

KANSAS

NEWBORN SCREENING PROGRAM

Financial Review

December 13-14, 2010



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KANSAS NEWBORN SCREENING PROGRAM FINANCIAL REVIEW AND CONSULTATION

Executive Summary

The following brief outline summarizes information in the report that follows. The points noted here should not be taken out of context without viewing the supporting report. Reference materials are included in the Appendices to aid the reader in achieving a fuller understanding of some of the issues discussed. In order to better understand the broad concepts of newborn screening (NBS) as a system that includes the public health program as one component, attention should be paid to Appendix 3 [*U. S. Newborn Screening System Guidelines: Statement of the Council of Regional Networks for Genetic Services (CORN). Screening 1992;1:135-147.*] and Appendix 4 [Executive Summary - *Serving the Family from Birth to the Medical Home: Newborn Screening a Blueprint for the Future. Pediatrics 2000;106(suppl 2):383-427.*].

- The current Kansas Newborn Screening Program (KNSP) includes screening for 29 of the 30 conditions now recommended by the Secretary of the U.S. Department of Health and Human Services. (Note: Severe Combined Immunodeficiency was added to the recommended panel in 2010 and pilot programs are currently being initiated in some states.)
- Current legislation allows for newborn screening (NBS) of “all infants born in the state” by the Kansas Department of Health and Environment (KDHE).
- The NBS program in Kansas and 3 other jurisdictions currently do not recover screening expenses through a fee. All other state NBS programs have fees ranging up to \$125.
- Fee revenue that sustains the NBS system generally comes from 3rd party payers (through maternity benefits packages as opposed to direct fee for service billing), including Medicaid, and no newborns are denied screening because of inability to pay.
- Kansas law provides for state payment for treatment/management of conditions identified through NBS on a sliding scale based on income.
- Kansas law does not currently require insurers doing business in the State to cover metabolic foods and formulas that are medically necessary for treatment of metabolic conditions identified through NBS. Such legislation exists in many other States. Legislation of this type might reduce the State’s expenditures for treatment/management.
- Screening services are currently offered 5 days a week despite the fact that expanded testing now includes several disorders that may have dire consequences within the first few days of life. A 6-day work week and adjusted holiday schedules are needed to meet detection and treatment norms for these conditions.
- A courier system that efficiently reaches specimen submitters would likely improve transit times for specimens from collection to testing. A system previously existed for other public health laboratory functions, primarily for county health departments, and it did not reach most NBS submitters.
- NBS data management is currently handled using multiple computer systems with data entered and transferred manually. An integrated data management system is needed to improve efficiency and decrease the chance of human errors in data management and

tracking, and to manage the legislatively required case registry.

- Long-term outcome information is not currently available to demonstrate the effectiveness of the current Kansas NBS program. Any future data system should have the ability to monitor long-term outcome indicators currently being developed nationally as a means of program evaluation and improvement.
- Data system integration with vital records is urgently needed for quality control measures to ensure that all Kansas newborns are receiving timely newborn screens. Integration with other systems (newborn hearing screening, immunizations, etc.) is also recommended.
- A NBS fee would provide a stable mechanism for sustainable funding, particularly since current funding comes primarily from Children's Initiative Funds (CIF), Medicaid, Title V Block Grant and general revenue.
- A fee would most efficiently be levied on NBS specimen collection kits at the time of ordering, and the amount should be sufficient to adequately cover system expenses as determined by the KDHE. Since only a single collection form would exist in a specimen collection kit, all specimens, including any additional NBS specimens subsequently collected on the same patient, would incur a fee. If a laboratory or shipping error resulted in the additional specimen collection, a refund or credit would be necessary.
- Based on current expenses and assuming similar expenditures annually, a fee of approximately \$72.59 would be necessary; however, in order to overcome program inefficiencies in data management, program evaluation and education, a fee of at least \$81.45 should be initiated (see table on next page for costing details).
- Fee amounts should be reviewed annually and adjusted as appropriate with concurrence of the NBS Advisory Committee and the Approval of the Secretary of KDHE.
- No newborn should be denied screening due to fee payment issues.
- Full Medicaid reimbursement (including both the federal and state matches) should be integral to any fee discussions.
- Medicaid funds currently dispersed to the laboratory are essential for its daily operation and fee disbursements should be carefully developed so that these funds are appropriately replaced.
- All funds generated by a fee-based system should be utilized for NBS system expenses first.

Estimated Costs for Fee Implementation

Item	Current Program Costs		Enhanced Program Costs	
	Comment	Cost	Comment	Cost
Staff – Salaries and Benefits	9 lab FTEs = \$563,146 1 data entry FTE = \$35,000 4 non-lab FTEs = \$222,532	\$820,628		\$820,628
Communications	Postage, phone, etc.	24,643		24,643
Freight and Express		2,453	<u>Courier (~\$64,500)</u>	<u>66,953</u>
Printing and advertising		12,577		12,577
Rents	Clinic sites; Facilities Mgt.	76,688		76,688
Repair and services		66,462		66,462
Travel		16,730		16,730
Fees – other services	Laboratory tests	33,403		33,403
Fees – professional services	Clinic contracts	139,182		139,182
Other contractual		318	<u>Computer Support</u>	<u>150,318</u>
Food for human consumption		1,350		1,350
Motor vehicle supplies		411		411
Office supplies		10,868		10,868
Professional supplies	Medical needs; lab reagents	1,164,910		1,164,910
Capital outlay	Laboratory	146,899		146,899
Other supplies		1,951		1,951
<u>*Billing Specialist</u>	<u>Laboratory kit fee recovery</u>	<u>41,654</u>	<u>Lab kit fee recovery</u>	<u>41,654</u>
<u>Education Specialist</u>			<u>Educational training</u>	<u>45,000</u>
<u>Laboratory Specialist</u>			<u>Weekend duties</u>	<u>45,000</u>
Operating total		2,561,177		2,865,995
Estimated indirects 25% **	0.25 x \$2,241,693 =	<u>560,423</u>	0.25 x \$2,546,511 =	<u>636,628</u>
Total program cost		\$3,121,600		\$3,502,623
Cost per newborn (43,000 births) ***		\$72.59		\$81.45

*One FTE needed to handle fee issues

**Fees – Professional services and capital equipment are excluded from indirect calculations.

***This report uses 43,000 as the approximate number of births. The latest NCHS data available are for 2008 and list KS as 42,568. The fees per newborn may vary slightly based on birth numbers and should be adjusted appropriately by the end user of this report.

Underlined items are not currently a part of the program.

Report

General Logistics

On December 13-14, 2010, a Newborn Screening Financial Review Team (Review Team) (brief resumes in Appendix 1) met with selected staff members of the newborn screening program of the Kansas Department of Health and Environment (KDHE) to review program finances and make related recommendations. This review service was provided by the National Newborn Screening and Genetics Resource Center (NNSGRC) through a cooperative agreement with the Genetic Services Branch of the Maternal and Child Health Bureau (MCHB), Health Resources and Services Administration (HRSA). This review was at the request and invitation of Marc Shiff, MPA, CPM, Director, Special Health Care Needs/NBS, Bureau for Children, Youth and Families. Also present via telephone was Gary Hoffman, special consultant to the KDHE on NBS financial issues.

Logistics Details

An intensive 1½ day interview session was held with selected KDHE staff members including: Linda Kenney (Bureau of Children, Youth and Families), Marc Shiff (Children and Youth with Special Health Care Needs), Linda Williams and Jamey Kendall (Newborn Screening – Follow-up), Stacey Sandstrom (Laboratory – Chemistry Supervisor), and Colleen Peterson (Laboratory – Newborn Screening Supervisor). Additional information was contributed by Liz Abbey (Sound Beginnings Program Coordinator), Eleanor Buffington (Laboratory Fiscal Analyst), and Shelley Russell (Hicks) (Fiscal).

All Review Team members had previously visited KDHE in 2005 as part of a program review and so their primary need was review and updating to reacquaint them with administration of the NBS program. Marc Shiff provided introductory comments and Linda Kenney provided a historical review of legislation affecting NBS, including in-depth information about funding. Additionally, she reviewed all NBS fiscally related issues within the follow-up and treatment program, and provided an overview of issues related to funding sources – primarily tobacco settlement funds [designated for Children’s Initiative Funds (CIF)], Title V, and Medicaid. Stacey Sandstrom provided similar information for laboratory operations. Linda Williams, Colleen Peterson, and Jamey Kendall answered technical question relating to their respective functions. As the interview process proceeded, Liz Abbey was requested to review the operations of the newborn hearing screening (NHS) program, particularly providing information relative to Medicaid reimbursement and hospital charges. Because a significant portion of the laboratory budget currently comes from Medicaid reimbursement for NBS and certain other services, Eleanor Buffington provided necessary clarifications. Financial questions about follow-up and treatment costs were answered by Shelley Russell.

The dedication of all staff associated with the screening program and their interest in providing a fiscally sound, quality newborn screening program that meets the needs of Kansas citizens was evident.

Newborn Screening Program Information

Newborn screening (NBS) is a preventive public health program focusing on early detection and treatment for congenital conditions that can cause mental and physical disabilities, including death. NBS began with legislation requiring screening for phenylketonuria (PKU) in 1965. It has expanded over the years until its current status as defined in Kansas Statutes Annotated (KSA) §65-180 (see Appendix 2 for full text). Since July 1, 2008, the education, screening, testing and follow-up NBS program has included the 29 conditions (including hearing screening) recommended by the Secretary of Health and Human Services prior to 2010. Additionally, the enabling legislation requires maintenance of “a registry of cases including information of importance for the purposes of follow-up services” and the provision of “necessary treatment products for diagnosed cases” limited to “appropriations available therefor.” Treatment services are provided by the state only after certain financial considerations have been met as defined in the statute. An Advisory Council appointed by the Secretary of Health and Environment advises the KDHE on the rules and regulations governing the program. The KDHE is tasked with periodically reviewing the program for efficacy and cost effectiveness, and to determine adjustments that may be needed in order to maximize program outcome.

There are approximately 43,000 births annually (42,568 in 2008 reported by the National Center for Health Statistics – latest figures available) in Kansas with a majority occurring in 10 birthing facilities. The number of out-of-hospital births is not known. Approximately 40% of all Kansas births qualify for Medicaid reimbursement. Over 90% of all births are recorded via electronic birth certification. NBS record keeping occurs through a combination of manual records and unlinked computer systems (one in laboratory and one in follow-up) that are limited to the most basic program needs. A comprehensive data system capable of managing patient and specimen information from the initial hospital patient encounter through maintenance of long-term outcome indicators, including linkage to birth records and other child health programs, is not available, primarily due to cost considerations. As a result, the program currently suffers from less than optimal record management and future electronic healthcare efforts targeting computerized medical information exchange will likely be impacted. Limited electronic capabilities also appear to contribute to the slow reporting of laboratory quality assurance data to the national reporting system (data has not been reported for the years 2008-2010) and may be limiting internal quality assurance efforts. Additionally, limited computer technology contributes to the fact that there is currently no efficient mechanism for comparing births to specimens tested to ensure that all Kansas babies are being screened. A NBS serial number is currently a part of the screening record of each newborn and could be included in the official birth record as a simple means of record linking. This linking concept has been favorably discussed internally, but there has been no action taken yet to implement it.

There is currently no NBS fee in Kansas, making it one of four programs without such a fee (NY, DC, PA, KS). It should be noted, however, that the programs in PA and DC include fees charged to patients for screening tests by hospitals. No data exists in Kansas regarding possible hospital charges for specimen collecting and administering the NBS program. At the present time, Kansas NBS funding arises primarily from CIF with some additional monies available from Medicaid, Title V Block Grant and general

revenue. Medicaid funds are only available in the laboratory and currently do not support any non-laboratory activities despite the fact that other activities comprise the newborn screening program [e.g. administration, education (patient, provider, and policy maker), follow-up, tracking, etc.]. In the case of newborn hearing screening (NHS), Medicaid costs for testing are reimbursed to hospitals depending on the type of testing provided (otoacoustic emission = \$54.58; auditory brainstem response = \$44.67). No data were available to the Review Team regarding total Medicaid expenditures for NHS.

KNSP Program Operations

The KNSP laboratory staff currently includes 9 personnel (2 Senior Laboratory Scientists, 2 Laboratory Scientists, 2 Microbiologists, 1 Chemist, 2 Laboratory Technicians, 1 Senior Administrative Specialist), exclusive of the data entry staff, who are in a separate unit (approximately 1 FTE is used for this activity). Testing in the laboratory occurs 5 days/week (40 hr. work week). The KNSP laboratory receives approximately 50,000 specimens annually (the number of specimens is much greater than the number of births because some pediatric practices routinely obtain a second specimen at the first outpatient visit, intensive care units may submit multiple specimens on the same baby, repeat specimens may be required to confirm initial findings, and specimens of unsatisfactory analytical quality may require recollection/testing). A state courier service, which collected specimens for other laboratory programs (primarily serving country health departments) previously transported approximately 10% of total specimens, but this service has been discontinued and essentially all specimens are now conveyed by the U.S. Postal Service. Bar coded serial numbers on specimen collection cards (kits) are linked to the birthing facilities receiving them as a means of monitoring collection kit inventories. These serial numbers are included in the data entered for each received specimen, and they enable automatic printing of facility mailing information when returning results. Phone calls are made to submitters in order to identify the newborn's physician on all specimens arriving without this information. Reports of laboratory results are provided to both the birthing facility and the physician identified on the submission form. For primary care physicians with fax numbers on file, reports are automatically faxed (approximately 60% are faxed). Most laboratory reports are finalized and submitted within two days of specimen receipt. All residual blood specimens are retained at -20°C for one month and then autoclaved and destroyed with one exception – specimen cards that exhibited abnormal results are retained indefinitely.

The non-laboratory part of the KNSP (administration/follow-up) consists of 4 personnel (2 Public Service Executive I, 1 Administrative Specialist, 1 Administrative Assistant). Several contracted medical consultants are available to the program to provide follow-up (including treatment/management. All (in addition to others) serve as program advisors. The administrative/follow-up staff is responsible for tracking all suspect screening results to ensure appropriate follow-up and confirmatory testing. These staff members also undertake public and professional education, and other system-wide administrative functions, including oversight of treatment/management protocols. As part of program services, metabolic foods and formula, and treatment drugs for other conditions, may be supplied to each patient on a sliding fee scale based on income criteria. While collection and evaluation of long-term outcome data are essential to evaluating and improving NBS, there are currently only limited efforts in this regard. Because long-term outcome follow-up requires funding, it must be included in costing

considerations.

KNSP Costs

As a part of this review, current cost information was supplied by both the laboratory and non-laboratory arms of the KNSP. These data have been summarized in Table 1. An additional employee identified as a “Billing specialist” has been listed as a laboratory expense (a necessary expense for fee administration identified by the program during the review). “Estimated indirect” expenses of 25% of direct costs (excludes professional fees and capital equipment) have also been included to give a total current expenditure estimate of \$3,121,650, or approximately \$72.59 per newborn (assuming 43,000 births), should a fee be established in the current program configuration. This fee assumes that professional fees, capital outlay, facilities management charges, etc. remain fairly consistent each year.

Table 1. Current Costs for Fee Implementation (supplied by program)

Item	Non-Laboratory Program Costs		Laboratory Program Costs	
	Comment	Cost	Comment	Cost
Staff – Salaries and Benefits	4 FTEs	\$222,532	9 FTEs = \$563,146 1 FTE (data entry pool) = \$35,000	\$598,146
Communications	Postage, phone, etc.	12,000		12,643
Freight and Express		500		1,953
Printing and advertising		3,000		9,577
Rents	Clinic sites	3,000	Div. of Facilities Mgt.	73,688
Repair and services		500		65,962
Travel	In-state = 3,000 Out-of-state = 9,381	12,381	Combined in-state and out-of-state	4,349
Fees – other services	Laboratory tests	33,153		250
Fees – professional services	Clinic contracts	137,874		1,308
Other contractual		318		
Food for human consumption		1,350		
Motor vehicle supplies		200		211
Office supplies		6,500		4,368
Professional supplies	Medical needs, inc. CYSHCN	154,000	Laboratory reagents	1,010,910
Capital outlay				146,899
Other supplies		<u>1,951</u>		
*Billing Specialist			Additional staff for fee recovery	<u>41,654</u>
	Operating total	589,259	Operating total	1,971,918
**Estimated indirects 25%	0.25 x \$418,232 =	<u>104,558</u>	0.25 x \$1,823,461 =	<u>455,865</u>
	Total	\$693,817		\$2,427,783

Total estimated expenditures for current program in fee configuration = \$3,121,650 or \$72.59 per newborn (43,000 births).

*One FTE needed to handle fee issues

**Fees – Professional services and capital equipment are excluded from indirect calculations.

The Review Team was asked to develop additional costing information for program improvements that would result in a more efficient and effective Kansas NBS program in today's electronic environment. In particular, there were major concerns raised during the interview process regarding the lack of a comprehensive computerized system of data management and record keeping, including proper maintenance of a statutorily required disease registry. Several commercial systems exist that might meet the needs of the KNSP. In general, these systems begin with an electronic record at the birthing facility and allow for patient information and specimen tracking through the screening process (specimen collection, transport to the screening laboratory, laboratory testing), short term follow-up to diagnosis, and long term follow-up through treatment/management and beyond (so that program and patient accomplishments can be evaluated). Additionally, an automated system should include 24 hour access to result reports either through voice response or secure on line reporting. Capability for internal linking to other related systems such as vital records should be available.

In order to obtain a system that best meets the needs of the program, multiple vendors should be asked to explain their current operations in other states and countries. From this information, the program will likely be able to generate a technical specification to guide the bidding process. Quick estimates obtained at the time of the Review Team visit seemed to indicate an initial outlay of approximately \$500,000 with recurring maintenance costs of approximately \$70,000. Subsequent information obtained by Review Team members suggests that annual costs of approximately \$100,000-\$150,000 per year long-term would be an appropriate estimate to use in fee considerations and would include amortized costing of both software and hardware, likely for 5-7 years, in some combination with ongoing maintenance. Because there are many ways in which to package a commercial system, it is difficult to estimate this cost, but it will likely be in the range of \$2.50- \$3.00 per specimen per year.

Two other major program enhancement areas involve testing service and education. Since many of the conditions now being screened as part of the KNSP involve disorders that have severe mortality and/or morbidity within the first few days of life, faster turnaround of results is essential. Two major influences on faster service involve specimen transit and laboratory testing. In the former instance, a courier service that reaches a large majority of the birthing centers is desirable. While a courier service for other laboratory testing programs was previously in place, it apparently did not efficiently reach the birthing facilities that submit NBS specimens. Careful attention to program needs should be addressed in any courier contract, including Saturday/holiday transport. The Review Team recommends an expansion of laboratory testing services and related follow-up to include weekends and holidays. In order to provide screening results within 5-7 days of birth for time critical disorders, a 6-day laboratory (and subsequent follow-up) work week for a portion of the testing staff is necessary. On special holidays, specimen transport may be problematic, but in no case should there be more than 2 consecutive days of laboratory closure. While scheduling can be challenging and staffing requirements may increase slightly, this type of testing service is needed since time-critical screening already exists in the KNSP. We estimate that a courier cost of approximately \$1.25-\$1.50 per patient would be required to meet the transit requirement and an increase of approximately 1 FTE in the laboratory (approximately \$45,000 or \$1.00 per newborn) would be required. Therefore, an increased cost of

approximately \$2.50 per newborn would be required to meet the overall service enhancement.

Education (including consumers, healthcare providers, and policy makers) was also considered to be an area of need and concern. Not only is it necessary to provide both pre-screening information about the program and the methodology for specimen collection and result reporting, but it is also increasingly important to provide post-screening information for physicians and family members about disorders that might have been detected, possible services available, and outcome measures that should be monitored. While residual NBS blood spots are not currently saved for research in Kansas, there is increasing interest among consumer groups regarding the ultimate disposition of their baby's blood specimen. Other programs are finding that education about disposition of residual dried blood spots requires increasing attention and the same is likely true in Kansas

Professional educational efforts can be combined with electronic quality assurance monitoring to improve program efficiencies. For example monitoring unsatisfactory/unacceptable specimen rates can identify submitters requiring additional specimen collection education where rates are the highest. This in turn can significantly reduce the number of babies requiring collection of a subsequent because of analytical quality issues. Fewer specimen rejections can enhance support of the program by the medical and consumer community. Proper attention to all of the educational needs identified will likely require the services of 1 additional FTE in the follow-up group at a cost of approximately \$45,000 annually or about \$1 for each newborn screened.

Based on the calculations above, an additional charge of approximately \$7 per newborn would cover most costs associated with program improvement. As treatment/management costs increase, it might be prudent to plan for some increase to cover these costs. On this basis, a fee of approximately \$82 would seem appropriate to cover the program and the improvements noted above. While this fee was calculated on the basis of 43,000 newborns, the usual number of specimens received would probably be higher (based on previous history). Any extra funds would likely be useful for replacement equipment or, perhaps, equipment for expansion. Of course, as national standards of practice change and new conditions are added to the screening program, cost adjustments will be necessary. Already it is anticipated that additional costs for adding Severe Combined Immunodeficiency (SCID) screening would increase costs in the neighborhood of \$5 per newborn.

As noted in the AAP Task Force Report {[*Pediatrics* 2000;106(suppl 2):383-427] – see Executive Summary in Appendix 4}, it is strongly recommended that the entire newborn screening system be supported in such a manner that if a fee exists, it should first pay for all newborn screening system expenses before it is absorbed into government general revenues for other programs. Table 2 (p. 12) provides a listing of the fees currently charged for newborn screening in all U.S. programs. The latest published information is used even though it exists in different years for different items. A comparison of fees in 2006 versus 2010 has been listed to show the tendency among programs to increase fees as program expansion needs are realized.

Table 2. Newborn Screening Births, Percent Medicaid Births, and Screening Fees (Latest published data)

State	Births (Occurrence) In 2008 ^a	Percent Medicaid births (2003 ^b)	Number of screens required	Fee 1/2006	Current Fee 1/2011	Notes
Alabama	63,450	45.5	1	\$139.33	\$125.57	Two screens strongly recommended.
Alaska	11,329	55.1	1	\$55.00	\$75.00	Fee includes any repeats. Two screens recommended.
Arizona	100,089	50.5	2	\$20.00	\$70.00	Separate fee for each mandated specimen.
Arkansas	39,502	51.7	1	\$14.83	\$89.25	
California	552,618	44.3	1	\$78.00	\$102.75	
Colorado	70,527	37.3	2	\$53.25	\$85.00	Fee includes 2 mandated specimens (2-part form).
Connecticut	40,930	28.4	1	\$28.00	\$56.00	
Delaware	12,545	41.0	2	\$64.00	\$98.00	Fee includes 2 mandated specimens and any repeats.
District of Columbia	14,499	33.7	1	No Fee	No Fee	Hospitals pay screening lab. District does not charge a fee.
Florida	231,652	49.6	1	\$15.00	\$15.00	
Georgia	147,799	50.0	1	No Fee	\$40.00	
Hawaii	19,463	27.2	1	\$47.00	\$55.00	
Idaho	24,676	39.7	1	\$23.00	\$30.00	\$46 for double kits if screening occurs prior to 48 hrs.
Illinois	173,410	39.9	1	\$47.00	\$59.00	
Indiana	89,345	41.2	1	\$62.50	\$85.00	Includes \$32.50 laboratory surcharge and all repeats.
Iowa	40,281	28.1	1	\$56.00	\$77.00	Fee includes any repeats.
Kansas	42,568	39.6	1	No Fee	No Fee	
Kentucky	56,621	43.7	1	\$14.50	\$53.50	
Louisiana	65,073	58.7	1	\$18.00	\$30.00	Fee expected to increase to \$40.00 later in 2005.
Maine	13,500	47.0	1	\$44.00	\$110.00	
Maryland	74,615	34.0	1	\$42.50	\$42.00	Fee includes repeats; 2 screens strongly recommended.
Massachusetts	77,543	29.5	1	\$54.75	\$54.75	
Michigan	120,172	35.5	1	\$55.72	\$87.03	Fee includes any repeats.
Minnesota	72,220	36.6	1	\$61.00	\$101.00	
Mississippi	44,139	60.0	1	\$70.00	\$70.00	
Missouri	81,992	45.4	1	\$25.00	\$65.00	
Montana	12,551	35.0	1	\$39.34	\$91.70	
Nebraska	27,082	39.6	1	\$30.75	\$38.50	
Nevada	39,192	21.0 ^c	2	\$60.00	\$60.00	Fee includes 2 mandated specimens (2-part form).
New Hampshire	13,630	23.3	1	\$18.00	\$71.00	
New Jersey	109,703	24.2 ^c	1	\$71.00	\$95.00	
New Mexico	29,572	51.7 ^c	2	\$32.00	\$89.00	Fee includes 2 mandated specimens (2-part form).
New York	252,360	40.5	1	No Fee	No Fee	
North Carolina	132,106	47.9	1	\$10.00	\$19.00	
North Dakota	10,312	30.0	1	\$36.00	\$60.00	
Ohio	149,346	32.1	1	\$33.75	\$55.16	
Oklahoma	53,720	49.5	1	\$75.59	\$120.33	Fee includes hearing screening.
Oregon	49,499	42.6	2	\$54.00	\$54.00	Fee includes 2 mandated specimens (2 -part form)
Pennsylvania	148,460	31.0	1	No Fee	No Fee	Most hospitals charge a fee for expanded testing. Fees vary.
Rhode Island	12,812	37.0	1	\$59.00	\$110.00	
South Carolina	60,401	55.3	1	\$42.00	\$68.51	
South Dakota	12,631	36.0	1	\$18.53	\$60.00	
Tennessee	90,885	46.2	1	\$47.50	\$75.00	
Texas	412,127	47.6 ^c	2	\$19.50	\$34.50	Separate fee for each mandated specimen.
Utah	56,787	30.2	2	\$31.00	\$75.00	Fee includes 2 mandated specimens (2-part form).
Vermont	5,957	47.6	1	\$33.30	\$95.00	
Virginia	104,990	27.6	1	\$32.00	\$53.00	
Washington	90,318	45.6	1	\$60.90	\$64.40	Fee includes repeats; 2 screens strongly recommended.
West Virginia	21,441	50.0	1	No Fee	\$45.00	
Wisconsin	71,272	38.1	1	\$65.50	\$69.50	\$30.00 laboratory surcharge included in fee.
Wyoming	7,444	46.0	1	\$45.00	\$70.00	

(a) Latest figures released by the National Center for Health Statistics

ftp://ftp.cdc.gov/pub/Health_Statistics/NCHS/Dataset_Documentation/DVS/nativity/UserGuide2008.pdf (accessed Jan 2, 2011)

(b) Latest published statistics - from Kaiser State Health Facts Online, <http://www.statehealthfacts.org/comparemaptable.jsp?ind=223&cat=4> (accessed Jan. 2, 2011)

(c) 2003 Medicaid statistics unavailable so statistics are taken from http://www.statemaster.com/graph/hea_as_per_of_sta_bir-births-financed-medicaid-percent-state (accessed Jan. 2, 2011).

Fee Collection

Several models for fee collection exist, but the most popular is the sale of newborn screening kits to birthing facilities and other specimen submitters. In this model, all system costs are calculated using standard accounting procedures and the cost of screening averaged across all births to calculate a screening kit charge. Kits are then sold (usually through an ordering and shipping process based in the screening laboratory) to specimen submitters. There are several methods for ensuring that the cost of the kits covers all system expenses (including patients who cannot pay, repeat specimens, ongoing testing improvement, future expansion, etc.) and that hospitals can recover appropriate costs from Medicaid. The important points are to ensure that the system costs are comprehensive and that Medicaid reimbursements for hospitals are appropriately determined. Because some physicians or birthing centers may prefer to send additional screening specimens on some patients and the number of such specimens is difficult to predict, it is strongly suggested that a fee be assessed for any specimen tested, unless it is the result of a laboratory or shipping error (in which case a refund or credit must be given). Experience in other programs has shown this billing process to be efficient and to assist in improving the quality of the specimens submitted (since poor quality specimens would result in an additional charge unless there was a laboratory or shipping error).

The Review Team recognizes that NBS Medicaid cost recovery issues in Kansas may be complex and, for KDHE laboratory services, may be linked to other funding issues. Medicaid funds currently contracted to the laboratory are essential for its daily operation and any fee and fee disbursement must be carefully developed so that these funds are appropriately replaced. This will likely require a reevaluation more comprehensive funding issues within the laboratory – complexities beyond the scope of this report. Laboratory fiscal information indicated a current annual Medicaid contractual income of \$322,654 based on the number of Medicaid patients served and program costs (per contract with the Kansas Public Health Authority). The mix of state and federal funds was unclear. No income from Medicaid was identified for the non-laboratory portion of the program. It was further noted that approximately 40% of Kansas births qualify for Medicaid. Likewise it was noted that there are Medicaid payments made for NHS, although a total amount of expenditure was not available. The state portion of the Medicaid match for Kansas is understood to be approximately 40%. Thus, assuming a current fee budget of \$3,121,650 (\$72.59 per newborn), Medicaid would presumably expend (through fee payments) \$1,248,660 (of which the federal portion would be \$749,196 and the state portion would be \$499,464). For the optimized program discussed, with a fee of \$81.45 per newborn (\$3,502,623 total budget), Medicaid payments would be expected to be \$1,401,049 (federal match = \$840,629; state match = \$560,420).

In the vast majority of state NBS programs, birthing facilities pay for screening kits as they are ordered and, in turn, recover their costs from insurers and Medicaid as part of their newborn delivery charges (including any additional administration or specimen collection fee). In the billing described here (sale of screening kits), Medicaid reimbursement schedules to hospitals will need to be adjusted so that hospitals do not incur unnecessary revenue loss. The KDHE will need to take a proactive role in assisting hospital in recovering both Medicaid and other third party payments. Likewise, Medicaid funds currently being accessed by the laboratory play an important role in overall

laboratory operations, and their replacement with fee funds must be a consideration in costing plans.

In order to ensure that program income is sufficient to cover all expenses, an annual cost accounting review should be conducted and its results reported to the NBS Advisory Committee and the Secretary of KDHE. If appropriate, fee adjustments should be considered. Rather than continually seeking Legislative approval for fee adjustments, program recommendations should be presented to the Advisory Committee for their concurrence, and a final recommendation should be presented to the Secretary of KDHE for approval and implementation. In cases where a new or improved process may require additional funds for implement not covered by the existing fee (either for personnel, equipment, reagents or any combination), then a temporary surcharge may be necessary. Absent a process for funding new or improved testing, including expansion to other disorders, it may be necessary to seek assistance from the Legislature.

Appendix 1

Brief Resumes of Review Team Members (Alphabetical Order)

BRIEF RESUME

WILLIAM HARRY HANNON

ADDRESS

4929 Duncans Lake Point
Buford, GA 30519
Email: whannon@bellsouth.net

EDUCATION

Georgia State University, B.S., 1965, Chemistry
University of Tennessee, Ph.D., 1972, Biochemistry
Oak Ridge National Laboratories, 1974, Post-Doctoral

PROFESSIONAL SOCIETY MEMBERSHIPS

Charter Member: International Society for Newborn Screening (ISNS) – 1987 - present;
Member of ISNS Executive Council (1999-2010); Vice-President (2002-2010)
Member of four other organizations.

PROFESSIONAL COMMITTEES

1991-present: International Quality Assurance Working Group - ISNS
1991-present: Newborn Screening and Genetics Committee –Liaison
Association of Public Health Laboratories
2005 – present: Member of HHS Secretary's ACHDNC, Subcommittee for Laboratory.
2010 – present: Member, Area Committee for Immunology and Ligand Assays
Clinical and Laboratory Standards Institute (CLSI)
Serves or served on 20+ other national and international committees for quality assurance,
and standards development committees for laboratory improvement, and 30+ organizing
committees for conferences and symposia.

EXPERIENCE

2010–present: Consulting services for newborn screening activities with NNSGRC.
1/5/2009 - Management/Consultant [McKing Consulting Corporation, Atlanta, Georgia] 9/1/2010
Provided management consultations and program support for the daily
operation of the Newborn Screening and Molecular Biology Branch,
Division of Laboratory Sciences (DLS), CDC and to the Acting Branch
Chief/Division Director, DLS for both scientific and administrative activities.
1/2/2009 Retired from Centers for Disease Control and Prevention with 40 years service.
2007-2008 Acting Branch Chief, Newborn Screening and Molecular Biology Branch, CDC.
1982-2007 Chief, Newborn Screening Branch, Division of Environmental Health
Laboratory Sciences, National Center for Environmental Health, Centers for
Disease Control and Prevention, Atlanta, Georgia 30341. Branch projects
involve operation of the National Quality Assurance Program for Newborn
Screening (NSQAP) for inborn errors of metabolism, sickle cell disorders
and other hemoglobinopathies, HIV antibody assays with dried-blood spots,
immunoassay and tandem mass spectrometry methods for fatty acid
oxidation disorders, amino acid disorders, organic acid disorders, and
assays for immune function disorders. NSQAP provides dried blood spot
proficiency testing and quality control materials, performance reports, and
technical consultations to 352 NBS laboratories in 53 countries.

PUBLICATIONS

Over 275 publications in scientific journals and proceedings with most of these concerning filter paper tests and newborn screening issues, e.g.,

Blood collection on filter paper for newborn screening programs: approved standard-fifth edition. Wayne (PA):CLSI; 2007 CLSI Document LA4-A5.

Using tandem mass spectrometry for metabolic disease screening among newborns. MMWR Recommendations and Reports April 13, 2001; 50:1-34.

Contribution of selected metabolic diseases to early childhood deaths-Virginia, 1996-2001. MMWR 2003; 52 (29):677-679.

Applying public health strategies to primary immunodeficiency diseases. MMWR Recommendations and Reports, 2004; 53:1-29.

Improved MS/MS analysis of succinylacetone extracted from dried blood spots when combined with amino acid analysis and acylcarnitine. Clin Chim Acta, 2009; 407:6-9.

Newborn screening systems performance evaluation program and assessment scheme (PEAS). Semin Perinatol 2010;34;105-120.

Development of a DNA-based cystic fibrosis proficiency testing program for newborn screening. Clin Chim Acta 2010; (submitted)

Author of 14 chapters in scientific books.

SPECIAL ACTIVITIES

Consultant – 2 international publications; *Screening and Infant Screening Newsletter*

Organizing committee – 30+ National and International Newborn Screening Symposia

Co-author for 2 World Health Organization guidelines on Prevention and Control of Congenital Hypothyroidism and Phenylketonuria.

Served as a member of the Expert Site Review Team for NBS – Reviewing over 34 state programs.

Serve on the Georgia Governor's Public Health Advisory Committee, 2009 to present.

AWARDS

Recipient of 35+ Public Health Service special recognition and service awards, e.g.,

1999 - ISNS Robert Guthrie Award for "Worldwide recognition of outstanding contributions to newborn screening."

2006 - Sigma Xi's Walter Dowdle Award for Achievements in Public Health Laboratory Science.

July 2006. For creating the Newborn Screening Quality Assurance Program and his many other contributions to the health and well-being of newborn infants.

2008 - Lifetime Achievement Award from Association of Public Health Laboratories (APHL)

for leadership in the field of public health laboratory science and influencing public health policy on a national and global level.

2008 - Russell J. Eilers Memorial Award, highest honor that Clinical and Laboratory Standards Institute (CLSI) confers, for outstanding contributions to consensus laboratory standards.

2008 - A global newborn screening award was named in my honor, "The Harry Hannon Laboratory Quality Improvement Award". The award is sponsored by APHL.

2009 - Dream Makers Award, "Pioneers in Newborn Screening," April 2009, Jeffrey Modell Foundation, New York City, NY

BRIEF RESUME

JULIE LUEDTKE

ADDRESS

Newborn Screening & Genetics Program
Nebraska Health & Human Services
301 Centennial Mall South
P.O. Box 95044
Lincoln, NE 68509-5044

EDUCATION

1983 B.S., University of Nebraska, Lincoln - Education, Community Health Education
1985-86 University of Nebraska, Lincoln – Graduate work – Human Development and the Family
2002-04 University of Nebraska Medical Center - Master's of Public Health work

EXPERIENCE

1/85-4/96 Developmental Disabilities Surveyor/Consultant, Nebraska Department of Health, Bureau of Health Facilities Standards
4/96-Present Program Manager, Newborn Screening and Genetics Program and State Genetic Coordinator, Nebraska Department of Health and Human Services, Division of Public Health, Lifespan Health Services

PROFESSIONAL ACTIVITIES

4/96-4/98 Member, GPGSN State Genetic Coordinators Committee
4/97-10/99 Member, GPGSN Newborn Screening Committee
10/97-04/00 Member, Nebraska Family Health Conference Planning Committee
10/97-present Member, Nebraska Chapter March of Dimes Program Services Committee
1/98-4/98 Member, Neb. Dept. of Health Public Health Week Promotion Committee
4/98-10/99 Chairperson, GPGSN State Genetic Coordinators Committee
1/99-4/02 Co-Chair, Nebraska Neural Tube Defects Prevention Campaign Coalition
11/00-01/03 Chair, Nebraska Chapter March of Dimes Program Services Committee
9/02-02/08 ACMG Work Group on Newborn Screening Follow-up and Diagnosis
1/03-05/04 Member, Planning Committee, National NBS and Genetics Symposium
9/04-12/05 NCCLS Subcommittee on NBS Follow-up Guidelines
9/04-present Member NNSGRC PEAS Committee to develop performance evaluation indicators for newborn screening
4/05-4/06 Member Nebraska Family Health Conference Planning Committee (2006)
9/05-12/08 Member - US HHS Secretary's Advisory Committee on Heritable Diseases in Newborns and Children, Subcomm. on NBS Follow-up and Management
2/05-present Member Advisory Council for the Heartland Regional Genetics Collaborative (HRGC)
10/05-2008 Member State Genetics Coordinators Committee of HRGC
10/05-present Member Newborn Screening Advisory Committee of HRGC
10/05-10/06 Executive Secretary to the Education Committee of HRGC
4/06-present Member Clinical Laboratory Standards Institute Subcommittee on Blood

- 5/07-present Collection on Filter Paper for Newborn Screening Programs, 5th Edition
Member Association of Public Health Laboratories Subcommittee on
Newborn Screening & Genetics in Public Health
- 7/07-present Co-Chair Clinical Laboratory Standards Institute Committee on Guidelines
for NBS Practices for Sick and Premature newborns in the NICU.
- 11/07-11/08 Member 2008 NBS Symposium Planning Committee
- 10/09-present National Coordinating Center for Translational Research Sub-Committee on
Long Term Follow-up Data Elements for Newborn Screening
- 10/09-present Member Nebraska Dept. Health & Human Services Legislative Liaison
Coordinating Council

INVITED PRESENTATIONS

- 10/2002 Great Plains Chapter of Clinical Laboratory Managers Association Fall
Meeting, Omaha, NE: “Nebraska Newborn Screening Program, Present and
Future.”
- 01/2003 3rd Annual MS/MS Program Implementation Meeting: Improving the Efficacy
and Effectiveness of Tandem Mass Spectrometry Screening, Berkeley
California: “Health education resources for families and health professionals.”
- 01/2003 3rd Annual MS/MS Program Implementation Meeting: Improving the Efficacy
and Effectiveness of Tandem Mass Spectrometry Screening, Berkeley
California: “Informed consent and informed decision making: Issues and
strategies for new MS/MS screening programs.”
- 04/2004 Great Plains Chapter of Clinic Laboratory Management Association Spring
Meeting, Omaha, NE: “Newborn Screening in Nebraska, An update”.
- 05/2004 APHL National NBS and Genetics Symposium:, Atlanta, Georgia, “State &
Private Partnerships for Newborn Screening, Nebraska’s Experience”.
- 04/2006 NBS Presentation at Central Plains Society of American Medical Technologists
- 06/2006 NBS Presentation at Kansas Governor’s Children’s Health Committee meeting
- 05/2007 APHL National NBS Symposium, presentations on using national newborn
screening data for quality improvement, and using the Performance Evaluation
and Assessment Scheme to improve newborn screening.
- 11/2008 Nat’l NBS Symposium, San Antonio: “CLSI, Newborn Screening Guidelines
for Premature, Sick and Low-Birthweight Newborns in the NICU - an Update”.
- 05/2010 Newborn Screening for Preterm, Low BirthWeight, and Sick Newborns;
Approved Guideline, Pre-Nat’l NBS Symposium, Orlando, FL: Pre-conference
workshop on quality assurance for NBS, “CLSI Newborn Screening Guidelines
for Premature, Sick and Low Birthweight Newborns in the NICU.”

POSTERS

- 11/2008 Nat’l NBS Symposium, San Antonio: “A Policy Analysis of Dried Blood Spot
Testing for CMV & Genetic Causes of Hearing Loss”
- 03/2010 Dried Blood Spot Testing for Congenital Cytomegalovirus and Genetic Causes
of Hearing Loss: A Public Health Perspective. Early Hearing Detection and
Intervention National Symposium, Chicago, IL Hoffman, J, Miller J, Johnson T,
Hoefler F.

BRIEF RESUME

BRADFORD L. THERRELL, JR.

ADDRESS

Director, National Newborn Screening and Genetics Resource Center
1912 West Anderson Lane Suite 210
Austin, Texas 78757

EDUCATION

B.S. 1966 - Chemistry - Mississippi College (Special Distinction, Honors)
M.S. 1969 - Inorganic Chemistry - The Florida State University
Ph.D. 1971 - Inorganic Chemistry - The Florida State University

CERTIFICATION

American Board of Bioanalysis - High-Complexity Clinical Laboratory Director

EXPERIENCE

1999 - Professor, Department of Pediatrics, UTHSCSA, San Antonio, Texas
1999 - Director, National Newborn Screening and Resource Center, Austin, Texas
1979 - 1999 Chemical Services Division Director, TDH Bureau of Laboratories, Austin, Texas
1974 - 1979 Clinical Chemistry Branch Supv., TDH Bureau of Laboratories, Austin
1971 - 1973 Project Director (Chemist) of Title XIX Laboratory Project

PROFESSIONAL ACCOMPLISHMENTS

1999-present Director, U.S. Nat'l Newborn Screening and Genetics Resource Center
1997-present Editorial Board - *JOURNAL OF MEDICAL SCREENING*
1997-present Editorial Board - *GENETIC TESTING*
1996-1999 Chairperson - TEXGENE Newborn Screening Committee
1995-present Expert Reviewer - International Atomic Energy Agency
1995-present NCCLS Subcommittee on Newborn Screening
1995-1998 Secretary of Policy-Council of Regional Networks for Genetic Services
1993-1999 President - International Society for Neonatal Screening (ISNS)
1991-1995 Chairperson - Newborn Screening Committee, CORN
1991-1996 Co-Editor - *SCREENING* (Journal of the ISNS)
1987-present U.S. Health and Human Services Select Panel on Neonatal Screening
1987-1999 Editor-in-Chief - *Infant Screening* (International Newsletter)
1987-1993 Secretary - International Society for Neonatal Screening (ISNS)

INTERNATIONAL ACTIVITIES

1982-present Over 75 invited lectures presented in foreign countries
1987-present Founding member, Secretary, and President of the ISNS
1988-present Member of organizing committees for 15 foreign screening meetings.
1991-present Faculty - Technology for Infantile and Neonatal Screening - Sapporo
1995-present Expert review activities for 15 foreign projects to improve infant health

PUBLICATIONS

BOOKS – Editor or co-editor of four books including:

Therrell BL Jr (ed) *Laboratory Methods in Neonatal Screening*. Washington, DC: American Public Health Association, 1993.

Therrell BL Jr (ed) *Advances in Neonatal Screening*. Amsterdam: Elsevier Science Publishers, 1987.

CHAPTERS – Author or co-author of five book chapters - Abbreviated titles:

Screening for congenital hypothyroidism, Automation and computerization, Laboratory methods for hypothyroidism, Hemoglobinopathy screening techniques for newborns, and Methods for phenylalanine analysis in newborns, Newborn Screening for CAH

MONOGRAPHS – Author or co-author of 6 monographs including 2 for the World Health Organization (*Guidelines for prevention and control of hypothyroidism, and Guidelines for prevention and control of phenylketonuria*).

ARTICLES – Author or co-author of over 115 scientific articles in the areas of: public health policy, computerization, automation, chemistry, microbiology, endocrinology, hematology, and newborn screening including:

Therrell BL Jr, Panny SR, et.al. U.S. Newborn screening system guidelines: statement of the Council of Regional Networks for Genetic Services. *Screening* 1992;1:135-147.

Therrell BL, Hannon WH, et.al. Guidelines for the retention, storage, and use of residual blood spot specimens after newborn screening analysis: Statement of the Council of Regional Networks for Genetic Services. *Biochem and Molec Med* 1996;57:116-124.

Meaney FJ, Kinney S, Therrell BL, et.al. Assessing genetic risks - implications for health and social policy: response from the Newborn Screening Committee of the Council of Regional networks for Genetic Services. *Screening* 1996;4:247-249.

Therrell BL, Berebaum SA, Manter-Kapanke V, et.al. Results of screening 1.9 million Texas newborns for 21-hydroxylase-deficient congenital adrenal hyperplasia. *Pediatrics* 1998;101:583-590.

Therrell BL. U.S. newborn screening policy dilemmas for the twenty-first century. *Molec Genetics and Metab* 2001 ; 74:64-74.

Larsson A and Therrell BL. Newborn screening: the role of the obstetrician. *Clin Obstetr Gynecol* 2002; 45:697-710.

Appendix 2

Kansas Newborn Screening Statute

Kansas Statutes Annotated (KSA) 65-180

Kansas Statutes Annotated (KSA) 65-180

Chapter 65: Public Health

Article 1: Secretary Of Health And Environment, Activities

Statute 65-180: Educational, screening, testing and follow-up program concerning phenylketonuria, congenital hypothyroidism, galactosemia, maple syrup urine disease and certain other genetic diseases; registry of cases; food and treatment products; reimbursement of cost; eligibility; newborn screening programs. The secretary of health and environment shall:

(a) Institute and carry on an intensive educational program among physicians, hospitals, public health nurses and the public concerning congenital hypothyroidism, galactosemia, phenylketonuria and other genetic diseases detectable with the same specimen. This educational program shall include information about the nature of such conditions and examinations for the detection thereof in early infancy in order that measures may be taken to prevent the mental retardation or morbidity resulting from such conditions.

(b) Provide recognized screening tests for phenylketonuria, galactosemia, hypothyroidism and such other diseases as may be appropriately detected with the same specimen. The initial laboratory screening tests for these diseases shall be performed by the department of health and environment or its designee for all infants born in the state. Such services shall be performed without charge.

(c) Provide a follow-up program by providing test results and other information to identified physicians; locate infants with abnormal newborn screening test results; with parental consent, monitor infants to assure appropriate testing to either confirm or not confirm the disease suggested by the screening test results; with parental consent, monitor therapy and treatment for infants with confirmed diagnosis of congenital hypothyroidism, galactosemia, phenylketonuria or other genetic diseases being screened under this statute; and establish ongoing education and support activities for individuals with confirmed diagnosis of congenital hypothyroidism, galactosemia, phenylketonuria and other genetic diseases being screened under this statute and for the families of such individuals.

(d) Maintain a registry of cases including information of importance for the purpose of follow-up services to prevent mental retardation or morbidity.

(e) Provide, within the limits of appropriations available therefor, the necessary treatment product for diagnosed cases for as long as medically indicated, when the product is not available through other state agencies. In addition to diagnosed cases under this section, diagnosed cases of maple syrup urine disease shall be included as a diagnosed case under this subsection. Where the applicable income of the person or persons who have legal responsibility for the diagnosed individual meets Medicaid eligibility, such individuals' needs shall be covered under the Medicaid state plan. Where the applicable income of the person or persons who have legal responsibility for the diagnosed individual is not Medicaid eligible, but is below 300% of the federal poverty level established under the most recent poverty guidelines issued by the United States department of health and human services, the department of health and environment shall provide reimbursement of between 50% to 100% of the product cost in accordance with rules and regulations adopted by the secretary of health and environment. Where the applicable income of the person or persons who have legal responsibility for the diagnosed individual exceeds 300% of the federal poverty level established under the most recent poverty guidelines issued by the United States department of health and human services, the department of health and environment shall provide reimbursement of an amount not to exceed 50% of the product cost in accordance with rules and regulations adopted by the secretary of health and environment.

(f) Provide state assistance to an applicant pursuant to subsection (e) only after it has been shown that the applicant has exhausted all benefits from private third-party payers, Medicare, Medicaid and other government assistance programs and after consideration of the applicant's income and assets. The secretary of health and environment shall adopt rules and regulations establishing standards for determining eligibility for state assistance under this section.

(g) (1) Except for treatment products provided under subsection (e), if the medically necessary food treatment product for diagnosed cases must be purchased, the purchaser shall be reimbursed by the department of health and environment for costs incurred up to \$1,500 per year per diagnosed child age

18 or younger at 100% of the product cost upon submission of a receipt of purchase identifying the company from which the product was purchased. For a purchaser to be eligible for reimbursement under this subsection (g)(1), the applicable income of the person or persons who have legal responsibility for the diagnosed child shall not exceed 300% of the poverty level established under the most recent poverty guidelines issued by the federal department of health and human services.

(2) As an option to reimbursement authorized under subsection (g)(1), the department of health and environment may purchase food treatment products for distribution to diagnosed children in an amount not to exceed \$1,500 per year per diagnosed child age 18 or younger. For a diagnosed child to be eligible for the distribution of food treatment products under this subsection (g)(2), the applicable income of the person or persons who have legal responsibility for the diagnosed child shall not exceed 300% of the poverty level established under the most recent poverty guidelines issued by the federal department of health and human services.

(3) In addition to diagnosed cases under this section, diagnosed cases of maple syrup urine disease shall be included as a diagnosed case under this subsection (g).

(h) The department of health and environment shall continue to receive orders for both necessary treatment products and necessary food treatment products, purchase such products, and shall deliver the products to an address prescribed by the diagnosed individual. The department of health and environment shall bill the person or persons who have legal responsibility for the diagnosed patient for a pro-rata share of the total costs, in accordance with the rules and regulations adopted pursuant to this section.

(i) Not later than July 1, 2008, the secretary of health and environment shall adopt rules and regulations as needed to require, to the extent of available funding, newborn screening tests to screen for treatable disorders listed in the core uniform panel of newborn screening conditions recommended in the 2005 report by the American college of medical genetics entitled "Newborn Screening: Toward a Uniform Screening Panel and System" or another report determined by the department of health and environment to provide more appropriate newborn screening guidelines to protect the health and welfare of newborns for treatable disorders.

(j) In performing the duties under subsection (i), the secretary of health and environment shall appoint an advisory council to advise the department of health and environment on implementation of subsection (i).

(k) The department of health and environment shall periodically review the newborn screening program to determine the efficacy and cost effectiveness of the program and determine whether adjustments to the program are necessary to protect the health and welfare of newborns and to maximize the number of newborn screenings that may be conducted with the funding available for the screening program.

History: L 1965,ch388,§1; L 1974,ch352,§49; L 1977,ch213,§1; L 1984,ch223,§1; L 1985, ch205,§1; L 1994,ch262,§4; L 1997,ch117,§1; L 2006,ch158,§1; L 2007,ch177,§23; May 17.

Appendix 3 – Article

U. S. Newborn Screening System Guidelines: Statement of the Council of Regional Networks for Genetic Services (CORN).

Screening 1992;1:135-147.

Screening, 1 (1992) 135-147

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SCREEN 0015

U.S. Newborn Screening System Guidelines: Statement of the Council of Regional Networks for Genetic Services

BL. Therrell, S.R. Panny, A. Davidson, J. Eckman, W.H. Hannon,
M.A. Henson, M. Hillard, S. Kling, H.L. Levy, F.J. Meaney,
E.R.B. McCabe, V. Mordaunt, K. Pass, E. Shapira and J. Tuerck

*CORN Newborn Screening Committee, Council of Regional Networks for Genetic Services, Cornell
University Medical College, New York, NY, USA*

(Accepted 21 February 1992)

The Council of Regional Networks for Genetic Services (CORN) was created in 1985 to provide a forum for information exchange among groups concerned with public health aspects of genetic services. The newborn screening committee includes representatives from each genetic region of the United States (equally divided among laboratories and administrators) and liaison members from related professional groups. State and regional newborn screening programs across the U.S. vary widely in most aspects of their organization and hence their program outcome. The Newborn Screening Committee of CORN has identified eight specific areas of mutual importance to all programs. These areas include: organization and administration; selection and evaluation of disorders for screening; communication; quality assurance; funding; diagnosis, management, treatment and counseling; program evaluation; and liability. Basic guidelines have been developed in these areas so that U.S. screening programs may begin to achieve uniform consistency in outcome. The guidelines are not intended to judge standard of care, but rather are meant as a framework about which to mold newborn screening programs.

Key words: Newborn; Screening; Guidelines; CORN

Background

The Council of Regional Networks for Genetic Services (CORN) is a federally funded project to improve the quantity, quality, and availability of cost effective

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genetic services in the United States. CORN was developed in 1985 in response to the need for an organization both to coordinate activities among federally funded genetic service networks encompassing the entire U.S. and to implement programs of national significance that emerge from regional initiatives in such priority areas as quality assurance, data collection and education. Two delegates from each network serve on the CORN steering committee with additional representation from the Alliance for Genetic Support Groups, national sickle cell disease programs and other organizations involved in genetic services. CORN members comprise a unique organization of genetic service providers, public health personnel and consumers. The organization maintains a strong focus on the public health components of genetic services in its goals and activities.

The Newborn Screening Committee of CORN was formed in 1987, to address newborn screening issues of regional and national concern regarding program implementation and facilitation. Members of this committee have served as expert reviewers on a federally sponsored panel assisting state newborn screening programs in self evaluation as an aid to improvement of service delivery. Observation of similar structural and functional problems among the state programs surveyed, coupled with a CORN initiative (assistance to local screening efforts through general administrative guidelines to increase national screening, uniformity and effectiveness), led to development of these guidelines. They are the result of over three years of committee deliberations and their continual review and update is intended to provide meaningful guidance to local newborn screening efforts both now and in the future. While this information may prove useful to newborn screening programs in other countries, these guidelines were developed for utilization only in the U.S. and consideration to geographic, political, and economic influences elsewhere were not a factor.

Introduction

Newborn screening is an essential preventive ~ public health program for early identification of disorders that can lead to potentially catastrophic health problems. Its efficient and productive outcome is governed by the smooth integration of specimen collection, laboratory analysis, follow-up contact and effective treatment.

The following basic program guidelines have been developed by the Newborn Screening Committee of the Council of Regional Networks for Genetic Services (CORN) so that screening programs across the United States may begin to achieve uniform consistency in outcome. Committee members recognize that geographic, political and economic factors affect the manner in which newborn screening programs function; however, guidelines considered both essential and achievable by a national, broad-based committee of professionals in this field should provide a basis on which to pattern a successful system. These guidelines should not be misused to judge standard of care, but rather should be used as a framework about which to mold the screening program.

From the outset, we must recognize that newborn screening is a system that

includes private medical practitioners, laboratory personnel, administrative follow-up personnel, tertiary care centers, third-party payers, and others with the same ultimate goal. This system must be designed to function smoothly and efficiently within the governmental/political framework which gives it life.

1.0. Organization and administration

1.1. Legislation

Where legislation is in place, it is preferable to authorize mandatory screening so that program changes may be made through board of health or other administrative action, without new legislation. Rules and regulations establishing responsibility for proper timing of specimen collection, specimen submission, record keeping, laboratory analysis, follow-up, and treatment should outline how the enabling legislation will be implemented. Although several states have no legislation requiring screening (voluntary programs appear to be reaching a high percentage of the desired population in these states), occasional system difficulties, such as financing problems, might be avoided if a legislative mandate for neonatal screening were in place. Some state programs have legislation specifically defining the intent of the program and the disorders that are included. In these programs, it may be extremely difficult to add or delete screening disorders. Responsibility for program details, therefore, should reside outside of the primary enabling statute.

1.2. Scope of responsibility

Documentation of the beginning and ending of organizational and individual responsibilities for medical, laboratory, and follow-up personnel must be clearly established and followed. Realistic, functioning procedural manuals for each of the responsible parties should be developed and combined into an overall newborn screening system manual. The procedures and protocols developed should be used in actual practice by those responsible. Whereas an ideal system may serve as a goal towards which protocols may be oriented, system manuals should define minimum standards and reflect actual practice, serving as usable reference sources that clearly define each step of responsibility. Starting and ending points of each function must be indicated so that smooth integration of system services is easily accomplished.

1.3. Advisory committees

The use of at least one advisory committee, including outside professional and consumer representation, is encouraged. Such committees) may be used to solicit administrative and other program advice as well as external advocacy. Involvement of persons independent of the responsible governmental body enhances the credibility of the program. This committee may be a subcommittee of a larger genetics advisory group, if appropriate, and should involve persons with suitable backgrounds and

interests to offer constructive aid to the system. Such areas as scope of responsibility of system components, screening protocols, and fiscal policy, may benefit from outside advice and advocacy. This committee should not assume responsibility for internal governmental matters or for technical decisions, but rather should act as a group of consultants that helps in developing approaches, planning future directions and problem solving.

1.4. Program centralization

The newborn screening program requires a strong, centralized, administrative staff, knowledgeable in all aspects of the program and concerned with efficient, effective program implementation. Some of the follow-up tracking may be decentralized, provided a central data bank is kept updated so that no children with disorders are lost to follow-up.

Consolidation of newborn screening laboratory testing is advised. Centralization promotes economy and efficiency in performing large numbers of screening procedures. Furthermore, the rarity of most screened disorders results in infrequent observation of abnormal tests, making it preferable to perform large numbers of analyses in order for the laboratory to observe 'real' cases from time to time. It has been suggested in one source that laboratories be required to test at least 50,000 samples annually [2]. States with smaller birth rates may find it efficient to combine efforts in a regional laboratory. It may not be practical to operate within this suggested quota of specimens analyzed. -In this case, it is essential that analytical proficiency be optimized through testing blinded control specimens, exchange of specimens with other laboratories, or external proficiency testing.

2.0. Selection and evaluation of disorders for screening

The disorders included in a screening program should be logically and systematically selected [7]. Some countries have developed priority listings for disorders to be considered for screening [4]; however, such rankings have not yet occurred in the United States. All U.S. programs currently perform screening for phenylketonuria and congenital hypothyroidism. It has been recommended by a National Institutes of Health (NIH) consensus panel, that newborn screening for sickle cell disease be mandated by all programs [4]. Thus, there is increased emphasis on sickle screening and approximately 80% of all U.S. programs include this testing.

2.1. Addition and deletion of disorders

To ensure that decisions to add or delete screening tests (see Sec. 1.1) are intelligent, and informed, demography (including genetic composition), methodology, outcome, and economics must be considered. Cost-effective, efficient screening should produce the desired effect on infant morbidity and mortality through a well designed pilot program and the experience of others. Before any screening disorders are added or

deleted, sufficient information should be gathered to support the changes. Specifically, incidence data and population statistics must indicate that screening for the disorder will result in detection of a reasonable number of cases. Additionally, the laboratory protocol proposed must be sufficiently sensitive to serve as a viable method for use with the specimens of interest. These specimens must be economically and technically feasible to collect, transport and analyze. Screening should be initiated only if effective intervention is accessible to all affected individuals.

2.2. Laboratory methods

Analytical methods should be of sufficient sensitivity and specificity, with adequate quality control, to ensure maximum disease detection with minimal false negative results. Low false positive rates are necessary to prevent overburdening of the follow-up system. Laboratory services should be centralized when possible and include multistate regionalization (see Sec. 1.4). Laboratories performing screening should adhere to professional guidelines in common usage by the College of American Pathologists (or other suitable accrediting body) concerning the type and frequency of use of analytical control material. Successful subscription to an external proficiency testing program is essential to demonstrating credibility of the procedures used in the screening laboratory. If no such program exists for the analyte and/or matrix involved, programs are encouraged to document reliable results on specimens exchanged with other well established laboratories in the field. Appropriate sensitivity and specificity limits are difficult to define; however, their importance cannot be overemphasized with respect to credibility and cooperation from the physician community in following up on abnormal results.

2.3. Follow-up

The newborn screening system must ensure follow-up of any positive, or potentially positive, result to the point of resolution. This is best accomplished when responsibilities and procedures for follow-up are assigned to a specific follow-up coordinator. All details of follow-up should be clearly defined. Data transfer must be timely and complete so that adequate information is available to the follow-up persons. Follow-up responsibility may reside within or outside of the laboratory; both types of follow-up are used in the United States. The beginning and ending of follow-up, along with organizational and individual responsibilities, must be clearly defined. These will usually include all activity from the time of notification of an abnormal result until the time the patient receives treatment. Initial notification to physicians that patients have medically significant abnormal results should be by telephone, if possible. A confirmatory letter should then be sent clearly outlining subsequent follow-up steps through case resolution. A successful follow-up protocol must include procedures for reaching a conclusion for each case, even if that conclusion is "lost to follow-up".

2.4. Repeat tests

Repeat testing should be performed whenever the first test result's reliability is questioned. Linkage of second test results to the initial screening specimen is important as a means of controlling quality and completing follow-up information. When transfusions or antibiotic treatment have been given before the appropriate laboratory specimen is collected, a mechanism for repeat testing should also exist. The physician or other person attending the newborn should be responsible for the timing and redrawing of repeat specimens; however, notification of the need for retesting is a program responsibility.

Routine second testing of all or selected newborns should be considered on the basis of resources available and cost effectiveness. To avoid biological false negative results in the early newborn period, a second specimen can be obtained later. The full benefit of detecting late onset disorders has not been measured.

3.0. Communication

Rapid, effective communication of abnormal results is essential and should include transmission of critical information, by telephone, at the earliest opportunity. Proper documentation is also necessary, given the medical-legal environment in which screening must function.

3.1. Documentation

Documentation of specimen collection (or refusal), laboratory analysis (including quality control), result reporting, and physician and patient contact are essential. Such documentation must withstand legal scrutiny and must be maintained until the legal statute of limitations expires. Many states operate under the concept of 'informed dissent' whereby documentation is necessary only if the screening service is refused by the patient. In these programs, the legislation or rules (regulations) governing screening require that all infants be screened except in specific instances such as religious conflict.

The person submitting the specimen should document the collection and transmittal of the specimen, and the laboratory should document its receipt and analysis. Receipt of results on each patient tested is proof that specimen transmittal has occurred. Programs must ensure rapid and direct communication of results from the laboratory to the persons responsible for the patient in all cases, regardless of the results - normal, abnormal or unsatisfactory. If abnormal results are obtained in the laboratory, proper transfer of these results to the follow-up coordinator must be documented. Likewise, transmittal from the follow-up coordinator to the person responsible for the infant's care must also be documented. Practitioners are responsible for documenting communications with the patients. Such documentation may include any of a number of forms such as paper or computer storage medium. Program officials should consult legal counsel for specific advice.

3.2. Computers

Computerization of the newborn screening program is cost-effective and provides better system control. Since the entire newborn screening system must be monitored by program administrators, computerization can provide a valuable management tool. Justifications for computerization include more efficient public service, through time savings, improved accuracy, and more extensive data assessment for program evaluation and improvement.

Proper design of the computer system's hardware and software can improve program efficiency and expand its scope. Computerized screening programs should strive to include on-site demographic data collection and submission, laboratory data management and reporting, follow-up result reporting and data collection, and analysis and program documentation. Programs should not expend unnecessary effort in developing elaborate systems without first surveying commercial and public newborn screening systems that have already been implemented. Some public programs are available at little or no charge to other newborn screening systems.

With careful thought and planning, program officials can develop a computer system that improves many aspects of the screening program. Developing computer systems that actually decrease program personnel is quite difficult; however, the available staff is often more efficiently utilized to accomplish expanded or multiple tasks within equivalent time periods. Ideally, the computerized newborn screening system includes demographic transmittal from the point of collection to the laboratory, data management within the laboratory, result reporting to the submitter and to the follow-up coordinator, generation of follow-up communications, documentation of all contacts on abnormal results (laboratory to follow-up, follow-up to physician, and extended follow-up), records for diagnosed patients, disease registries and linkage to birth records.

3.3. Education

Public awareness coupled with professional and patient education are significant program responsibilities that must be part of the complete screening system. The importance and intent of the screening program must be properly and adequately communicated to all persons involved. Educational materials for parents should be available along with similar information for concerned professionals. Parent information is most effective when developed on an elementary level, with appropriate ethnic and cultural sensitivity. Professional literature should be more technical. Some programs have used manuals of protocol and responsibilities as a portion of professional education. Sound-slide and video tape presentations have also been developed. While audio-visual aids may augment educational efforts, personal contact and demonstrations have been found to be most effective. This is particularly true in educating specimen submitters about proper collection technique. Simple flow charts of both laboratory and follow-up protocols offer handy references for all professionals participating in the system. Funding for educational expenses is often overlooked but should be included as an integral part of the overall financing system.

4.0. Quality assurance

The performance of each component of newborn screening and its contribution to the overall system must undergo quality assurance and monitoring. Criteria for adequate performance must be established for each functioning unit of the system. Ideally, a blinded specimen should be prepared and submitted to periodically check the entire system from the submission of specimens to follow-up contact.

4.1. Submitter

The submitter is responsible for assuring and documenting the quality of specimen collection and patient data. The newborn screening system must begin with a specimen of adequate quality from every newborn. The specimen submitter must ensure that there is sufficient blood uniformly distributed in each target circle on the filter paper collection form and that the specimen is collected according to program guidelines. An approved standard on collection of blood on filter paper has been set forth by the National Committee for Clinical Laboratory Standards (NCCLS) [5], and its adoption by all screening programs is recommended. Similarly the American Academy of Pediatrics (AAP) [1] has set forth guidelines concerning such issues as early discharge, and intensive care situations, which also should be incorporated into screening practice.

The demographic data accompanying each laboratory specimen must be correct and complete. Failure of the submitter to provide accurate complete data, can result in difficulties in analysis, interpretation, and follow-up. Incorrect or incomplete data can result in damage to the newborn and increased legal exposure of the submitter should diagnosis and treatment be impaired. Some programs practice demographic and specimen collection error surveillance; computers can increase efficiency and facilitate this activity. Detection of submission errors, however, should result in an educational/corrective action in order for surveillance to be of benefit.

4.2. Laboratory

Successful participation in a recognized proficiency testing (PT) program along with appropriate quality control must be practiced and documented by the analyzing laboratory. A routine PT specimen analyzed quarterly for each analyte is sufficient. With new procedures or analytes, or in response to PT misses, such testing should be performed as often as necessary in order to establish reliable performance. The laboratory must document analysis, frequency of quality assurance specimens, and appropriate use of standards (or calibrators traceable to a primary standard source). The laboratory should be able to provide adequate documentation of analytical results of unknown specimens and controls upon request. Program officials should seek legal advice about the amount of documentation needed, its length of retention, and time frame in which retrieval must occur. Laboratory protocols also must be documented and updated when procedural changes occur. Such documentation

should include a clear definition of where the laboratory's responsibility begins and ends within the screening system.

4.3. Follow-up

Regular review of documentation should be included in all newborn screening systems. Procedures to ensure complete and thorough follow-up of all positive screening findings and inadequate specimens must be in place and subject to random checks to assure proper functioning. In addition to the normal flow diagrams for follow-up activities found in most programs, a complete procedure manual must be developed to clearly indicate follow-up protocols for all combinations of abnormal laboratory results. It must define where follow-up begins, the level of activity required according to the disorder and analytical result, strategies to follow as a secondary protocol, types of documentation necessary, and how to achieve final disposition. This procedure manual must be realistic and carefully followed in order to minimize legal exposure. Some programs have found computerization and linkage to birth records (see Sec. 3.2) to be of major benefit in assisting with tracking. Cumulative listings by disorder, submitter, and region are also possible; thus, program statistics may be accurately and easily monitored and problem submitters may be identified and targeted for enhanced educational efforts.

4.4. Treatment and management

Treatment and management should lead to a partnership between the patient's primary physician, the treatment center and the newborn screening program. Many programs have found success in using specialists and consultants to assist with patient treatment and act as intermediaries with the patient's private physician. The newborn screening staff must be aware of screening outcome; therefore, data regarding treatment and management should be periodically reviewed. Only through evaluation of outcome data may program effectiveness be judged. This information may be obtained in a variety of ways, such as return mail or telephone, depending on program size.

5.0. Funding

Funding is the most difficult problem confronting newborn screening systems. Not only is there a question as to source of funds, but also there are questions related to program scope and methods of reimbursement.

5.1. Sources

Sources of funding should not necessarily be limited to state tax revenues. Federal grants, private foundations, and fees for service are viable alternatives in addition to funding through Maternal and Child Health, Women, Infant and Children (WIC),

and Crippled Children Services. Many state programs have developed a fee for services. Those unable to pay for service must be supported by the program's income from fees. In some states, financial support for all or portions of the program may be obtained from federal grants or other related programs such as WIC. In rare instances, support for particular programs has been supplemented from private sources.

All program costs should be included in any fee calculated (see Sec. 5.2). System funding should include screening laboratory analysis, follow-up services (including education), computerization, metabolic food supplements, and other treatment necessities if appropriate. Because program fees may be tied to laboratory collection kits, other funding items are sometimes overlooked. As a result, follow-up and education may be weak and fragmented. Because newborn screening encompasses many disciplines to create an efficient system, all system components must be considered in the financing structure.

5.2. Cost analysis

The societal benefits resulting from screening should be included in any cost accounting in order to fully reflect actual program benefit. A uniform method of determining program costs should be developed nationally. The recently published fiscal study prepared by the Office of Technology Assessment [6] offers some assistance.

6.0. Diagnosis, management, treatment and counseling

Diagnosis, management, treatment, and counseling must be included as newborn screening system functions. Otherwise, there is no real purpose to screening.

6.1. Diagnosis

Screening does not equate to diagnosis; therefore, detection of some cases of disorders included in newborn screening will not occur, even in a quality program, due to biological variables and other factors. Confirmation and diagnostic follow-up must be included in a complete screening system. Clinical judgment must play an important role in the system. Testing for bipterin problems should be carried out on all patients with phenylketonuria or hyperphenylalaninemia. Similarly, hematologic examinations for clinically significant hemoglobinopathies might also be appropriate. Related procedures consistent with diagnostic follow-up of other screening disorders may be of equal importance.

6.2. Consultant resources

Pediatric subspecialists may be included as resource personnel who can act as liaisons between the governmental screening agency and the private physician. Many successful screening systems report abnormal results to the infant's physician and to consultants

specializing in the disorder of interest who are willing to offer their professional assistance. Consultants can contact the primary physician to offer assistance, if needed. This can provide an excellent feedback mechanism for the follow-up coordinator. Some state programs use pediatric endocrinologists for hypothyroidism and congenital adrenal hyperplasia, pediatric hematologists for sickle cell disease, and metabolic subspecialists for the inborn errors of metabolism. Open communication and planning can help to alleviate problems from competition related to service areas.

6.3 Long term tracking and outcome

Long-term tracking and outcome evaluation for each screening disorder should be maintained and updated periodically. In a highly mobile society, interprogram communication is essential. Such tracking can serve as an extension of follow-up with potential benefit for long-term case management and for minimizing program losses due to relocation. Particular attention must be paid to patient confidentiality and limitations placed on data availability. As medical benefits develop for patients later in life, outcome data can provide the vital patient information necessary for tracking and health care. Benefits of this type are currently recognized for maternal hyperphenylalaninemia. Active maintenance of hyperphenylalaninemia registries (see Sec. 7.2) have assisted in locating women of childbearing age for educational follow-up. Program personnel must be advised of the latest developments with maternal hyperphenylalaninemia follow-up so that maximum benefit of this research may be realized. Cooperation with the national Maternal PKU Collaborative Study (MPKUCS) is encouraged.

6.4 Carrier counseling

Counseling programs, if appropriate, should be developed before screening in order to augment follow-up. This is particularly important in hemoglobinopathy screening. It is of primary importance that sickle cell disease be detected and treated; however, as a consequence of screening, parents and infants with sickle trait also will be identified. Because the number of carriers is extremely high relative to persons with the disease, resources for quality counseling are generally scarce. Many programs therefore, are developing liaisons with community-based sickle cell organizations in order to augment the counseling aspect of screening. All qualified, existing resources should be fully utilized to provide counseling and education to the large numbers of parents of carrier infants.

Minimum standards of professional qualification, quality assurance, and continuing education must be developed and required of all counselors. Evaluation of counseling and educational approaches should include assessment of information retention, impact on reproductive choices and cost-effectiveness.

7.0. Program evaluation

Programs should evaluate their effectiveness through documentation of the public health impact. Ongoing or periodic evaluations should be performed and monitored regularly to assess the quality of the program.

7.1. Data collection

Minimal data elements appropriate for the specimen collection form have been determined by the NCCLS j5J. Although additional data may be collected to evaluate certain program aspects, these data should be consistent with the primary objectives of the screening program. Data integrity of the newborn screening system should not be compromised by epidemiologic studies for secondary research purposes.

7.2. Outcome data

Timely, accurate reporting of outcome data should be a program priority to ensure screening effectiveness. Periodic follow-up of the status of diagnosed cases not only provides evaluation data but also allows a mechanism for patient re-entry into the medical care system, should those cases be lost. In programs where follow-up and laboratory services are decentralized, a central depository for follow-up data should be established.

8.0. Liability

Liability must be considered when planning and conducting newborn screening. It is prudent to address liability issues in making program decisions since financial losses resulting from lawsuits could have devastating effects on the screening system.

8.1. Documentation

All responsibilities within the system must be clearly defined and their completion appropriately documented (see Sec. 3.1 J. Program leaders must not assume that all tasks have been completed and all aspects of screening have been carried through without proof. Documentation must be available. The physician or hospital must be able to show evidence of a proper screen. The laboratory must record its test results along with evidence of proper standardization and control. Follow-up must be carefully documented. A contact log should be kept, listing the person contacted, date and nature of the contact, and the person making the contact. Letters sent informing physicians of the need for follow-up action preferably should be sent by certified mail. All records must be kept for a period of time in accordance with state regulations regarding medical result records for children. Program officials should consult with legal counsel on this point.

8.2. Legislation

Statutes pertaining to newborn screening within a state must be carried out. If rules and regulations are required, they should be molded for maximum program benefit. All loopholes should be closed, and all legal responsibilities carefully defined. Performing beyond the law may increase legal exposure. Programs should review the appropriate literature concerning liability issues (see entire publication in Ref. 2).

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Appendix 4

Executive Summary -
Serving the Family From Birth to the Medical Home
Newborn Screening: A Blueprint for the Future

Pediatrics 2000;106:386-8.

Serving the Family From Birth to the Medical Home

Newborn Screening: A Blueprint for the Future

Executive Summary: Newborn Screening Task Force Report

Approximately 4 million infants are born yearly in the United States (US), and are screened to detect conditions that threaten their life and long-term health. Newborn screening is a public health activity aimed at the early identification of infants who are affected by certain genetic/metabolic/infectious conditions. Early identification of these conditions is particularly crucial, as timely intervention can lead to a significant reduction of morbidity, mortality, and associated disabilities in affected infants.

Newborn screening has been universally accepted for the past 3 decades. It represented the first population-based genetic screening program, and signaled the integration of genetic testing into public health programs. Today, advances in technology are making possible new forms of newborn screening programs, such as newborn hearing screening. These technological advances will continue to have a significant impact on the sensitivity, specificity, and scope of newborn screening programs, including newborn heelstick screening.

Challenges are anticipated with technological advances. It is likely that public pressure to deploy new diagnostic capabilities, such as DNA-based technology, will increase despite limited knowledge of potential risks and benefits. In addition, the ability to detect individuals with conditions for which there is no effective or necessary treatment is likely. Further, as the Human Genome Project is completed, the impetus and opportunity for the transition of genetic technology into practice will increase. These and other challenges will affect not only newborn screening tests, but also the entire newborn screening system, which includes short-term follow-up, diagnosis, treatment/management, and evaluation. Inherent to each of these components is an education process. A national dialogue and process is needed to support state newborn screening systems as they try to keep pace with new technology.

To address these and other issues, a national Task Force on Newborn Screening (Task Force) was convened by the American Academy of Pediatrics (AAP) with funding from and at the request of the Maternal and Child Health Bureau (MCHB), Health Resources and Services Administration (HRSA), US Department of Health and Human Services (HHS). The AAP was asked to convene the Task Force in recognition that pediatricians and other primary care health professionals must take a lead in partnering with public health organizations to examine the many issues that have arisen around the state newborn screening programs.

To ensure that children who are screened are linked to a medical home, it was essential that pediatricians and other primary care health professionals be involved. The AAP defines the medical home as care that is accessible, family-centered, continuous, comprehensive, coordinated, compassionate, and culturally competent. A child who has a medical home has a pediatrician or other primary care health professional who is working in partnership with the child's family to ensure that all medical, nonmedical, psychosocial, and educational needs of the child and family are met in the local community.

Task Force members were appointed to represent many perspectives among those who operate programs, conduct research, and are affected by newborn screening systems. The co-sponsors of this effort were: other HHS agencies including the National Institutes of Health (NIH), the Centers for Disease Control and Prevention (CDC), and the Agency for Healthcare Research and Quality (AHRQ); the Genetic Alliance, which is a consortium of consumer groups; and national public health organizations including the Association of State and Territorial Health Officials, the Association of Maternal and Child Health Programs, and the Association of Public Health Laboratories. This report has been approved by the AAP Board of Directors. It does not necessarily reflect the viewpoints of sponsoring organizations or the organizations represented by members of the Task Force.

The purpose of the Task Force was to review issues and challenges for state newborn screening systems. The review process was structured to further expand representation. Task Force members were divided into 5 work groups, and additional individuals were invited to participate in each work group's examination of key issues. Over the course of 6 months, questions, concerns, and issues were collected from state public health agencies, state public health laboratory directors, maternal and child health programs, pediatricians, and other primary care health professionals who care for children, families and other consumers, bioethicists, scientists, and health services researchers. Each work group formulated conclusions and developed consensus recommendations. On May 10–11, 1999, the Task Force heard presentations from the 5 work groups, along with public comment on the reports and recommendations. A set of recommendations was developed incorporating key elements of the work group reports, issues raised by the public, and other related information. This document summarizes the Task Force recommendations.

The Task Force has outlined a national agenda for strengthening each "state" newborn screening system. ("State" newborn screening systems refer to state and territorial programs for heelstick newborn screening.) The Task Force believes that public health agencies (federal and state), in partnership with health professionals and consumers, should continue to:

- Better define public health responsibilities for federal and state public health agencies;
- Develop and disseminate model state regulations to guide implementation of state newborn screening systems (including disease and test selection criteria);
- Develop and evaluate innovative testing technologies;
- Design and apply minimum standards for newborn screening activities (eg, sample collection, laboratory quality, sample storage, and information systems);
- Develop and disseminate model follow-up, diagnosis, and treatment guidelines and protocols for health professionals, and other participants in the newborn screening system;
- Design and evaluate model systems of care with services and supports from infancy to adulthood that are consistent with national guidelines for children with special health care needs (ie, family-centered, community-based, and coordinated systems of care).
- Design and evaluate tools and strategies to inform families and the general public more effectively; and
- Fund demonstration projects to evaluate technology, quality assurance, and health outcomes.

KEY RECOMMENDATIONS

I. Effective Newborn Screening Systems Require an Adequate Public Health Infrastructure and Must Be Integrated With the Health Care Delivery System

- Federal agencies must take action to strengthen the public health infrastructure for newborn screening.
 - The federal government—acting through the HRSA, CDC, Health Care Financing Agency (HCFA), AHRQ, NIH, and other agencies—should collaborate to provide ongoing leadership and support for development of newborn screening standards, guidelines, and policies.
 - As the federal unit with most responsibility for newborn screening system development, the HRSA should engage in a national process involving government, professionals, and consumers to advance the recommendations of this Task

Force and assist in the development and implementation of nationally recognized newborn screening system standards and policies.

- Federal resources should be identified to sustain a Newborn Screening Quality Assurance Program to assist state public health laboratories. Such assistance must be both sustained and expanded as states adopt new screening technologies and modalities.
- The HRSA's MCHB should strengthen current mechanisms to improve coordination of infant health programs and initiatives within the state and/or between states, including continuation of funding in support of newborn screening program reviews.
- State public health agencies should direct their newborn screening program to be consistent with professional guidelines and recommendations. Each state public health agency should take responsibility for systems development. Specifically, states and their agencies have responsibility to:
 - Design and coordinate the newborn screening system;
 - Adhere to nationally recognized recommendations and standards for the validity and utility of tests. State newborn screening systems have a responsibility to review the appropriateness of existing tests, tests for additional conditions, and new screening technology and modalities; and
 - Adopt standards for laboratories, health professionals, and health care financing plans based on nationally recognized standards and guidelines for follow-up, diagnosis, and treatment.
- State public health agencies, working under legislative authority, have the ongoing responsibility to ensure quality and evaluate program effort. States and their state public health agencies should:
 - Maintain a newborn screening system that has appropriate evaluation, performance monitoring, and quality assurance activities from initial screening, through follow-up, diagnosis, treatment, and services through adolescence and adulthood;
 - Conduct oversight of program operations, including those outside the public health agency, such as test analysis and tracking, private sector collection and transmission of screening data, laboratory quality, and the quality of the diagnostic procedures and treatment programs at pediatric subspecialty clinics; and
 - Monitor and evaluate program performance through collection, assembly, analysis, and reporting of data, including outcome evaluations.
- States and state public health agencies should implement mechanisms to inform and involve health professionals and the public. Each state should:
 - Develop a program advisory board that is multidisciplinary, involves pediatricians and other primary care health professionals who provide medical homes for children, pediatric subspecialists, and has meaningful representation of families and the general public; and
 - Design and implement public, professional, and parent education efforts regarding newborn screening.
- States and state public health agencies should provide support for coordination and integration of program activities, including information and services. This will require public—private, federal—state, and intrastate partnerships. States should:
 - Use public and private resources to fund demonstration programs that can serve as a testing ground for linking information and services in ways that improve the newborn screening system; and
 - Structure interagency coordination to maximize resources and to improve the efficiency and effectiveness of newborn screening systems.

II. Public Health Agencies Must Involve Health Professionals, Families, and the General Public in the Development, Operation, and Oversight of Newborn Screening Systems

- The pediatrician or primary care health professional who, in partnership with parents, is the source of the child's medical home, should:

- Ensure that all newborns admitted to their practice have received adequate newborn screening, and that appropriate documentation of testing is present;
- Follow positive screening results to diagnosis (ie, confirmed or excluded), including repeated screening and diagnostic testing;
- Coordinate a seamless system of care with pediatric subspecialty clinics, tertiary care centers, and/or community-based providers, when a child is diagnosed with a disorder through newborn screening;
- Maintain a central record and database containing all pertinent medical information about the child. This record should be accessible to the family and others involved in the child's care, but confidentiality must be ensured; and
- Assist the family in understanding the diagnosis, symptoms, and potential implications of a diagnosed genetic/metabolic condition, as well as the availability of genetic counseling, family testing, and other family support services.
- Parents should receive information (on behalf of their children) about newborn screening.
 - Prospective parents should receive information about newborn screening during the prenatal period. Pregnant women should be made aware of the process and benefits of newborn screening and their right of refusal before testing, preferably during a routine third trimester prenatal care visit.
 - Parent knowledge should be reinforced after delivery by educational materials and discussion as needed by the infant's pediatrician or primary care health professional and/or knowledgeable hospital staff.
 - Prenatal health care professionals as well as the infant's primary care health professional should be knowledgeable about their state's newborn screening program through educational efforts coordinated by the state's newborn screening program in conjunction with a newborn screening advisory body.
- Written documentation of consent is not required for the majority of newborn screening tests, for example, those tests of proven validity and utility.
 - Parents should always be informed of testing and have the opportunity to refuse testing.
 - If after discussions about newborn screening with health professionals, parents refuse to have their newborn tested, this refusal should be documented in writing and honored.
 - If a newborn screening test is investigational or in the process of being developed, the benefits or potential risks have yet to be demonstrated, and identifiers are not removed from the specimen, informed consent should be obtained from parents and documented.
- Studies should be performed to broaden understanding of the ways in which communication can be performed more effectively for the benefit of consumers.
 - Pilot studies and evaluation research should be conducted to assess the potential impact of revised parental permission and informed decision-making policies.
 - Each state or region should, with input from families who have children with special needs and/or parent information centers, develop and provide family educational materials about newborn screening.
 - Evaluation of materials should be ongoing, particularly because of the changing demographics of childbearing, cultural changes, and rapid developments in genetic science.
- Parents have a right to confidentiality and privacy protections for the medical and genetic information in any type of newborn screening results. Based on nationally recognized standards and guidelines, each state should have appropriate policies and mechanisms in place to ensure families' privacy and confidentiality. Laws to guarantee genetic privacy and protect against genetic discrimination should benefit patients identified by newborn screening.
- States and the federal government should include public participation in medical policy-making. The Secretary's Advisory Committee on Genetic Testing provides a mechanism for public participation in genetic policy development at the federal level. Each state should establish and fund a newborn screening advisory body with public participation to advise on newborn screening policy developments.
 - Such an entity should include a broad range of public advisors representing parents, health professionals, third-party

- payers, appropriate government agencies, and other concerned citizens.
- Such an entity should be empowered to advise state officials about screening for particular conditions based on accepted standards and be consulted about the development of related state regulations.
- Such an entity should be involved in the review of new tests under consideration by the state and in the development of pilot programs for new tests.
- Such an entity should be involved in the ongoing evaluation of all aspects of the state's process for newborn screening. Oversight activities should include a review of: testing, follow-up, and treatment efforts; the impact on families of receiving a false-positive screening result; and the state's process for handling consumer input including grievances.

III. Public Health Agencies Must Ensure Adequate Infrastructure and Policies for Surveillance and Research Related to Newborn Screening

- State Maternal and Child Health (MCH) programs should conduct a review of the newborn screening system and its relationship to the HRSA MCH Block Grant Performance Measures and evaluate the quality of data of the newborn screening-related performance measures.
- The federal HCFA should develop Health Plan Employer Data and Information Set (HEDIS) measures to evaluate the health plans' performance within the newborn screening system.
- A federally-funded newborn screening research agenda should be outlined that aims to: develop better tests (more sensitive, more specific, and less costly); assess the validity and utility of new technologies (eg, tandem mass spectrometry, DNA-based testing, and other evolving technologies); and define appropriate uses of residual biologic samples for population-based research and surveillance.
- The HRSA's MCHB should provide grants to states to stimulate development of newborn screening information systems, with a focus on newborn screening systems that are connected to the medical home, newborn screening system process and outcome evaluation, development of standardized data sets, analyses of cost-efficiency and effectiveness, and integration with other public health data systems. Support for technological innovation (ie, new test technologies) should include these measures.
- Pediatricians, pediatric subspecialists, and other health professionals who care for children should contribute to newborn screening data collection to advance knowledge about health outcomes and intervention effectiveness. Professional associations, the HRSA-funded National Newborn Screening and Genetics Resource Center, and state newborn screening programs should develop strategies to assist health professionals in their efforts to participate in and learn from newborn screening information systems.
- Pilot studies should be undertaken to demonstrate the safety, effectiveness, validity, and clinical utility of tests for additional conditions and new testing modalities. Informed consent of parents is called for in all such pilot studies. These studies might be undertaken by individual states, regional or nationwide groups of states, or through federal grants provided to research institutions across the country.
- Federal and state public health agencies, in partnership with health professionals, families, and representatives of ethnic, minority, and other diverse communities should:
 - Develop model legislation and/or regulation that articulates policies and procedures regarding utilization of unlinked and identifiable residual samples for research and public health surveillance. This process should include review and consideration of the recent recommendations to the President set forth by the National Bioethics Advisory Commission (NBAC) for research involving human biological materials;
 - Develop model consent forms and informational materials for parental permission for retention and use of newborn screening samples;

- Develop educational materials for parents that includes information regarding the storage and uses of residual samples;
- Organize collaborative efforts to develop minimum standards for storage and database technology to facilitate appropriate storage of residual newborn screening blood samples at the state level; and
- Consider creating a national or multi-state population-based specimen resource for research in which consent is obtained from the individuals from whom the tissue is obtained. Such a resource could be an alternative to retaining newborn screening samples for potential use in research.
- Using national recommendations, each State program should develop and implement policies and procedures for retention of residual newborn screening blood samples that articulate the rationale and objectives for storage, the intended duration of storage, whether storage is with or without identifiers, and guidelines for use of identifiable and unlinked samples. An advisory group for newborn screening programs with broad health professional and family/community representation is a valuable resource in developing policies and procedures and in reviewing applications for use of retained samples. The advisory body also could determine priorities for use.

IV. Public Health Agencies Should Ensure Adequate Financing Mechanisms to Support a Newborn Screening Program

- States should ensure adequate financing of all parts of the newborn screening system: screening, short-term follow-up, diagnostic testing, comprehensive medical care/treatment, and evaluation of the system. If newborn screening fees are not adequate, funding of all components of the system could be accomplished with other public health dollars or by third-party payers. Other uses of newborn screening fees should not be considered until all of the components of the newborn screening system are fully funded.
- States should take responsibility for blending resources available through Title XIX (Medicaid), Title V (MCH Block Grant), Title XXI (State Children's Health Insurance Program) [SCHIP], and private insurance to guarantee necessary coverage and financing for all children and adolescents with a condition diagnosed through the newborn screening system.
- State contracts for publicly-subsidized third-party insurance plans that cover children (eg, Medicaid and SCHIP) should explicitly require coverage for newborn screening and those services and treatment related to disorders identified by newborn screening. State contracts also should require that third-party payers ensure access to health care professionals with appropriate pediatric expertise within the network or through out-of-network referrals.
- States, in cooperation with health professionals and payers, should put mechanisms in place to identify the third-party payers for newborns immediately following birth. For example, all states should operationalize the automatic newborn eligibility requirements under Medicaid and the Health Insurance Portability and Accountability Act (HIPAA) newborn coverage provisions that require infant coverage and prohibit preexisting condition exclusions for newborns.
- Purchasers—public and private—should ensure that the benefits package they pay for includes the care and services defined by the AAP Scope of Health Care Benefits Statement and the Council of Regional Networks for Genetic Services Guidelines.
- In the Supplemental Security Income (SSI) program, the federal government should review the technical appropriateness of guidelines, and evaluate the consistency of their application, for children with conditions identified through newborn screening.

Appendix 5

***Article - Data Integration and Warehousing: Coordination Between
Newborn Screening and Related Public Health Programs***

**Southeast Asian Journal of Tropical Medicine and Public Health
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DATA INTEGRATION AND WAREHOUSING: COORDINATION BETWEEN NEWBORN SCREENING AND RELATED PUBLIC HEALTH PROGRAMS

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Abstract. At birth, patient demographic and health information begin to accumulate in varied databases. There are often multiple sources of the same or similar data. New public health programs are often created without considering data linkages. Recently, newborn hearing screening (NHS) programs and immunization programs have virtually ignored the existence of newborn dried blood spot (DBS) newborn screening databases containing similar demographic data, creating data duplication in their 'new' systems. Some progressive public health departments are developing data warehouses of basic, recurrent patient information, and linking these databases to other health program databases where programs and services can benefit from such linkages. Demographic data warehousing saves time (and money) by eliminating duplicative data entry and reducing the chances of data errors. While newborn screening data are usually the first data available, they should not be the only data source considered for early data linkage or for populating a data warehouse. Birth certificate information should also be considered along with other data sources for infants that may not have received newborn screening or who may have been born outside of the jurisdiction and not have birth certificate information locally available. This newborn screening serial number provides a convenient identification number for use in the DBS program and for linking with other systems. As a minimum, data linkages should exist between newborn dried blood spot screening, newborn hearing screening, immunizations, birth certificates and birth defect registries.

INTRODUCTION

At (and sometimes before) birth, patient demographic and health information begin to accumulate in varied databases. Even in public health programs that reside within the same agency, there are often multiple sources of the same or similar data. It is not unusual for these databases to exist separated from one another, often without the responsible parties having knowledge that other databases containing similar elements exist. In cases where such knowledge is present, there are often political or administrative reasons that influence the creation of separate data "silos." Additionally, new public health programs are often created without considering possible linkages to data with common elements that might exist in another program. Sometimes, persons developing new public health programs may not even think to consider the existence of databases that could be useful in supporting the new program.

For example, in recent years newborn hearing screening (NHS) programs and immunization programs seeking to develop databases for patient tracking have virtually ignored the long time existence of newborn

dried blood spot (DBS) newborn screening databases containing relevant demographic information eventually duplicated in their 'new' data systems. At a minimum, all of these programs (NHS, DBS, and immunizations) have some basic newborn patient demographic information in common since all three deal with the newborn soon after birth. In addition, common demographic elements exist between these three programs and birth certification programs, among others. It is a waste of time and effort when all of these systems duplicate data fields that could be easily shared if an overall data integration plan were thoughtfully developed and implemented.

In order to decrease duplicative data entry, database maintenance, and human errors in data handling, some progressive public health departments are developing data warehouses of basic, recurrent patient information, and linking these databases to other health program databases where programs and services can benefit from such linkages. Thus, in data warehouses, patient demographic data (and possibly other common data elements) are uploaded from any one of a number of possible sources to reside in a centrally accessible database (ie the data warehouse)

potentially accessible to multiple users. These individual users (ie other health programs) may have separate specialized databases and/or software for unique program-specific purposes separate and apart from the data warehouse, but access to basic patient information available in the data warehouse makes duplicate entry of this information unnecessary. Demographic data warehousing not only saves time (and money) by eliminating duplicative data entry for programs utilizing it, but it also reduces the chances of errors within the data since human interactions with data entry (the largest source of potential error) are minimized or eliminated.

In most US newborn screening programs, the DBS specimen is collected within the first 1-3 days after birth. Thus, newborn screening information is usually the first patient demographic data available, even earlier than official birth certificate information in most systems, including electronic birth certificate systems. In a data warehousing system, therefore, it is logical to construct the system to take advantage of these early data. While newborn screening data are usually the first data available, they should not be the only data source considered for early data linkage or for populating a data warehouse. Birth certificate information should also be considered along with other data sources for infants that may not have received newborn screening, who may have been born outside of the jurisdiction or whose birth certificate information is not locally available. In any case, it is prudent to consider the possible linkages of newborn screening information with other newborn or child health programs and birth certificates in order to improve overall public health program efficiency and cost effectiveness.

NEWBORN SCREENING SYSTEMS

Newborn dried blood spot screening

In processing DBS newborn screening specimens, the first step is to obtain blood from the newborn for the required testing and submit the specimen with identifying information to the newborn screening laboratory. In the US, there is a published national standard that describes this process in detail including: quality assurance steps, minimal data to be obtained, sampling procedures, and related processes (Hannon *et al*, 1997). Newborn demographic information is usually printed by hand onto the collection device. In some electronically advanced newborn screening/hospital systems, the patient demographic information may be internally downloaded from admissions records to the facility laboratory or nursery, the serial number of the DBS collection card and the date of collection added to the data, and a label printed and attached to the collection card prior to mailing. Thus,

the manual step of completing the demographic information portion of the dried blood collection device is eliminated. Often, for quality control purposes, a handwritten or electronically maintained logbook is also kept in the newborn nursery or birthing facility laboratory, into which information tracking the newborn screen is maintained including date of sample collection and result of the testing when it has been completed. Soon after birth (usually 24-72 hrs), a DBS sample is collected, dried, packaged for shipment, and sent to the screening laboratory by mail or courier.

Upon receipt at the testing laboratory, the patient identifying information is usually keyed into a laboratory database [or uploaded from electronic transmittal systems, in more technologically advanced systems usually linking on collection device serial number], the laboratory tests are performed, the results are recorded, and the test results are reported back to the submitting facility and/or to the physician of record, depending on program rules and regulations. If abnormal or unsatisfactory results are obtained, then notification is also given to a follow-up coordinator responsible for ensuring that confirmatory diagnostic or repeat testing occurs, depending on the urgency of the testing results. This coordinator may contact subspecialists for assistance, or in emergency situations, may directly contact the patient's family.

Newborn hearing screening

In a typical NHS system, the newborn demographic information in the hospital admissions database is accessed for input into the hearing testing equipment in the newborn nursery [alternatively, this information may be obtained directly from the parent(s)]. Testing is performed and the testing results are recorded in the patient's chart (and may also be stored in a database at hospital or associated with the testing equipment). If a second test is required by the testing protocol, then these test results are also entered into the patient's chart (and database). If further testing is needed, then this information is usually shared with the parents and audiological follow-up is recommended (perhaps even scheduled). In many hospital-based NHS systems, the hospital is responsible for following up on results of the confirmatory testing and ensuring that the patient is aware of any needed intervention services. The process of obtaining intervention (hearing aids, etc.) may also be a hospital responsibility, but often this step in the system is left to others. If centralized data reporting is required by the government agency overseeing NHS, then the hospital is responsible for maintaining and transferring testing results from the hospital database to a central data repository.

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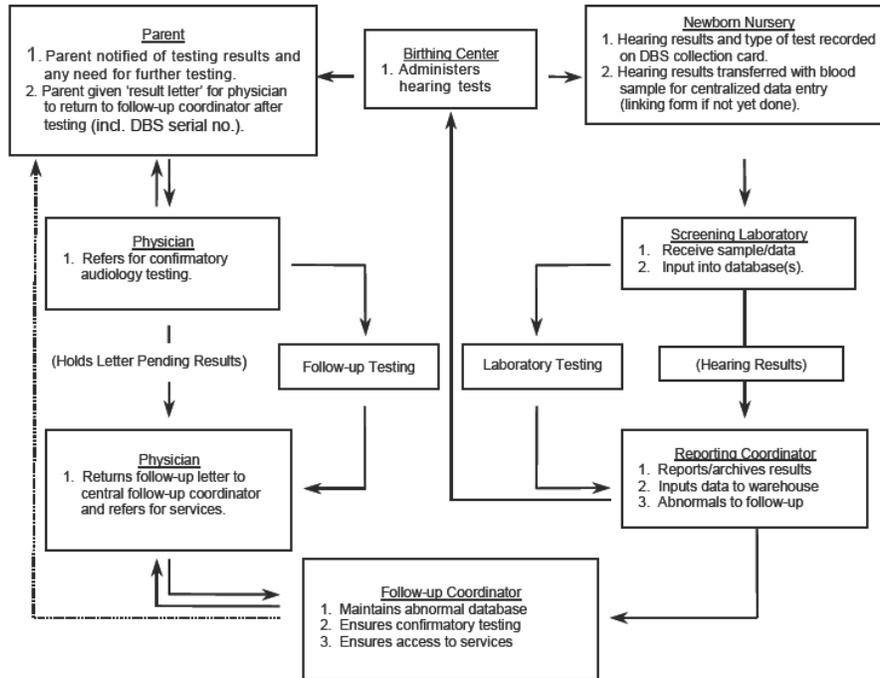


Fig 1. Diagram showing one mechanism of integrating newborn hearing screening and data into an ongoing dried blood spot (DBS) screening program.

Alternatively, as an aid to data management and centralization of the data, some NHS programs have added a limited number of extra data fields to the DBS collection form so that the hearing screening data can be recorded and transferred with the DBS data (see Fig 1). In such systems, NHS results are recorded on the DBS collection card, along with the patient's demographic information, and submitted to the central newborn screening testing laboratory.

Here they are recorded along with the other patient information and then transferred to a hearing follow-up data system. If the hearing results are not available in the nursery at the time the DBS sample is submitted, a tear out sheet is usually included as part of the DBS collection device so that this sheet can be removed and submitted later after hearing testing has been completed. The tear out sheet contains the NBS serial number for linking to patient's demographic information and can be color-coded to aid in recognition. In programs where NHS and DBS follow-up are combined, the data may reside in the same system, but this is not necessary if follow-up and program evaluation are facilitated through other databases. The DBS card merely provides a mechanism for transmitting data in a fast and efficient way to a central database. The

addition of NHS data fields on the DBS card also serves as an educational reminder to hospitals not yet performing newborn hearing screening. Data submitted in this way, cover essentially all newborns in the jurisdiction and provide not only a mechanism to enhance follow-up, but also allow for a count of patients not yet receiving NHS services.

DISCUSSION

A unique linking number is often cited as the critical missing element in linking or integrating data systems. Patient names or other data elements or combination of data elements have been used for linking, but they are usually complex and subject to a number of different caveats in order to make them useable. For example, if a name is used, then the spelling must be exact. Some programs have tried to develop unique linking numbers from selected information fields in the data, and some have even tried to create pseudo-social security numbers, based on the mother's social security number, but problems have arisen in the case of multiple births and/or early infant deaths. The simpler and more effective approach to data linking has been to provide and use the unique serial numbers preassigned and preprinted on the DBS newborn screening collection devices. In addition to providing a progressive

sequential numbering system, 'smart' DBS serial numbers can include data elements for other program components including the year in which the cards were ordered and the birth location (ie the particular state in which the birth occurred). A check sum character can also be included at the end of the serial number to ensure the accuracy of the number when it is entered into a computer system. Newborn screening collection devices can be manufactured with multiple serial number stickers printed and attached to the collection devices. These stickers can be formatted with serial numbers and/or barcodes, and can be easily peeled away from the DBS collection device and attached to other paperwork. In this way, the identification number can be easily appended to other linkage documents. For example, the linking serial number can be affixed to the birth certificate, to the patient's medical record, and/or to the tabular listing of patients maintained in the nursery for newborn screening submission and result record keeping. In cases where newborns receive more than one newborn screen, the initial screening collection device provides the permanent tracking number to which all subsequent specimens from that patient are linked.

In the US, essentially all programs now include a serial number on the DBS collection device, and many have it bar coded for quick entry into automated data systems. While many programs use the DBS serial number as a means of tracking the patient's collection device from the birthing facility to the testing laboratory and beyond, the DBS serial number was originally created for use in inventory management. In inventory management systems, the serial numbers of collection devices shipped to various birthing facilities are recorded so that the facility can be easily identified should its identification fail to be recorded on a submitted specimen (eg if a facility loaned some collection devices to another facility). In cases where such an inventory control system is linked to the laboratory data management system, submitter data (uploaded from the inventory system) can be automatically supplied to the data management system at the time the serial number is entered. In this way, the keystrokes necessary to type the submitter's identity and address can be conserved during the data entry process. Editing overrides can be provided in data entry software so that address changes can be made if the submitter information on the form differs from that contained in the inventory control system. Additionally, bar coded serial numbers that include check sum characters automatically validate the number at data entry so that bar codes not only speed the entry process but also improve the accuracy of data entry.

As noted earlier in this report, the current NHS environment includes a large number (approximately 50%) of newborns with positive hearing screening tests who do

not receive confirmatory testing and/or intervention services. By integrating newborn hearing results with DBS newborn screening programs already in place, and by using existing DBS follow-up systems as a model, NHS follow-up should be able to improve in effectiveness and efficiency (American College of Medical Genetics, 2000). In NHS programs the overall data flow is similar to that of DBS screening. While many NHS programs are organized so that screening and follow-up responsibilities reside with the birthing facility, especially in small states and in pilot programs, the need for improved follow-up, monitoring of service access and delivery, and centralized program evaluation data creates a data flow that is operationally similar to that of DBS programs. Thus, it seems reasonable to consider using the DBS collection device as a mechanism for transmitting NHS data to a central database utilizing the data capturing processes already in place in an existing DBS program. Indeed, some state NHS programs have already taken advantage of this mechanism for data transmittal. Currently, data fields for NHS information are included on the DBS collection devices in: Delaware, Maryland, Michigan, Minnesota, Mississippi, Missouri, Nevada, Oklahoma, Oregon, Tennessee, Washington, and Wisconsin. Basic information such as type of test and test results can be easily added to most collection forms using 1-digit coded responses, and entry of these data have little impact on the data entry workload. For example, minimal data may include recording a coded response for whether a test was performed, the hearing results for each ear, and/or the equipment used for testing. While modification of an existing DBS data system to accommodate added data fields is often required, the modifications are usually straightforward, easily accommodated, and advantageous to the overall screening system. At least one state, Utah, has initiated data linking/integration using the birth certificate as the primary data source to which all programs link, but utilizing the DBS serial number as the unique linking number. While the experiences thus far in Utah have been positive, in some states where a provision for recording the DBS serial number on the birth certificate exists, completion of this data field has not been required, and therefore its potential for linking has been limited because the field is often left blank.

The patient demographic information required for newborn hearing follow-up is similar to that required for routine DBS follow-up. The minimal data elements suggested for DBS newborn screening are specified in a national standard (now in its fourth revision) and are limited to the essential data elements needed for identifying patients considered at risk as a result of screening (Hannon *et al.*, 1997). Already captured in most DBS databases are: infant's name, address, phone number, physician of record,

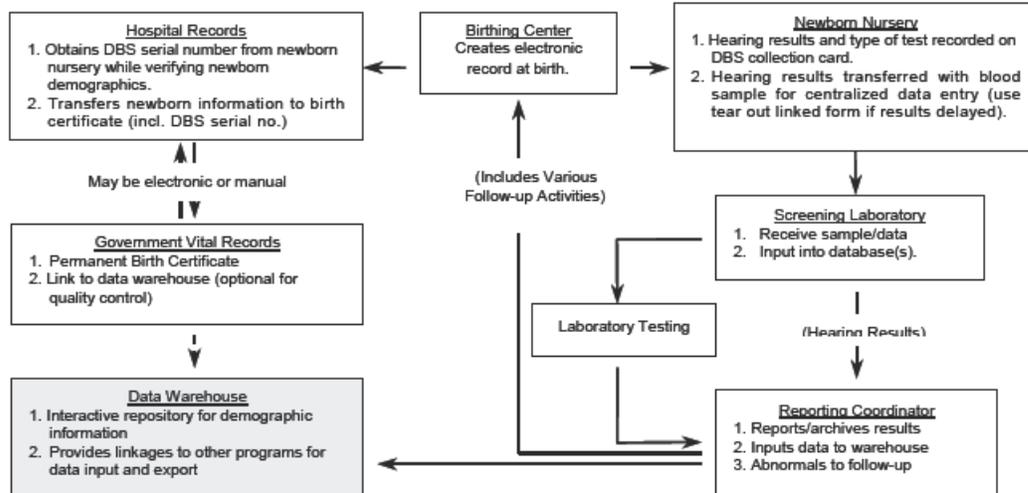


Fig 2. Diagram of newborn screening data flow using the warehousing concept and linkages with vital records as a means of ensuring that all newborns receive both a newborn screen and a birth certificate.

physician's phone number, etc. By limiting the information captured on all patients to essential elements, the mass of population data entered into the data system is minimized. Case specific information on the small number of patients with abnormal test results can be obtained as part of the follow-up process. Limiting the case specific data in this way adds to the overall efficiency of the data collection/data entry part of screening by leaving non-critical information to be obtained only on the less than 1% of patients for whom it is needed. Thus, for example, additional data elements

specific to hearing loss can be recorded in a case management database (or other appropriate file), in a process similar to that generally employed in the DBS newborn screening follow-up. In cases where a screening program may wish to monitor data on risk factors, additional data elements could be added to the DBS form. However, care must be taken to ensure that the information anticipated from added data is valid and useful, since data entry expenses will be increased by any additional information that must be input. Overall, the logistics of recording NHS results on the DBS form at

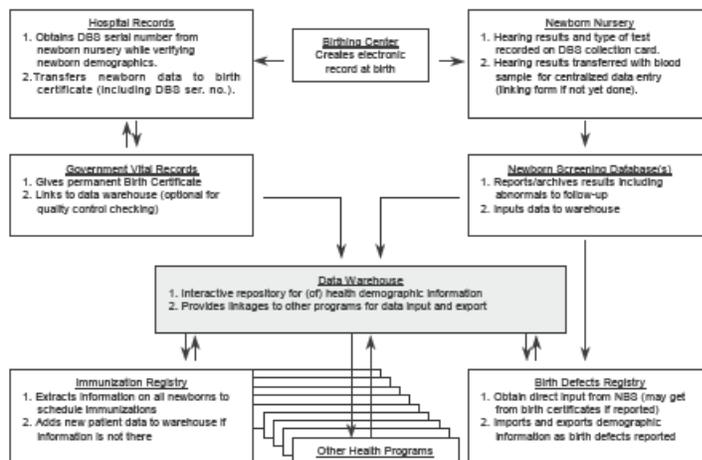


Fig 3. Diagram showing data flow into and out of a data warehouse, with particular attention to interactions with newborn screening, birth certificates, immunization registries, and birth defects registries.

the newborn nursery may be more complex than a stand-alone system for NHS, but data transfer between the screening systems can be managed by most hospitals with little additional effort, and the system savings in reducing duplication of effort is cost beneficial.

Timely information available from DBS and NHS early screening can also provide demographic information that could be useful for databases associated with birth certificates, childhood immunization programs and birth defects registries, among others. If newborn screening data are not used to populate the birth certificate database, they can still be used alternatively as a quality control check to ensure that birth certificates exist for each newborn receiving a newborn screening test (see Fig 2). Reverse validation may also be beneficial in assuring that each recorded birth has received an appropriate newborn screening test (although programs should be sensitive to the fact that birth certificate information is not collected to be used punitively).

A truly comprehensive data warehousing system would theoretically include mechanisms for integrating initial patient information from any program that may have the data available, whether or not it originated in the newborn screening program (see Fig 3). Thus, for example, if a child was to be given an immunization, an inquiry of the warehouse should indicate whether or not there was basic demographic information available, and additionally whether or not there was an immunization history. If demographic data were missing, then they would be input at that time and would be available for future inquiries, whether or not the inquiry originated with the immunization program.

In the typical birthing facility billing (and tracking) system, demographic information on the newborn is available almost immediately following the birth (technically a new hospital admission), and this information is available to hospital personnel in both the nursery and the hospital laboratory through internal communication pathways. This information has the potential to form the nucleus of a patient information data record from which basic patient information can be accessed and extracted as health-related encounters occur within the hospital. Therefore, these data offer the potential for uploading pertinent patient information to the DBS request card, the NHS data form and the birth certificate. Even though most hospitals now have electronic databases of newborn demographic information, and could share this information electronically within the hospital, it is still the practice in most birthing facilities to manually record patient

information on the newborn DBS screening collection device or other patient records. Additionally, many facilities also keep manual tracking records of DBS specimen submissions in order to ensure that results are received on all newborns transiting the newborn nursery.

Birthing facilities maintain a supply of DBS collection cards, and in almost every state, the DBS collection card contains a preprinted unique serial number defined by the state newborn screening program, primarily for inventory purposes and patient tracking within the system. This serial number provides a convenient identification number for use not only in the DBS program, but also for patient linking to other systems. It is usually the case, that other health program data systems also have identification numbers that could be used in such a linking system or data warehousing system, but since the DBS serial number is the first one to be used chronologically, and since it can exist as multiple preprinted labels that could be peeled off the DBS card and affixed to other patient records, it seems to be a simple solution to the problem of an identifier that can link systems to the patient.

While to some it may be appealing to develop new programs from the ground up without interference from outside influences, it is usually the case that new programs can benefit from the experiences of older programs. This is particularly true of data systems in which there has historically been a rapid change in data technology resulting in systems modifications that are expensive and complex. The experiences of 'old' data systems such as those associated with DBS newborn screening provide a wealth of development experience, and the opportunities for linkage and 'exploitation' by other health systems with similar data needs should not be overlooked. It is incumbent on all public health programs to continue to explore ways in which data duplication can be minimized with consequent savings in funds previously used for data entry so that health monies can be better utilized for service delivery.

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Appendix 6

Standardizing Newborn Screening Results for Health Information Exchange

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<http://www.lhncbc.nlm.nih.gov/lhc/docs/published/2010/pub2010037.pdf>

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Standardizing Newborn Screening Results for Health Information Exchange

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Abstract

Newborn screening is a complex process that has high-stakes health implications and requires rapid and effective communication between many people and organizations. Currently, each state program has its own method of reporting results, with wide variation in content and format. Pediatric care providers receive reports by mail, email, fax or telephone, depending on whether the results are normal or abnormal. This process is slow and prone to errors, which can lead to delays in treatment. Multiple agencies worked together to create national guidance for reporting newborn screening results with HL7 messages that contain a prescribed set of LOINC and SNOMED CT codes, report quantitative test results, and use standardized units of measure. Several states are already implementing this guidance. If the guidance is used nationally, office practice systems could capture NBS results more efficiently in EHRs, and regional and national registries could better analyze aggregate results to facilitate further research for these rare conditions.

Introduction

Newborn screening (NBS) is a vital process that identifies apparently healthy infants with serious medical conditions so they can be treated before they suffer significant morbidity or mortality. NBS includes both dried blood spot (DBS) and hearing tests. Most NBS conditions are rare and comparing data across states is necessary to optimize screening protocols and assess screening outcomes. Until this project began, there was no standard for reporting NBS results, and therefore no way to efficiently transmit data to pediatric care providers, or to reliably compare or pool data across states. In this report we describe a standard way to deliver newborn screening results in a Health Level Seven (HL7) message.

Background

In the United States, NBS programs are operated by fifty states plus the District of Columbia, certain U.S. territories and the military. Almost all of the programs test for the 29 core conditions defined by the Recommended Uniform Screening Panel of the Secretary's Advisory Committee on Heritable Disorders in Newborns and Children.¹ Many also screen for various additional conditions. Most programs recommend DBS screening at 24-48 hours after birth, and hearing screening at least 24 hours after birth but before hospital discharge. Nine states require, and others recommend, a repeat DBS screen at one to two weeks of age such that about 25% of US newborns receive two screens. The newborn's blood is obtained by heel stick, and collected on special filter paper attached to a collection card that includes questions ("card variables") about the newborn and mother. Metabolic, hematologic, endocrine and other abnormalities are screened using various tests on the DBS. Congenital hearing loss is screened by physiologic measures.

Programs differ not only in the number of conditions screened, but also in how the results are reported. Each program uses its own local non-standard codes for tests and results. Some programs report only qualitative interpretations of results ("normal"), while others use various combinations of narrative, qualitative and quantitative reporting. Even quantitative results can vary – some programs report numeric values, while others result ranges (e.g. 2.3 vs. <10 or 5-7) or percentiles. Some programs report results for individual conditions or the individual test measure, while others group results based on disorder categories, with some variations in grouping among states (e.g. amino acids, fatty acids and acylcarnitines). Given all of this variability, it is very difficult for office practice system developers to capture NBS results efficiently into electronic health records (EHRs), and for regional or national registries to collect and analyze NBS information.

NBS programs report positive DBS results to pediatric care providers by phone due to the urgent need for follow up and treatment. Currently, most NBS programs use postal mail to send normal NBS reports to the birthing facility or pediatric care provider designated when the baby is screened; however, the provider who sees the baby in the hospital is often not the same one that follows the baby long-term. Some states do not send negative results as timely as they could, which can cause confusion and delay. In one survey of pediatricians, 26% reported they were not routinely notified of the screen-negative results. When asked hypothetically if they would actively track down NBS results for a 2-week-old infant with a normal exam, 28% reported they would not either because they assumed “no news is good news,” the state does a good job, or a combination of “the infant is healthy and lack of report implies the test results were negative.”² Although a few states provide websites or automated voice response systems where physicians can obtain screening results,³ tracking down newborn screening results can require many attempts per baby, which is a burden on the office staff.

Newborn hearing screening results are hospital-based, not laboratory-based. The mechanism for reporting hearing screening results depends on the jurisdiction and, in some cases, hospital-level policy and procedures. Hospitals can communicate hearing screening results in various formats to stakeholders such as the family, the state Early Hearing Detection and Intervention (EHDI) program, and audiologists. This non-uniformity of communications processes is one barrier to effective hearing screening follow-up.

Methods

Our goal was to develop a template that could carry the DBS screening results and accommodate state variations in hearing screening and reporting styles. We used a hierarchy of nested Logical Observation Identifiers Names and Codes (LOINC®)⁴ panels to create this template, following an approach that has been successful for many other complex LOINC data capture processes.⁵ This approach provides a way to organize variables in a nested structure with their associated attributes: data type, cardinality,⁵ UCUM®⁶ units of measure (for numeric variables), answer lists (for categorical variables), descriptions

and help messages. The contents of this structure can be mapped to an HL7 message with each LOINC code and its corresponding test value carried in one OBX (observation result) segment within the HL7 message. Nesting can be reflected in the message by incorporating an OBR (observation request) segment for each node in the hierarchy.

The information in a LOINC panel can be represented by three relational database tables. One table carries a record for each LOINC term used in the panel with all of that term’s attributes. The second describes the relationship of a nested LOINC panel to its observation codes as a hierarchy. Each record in the second table contains a link to a parent LOINC term in the first table and other attributes that vary for a given LOINC term across panels. The third table contains answer lists for all of the categorical questions in the panel.

We designed an all-inclusive LOINC NBS panel – called the American Health Information Community (AHIC) Newborn screening panel – based on the above structure so that a given NBS program can select the variables it needs for reporting conditions screened. Therefore, different states can include different subsets of tests in their test reports, but any result for a test that is the same across more than one state will be reported using the same LOINC code in the same format.

The U.S. Department of Health and Human Services Office of the National Coordinator for Health IT (ONC) obtained and analyzed DBS cards from all U.S. NBS DBS programs, and we developed a condensed set of questions and answer lists that covered the content represented in these cards. This standardized content included demographic information (such as baby and mother’s name and contact information – which go into the HL7 Patient Identification (PID) and Next of Kin (NK1) segments respectively), as well as birth history information that laboratories and clinicians may use to interpret and analyze screening results (such as history of blood transfusion or antibiotic administration prior to specimen collection). We worked with many organizations and individuals to develop and refine the answer lists for card variables, overall screening impressions, hearing loss risk indicators, hemoglobin disorders and more.

The Centers for Disease Control and Prevention’s (CDC) EHDI Program helped develop a single set of LOINC answer codes for hearing screening methodology, results, and hearing loss risk indicators. A LOINC answer list includes all of the hearing loss

⁴ Including HL7 clinical genomic reporting, many large and complex Medicare forms (OASIS, MDS, CARE), HEDIS quality measures, laboratory test panels and standardized research measurements (PhenX and PROMIS).

⁵ Cardinality specifies whether the field is required, and whether you can have multiple repeated values.

risk indicators identified by the Joint Committee on Infant Hearing (JCIH) 2007 Recommendations.⁶ In an HL7 message, a single LOINC code for “hearing loss risk indicators” can repeat as necessary across many HL7 OBX segments to carry information about multiple risk factors. When no risk factors are identified, a single OBX segment should be used with the answer code for “None.”

The National Library of Medicine (NLM) also worked with the CDC National Center for Health Statistics Division of Vital Statistics to create LOINC codes and corresponding answer lists for several card variables that reflect information contained in the 2003 revisions of the U.S. Standard Certificate of Live Birth. These variables include date of birth, time of birth, obstetric estimation of gestational age and mother’s education. Everything we did was reviewed and refined via feedback from many NBS experts and agencies as well as input during a formal Health Information Technology Standards Panel (HITSP) public comment period.

Regenstrief Institute assigned to all of the variables a unique LOINC code, units of measure, and cardinality as appropriate. For categorical variables, we defined formal answer lists and assigned each answer a placeholder LOINC answer code. We also included SNOMED CT codes (with permission from the International Health Terminology Standards Development Organisation) where available for the answers that represent the conditions, and, as they become available, we will add new SNOMED CT codes to other answer lists to facilitate universal interpretation.

The Interim Final Rule on Health Information Technology specified that electronic laboratory reports be transmitted as HL7 2.5.1 messages.⁷ The AHIC Personalized Healthcare Workgroup’s NBS Subgroup, with special help from the American College of Medical Genetics (ACMG), HRSA, the CDC EHDI Program, and the National Newborn Screening and Genetics Resource Center (NNSGRC), developed initial tables of NBS conditions screened in any state, associated measurements, and condition details.⁸

Finally, NLM worked with the Health Resources and Services Administration (HRSA) to develop guidance specifying how to construct HL7 newborn screening messages using the codes in the LOINC NBS panel, and developed an annotated example HL7 message as an embodiment of that guidance. This guidance harmonizes with the Public Health Informatics Institute Implementation Guide,⁹ which focuses more

on the administrative HL7 segments (e.g. MSH, PID, NK1), whereas this project focuses on detailed codes for the results “payload.”

Beyond the organizations mentioned above, the effort to produce a standard NBS message also required the expertise and guidance of the HITSP Population Perspective Technical Committee, lab system vendors, and state NBS programs (hearing and DBS).

Results

The LOINC NBS panel includes a total of 219 LOINC codes including 18 panel codes (used to group LOINC codes), 153 codes representing measured results or calculations and 30 codes for reporting interpretations of, or comments/discussion about, NBS results. In addition to individual analyte measurements and interpretations, the LOINC NBS panel contains summary interpretations (Figure 1) and card variables. The specification provides coded OBX segments for transmitting comments, instead of note (NTE) segments. Three of the summary variables (overall interpretation, reason for lab test in dried blood spot, and sample quality of dried blood spot) have specific answer lists, which are based on recommended practices and federal reporting standards, and each answer has its own LOINC answer code (Figure 2).

LOINC code	LOINC Name of variable (question)	R/O	Card	v	Data type
57128-1	Newborn Screening Report summary panel	R	1..1		
57721-3	Reason for lab test in Dried blood spot	R	1..1		CE
57718-9	Sample quality in Dried blood spot	R	1..1		CE
57130-7	Newborn screening report – overall Interpretation	R	1..1		CE
57131-5	Newborn conditions with positive markers [Identifier] in Dried blood spot	R	1..n		CE

Figure 1. Excerpt of the LOINC hierarchy showing codes and attributes (required/optional, cardinality and data type) for four of the eight variables in the Newborn Screening Report summary panel.

SEQ #	Answer	SNOMED CT Code	LA code
31	GA-1	76175005	LA12493-5
32	GA-1 (mat)	76175005	LA12494-3
33	GA-2	22886006	LA12495-0
34	HCY	24308003	LA12496-8
35	HHH	30287008	LA12497-6
36	HIS	124628005	LA12498-4
37	HMG	124611007	LA12499-2
38	H-PHE	68528007	LA12500-7

Figure 2. Answer list excerpt for “Conditions tested for in this newborn screening study,” with sequence numbers, SNOMED CT and LOINC answer codes.

The LOINC NBS panel also includes 12 card variables (e.g. state of origin, date of birth, time of birth, birthweight, and unique serial number of current sample), with individual answer lists for the

categorical variables (birth plurality, clinical events that affect newborn screen interpretation, hearing loss risk indicators, and mother's education). The full LOINC NBS panel is most easily reviewed by downloading the PDF from <http://newbornscreening.codes.nlm.nih.gov/nb/sc/constructingNBSHL7messages/>. You can also obtain a spreadsheet version from the same web site.

The LOINC NBS panel can accommodate NBS results from all of the U.S. NBS programs. It specifies the codes for an NBS HL7 message. To show how these codes load into such a message, we created an annotated example HL7 v2.5.1 NBS message.¹⁰ The example message includes segments for reporting NBS data including all of the card variables and summary reports, and some of the condition impressions and quantitative results. There are at least four potential destinations for newborn screening result messages: 1) the birth hospital, 2) the physician responsible for the infant's on-going care, 3) the state NBS and state EHDI programs and/or public health department, and 4) national and/or regional registries of NBS data. The message was designed to be used to send data to all such recipients with tailoring where needed, e.g. removing identifying data before sending to central registries.

The 50-plus NBS DBS programs are served by some 36 NBS laboratories, and there are only a few main commercial information system vendors, plus some internal state computer information departments. Because the numbers of involved organizations are limited, relative to other health information exchange contexts, rapid adoption of this standardized HRSA/NLM approach to NBS results messaging is possible. Indeed 15 months after the AHIC report to the HHS Secretary,⁸ three major NBS lab system vendors (Natus/Neometrics, PerkinElmer and Oz Systems) can demonstrate early versions of HL7 messages that meet this specification, and at least one laboratory is already sending NBS HL7 messages (Figure 3).

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OBX|26|SN|31144-9^Thyroxine^LN||^21.3^^|ug/dL^
microgram/deciliter^UCUM|^>5.0 ug/dL||||F|||200703311010

OBX|27|SN|54084-9^Galactose^LN||^1.9^^|mg/dL^
milligram/deciliter^UCUM|^<14 mg/dL||||F|||200703310904

OBX|28|SN|33288-2^Galactose 1 phosphate uridylyl transferase^LN||
^^^|U/g(Hb)^units/gram Hb^UCUM|Enzyme Present||||F
|||200703310904

OBX|29|CE|54105-2^Hemoglobin Pattern^LN||LA11974-5^Hb F,A
(normal)^LN||||F|||200704031215
```

Figure 3. Excerpt from prototype Pennsylvania HL7 message, being developed by PerkinElmer and Oz Systems.

Discussion

The HRSA/NLM HL7 message guidance provides a uniform way to communicate newborn screening results in a computer-understandable form. As hospitals and office practice EHR systems adopt this guidance, they will solve some of the current problems with reporting NBS results. Most commercial EHRs already come equipped with HL7 inbound interfaces, and the Standards and Certification Criteria Interim Final Rule requires the support of LOINC and encourages the use of SNOMED CT in laboratory messages to meet the definition of meaningful use.⁷ The Centers for Medicare and Medicaid Services is also considering expanding the Medicaid EHR incentive programs to include NBS documentation as a pediatric clinical quality measure.¹¹ If all U.S. NBS laboratories adopted the standard described here, EHRs could be designed to accept these messages out of the box, with no need to individually map and customize what would otherwise be large differences in NBS reporting formats, by state.

Some regional health information exchanges provide web-based report delivery systems that accept lab results messages from many sources (e.g. hospital laboratory, stand-alone radiology services) and deliver them in a uniform format to physician offices. Such systems, which provide another vehicle to deliver NBS results to care providers, already operate in Indiana, parts of Ohio, and Ontario, Canada (eCHN). Kentucky is developing a statewide health information exchange that will use HL7 messages to provide NBS results as its inaugural effort.

Having a standard message will also make it easier to collect regional and national data. Many of the conditions are extremely rare, with incidences of 1 in 100,000 births. Therefore, pooled data for all newborns screened are needed to study the effects of NBS follow-up programs and potential health interventions. These collections should contain quantitative results for negatives as well as positives. With such collections of quantitative data, researchers can improve screening methods and reduce false positive rates.

There have been differences of opinion in the NBS community about reporting numerical results as well as interpretations to pediatric care providers when screening tests are positive for a given condition. We support reporting numerical screening results whenever they can be reliably reported as the result of a standardized process. "Less than" or "greater than" results should only be reported when specific

results are outside of the analytical range of the measurement. In the case of tests that produce non-numeric results, such as hemoglobinopathy screening and DNA mutation analysis, the specific hemoglobin or mutation observed should be reported as opposed to a qualitative interpretation such as "positive." If cut-offs are obtained by evaluating percentiles rather than averages of analyte concentrations the limitations of that approach should be explained

Discussions with local pediatricians suggest that they tend to prefer qualitative reporting for negative NBS results because they are quick to read and digest. On the other hand, they prefer to get numerical values for the positives derived from quantitative measures, because the numerical values cue them to the likelihood that the positive is a true positive, needing close follow-up. Having the numerical results also makes it easier to discuss the results with the family.

Though challenges remain – including the unavailability of the follow-up physician's name at the time of initial screening and a lack of electronic and automated linkages to vital records (and other systems that could help assure that all infants are tested and receive appropriate follow up) – we are encouraged that standardized NBS messaging is being embraced so rapidly. This early success is testimony to the great cooperation among many organizations in the NBS community and their keen interest in the health of newborns.

Disclaimer

The findings and conclusions in this paper are those of the authors and do not necessarily represent the official position of the CDC, HRSA, NIH, NLM, or the Department of Health and Human Services.

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