STATE OF MARYLAND
DHMH
Maryland Department of Health and Mental Hygiene
201 W. Preston Street • Baltimore, Maryland 21201
Martin O'Malley, Governor – Anthony G. Brown, Lt. Governor – John M. Colmers, Secretary

JAN 0 5 2009

The Honorable Thomas M. Middleton
Chair
Senate Finance Committee
3 East, Miller Senate Building
Annapolis, MD 21401-1991

The Honorable Peter A. Hammen
Chair
House Health & Government Operations
Committee
241 House Office Building
Annapolis, MD 21401-1991

RE: HB 216 (Ch. 256) of the Acts of 2008 -
2008 Legislative Report on Whether A Coordinated Statewide System For Screening Newborn Infants Be Applied To All Newborn Infants In Maryland

Dear Chair Middleton and Chair Hammen:

Pursuant to House Bill 216 which was enacted during the 2008 legislative session, the Department of Health and Mental Hygiene is directed to submit this one-time legislative report that addresses whether a coordinated, Statewide system for screening newborn infants in the State for certain hereditary and congenital disorders should be applied to all newborn infants in the State.

If you have any questions about this report, please contact Ms. Anne Hubbard, Director of Governmental Affairs, at 410-767-6481.

Sincerely,

John M. Colmers
Secretary

Enclosure

cc: Frances B. Phillips, R.N., M.H.A.
Russell W. Moy, M.D., M.P.H.
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MARYLAND DEPARTMENT OF HEALTH AND MENTAL HYGIENE

2008 LEGISLATIVE REPORT

SHOULD A COORDINATED STATEWIDE SYSTEM FOR SCREENING NEWBORN INFANTS BE APPLIED TO ALL NEWBORN INFANTS IN MARYLAND?

Martin O'Malley	Anthony G. Brown	John M. Colmers	Frances B. Phillips
Governor	Lt. Governor	Secretary	Deputy Secretary Public Health Services

Introduction

House Bill 216-2008 directs the Department of Health and Mental Hygiene (DHMH) to study whether a coordinated statewide system for screening newborn infants in the State for certain hereditary and congenital disorders should be applied to all newborn infants in the State. HB 216-2008 further requires DHMH to report the conclusions from the study to the Senate Finance Committee and the House Health and Government Operations Committee.

Survey of Newborn Screening Programs throughout U.S.

To determine whether all infants in Maryland should be required to undergo newborn screening, the DHMH first researched the newborn screening practices in all states. A survey of state newborn screening personnel regarding parental consent and refusal policies was conducted by DHMH in 2008 for this report (see Appendix A). Data from several previous surveys in other states were also reviewed (see Appendix B) and it appears the data are very consistent.

All states have newborn screening procedures. Every state except one allows an infant's parents to refuse newborn screening in some manner. The only state that requires mandatory newborn screening with no exceptions is Nebraska. Wyoming and, historically, Maryland require written informed consent for newborn screening. Eleven other states and Washington, DC allow parents to object for any reason. Five states do not mention the possibility of parental refusal in their laws, but refusal is allowed and documented in these states. Thirty-one states allow parents to refuse screening for religious reasons and a few parents do refuse in each of these states. Massachusetts is the only state that routinely has both mandatory screening with a religious exemption (for a screening panel meeting classical screening criteria for well-established disorders) and informed consent (for an optional screening panel involving less well-validated disorders).

National Recommendations Support Voluntary Newborn Screening

The major national professional groups and federal agencies recommend that parents be allowed to refuse newborn screening. In 2000, a very influential national *Newborn Screening Taskforce* was convened by the American Academy of Pediatrics (AAP) and funded by the Maternal and Child Health Bureau (MCHB) of the federal Health Resources and Services Administration (HRSA) to make recommendations for the future of newborn screening. This taskforce was co-sponsored by the Agency for Healthcare Research and Quality (AHRQ), the Association of Maternal and Child Health Programs (AMCHP), the Association of Public Health Laboratories (APHL), the Association of State and Territorial Health Officials (ASTHO), the Centers for Disease Control and Prevention (CDC), the Genetic Alliance and the National Institutes of Health (NIH). The report of this taskforce, entitled "*Serving the Family from Birth to the Medical Home - Report of the Newborn Screening Taskforce,"* was published in Pediatrics 106: 383-427 (2000).

The pertinent recommendations of this taskforce are that: 1) parents should be informed of testing and have an opportunity to refuse testing; and 2) if after discussions about newborn screening with health care professionals, parents refuse to have their newborn tested, the refusal should be documented in writing and honored. (Pediatrics 106: 411 (2000). The recommendations of this taskforce were then endorsed by the major health-related federal agencies (CDC, NIH, AHRQ), the major associations of public health professionals (AMCHP, APHL, ASTHO), and the Genetic Alliance, a broad-based association of genetic support groups and genetics-oriented consumer organizations.

Another recommendation of the *Newborn Screening Taskforce* was that a uniform screening panel of disorders to be included in newborn screening programs be developed and used by all states. HRSA then contracted with the American College of Medical Genetics (ACMG) to develop a methodology for including conditions in newborn screening and to develop the uniform screening panel. The results of this work, *Newborn Screening: Torward a Uniform System and Panel,* were published in Genetics in Medicine 8: 1s-11s (2006). A uniform screening panel was developed and has been endorsed by the Health Resources Services Administration (HRSA) and is now standard of care.

Policy Arguments

Parental refusal of newborn screening is clearly rare throughout the U.S. However, all states have a few families who do refuse, including Nebraska. Only Nebraska takes legal action against parents who refuse. Many states do not track refusals but 24 states provided data on their refusal rates. Very few states have the ability to match in real time birth certificates and newborn screening submissions and so cannot document refusals in that way. Documented refusal rates range from 0.004% to 0.8%, with most of these states having less than 0.2% refusals (see Appendices A & B). In 2007, the DHMH found that 78,738 infants were screened by the Maryland Newborn Screening Program and there were only three refusals. (See Appendix C for the methodology used for determining the number of refusals and for the methodology for calculating the number of infants screened.)

There are very few known cases where an infant affected by one of the disorders in the newborn screening panel was missed because the parents refused screening. One recent survey focused on cases that were missed because of parental refusal. (see Appendix B) Thirty-three state newborn screening programs responded to the survey. Nine infants (including two sets of twins) were identified as having been missed because of parental refusal between 1990 and 2007. Approximately 71 million babies were born between 1990 and 2007. This is approximately one in 10,000,000 or 0.00001 % of babies born. It is not surprising that relatively few missed cases were identified because very few parents refuse and because the disorders themselves are quite rare. More babies are missed because of false negative results. The false negative rate for disorders detected by tandem mass spectrometry (a relatively new multiplex testing technique which makes it possible to screen for a number of disorders that could not be detected by previous methods) is probably in the area of 1 in 100,000 but the false negative rate for some disorders is very high (10% for cystic fibrosis, 7 to 12 % for hypothyroidism).

If the parents wish to refuse screening, the rights of the parents must be balanced with the rights of the newborn infant. The United States has sturdy societal values supporting individual liberty, including the right to parent, the right of an individual to decide what treatments they will have, whether they will participate in research, and what can be done with their personal information and their bodily tissues, including blood samples. In these matters, parents are normally considered the appropriate individuals to make decisions on behalf of their children. However, individual liberty can be overridden by society if the individual's action or negligence poses a significant risk of harm to another person. When parents refuse standard treatment for a child who will die or become seriously ill if he or she is not treated, the state properly intercedes to ensure treatment according to the doctrine of parens patriae. However, in general for the state to intercede there has to be imminent serious harm. In the case of newborn screening, however, the baby is not sick and the chance that the baby will not have one of the disorders in the screening panel is greater than 99.5 %. (See Appendix C for calculation.)

In Maryland in 2007, there were 197 infants with some disorder or variant identified through newborn screening out of 78,738 infants screened. Many of these infants had a disorder that does not cause death or mental retardation. Over half of these infants (104 infants) had a hemoglobin disorder, for which treatment does not have to be started until three months of age. A newborn infant who is not affected by any of the disorders in the screening panel does not actually derive an overriding benefit from screening.

A prevailing concern about the effects of mandating screening is the intrusion of government into medical care and the family. What is the impact of mandating newborn screening? Fears persist that mandatory newborn screening could set a precedent for mandatory governmental programs in other areas where it is clearly inappropriate. There is also concern that families wishing to refuse screening will choose to forgo routine obstetrical and newborn care and will have their infants at home without a birth attendant. Arguments also arise over the propriety of mandatory testing since the test results have limited predictive power both with respect to whether an infant will actually develop a certain genetic disease and whether medical outcomes will be favorably impacted. Moreover, there is also an increasing number of test panels to include less-validated disorders for which no effective treatment exists.

Conclusion

The conclusion of this report is that the coordinated statewide system for screening newborn infants should be applied to all newborn infants unless the parents or guardians of the infant object.

The report further concludes that the policy of the State of Maryland should still require that parents be informed about the purpose of newborn screening. Maryland's parent education materials can still strongly recommend newborn screening. The DHMH recommends that written documentation of consent should no longer be required, lightening the paperwork burden on hospitals and providers. This policy would be consistent with the policy set by the statute establishing the Advisory Council on Hereditary and Congenital Disorders (Health-General Article, §§ 13-101-110, et. seq.). No new legislation is required to adopt this policy and COMAR 10.10.13 and 10.52.12 is compatible with this policy.

As previously stated, the federal recommendation is that parents should be allowed to refuse newborn screening and that all refusals of newborn screening be recorded in writing. All states except Nebraska allow parents to refuse. Requiring and enforcing that all babies be screened would result in a significant government intrusion into the family, and into the relationship between the family and their medical providers, for a relatively remote chance that a baby will have one of the disorders in the screening panel. This would not meet the usual threshold for invoking the principle of parens patriae. The public health mission is not damaged by allowing a very small number of families to refuse. Nationally less than 0.02% of families refuse and even fewer Maryland families refuse, 0.004% in 2007.

Adoption of this policy will bring Maryland policy in line with that of the vast majority of states and current national recommendations.

Appendix A

Survey of State Newborn Screening Parental Consent Policies 2008

Survey of State Newborn Screening Parental Consent Policies 2008						
State	**Actual Refusals Allowed?**	**How Many Parents Refuse?**	**How Documented?**	**Refusal Mentioned in Educational Materials?**	**Can Parents Refuse Part of the Screen?**	**Can Parents Refuse Storage of Residual Sample?**
States with Laws Requiring Informed Consent						
Maryland	yes	0.004%	Consent form	yes	no	no
Wyoming	yes	unknown	Consent form	unknown	unknown	unknown
States with No Exemptions In Law						
Arizona	yes	unknown	written petition and check box on lab slip	no	no	no
Michigan	yes	10 per year	generic form that hospitals can adapt	no	no	yes
Montana	yes	0.01%	attendant/hospital responsible for having a signed refusal form	no	no	no
South Dakota	yes	0.08%	Internal program follow-up form	no	no	no
West Virginia	yes	no actual refusals known	documented in medical record	no	no	no

Survey of State Newborn Screening Parental Consent Policies 2008

State	Actual Refusals Allowed?	How Many Parents Refuse?	How Documented?	Refusal Mentioned in Educational Materials?	Can Parents Refuse Part of the Screen?	Can Parents Refuse Storage of Residual Sample?
States with No Exemptions In Law and Enforcement Provisions						
Nebraska	no	0.004% (1every other year)	reported to County Attorney	no	no	no
States with Laws Allowing Parental Refusal for Any Reason						
Alaska	yes	10-20 per year	Form	no	no	no
Arkansas	yes	don't know exactly	documented in the medical record	no	no	no
Colorado	yes	no data	recommend documenting in the medical record	yes	have not heard of that	may request and receive residual specimen
Florida	yes	<1%	Written dissent	no	no	no policy
Louisiana	yes	no reliable data	no state requirement	unknown	unknown	no policy
Iowa	yes	unknown	Unknown	yes	unknown	unknown
Minnesota	yes	0.07%	Form	yes	no	yes
New Hampshire	yes	0.01%	Form	no	no	no policy
New Mexico	yes	0.05%	Form	no	no	no
Nevada	yes	unknown	Unknown	unknown	unknown	unknown
Vermont	yes	0.05%	Form	no	would consider	yes
Washington DC	yes	unknown	Unknown	yes	unknown	unknown

Survey of State Newborn Screening Parental Consent Policies 2008

State	Actual Refusals Allowed?	How Many Parents Refuse?	How Documented?	Refusal Mentioned in Educational Materials?	Can Parents Refuse Part of the Screen?	Can Parents Refuse Storage of Residual Sample?
States with Laws Allowing Parental Refusal for Religious Reasons						
Alabama	religious	rare	Form	not answered	no	no
California	religious	0.12%	Form	yes	no	yes
Connecticut	religious	0.01%	Form	yes	no policy	no policy
Delaware	religious	0.02%	Form	no	no	no
Georgia	religious	do not track	varies by hospital	no	no	no
Hawaii	religious	0.02%	Form	yes	unknown	no
Illinois	religious	rare, no reliable data	varies by hospital	no	no	no
Indiana	religious	0.02%	Form	no	yes	no
Kansas	religious	0.06%	Recommended form	no	no	no
Kentucky	religious	0.02%	Recommended form	no	yes	no
Maine	religious	0.05%	Form	yes	no	yes
Massachusetts	religious	rare	Documented	yes	no	no
Michigan	religious	10 per year	form recommended	yes	no	yes
Mississippi	religious	0.01%	Form	no	no	no
Missouri	religious	10 per year	Documented	no	yes	no
New Jersey	religious	rare	Documented	no	no experience with this	no experience with this
New York	religious	rare, few times a year	Recommended form	no	no	yes
Oklahoma	religious	1-2 per year	Form	no	no	no
Ohio	religious	0.02%	Form	no	no	no
Oregon	religious	0.03%	Form	no	no	no experience with this
Pennsylvania	religious	0.01%	written statement of the objection	yes	no	no policy
Rhode Island	religious	0.01%	Form	yes	no	yes
Texas	religious	unknown/rare	Recommended form	no	no	yes
Utah	religious	<1%	Form	no	no	no
Virginia	religious	rarely (4/year)	Form	yes	no	no experience with this
Washington	religious	0.28%	Form	yes	no	yes
Wisconsin	religious	unknown/rare	form recommended	yes	no	no

APPENDIX B

Florida and Nebraska Surveys on Parental Consent and Missed Cases

Florida Data Sharing Survey 2008

Lois Taylor, RN, BSN, CPM, Director of the Florida Newborn Screening Program did a survey in early 2008 to find out if newborn screening programs were sharing data with their state birth defects registries. In the course of the survey she asked state newborn screening programs if they got written informed consent for newborn screening. Her results are consistent with those in the 2006 article by Dr. Bradford L Therrell, Director of the National Newborn Screening and Genetics Resource Center. The article, Status of Newborn Screening Programs in the United States, was published in Edwards ES, Howell RR and Lloyd-Puryear A, Eds. A look at newborn screening: Today and tomorrow. Pediatrics 117: S193-S350 (2006).

Ms. Taylor polled all 50 states plus the DC program to see if they had changed their process since the article by Dr. Therrell. Only two states, Maryland and Wyoming, were found to require written consent for the newborn screening tests. Neither state confirms whether the consent is completed in all cases. They rely on the hospital to obtain it and maintain the form in the medical record. One state requires verbal consent for the MS/MS disorders. Massachusetts does not require written consent for the mandatory disorders but does require that parents give a "verbal" consent. If they do not, the specimen card is marked and the testing for the 19 MS/MS disorders is not performed by the lab. The Florida survey follows:

Other States' Experience with Written Consent and the Sharing Of Newborn Screening Data with Other Programs.

State	*Share Newborn Screening Data with other programs*	*Comments*	*Require Written Consent for NBS test*	*Comments*
Alabama	No		No	
Alaska	No/Yes	Could not share data with BDR (in same division) without MOA	No	
Arizona	No	BDR doesn't want it because the data "is bad" with too many errors	No	
Arkansas	No	There is limited sharing of data with NNSIS	No	
California	No	Unsure if they can.	No	
Colorado	Yes	Shares with BDR. There is linking between Vital Stats. IT actively cleans up data.	No	
Connecticut	Yes	Child health profile regular collaborative partners	No	
Delaware	No	Unsure if they can.	No	
DC	Yes	Tries to share within department	No	
Florida	Yes	Will begin sharing with BDR in 2008	No	
Georgia	Yes		No	
Hawaii	No	Can't because of confidentiality clause in NBS rule	No	

State	*Share Newborn Screening Data with other programs*	*Comments*	*Require Written Consent for NBS test*	*Comments*
Idaho	No	Birth certificate info is shared with NBS if box for NBS not checked. (All birth certificates have NBS info checkbox.)	No	
Illinois	Yes	Will begin to share with BDR soon.	No	
Indiana	Yes	Shares with BDR because it NBS disorders are reportable conditions in BDR law.	No	
Iowa	Yes	Has agreement with BDR.	No	
Kansas	No	Unsure if they can.	No	
Kentucky	Yes	Both NBS and BDR housed together.	No	
Louisiana	Yes	Shares with BDR. Has HRSA grant to share data but has MOU to protect data.	No	
Maine	Yes	New England has grant to collect long term data. BDR in Title V agency. NBS has integrated database with BDR and hearing. Has MOA with Part C/DOE.	No	
Maryland	No		Yes	Written consent form developed by MD NBS is completed by hospital. Kept in medical records. Have never checked it, takes hospital's word that it was done.
Massachusetts	No	But law allows research with data and dried blood spots	No/Yes	No for "old" disorders. Yes for the new 19 disorders, parent must give "verbal consent". Specimen card will be "X'd" out if parent refuses.
Michigan	Yes	Shares with BDR	No	
Minnesota	No/Yes	Not sharing data but would give it to them if they asked for it.	No	
Mississippi	Yes	BDR and NBS are housed together.	No	
Missouri	Yes	Will do in future, have in past. Hearing info is given to Child Health Profile	No	
Montana	No	But does give NBS info to the Title V program for follow-up purposes.	No	
Nebraska	No	Statute limitations.	No	
Nevada	No	BDR shares with NBS but not vice versa.	No	
New Hampshire	No		No	

State	*Share Newborn Screening Data with other programs*	*Comments*	*Require Written Consent for NBS test*	*Comments*
New Jersey	Yes	To verify and register cases.	No	
New Mexico	No/Yes	But is getting database together	No	
New York	No	Probably wouldn't share but currently does compare info with hepatitis and HIV programs	No	
North Carolina	Yes	But limited sharing of info.	No	
North Dakota	No		No	
Ohio	No/Yes	But would if they asked.	No	
Oklahoma	No	But may share with VS, bar code is on the birth certificate.	No	
Oregon	No/Yes	If they were asked.	No	
Pennsylvania	No		No	
Rhode Island	Yes	Have integrated child health information system.	No	
South Carolina	No/Yes	But they haven't asked yet.	No	
South Dakota	No		No	
Tennessee	No/Yes	Not sharing with BDR yet but NBS # is on birth certificate.	No	
Texas	No		No	
Utah	No		No	
Vermont	No	Have concerns about sharing data.	No	
Virginia	Yes	Sends NBS to BDR	No	
Washington	No	Notifies child profile of deaths	No	
West Virginia	No/Yes	Would share but they haven't asked	No	
Wisconsin	No/Yes	But will give them data if they ask.	No	
Wyoming	No	Matches birth certificate with NBS by hand	Yes	Consent form is completed at hospital. Forms are not checked by NBS but they could be asked by hospital licensing agency.

M:\Lois\Newborn Screening\2008 survey re consent sharing data.doc

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Nebraska Survey on Parental Consent and Missed Cases

Julie Miller, the Newborn Screening Coordinator in Nebraska polled the states as to their parental consent practices when Nebraska was evaluating its mandatory screening policy. Her study follows:

The Risk of Missing a Newborn When Parental Refusal Is Allowed

Julie Miller, B.S., Nebraska Newborn Screening & Genetics Program.

The question of parental rights to refuse newborn screening testing has been debated as long as newborn screening has been done. The vast majority of programs allow parents to sign a "waiver" objecting to and refusing screening based on either religious or other reasons. Only four State's laws governing newborn screening or other medical care do not include a provision allowing refusal for any reason, (Nebraska, Montana, South Dakota and West Virginia).[1] Of these four, only Nebraska's includes an enforcement provision. So what is the best policy?

A 2006 informal survey of Newborn Screening Programs via the National Newborn Screening & Genetics Center list-serve found most programs that allow refusal did not routinely provide a precise number or percentage of parents who refused. (Thirty three U.S. programs and two international programs responded to this survey). This may have been due to: data not being readily retrievable; not requiring the refusal documentation to be submitted to the State program in order to account for all refusals, or; because the program did not routinely do a matching process between the birth records and newborn screening records to account for every birth in their state via a record of newborn screening test results, or proof of parent refusal. The percent of parents refusing on behalf of their newborns ranged between 0.01% < 5.0%. Due to several non-specific responses such as < 1%, the mean and median refusal rates could not be determined.

State refusal policies March 2006 survey

State	Documentation required	Percent or # refusing
Reason accepted for refusal *Any reason*		
Colorado	Nothing required in writing	2.5% not screened, but unknown # of refusals
District of Columbia	Each hosp decides how to document parent dissent	> 1%
Florida	Sign a form provided by the hospital	unknown
Iowa	Sign pre-existing form	<0.5%
Maryland	Sign pre-existing form	0.01%
Minnesota	Sign pre-existing form	<1%
Netherlands	Nothing required in writing	0.01%
New Hampshire	Sign pre-existing form	unknown
New South Wales	Sign pre-existing form, hospital submits "refusal" on filter paper	0.06%-0.1% last 6 years
Reason accepted for refusal: *No exceptions in law*		
Michigan	Sign pre-existing form (state-provided model consent)	unknown < 1/month
Nebraska	Refusal not allowed. Law includes enforcement provision.	2 in last 3 years
Reason accepted for refusal: *Religious only*		
Alaska	Sign pre-existing form	<1%
California	Sign pre-existing form	500/yr <0.1%
Delaware	Sign pre-existing form	1-2%
Georgia	Sign a statement, (not a form statement)	<1%
Hawaii	Sign pre-existing form	0.1%
Illinois	Nothing required, recommend hospital document	unknown
Indiana	Sign pre-existing form	4-5 per year
Kansas	Sign pre-existing form made available	<5%
Kentucky	Nothing required, recommend hospital obtain signed refusal	unknown
Massachusetts	Nothing required in writing, recommend physician assure understanding for required / (<2% dissent from optional testing	no response

Missouri	Write out their refusal, keep in medical record and to DOH	≅ 5/year
New Jersey	Hospital choice	unknown
New York	Sign pre-existing form	5 per year
North Carolina	Nothing required in writing, request physician send note	<1%
North Dakota	Sign pre-existing form	unknown 0%
Ohio	Sign pre-existing form	0.02%
Oregon	Sign pre-existing form	< 0.03%
Pennsylvania	Physician documents in record	<1%
Rhode Island	Sign pre-existing form, file med record and with state program	no response
South Carolina	Sign pre-existing form	0.04%
Texas	Write out /sign their refusal, form available	unknown, < 1%
Virginia	Physician sends to state in writing	unknown
Washington	Sign pre-existing form (back of filter paper)	0.045%
Wisconsin	Nothing required in writing, form made available	0.005%

If it is true that so few parents refuse, statistically the odds of missing a rare condition should be extremely rare given the additional factor that such conditions individually are quite rare. For example the AAP Newborn Screening Fact Sheets[2] estimate the incidence rates for Galactosemia at 1:47,000, PKU at 1:19,000 to 1:13,500, MCAD at 1:46,000-1:6,400, and Congenital Hypothyroidism at 1:4,000 to 1:3,000.

However, a review of collective incidence rates when a State Program screens for the recommended core panel of 29 disorders, finds a much higher incidence rate. In Nebraska alone, with less than 27,000 births per year, incidence rates have been about 1:850 [3] for clinically significant conditions in this panel. (99.77-99.81 % of newborns received the required panel, and more than 96% receive the rest of the disorders screened by tandem mass spectrometry which has been optional since July of 2003).

It is less surprising then to learn of several missed cases due to parent refusal of newborn screening. The data below was received from an informal survey conducted on the National Newborn Screening & Genetics Center's list serve in January of 2007. State Newborn Screening Program representatives responded to the question "Is anyone aware of any missed cases due to parent's refusal to have the screen?" Responses were received from 30 States. Twenty states reported no known missed cases due to parental refusal. However, 9 babies were reported to be affected or "missed" due to not being screened because of parental refusal, and 4 additional babies were reported as "near misses," from parents initially choosing to refuse, but who ultimately were convinced to have their newborn tested.

"Missed cases" reported on 2007 NBS List-serve survey:

State	Year	Condition	Age at diagnosis	Circumstance
California	2004	2 with PKU (siblings)	1 @2 years, 1@ 5 months of age	Father had refused screen. Finally consented to screen when midwife noticed delays in the older child.
Hawaii	1993	1 with Congenital Hypothyroidism	6 months of age	Parent refused screen
Indiana	2003	1 with Galactosemia	Patient expired at 8 days of age	Parent refused screen
Maine	1992	1 with Congenital Hypothyroidism	4.5 months of age	Parent refused screen
Michigan	Not reported	2 with PKU (siblings)	Not reported	Parent/midwife refused screen. Older child diagnosed after

				second child. Midwife now a strong advocate for screening.
Missouri	1990's*	1 with Congenital Hypothyroidism	Not reported	No formal refusal, but Mennonite family, and child had not been screened.
Washington	Not reported	1 with Congenital Hypothyroidism	14 months	Parent refused screen

*exact year not reported

Given the continuing advances in newborn screening technologies and treatment, there is every likelihood that newborn screening panels will continue to expand. With additional conditions added to the panel, the collective incidence rates get higher, as does the risk of missing affected newborns. Perhaps it is time to reconsider which should weigh the most in policy and practice decisions.

[1]"Status of Newborn Screening Programs in the United States," Therrell, Bradford L, et.al., *PEDIATRICS, A SUPPLEMENT TO PEDIATRICS, A Look at Newborn Screening: Today and Tomorrow*, May 2006, Volume 117, Number 5, S212-S252

[2]"Newborn Screening Fact Sheets," Kaye, Celia I and the Committee on Genetics, *PEDIATRICS* 2006; 118;934-963, DOI: 10.1542/peds.2006-1783

[3] " Newborn Screening in Nebraska" Annual Report 2004, 2005, 2006, http:www.hhss.ne.gov/nsp

"Near Misses" reported on 2007 NBS List-serve survey:

State	Year	Condition	Circumstance
California	Not reported	2 with MSUD (twins)	Initially parents refused, but midwife encouraged them for almost two weeks till they consented.
Colorado	2001	PKU diagnosed at 28 days	Parent refused screening, but physician convinced them to rescind this at a follow-up visit.
Texas	Not reported	Galactosemia	Parent wished to leave hospital without the screen. Nurse begged them to stay a few more minutes to have the test. Parents now strong advocates for screening.

Given the continuing advances in newborn screening technologies and treatment, there is every likelihood that newborn screening panels will continue to expand. With additional conditions added to the panel, the collective incidence rates get higher, as does the risk of missing affected newborns. Perhaps it is time to reconsider which should weigh the most in policy and practice decisions.

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APPENDIX C

Methodology for Determining the Number of Babies Screened
Methodology for Determining the Number of Refusals
Diagram of Population

Methodology for Determining the Number of Refusals

Informed consent is required for newborn screening. When parents refuse screening, it is documented on the Informed Consent Form provided by the DHMH as the last page in the detailed parent information brochure "Screening Tests for Your Baby." This form goes into the baby's permanent medical record. In addition, the hospital, or the person required to file the certificate of birth on an out of hospital birth, is required to fill out a newborn screening filter paper blood-spot collection test requisition card (bloodspot card) and send it in to the DHMH without the blood sample and marked refusal. When the card is received in the laboratory, the information on the card is entered into the newborn screening data base (in the same way as the information on all other cards) and the results are entered as unsatisfactory 12 (UNSAT 12) (no blood). There may be other reasons for a card being submitted without blood. For example, up until August of this year, when the Web-based infant hearing screening database went live, infant hearing screening test results were reported on the bloodspot cards. Birthing facilities tried to coordinate the hearing screening tests and the bloodspot collection so both could be sent in on the same card; however, sometimes the hearing screening test results were not yet ready when the bloodspot card had be sent to the laboratory. In that case, the hearing screening test results were sent in on a separate card, with no blood.

In order to assess the number of refusals, the DHMH examined all records with UNSAT 12 results for the year 2007. The entire newborn screening record of each baby was examined to determine whether the baby had been adequately screened. Each UNSAT 12 card was also visually inspected to see what was written on the card. In all but three cases, examination of the baby's complete record in the newborn screening database, showed that the baby had been adequately screened on samples submitted on other cards. The conclusion is that **there were three refusals in 2007**. Previous studies had shown an average of five refusals a year. There is no way to absolutely assure that a card was submitted for all babies whose parents refused screening. The DHMH does receive calls from birthing facilities about parents wanting to refuse screening. Birthing facility staff devote extra attention to parents wishing to refuse to be sure that they understand the program, employing interpreters when necessary. The newborn screening follow up personnel are frequently asked to speak with the family. In almost all cases, when newborn screening is explained to the parents, they change their minds. If the family still wishes to refuse, their refusal is honored and documented.

Methodology for Determining the Number of Babies Screened

Albert Einstein said, "Make things as simple as possible, but not simpler." Determining the number of babies screened, as opposed to the number of specimens analyzed, is complex. This requires an understanding of the population utilizing the program, an understanding of Maryland's two specimen newborn screening system and an understanding of the difficulties introduced by having two competing laboratories for the last five years. An overly simplistic interpretation of the data leads to erroneous conclusions.

The Population Utilizing the Maryland Newborn Screening Program

Maryland has "leaky borders". This is a common problem in areas like the north eastern region of the U.S., where states are small and the nearest major city maybe in another jurisdiction. In these regions of the country it is common for people living in one state to use hospitals and health care providers in another jurisdiction. In 2007, there were 74,939 babies born in Maryland, 71,214 of these babies were born to Maryland residents but 3,725 of these babies were born to families who are not Maryland residents. All these babies are screened in Maryland. In 2007, there were 78,054 babies born to Maryland residents, 71,214 of these babies were born in Maryland but 6,840 of these babies were born to Maryland residents in other jurisdictions. Some but not all of the 6,840 babies born out of State are screened by Maryland. (See diagram in this Appendix) Indeed some of these babies are screened in two jurisdictions, where they were born and then again in Maryland. Maryland has screened more babies than are physically born in Maryland for many years. (There are numerous reasons for a Maryland resident baby born outside Maryland to be screened in Maryland. In recent years, the deterioration of hospital medical records systems, misinterpretations of HIPAA, and the changes in the periodicity schedule of the American Academy of Pediatrics recommending that babies have a pediatric office visit at three to five days of age, have resulted in many providers sending a newborn screening specimen on a baby less than a week old from the office because it is so difficult to get the results of the specimen sent from an out of state hospital.)

Maryland's Two Specimen Newborn Screening System

Maryland and 14 other states use a two specimen system. (The second specimen is mandated by law in eight states and recommended in seven states). The first specimen is usually drawn in the hospital after 24 hours of milk feedings and the second specimen is drawn between one and 4 weeks of age. It is usually ordered by the pediatrician but often drawn by a laboratory provider designated by the baby's health insurance. (It is not always possible to tell if a specimen is sent from a hospital or from a pediatric office. In addition, close to 15% of babies are in the neonatal intensive care unit for some period of time and many are transferred to hospitals with more sophisticated intensive care nurseries, causing confusion about the hospital of birth.)

There are certain disorders that are better detected on a specimen drawn after the first week of life, for instance homocystinuria and hypothyroidism due to ectopic hypoplastic glands. There are disorders that require two specimens, separated in time, to complete screening, for instance cystic fibrosis by the IRT/IRT method. In addition, a second specimen gives the program a second chance to detect all the disorders in the screening panel. A two specimen system complicates record keeping when there are two competing newborn screening laboratories, because a baby may have the first specimen sent to one laboratory and the second specimen sent to the other laboratory. The federal definition of "screened" is at least one sample, so both laboratories would claim to have screened that baby. Clearly the data from each laboratory has to be looked at in greater detail to determine the number of babies screened.

The Best Estimate of the Number of Babies Screened

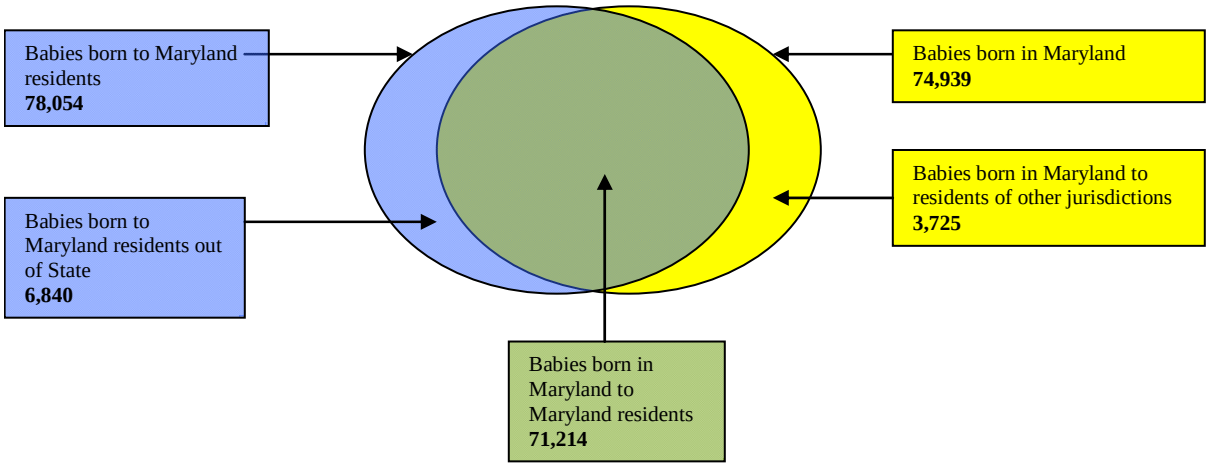
The number of infants screened was determined by counting only specimens screened by each laboratory when the infant was less than one week old. (This is the usual definition of an initial screen.) **The best estimate of the number of individual babies screened is 78,738.** There are two labs screening and we do two routine screens. The State lab screened 73,370 infants; 59,850 as newborns less than 7 days old and 13,370 screened for the first time as subsequents older than 7 days. The state lab screened a total of 65,759 babies as subsequents. The commercial laboratory screened 19,756 babies; 18,888 screened as newborns and 868 screened for the first time as subsequents. The commercial laboratory screened a total of 11,705 babies as subsequents. Every effort, within our limited IT resources, was made to match babies from both laboratories to avoid duplication but names, addresses and phone numbers change so frequently between birth and the first pediatric visit that complete deduplication is impossible.

National Newborn Screening and Genetics Resource Center Specific Disorder Reports

The specific disorder reports of the National Newborn Screening and Genetics Resource Center do not accurately reflect the number of Maryland babies screened from 2003 to 2008. These reports require detailed data on all “not normal” results as well as data on presumptive positive results. During this period the State Public Health Laboratory screened the majority of the babies and a commercial laboratory screened the remainder. The specific disorder reports reflect only the data on the babies screened by the State Laboratory. The commercial laboratory only provided data on presumptive positive results which was insufficient for inclusion in the specific disorder reports. The babies “missing” from the specific disorder reports for those years were indeed screened but were screened by the commercial laboratory.

Population of Babies Screened in the Maryland Program

CY 2007 Births



Babies receiving blood spot newborn screening- includes all births in Maryland and some babies born to Maryland residents out of State
78,738

National reporting convention for blood spot newborn screening:

Numerator: **number of babies screened**

Denominator: **number of babies born in Maryland**

Therefore in CY2007, Maryland screened 78,738 / 74,939 or 105%.
(In these cases, it is usually reported as ~ 100%.)
Specimens are sent in by courier or mail.

Number of Babies with Disorders Identified by Newborn Screening

In Maryland in 2007, there were 197 babies with some disorder identified through newborn screening out of 78,738 babies screened. About 1 in 400 babies had some disorder. Over one-half of these babies (104) had a hemoglobin disorder, which is to be expected because of Maryland's large African-American population. Historically, 5% of the 104 babies with hemoglobin disorders would die before age two years without treatment; however, treatment does not have to be started until three months of age. Ninety-three babies, approximately one in 1000 babies, had something other than a hemoglobinopathy. Three had cystic fibrosis, four had congenital adrenal hyperplasia, 26 had hypothyroidism, 13 had the transient hypothyroidism of the premature and 47 had "metabolic disorders" (including 15 non-clinically significant variants and 32 clinically significant cases). The 32 clinically significant cases include five with 3MCC (3 methylcrontoyl-CoA carboxylase deficiency) which rarely causes serious illness or mental retardation. However, 78,542 infants were unaffected by any disorder in the screening panel (99.75% of the infants screened).

If all the significant metabolic cases and all the clinically significant cases of hypothyroidism are counted, there were 58 infants at risk for mental retardation. This is approximately 1 in 1,350 at risk for mental retardation. Twenty-one of the "metabolic" cases (fatty acid oxidation disorders, organic acidurias and galactosemia) and the four cases with CAH were at risk from death in infancy. This is approximately 1 in 3,333 infants at risk for death in infancy. (Some infants were at risk for both metal retardation and death.) For these 62 Maryland infants (1 in 1,282) born in 2007, newborn screening provided a benefit of overwhelming importance.

APPENDIX D

Decision of the Nebraska Supreme Court on Improper Enforcement of the Mandatory Newborn Screening Law

http://www.firstamendmentcenter.org/news.aspx?id=20958

First Amendment Center - Nashville, Tennessee - USA; December 5, 2008
Nebraska High Court Upholds Newborn Screening Law

NEBRASKA HIGH COURT UPHOLDS NEWBORN SCREENING LAW

By The Associated Press

LINCOLN, Neb. — Nebraska's newborn-screening law didn't violate the right to freely practice religion, but state officials crossed a line when they took an infant from his parents for a week last year, the state Supreme Court ruled today.

The state high court upheld the law requiring blood to be drawn from infants to test for rare and deadly diseases. But the court also said an Omaha infant was hurriedly, and improperly, taken from his parents by officials who said the parents weren't properly caring for him and that he was in danger because the tests weren't performed.

Said the infant's mother, Mary Anaya, as she wept today: "I'm relieved to know if we find ourselves in the same situation we don't have to be violated by having (police) sweep into our home and terrorize our children."

But, she added, "What's done can't be undone. It's only a small satisfaction that what was done to us was found to be wrong when there's no consequences ... it's a small consolation."

She and her husband, Josue, believe that, in accordance with the Bible, blood shouldn't be deliberately drawn and that doing so can shorten a person's life. Nebraska is one of four states — South Dakota, Michigan and Montana are the others — that doesn't offer a religious exemption for parents who don't want the test performed.

The test involves a pinprick and checks for eight metabolic and genetic disorders, including cystic fibrosis.

In its opinion today, the high court recalled a ruling it made three years ago in the case of another Anaya child, Rosa. In that case, the couple argued that the newborn-testing law violated the First Amendment of the U.S. Constitution, which says laws can't prohibit the exercise of religion.

The state Supreme Court eventually turned down their arguments, but Rosa never was tested.

In the new case, the Anayas challenged the screening law by saying it violates a clause of the Nebraska Constitution that says people can worship God according to their own consciences. The Anayas argued that the state law sets a higher standard than the U.S. Constitution.

But the high court disagreed, saying similarities between the two constitutional clauses required it to review the law's compliance with the U.S. Constitution. The court repeated its opinion from three years ago that the law does not specifically aim to infringe on religion, pointing out the law does not include exemptions for some children to be excused from testing.

The case decided by the high court today began after Mary Anaya gave birth to Joel at home on Sept. 2, 2007. The Anayas received a letter and phone call weeks later asking whether they would have Joel tested.

The letter said that if they did not, the county attorney would be notified.

Unlike in 2003, when Rosa wasn't tested, state officials wanted to make sure testing was completed. They got an order from a juvenile court judge to take the baby.

Sheriff's deputies went to the Anayas' home Oct. 11, 2007, to take Joel, who was almost 6 weeks old.

Mary Anaya said four armed deputies came into her house that day. She said she tried to keep them outside, even using physical force, but they came in and took Joel from the arms of her 12-year-old child.

"It was terror," she said.

He was placed in the custody of the state Department of Health and Human Services.

A Douglas County juvenile court judge ordered the next day that the baby remain in foster care until the preliminary results came back and confirmed further testing wasn't needed. Joel was returned to them Oct. 16, when the tests came back negative.

The high court, in the opinion written by Justice Lindsey Miller-Lerman, criticized the decisions that were made. While failure to do the required tests can be considered with other actions to conclude a child is being neglected, "failure to test under the newborn screening statutes, standing alone, does not establish neglect," the judge wrote.

"There simply was no legal, factual, or logical basis to keep Joel in state custody after the blood sample was taken," today’s opinion continues.

The high court pointed out that the newborn-testing law directs officials to use district courts to force parents to comply with the law.

Jefferson Downing, attorney for the Anayas, said: "The use of the juvenile code was absolutely improper. The manner in which they went about enforcing the statute broke this family for a week."