

**Winter
2009/2010**



Newborn Screening 411

RGHSD

Newborn Screening
Area Service Center

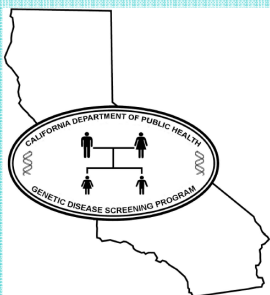
What's new?

- The newborn screening methodology has been refined to better detect Tyrosinemia type I, with the addition of succinylacetone (SA) to the analytes currently used. Tyrosinemia type I is the result of a deficiency of the enzyme fumarylacetoacetate hydrolase (FAH), which is responsible for breaking down the amino acid tyrosine. The disorder often presents as severe liver disease in infants. Other symptoms may include diarrhea and bloody stools, vomiting, poor weight gain, lethargy, irritability, and a "cabbage-like" odor to the skin and urine. If not treated, liver and kidney damage, neurologic crises, and death may occur. Treatment typically includes the implementation of a low-tyrosine diet and the medication nitisinone to help prevent liver and kidney damage.
- The Newborn Screening Program recently changed tandem mass spectrometry methodology. Consequently, the cutoffs/reference ranges were modified in order to maintain the same sensitivity and specificity. The new ranges may be found online at:

<http://www.cdph.ca.gov/programs/nbs/Documents/NBS-TestRangesCutoffs121509.pdf>

Important Reminders

- Transport logs: please ensure the transport logs are completed accurately and that the log reflects the specimens in that group. We have seen an increase in "missing" specimens (specimens listed on the transport log, but not included with the shipment) as well as "extra" specimens (specimens not listed on the transport log, but included in the shipment) at our NAPS labs.
- If you need more transport logs, they may be obtained from the NAPS lab that processes your newborn screening specimens. For facilities in Riverside County, the NAPS lab may be reached at 559-459-6273. For facilities in San Diego, Orange, and Imperial Counties, the NAPS lab may be reached at 562-933-3911.
- Please do not "batch" specimens (do not wait until several are accumulated before sending them to the NAPS lab). Specimens should be sent from your facility with the first available shipment or courier pick up.
- It's a new year! Please remember to correctly record the birth year and year of collection on the TRF as 2010.
- The ideal time to collect a newborn screening specimen is at 24-48 hours of life.
- Call your Area Service Center at 1-800-793-1313 with any questions or concerns.



RCHSD

Newborn Screening Area

Service Center

LOCATED AT:

7920 Frost Street, Suite 302
San Diego, CA 92123

MAILING ADDRESS:

3020 Children's Way
Mail Code 5116
San Diego, CA 92123

PHONE:

(858) 966-8708

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(858) 966-8709

For more information visit:

www.dhs.ca.gov/NBS/



A baby has a special way of adding joy in every single day.

-- Author Unknown

Site Visits and In-Services

As we begin the new year, we are scheduling the State-mandated site visits and in-services for our facilities. If you have new staff, would like a refresher on the best practices for newborn screening collection and handling, or have another reason you would like us to visit your facility sooner rather than later, please call us at 1-800-793-1313. Otherwise, we will be contacting you to arrange your next scheduled visit.

Hospital Newborn Screening Protocols

Every department involved with newborn screening specimens and/or results should have an up-to-date protocol for each aspect of screening that occurs in that department. Please have your protocols available for reference during site visits. We will gladly review them for content and completeness.

If you need guidance regarding what to include in your protocols, or information about the regulations mandating the California Newborn Screening Program, please contact the Area Service Center Project Director, George Knapp, at 1-800-793-1313.

Question of the Quarter

Question: The State regulations require that newborn screening specimens be collected after 12 hours of age. Is it OK if we collect the specimens just a little early (e.g. 11 hours and 45 minutes)?

Answer: No, it is not permissible to collect a newborn screening specimen before 12 hours of age.* Even a specimen collected at 11 hours and 59 minutes will require a second collection. Please ensure specimens are collected after 12 hours of age (ideally at 24 to 48 hours) to minimize the risk for inaccurate results, inconvenience to the family (by having to return to the facility for the collection of a second specimen), and an unnecessary second heel-stick for the baby.

**There is one exception: If the baby will receive a red blood cell transfusion before 12 hours of age, collect a specimen before the transfusion, regardless of age, and collect a repeat specimen at least 24 hours post-transfusion.*

Thank You and Happy New Year

In 2009 we saw overall reductions in the number of errors made by our facilities, including inadequate specimens due to poor collection technique and specimens collected early in error (collected at less than 12 hours of age). We have also seen increased compliance with chart reviews. We would like to thank you for your hard work and a successful 2009. With your continued support and effort, we look forward to an even more successful 2010!