder of creatine synthesis, in which an infant is born lacking the GAMT enzyme. As a result, creatine does not reach organs or the brain. Low levels of GAMT in the brain lead to serious developmental delay, autism-like behavior, seizures, and speech delay. Treatment is highly effective and inexpensive.

The additional test will not require an increase in newborn screening fees and no additional blood is necessary on the newborn screening card. Testing for GAMT will be part of tandem mass spectrometry which is currently utilized to test all other acylcarnitine and amino acid disorders.

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