

Newborn Screening Update



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Impact of Serial Screening of NICU babies

In July 2011, Nebraska implemented changes in regulations requiring admission screening for premature, low birth weight and sick infants admitted to the NICU. These were based on consensus guidelines of the Clinical and Laboratory Standards Institute. Admission screening is defined as “prior to the infant receiving any treatments with the exception of respiratory treatments”. Given the vast number of conditions screened,

complexity of treatment, and biological state of the premature, low birth weight and sick infants, multiple things can conspire to interfere with getting acceptable screening results. The admission/pre-treatment screen should be followed by a 48-72 hour screen if the first one was done prior to 24 hours of life.

Babies weighing less than 2000 grams, should have a 3rd screen upon discharge or at 28 days which-

ever is first. The primary rationale for this specimen is to allow the HPT axis to mature so a more reliable screen for congenital primary hypothyroidism can be done.

One change physicians will notice is lab result reports on repeats collected because the initial specimen was drawn early (< 24 hours) will now include results for all conditions.

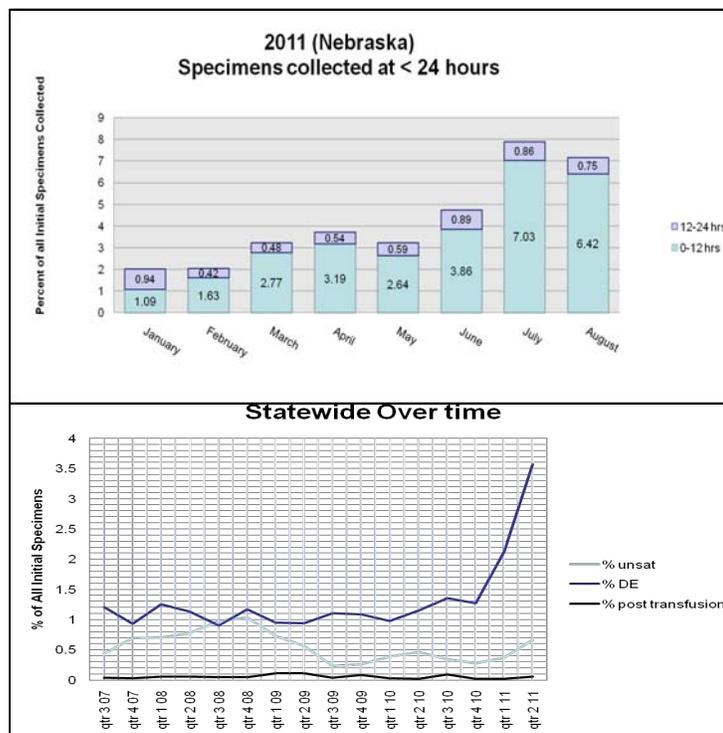
Along with the increase in early draws,

especially for 0-12 hour specimens, data is showing an increase in unsatisfactory specimens, although not proportional to the increase in drawn early specimens.

The serial screening guidelines and regulations were not meant for the babies who are in the NICU for close observation and are not receiving treatments that can interfere with screen results such as blood transfusions or hyperalimentation. The ideal scenario is when the specimen can be collected at between 24-48 hours of life and before any treatments. Screening specimens from babies in “observation” should usually be able to be collected in this manner.

Another change to lab reports you will see (effective 10/3/11) is testing for CPH and CAH will no longer be done on less than 24 hour specimens. The greater than 24 hour repeat specimens must be collected, and will have all the tests done.

The laboratory does not charge the submitter for requested repeats necessary to fulfill the agreed upon protocol.



Newborn Screening Update



Screening for SCID

In May 2010 Secretary Sebelius endorsed the recommendation of the Advisory Committee on Heritable Diseases in Newborns and Children that Severe Combined Immune Deficiency(s) should be added to the Recommended Universal Screening Panel (RUSP). Following that endorsement, a pilot study was conducted in California, New York, Massachusetts and Wisconsin including sub-sets of births from Louisiana and Puerto Rico. Over 1 million babies have now been screened. Several infants with various T-cell deficiencies have been identified, some with complete deficiency have been transplanted and successfully engrafted. Interestingly the previously thought incidence of 1:100,000 was proven to be closer to 1:30,000 to 35,000.

In the ensuing months a workgroup of the Nebraska Newborn Screening Advisory Committee (NBSAC) began meeting to evaluate what resources would be necessary to successfully add SCID to our screening panel. Of primary concern was the ability to get the recommended treatment (e.g. protective isolation) covered while further diagnostic testing and bone marrow stem cell transplant donors were sought. The children with the best outcomes (complete reconstitution) appear to be those transplanted before 3 months of age, and who have had infections prevented.

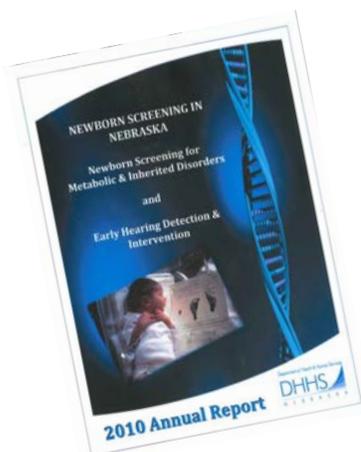
In July 2011, the Nebraska NBSAC made the recommendation to add SCID to Nebraska's required screening panel. The request for permission to draft regulations to add SCID has been submitted. The Department is considering a wide range of factors with the input of insurers and others.

Key decision points include:

- An inexpensive sensitive and specific test is available. The TREC assay (T-Cell receptor excision circles) at PerkinElmer Genetics, Inc. is a DNA (PCR) based test, that has been validated. The laboratory reports having optimized the test to maximize sensitivity and specificity. Based on the experience in California where PerkinElmer implemented the method, and Nebraska's birth rate, we should have about 3 presumptive positive newborns per year requiring further diagnostic evaluation by flow cytometry. In order to add SCID to the Nebraska screening panel, \$5 would be added to the current \$38.50 charge to hospitals by the laboratory.
- The condition is not otherwise detected before damage has occurred and the effects are significant. Patients with SCID frequently present with common childhood illnesses, frequent and lasting infections and may go undiagnosed despite numerous tests and hospitalizations for a couple years or may succumb to the illness before diagnosed. Late diagnosed and transplanted SCID patients have much higher mortality than those transplanted by 3 months of age.
- Screening/Treatment is cost effective. Reports from the Wisconsin Public Health Laboratory (JAMA Oct-27-2010) found costs to treat infants identified early through screening were as low as \$250,000-300,000 but for children not identified at birth was about \$2 million. Other reports show treatment at even less than \$100,000. Under best conditions (early diagnosis, prevention of infection, transplant by 3 months), treatment is curative.

2010 Annual Report Available—New Web URL

The Annual Report on Newborn (Blood-spot) screening, and the Early Hearing Detection and Intervention Program has been published to the Newborn Screening Web-site. The report highlights 2010 events, and provides data on the processes and outcomes in newborn screening such as the 40 babies who had clinically significant conditions identified in time to prevent damage. The web page can be found at www.dhhs.ne.gov/nsp. This URL will be changing in mid-November to: <http://dhhs.ne.gov/publichealth/Pages/nsp.aspx>. Remember to subscribe for updates, it's an easy way to stay up to date.



Newborn Screening Update

Prenatal Education About Newborn Screening

In 2011 the American College of Obstetrics and Gynecology published a new position statement supporting education about newborn screening during the third trimester.

In an effort to help obstetric providers meet this recommendation, the Newborn Screening Program developed a one-page information sheet introducing newborn screening concepts with a few goals; 1) Provide expecting parents a web site where they could get more information so the obstetrician would not be faced with

questions they may not be prepared to answer, and 2) introduce the idea of looking for a doctor for their baby, and making this the medical home for their new baby.

The one-pager was sent out with a questionnaire to OB/GYN practices across Nebraska, and to a group of family physicians providing obstetric services according to responses from hospital mother/baby unit coordinators.

Seventeen percent or 10/59 OB practices responded they'd be very likely to use the one pager for parent education. These rep-

resented 21 physicians in 6 Nebraska cities/communities. Only one responded negatively, but noted they already provide the full newborn screening brochure.

Twenty nine percent or 23/72 family physician obstetric clinics responded they'd be likely or very likely to provide the materials. Only one responded unfavorably. They represented 72 practitioners from 17 cities/communities. A supply of the one pagers were sent to positively responding clinics. For a free supply of this resource contact 402 471-9731.



Newborn Screening Quality Assurance at work for you

Quality assurance is a key element to the success of screening in Nebraska. Our NBS program staff work closely with the PerkinElmer Genetics (PEI) lab staff, particularly the IT team for QA report development and with consultant Dennis Freer, PhD. While PEI conducts testing for several clients, Nebraska's size and expertise uniquely position us to pick up trends or potential problems quickly. That was the case this

summer when a sudden increase in inconclusive screen results for biotinidase was noticed in a two day period. Knowing that Nebraska had not experienced extreme heat that can cause the enzymes to denature and appear inconclusive or positive, we alerted the laboratory and pressed for investigation. Concurrently we reminded hospitals to be certain specimens were completely dry before shipping. After ruling

out a suspected problem with the performance of the filter paper, and investigating if any changes had occurred with the overnight shipping of samples, the laboratory evaluation concluded the problem was with a particular lot of plates used for processing specimens. Now the laboratory routinely conducts QC for each new lot of these plates., and is back to a well performing assay.

Testing Changes for “Drawn-Early” Specimens

As mentioned in the article on page one, specimens that are collected at less than 24 hours will no longer be tested for congenital adrenal hyperplasia and congenital primary hypothyroidism.

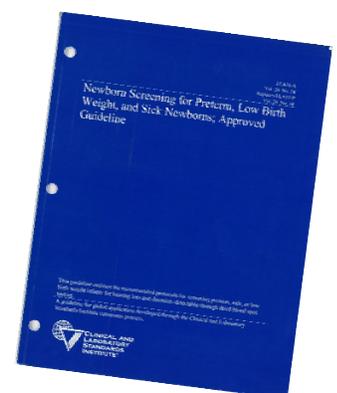
With the implementation of the admission/pre-treatment screen of the premature, low birth weight and sick infants, there has been a dramatic increase in drawn early specimens. Inconclusive results

for CPH and CAH go hand in hand with prematurity and with early drawn specimens. We determined those screening results were not meeting our expectations for clinical utility and perhaps were creating more anxiety for parents and clinicians than warranted.

Therefore, after substantial consideration by a workgroup of the newborn screening advisory com-

mittee including neonatologists, pediatric endocrinology, the laboratory and program staff, the workgroup determined the T4/TSH for CPH and I7-OHP for CAH, would no longer be run on specimens collected at less than 24 hours of age.

The lab reports will reflect this in the comments. It remains essential to get the repeat specimen after 24 hours.



Newborn Screening Program

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Nebraska Newborn Screening Program

Visit us on the web. We have a wealth of information here about Nebraska's program, requirements and many links to trusted sources.

WWW.DHHS.NE.GOV/NSP

Changing in MID-November to:

<http://dhhs.ne.gov/publichealth/Pages/nsp.aspx>

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