2011 SENATE HUMAN SERVICES

SB 2067

2011 SENATE STANDING COMMITTEE MINUTES

Senate Human Services Committee
Red River Room, State Capitol

SB 2067
1-12-2011
Job Number 12816

☐ Conference Committee

Committee Clerk Signature

Explanation or reason for introduction of bill/resolution:

Relating to newborn disease screening and research regarding metabolic and genetic diseases.

Minutes: Attached testimony.

Senator Judy Lee opened the hearing on SB 2067.

Kim Mertz, director of the Division of Family Health for the ND Department of Health, testified in support of SB 2067. See attached testimony #1.

JoAnn Brager, VP of Public Policy for the ND Association for the Education of Young Children, testified in support of SB 2067. Attachment #2

There was no opposing or neutral testimony.

The hearing on SB 2067 was closed.

Senator Tim Mathern moved a **Do Pass.**

Seconded by **Senator Gerald Uglem**.

Roll call vote 5-0-0. **Motion carried.**

Carrier is **Senator Dick Dever**.

Date: 1-12-2011

Roll Call Vote # 1

2011 SENATE STANDING COMMITTEE ROLL CALL VOTES

BILL/RESOLUTION NO. 2067

Senate **HUMAN SERVICES** Committee

☐ Check here for Conference Committee

Legislative Council Amendment Number _____

Action Taken: ☒ Do Pass ☐ Do Not Pass ☐ Amended ☐ Adopt Amendment

☐ Rerefer to Appropriations ☐ Reconsider

Motion Made By **Sen.** Mathern Seconded By **Sen.** Uglem

Senators	Yes	No	Senators	Yes	No
Sen. Judy Lee, Chairman	✓		Sen. Tim Mathern	✓	
Sen. Gerald Uglem, V. Chair	✓				
Sen. Dick Dever	✓				
Sen. Spencer Berry	✓				

Total (Yes) 5 No 0

Absent 0

Floor Assignment Sen. Dever

If the vote is on an amendment, briefly indicate intent:

REPORT OF STANDING COMMITTEE

SB 2067: Human Services Committee (Sen. J. Lee, Chairman) recommends **DO PASS** (5 YEAS, 0 NAYS, 0 ABSENT AND NOT VOTING). SB 2067 was placed on the Eleventh order on the calendar.

2011 HOUSE HUMAN SERVICES

SB 2067

2011 HOUSE STANDING COMMITTEE MINUTES

House Human Services Committee
Fort Union Room, State Capitol

SB 2067
March 9, 2011
Job #15156

☐ Conference Committee

Committee Clerk Signature Vicky Crabtree

Explanation or reason for introduction of bill/resolution:

Relating to newborn disease screening and research regarding metabolic and genetic diseases.

Minutes: See attached Testimony #1

Chairman Weisz: Opened the hearing on SB 2067.

Barb Schweitzer: Director of ND Newborn Screening Program for Dept. of Health testified in support of the bill. (See Testimony #1.)

Rep. Devlin: Do the parents any say in whether they want this testing done or not?

Barb: Yes. We have a refusal form they can sign.

Rep. Porter: How are we expanding this education program without a fiscal note? There must be a cost to the department to do some of the things that you are saying.

Barb: We are funded by the MCH (Maternal Child Health) Title V Block Grant and if and when we add new disorders or diseases, we go through the ND newborn screening advisory committee, the state health council as well as the legislative council for any additional disorders or diseases that we are going to add. Our fee is $50 a screening.

Rep. Porter: As we the legislature are added it isn't there an upfront cost that should be identified on a fiscal note for the printing of new materials and for new tests that are going to be ordered; then offset by special fund grants so we can see what it is going to cost?

Barb: May I defer to that to Kim Mertz my supervisor?

Rep. Porter: Sure.

Kim Mertz: Director for the Division of Family Health. Currently we do utilize the MCH or the Title V Block Grant for the educational component of this program. The MCH Block Grant is really money that comes into our state to serve the MCH population. This program fits very nicely into the objectives of that grant. It is true however, that the grant has been

leveled funded for many years and is slated for reduction at the federal level. In the past we have had what we consider adequate funding to fund this program. It will become difficult when federal funds receive a reduction. The actual screen itself is $60 and this is third party reimbursement. Does that answer your question?

Rep. Porter: No it didn't. There is going to be a cost to this program that is being presented in this bill. We are not seeing what the cost is or where the funds are coming from like the normal fiscal note process. If those funds are flat and there is a possibility that they aren't going to be part of that Title V Block Grant, then it has to come from some place. It would be the general fund or you are moving funds inside of the agency or you are doing something to pay for what you are asking us to do here. I'm not comfortable doing that without knowing how this is going being funded in the normal fiscal note scheme of things.

Kim: This bill does nothing to add screening. Basically what this bill is, is add the word genetic. It really is a cleanup language that we need to move forward. There is no more screening added in this bill. This bill in itself will not add anymore fiscal responsibility to the Dept. of Health. In the administrative rule process in the future as we add new screenings that is when the additional cost will occur.

Rep. Porter: Then in section 3 by adding genetic disease into the development and implementation of educational program, you don't have to reprint anything or print new brochures? This new program is already printed and in place and already going?

Kim: For the majority yes, it is ongoing. If we add new diseases we would have to make a new protocol for that. At this time we really do feel like we'd be able to add that additional educational resources within the current MCH Block Grant. We have a partnership with the University of Iowa and we have a joint educational component that we work with Iowa.

Rep. Porter: One of the concerns brought forward to us as we look at these programs that are funded through grants where the grants are running out or stagnant or new programs that grants are being sought; then all of a sudden they are responsibility to the general fund. We don't hear about them while we are in session and all of a sudden they pop through administrative rules through the budget section and then we have this new obligation to worry about as we go to the next session. I am concerned that we add language that we are going to develop and implement and have a genetic disease educational program. How can you add genetic diseases to an educational program without having to reprint everything you have to get that to get that educational information out. So we are authorizing you, but you aren't telling us what it is going to cost to do that or where the funding is going to come from.

Kim: I agree with you and that is a legitimate concern. The MCH Block Grant does require state match. We do have state funds going into this program as well.

Rep. Hofstad: Can you tell me in the identification of these diseases and now expanding that spectrum, does that lead to more abortive pregnancies? Do you have statistical information that would suggest or support that or dispel that?

Barb: I'm not aware of that at all. The new born screening is a blood spot that gets collected 24 hours after birth.

Rep. Hofstad: This is after birth and not during pregnancy.

Barb: No it is not.

OPPOSITION

Beth Nodland: Testified in opposition of the bill. (See Testimony #2.)

Vice-Chair Pietsch: Closed the hearing on SB 2067

2011 HOUSE STANDING COMMITTEE MINUTES

House Human Services Committee
Fort Union Room, State Capitol

SB 2067
March 15, 2011
Job #15442

☐ Conference Committee

Committee Clerk Signature Vicky Crabtree

Minutes: You may make reference to "attached testimony."

Chairman Weisz: Let's take up 2067. There was opposition on this one. Designer Genes has 5 points of concern. All the bill is doing is adding the language genetic to metabolic that does open up a whole new area. I am surprised there is not a fiscal note in here. They already have an educational program set up for the metabolic.

Rep. Conklin: I move a Do Pass.

Rep. Holman: Second.

Rep. Devlin: We talked about the parent's right to opt out and whether we needed to look at that. I don't know if we want it any broader than having it conflict with their religious tenets and practices.

Rep. Hofstad: How about parents opting in. Do they not have the ability right now to get their newborn genetically tested?

Chairman Weisz: Yes the can. The point of this is the Dept. of Health wants it for a data base. Here we are looking at requiring the tests so they can develop the data base like they are for the metabolic disorders. Is that the road we want to go down or not? There are some who have concerns about having this genetic information on the data base somewhere. Didn't somebody offer an amendment for (drops sentence).

Rep. Schmidt: (Has microphone off and is inaudible.)

Rep. Paur: I have no opinion on this, but did ask Chris Dobson about the genetic and he said as long as it is done after the child is born he had no philosophical objection to it. But, whether it is a good idea, I don't know.

Chairman Weisz: Part of the reason for it is to be proactive. Some genetic conditions are not apparent at birth and don't show up until whenever. If they would have known earlier they could have done things earlier that would have helped the condition.

Rep. Hofstad: I agree with Beth's suggestion that we put a period after the word "testing" because to distinguish an objection because only of religious grounds is a bit too restrictive.

Chairman Weisz: You would prefer the language on 7 and 8 say, "the testing requirements do not apply if the parents object to the testing".

Rep. Conklin: Withdraw motion.

Rep. Holman: Withdraw my second.

Chairman Weisz: Just so we are clear, it will read starting on line 6, "the testing requirements of this section do not apply if the parents of a newborn child objects to this testing." The rest of the language will be deleted. Correct Rep. Hofstad?

Rep. Hofstad: That is right.

Rep. Kilichowski: I was just going over the testimony and this started back in 1964. Has there ever been any problems with people refusing. Is it automatically done now after the baby is born? Is there a cost involved?

Chairman Weisz: It is being done now. I don't know if there is any cost from the doctor's side. Obviously it costs to do the screening.

Rep. Kilichowski: Is it automatically being done? Are the parents informed that they have a choice?

Chairman Weisz: There is testimony here that implies that you are assumed in and so there isn't anything telling you that you don't have to do it. This amendment wouldn't even change that. If they want to object to the testing, they have to say it up front.

Rep. Devlin: I'm surprised there is no fiscal note. They talked about it being a maternal and child healthcare grant Title V. Four federal dollars and three state dollars and talked about a $60 fee up front when done by private screeners. Do we want to change the parent opt out for what we are already doing? I was thinking more of doing it for the genetic diseases. I think this is where the fight is going to be.

Chairman Weisz: You are saying leave the language as is for the metabolic and merely change it for the genetic?

Rep. Devlin: Yes. I think that is where the argument is going to be.

Rep. Holman: It might end up a money issue and then we will decrease the amount of testing if it is not a covered charge by somebody. I agree with Rep. Devlin.

Rep. Paur: You were saying this could be used for determining early problems with a child, but is that the intent of the bill? But, this is for research purposes.

Chairman Weisz: If you read the bill it is one, for a registry that can be used. It says here, "follow-up with attending physician cases with positive tests for metabolic diseases or genetic diseases". It is intended if something is screened to relay the information to

appropriate (drops sentence). So care can be taken. It says, "early detection of these diseases so that proper measures may be taken to reduce mortality, morbidity and associated disabilities". That is on top of page 2, in subsection 1. There are multiple reasons for this.

Rep. Paur: Wouldn't the test be paid for by the insurance or whatever and just use the information?

Chairman Weisz: I don't think it is clear who is paying for the tests. The only thing it says is the state health council may adopt rules that establish reasonable fees and may oppose those fees to cover the costs of administering tests under this chapter. All test fees collected by the state must be deposited in the State Health Department's operating accounts. Do you need a specific answer to that question before we take this up? If you do I will get it. Rep. Hofstad, do you want to modify you amendment?

Rep. Hofstad: I don't know how to do it.

Chairman Weisz: I think you would take "genetic" out on line 8 and the "or both" and leave that language as is. I think you would say, "the testing requirements of this section do not apply for the testing of genetic diseases if the parents of the newborn child object to the testing or for metabolic disease if it conflicts with their religious tenets and practices". You could change the language to be that.

Rep. Hofstad: I think that is appropriate and I modify my motion to reflect that.

Rep. Schmidt: Second.

Chairman Weisz: Starting on line 6, "The testing of these requirements of this section, do not apply if the parents of the newborn child object to the testing of genetic diseases or testing for metabolic diseases if it conflicts with their religious tenets and practices". You can object to genetic diseases and with metabolic you would keep current law in place.

Rep. Devlin: When we did the metabolic we gave everybody an opt out when we said, against my religious practices. We might be trying to reinvent the wheel here.

Chairman Weisz: I don't know if anyone has ever objected. I'm guessing Jehovah Witness wouldn't allow it.

Rep. Louser: Where is it that the parent is notified in ND that this is now going to happen?

Chairman Weisz: There is no need to inform.

Rep. Louser: If I wasn't on this committee I wouldn't know about this. And if I or my wife object for whatever reason we are not even going to know we had that option and may not even know the test was taken.

Chairman Weisz: I don't remember anyone telling us they were going to screen our kids. I wouldn't have been aware of it either.

Rep. Damschen: In one part of the bill on the first page lines 14-16, metabolic and genetic disease mean the same thing. Then in the rest of the bill they are separated or referred to individual or both. One sense it makes sense and then in another way it totally confuses me.

Chairman Weisz: Are you talking about in section 2, number 3 where it basically defines them as the same?

Rep. Damschen: Yes.

Chairman Weisz: We are in the sense giving them the same definition at a standpoint of how they are used in the bill. We do have a motion in front of us and we will vote on it. The motion would be to change the opt out language.

VOICE VOTE: MOTION FAILED

Rep. Devlin: If you want to give them the opt out, you can take out the last four words on the last page; "religious tenets and practices" and put the word "beliefs" after "there".

Chairman Weisz: Do we want some informed consent language? The physician has to tell you they are doing the genetic and metabolic testing?

Rep. Paur: I like Rep. Louser's observation and don't think it will hurt at all.

Rep. Devlin: I move we eliminate the last four words, "religious tenets and practices" and replace those words with the word "beliefs".

Rep. Anderson: Second.

VOICE VOTE: MOTION CARRIED

Chairman Weisz: Now informed consent so to speak.

Rep. Hofstad: Can we look at an admission form and see what it says, if there is consent there?

Chairman Weisz: I'll find out from the Health Dept. and talk to the medical community and find out what there is and isn't. Maybe we can kick out this afternoon.

Rep. Porter: A few sessions back we were dealing with mandatory requirements for vaccinations and the same language was in there about just for religious purposes. And the question came up that it is not always a religious reason why a parent would want to opt out of the procedure. We changed the language then if the parent had an objection to their child having the vaccine and didn't restrict them from attending school. If there was an outbreak of measles then the child would not be allowed to attend school until the outbreak had subsided. That has been about 10 years now it seems to be working out just fine. I don't agree with the idea that they come and take your child and say they are going back to

the nursery and then are secretly doing tests on your child. I think we need in this that the testing requirements of this section require informed consent also. The parents are told up front that they are doing these tests. This is still your child and you are the parent and they should have to ask you before they start poking and prodding your kid. I bet those tests were done on both of my kids and I never knew it was being done.

Rep. Holman: We should find out the process of this before we act on this.

Chairman Weisz: We will get some information.

Rep. Porter: In my mind it doesn't matter how they are doing it now. I don't' think it is going to hurt to make it an informed consent requirement and the language is broader so a person can object to it.

Rep. Devlin: If we have language in there for the vaccines we could use that to be consistent.

Chairman Weisz: We can look that up. We are recessed until 2:15 p.m.

2011 HOUSE STANDING COMMITTEE MINUTES

House Human Services Committee
Fort Union Room, State Capitol

SB 2067
March 21, 2011
Job #15706

☐ Conference Committee

Committee Clerk Signature Vicky Crabtree

Minutes: You may make reference to "attached testimony."

Chairman Weisz: Taking up SB 2067.

Rep. Paur: Wasn't Rep. Holman going to check on authorization? I thought somebody was about notifying the parents.

Chairman Weisz: There was a question on, should there be notification language? That's right.

Rep. Porter: On page 3, line 7, I put a period after testing and started a new sentence. The tests required informed consent prior to testing.

Chairman Weisz: Do you want the language in there for who the religious, philosophical and moral beliefs are opposed.

Rep. Porter: Nope, I don't think we need it. It just says, the tests of this do not apply if the parents of the newborn child object to the testing. I don't think they need a reason to object. If they are going to do the test they need to inform the parents prior to doing the testing.

Chairman Weisz: Testing requirements do not apply if the parents of the newborn child object to the testing (stops)

Rep. Porter: Period.

Chairman Weisz: Period and then you would add the language, the tests (interrupted).

Rep. Porter: The tests requires informed consent prior to testing.

Chairman Weisz: Ok, that is clear enough.

Rep. Holman: I have a note here that says Hofstad's amendment, but nothing written.

Chairman Weisz: On page 3, line 7, after the first testing there will be a period and then delete the language after that. Overstrike all of that. Then say, the test requires informed consent for prior to testing.

Rep. Schmidt: Do you mean to have parental before consent?

Chairman Weisz: They are the only ones who can give consent.

Rep. Porter: I move we adopt the amendment.

Rep. Schmidt: Second.

Voice Vote: Motion Carried

Rep. Paur: I move a Do Pass as amended.

Rep. Anderson: Second.

VOTE: 12 y 0 n 1 absent – Rep. Hofstad

Bill Carrier: Rep. Paur

Date: 3-15-11
Roll Call Vote # 1

2011 HOUSE STANDING COMMITTEE ROLL CALL VOTES
BILL/RESOLUTION NO. 2067

House HUMAN SERVICES Committee

☐ Check here for Conference Committee

Legislative Council Amendment Number _____

Action Taken: ☐ Do Pass ☐ Do Not Pass ☐ Amended ☒ Adopt Amendment

☐ Rerefer to Appropriations ☐ Reconsider

Motion Made By Rep. Hofstad Seconded By Rep. Conklin

Representatives	Yes	No	Representatives	Yes	No
CHAIRMAN WEISZ			REP. CONKLIN		
VICE-CHAIR PIETSCH			REP. HOLMAN		
REP. ANDERSON			REP. KILICHOWSKI		
REP. DAMSCHEN					
REP. DEVLIN					
REP. HOFSTAD					
REP. LOUSER					
REP. PAUR					
REP. PORTER					
REP. SCHMIDT					

Total (Yes) _____ No _____

Absent _____

Floor Assignment _____

If the vote is on an amendment, briefly indicate intent:

Voice Vote
Motion failed

Date: 3-15-11
Roll Call Vote # 2

2011 HOUSE STANDING COMMITTEE ROLL CALL VOTES
BILL/RESOLUTION NO. 2067

House HUMAN SERVICES Committee

☐ Check here for Conference Committee

Legislative Council Amendment Number _____

Action Taken: ☐ Do Pass ☐ Do Not Pass ☐ Amended ☒ Adopt Amendment

☐ Rerefer to Appropriations ☐ Reconsider

Motion Made By Rep. Devlin Seconded By Rep. Anderson

Representatives	Yes	No	Representatives	Yes	No
CHAIRMAN WEISZ			REP. CONKLIN		
VICE-CHAIR PIETSCH			REP. HOLMAN		
REP. ANDERSON			REP. KILICHOWSKI		
REP. DAMSCHEN					
REP. DEVLIN					
REP. HOFSTAD					
REP. LOUSER					
REP. PAUR					
REP. PORTER					
REP. SCHMIDT					

Total (Yes) _____ No _____

Absent _____

Floor Assignment _____

If the vote is on an amendment, briefly indicate intent:

Voice Vote
Motion Carried

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Title.02000

Adopted by the Human Services Committee

March 21, 2011

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3/21/11

PROPOSED AMENDMENTS TO SENATE BILL NO. 2067

Page 3, line 7, overstrike "on the grounds that testing for metabolic"

Page 3, line 7, remove "or"

Page 3, line 8, remove "genetic"

Page 3, line 8, overstrike "diseases"

Page 3, line 8, remove ", or both,"

Page 3, line 8, overstrike "conflicts with their religious tenets and practices"

Page 3, line 8, after the period insert "The testing requires informed consent before the testing."

Renumber accordingly

Date: 3-24-11
Roll Call Vote # 1

2011 HOUSE STANDING COMMITTEE ROLL CALL VOTES
BILL/RESOLUTION NO. 2067

House HUMAN SERVICES Committee

☐ Check here for Conference Committee

Legislative Council Amendment Number _____

Action Taken: ☐ Do Pass ☐ Do Not Pass ☐ Amended ☒ Adopt Amendment

☐ Rerefer to Appropriations ☐ Reconsider

Motion Made By Rep. PORTER Seconded By Rep. Schmidt

Representatives	Yes	No	Representatives	Yes	No
CHAIRMAN WEISZ			REP. CONKLIN		
VICE-CHAIR PIETSCH			REP. HOLMAN		
REP. ANDERSON			REP. KILICHOWSKI		
REP. DAMSCHEN					
REP. DEVLIN					
REP. HOFSTAD					
REP. LOUSER					
REP. PAUR					
REP. PORTER					
REP. SCHMIDT					

Total (Yes) _____ No _____

Absent _____

Floor Assignment _____

If the vote is on an amendment, briefly indicate intent:

motion carried

Date: 3-24-11
Roll Call Vote # 2

2011 HOUSE STANDING COMMITTEE ROLL CALL VOTES
BILL/RESOLUTION NO. 2067

House HUMAN SERVICES Committee

☐ Check here for Conference Committee

Legislative Council Amendment Number _____

Action Taken: ☒ Do Pass ☐ Do Not Pass ☒ Amended ☐ Adopt Amendment

☐ Rerefer to Appropriations ☐ Reconsider

Motion Made By Rep. Paur Seconded By Rep. Anderson

Representatives	Yes	No	Representatives	Yes	No
CHAIRMAN WEISZ	✓		REP. CONKLIN	✓	
VICE-CHAIR PIETSCH	✓		REP. HOLMAN	✓	
REP. ANDERSON	✓		REP. KILICHOWSKI	✓	
REP. DAMSCHEN	✓				
REP. DEVLIN	✓				
REP. HOFSTAD	A				
REP. LOUSER	✓				
REP. PAUR	✓				
REP. PORTER	✓				
REP. SCHMIDT	✓				

Total (Yes) 12 No 0

Absent 1

Floor Assignment Rep. Paur

If the vote is on an amendment, briefly indicate intent:

REPORT OF STANDING COMMITTEE

SB 2067: Human Services Committee (Rep. Weisz, Chairman) recommends **AMENDMENTS AS FOLLOWS** and when so amended, recommends **DO PASS** (12 YEAS, 0 NAYS, 1 ABSENT AND NOT VOTING). SB 2067 was placed on the Sixth order on the calendar.

Page 3, line 7, overstrike "on the grounds that testing for metabolic"

Page 3, line 7, remove "or"

Page 3, line 8, remove "genetic"

Page 3, line 8, overstrike "diseases"

Page 3, line 8, remove ", or both,"

Page 3, line 8, overstrike "conflicts with their religious tenets and practices"

Page 3, line 8, after the period insert "The testing requires informed consent before the testing."

Renumber accordingly

2011 SENATE HUMAN SERVICES

CONFERENCE COMMITTEE

SB 2067

2011 SENATE STANDING COMMITTEE MINUTES

Senate Human Services Committee
Red River Room, State Capitol

SB 2067
4-8-11
Job Number 16446

☒ Conference Committee

Committee Clerk Signature

Explanation or reason for introduction of bill/resolution:

Relating to newborn disease screening and research regarding metabolic and genetic diseases.

Minutes: Attachments.

Senator Judy Lee brought the conference committee on SB 2067 to order.
All members were present: Senator Judy Lee, Senator Spencer Berry, Senator Tim Mathern, Rep. Chuck Damschen, Rep. Gary Paur, and Rep. Tom Conklin.

Senator Judy Lee asked the House to explain the amendments that were made in the House.

Rep. Damschen explained that their amendment was pretty simple. The intent was that they felt the parents should be informed before the testing. They definitely support the testing.

Senator Judy Lee asked the Department of Health for information that might help the committee understand better.

Barb Schweitzer, Department of Health, provided information for the committee members on what newborn screening is and how it can save lives, and their opinion of the opt in and opt out. Opt in has been tried in several states and they have gone back to the opt out only. She said that she had talked to a colleague in another state who told her that with the opt in provision about 10% of the babies are not getting screened.
She explained forms in the packet provided. Attachment #1 Currently there is a refusal form for parents to fill in and submit to refuse the newborn screening. She gets about 1 or 2 per year.

Senator Judy Lee asked if it is for religious reasons those 1or 2 a year refuse screening.

Ms. Schweitzer said they check the religious tenets but the law does not define what that means.

She also pointed out that they do a lot of education in the facilities – an annual visit to the providers as well as the facilities, parents get a brochure in the prenatal period identifying what newborn screening is and the 40 disorders they are screening for.
She also pointed out that newborn screening saves lives and money.

Rep. Paur asked where she got her number of opting in resulting in about a 10% reduction in testing.

Ms. Schweitzer responded that it was from the colleague she visited with in the state of Nebraska. It is his opinion and experience.

Rep. Paur – The Committee on Bio Ethics of the American Academy of Pediatrics in 2001 said that in a study of newborn screening in Maryland involving informed consent, the majority of women preferred that permission be asked before screening and the informed refusal rate was only 5/1000 infants.

Ms. Schweitzer responded that the state of Maryland used to have the opt in and in 2002 went back to opt out.

Rep. Paur said that currently MN and Texas are looking at the opting in.

Senator Spencer Berry asked if there have been any problems or complaints mentioned to the department as it relates to the current opt out.

Ms. Schweitzer has not heard of any complaints about the way the present system is running.

Rep. Damschen said they hadn't done an in depth study but they don't know if all the parents of newborns are being properly informed. They don't consider a heel stick an invasive process. His doesn't think it would be a problem to at least inform the parent in prenatal visits.

Senator Judy Lee pointed out that they are getting that information.

Rep. Damschen wasn't sure that everyone is getting it, reading it or having it explained.

Senator Judy Lee asked Sen. Berry if he could offer some information as a physician.

Senator Spencer Berry offered information that a prenatal packet is handed out to women at their first visit. It contains a variety of information on things such as newborn screening and breastfeeding. Patients are instructed to please go through the material, read it and, if there are any questions, that would be revisited at subsequent visits.
He went on to explain the importance of newborn screening.
He felt that it is a good system that is in place and there aren't complaints coming in. He has been in communication with representatives of organizations in the state that are worried if the state would go to an opt in versus an opt out that diagnosis, at the very minimum, will be delayed. That would be to a significant detriment to the children.

Senator Tim Mathern was hearing the House agreeing with the Senate about the value of the testing. The concern he was hearing was whether or not people are getting this material in a framework or presence of mind to digest it. He wondered if there was a way to assure that every woman getting prenatal care gets the information and if there was any data on that.

Ms. Schweitzer responded that the facilities and providers do call the health department for brochures on a regular basis. They monitor that. If a facility has not ordered for awhile she calls them to make sure they haven't forgotten. The department distributes about 12,000 brochures a year – births are about 10,000 a year. Each office has a CD available for the parents as well as for the lab techs and nursing staff to explain what newborn screening is.

Senator Judy Lee asked if that information is also available on line.

Ms. Schweitzer said it is on their website.

Senator Tim Mathern asked if this screening is part of the medical training of physicians and people who are generally involved in pre natal care.

Ms. Schweitzer replied that she has gone to the clinics to provide education, specifically the center for rural health for Bismarck and Minot.

Senator Spencer Berry affirmed that, when physicians are taking pediatric courses and are involved in the pediatrics portion of residencies, newborn screening is a very important part of that training.

Rep. Damschen pointed out that the House pretty much agrees with the benefits and would be satisfied in knowing at some point before the test that the parents were presented with the opt out form and told that it was the information about testing for genetic diseases and if they wish to opt out this is the form they need.
The intent is not to prevent the testing but to be certain the parents of the child are informed before the test is done.

Senator Judy Lee asked if he was interested in seeing a packet of information the physician or family practitioner might offer. Her reason being that this is only one thing – she went on to explain other tests and things that need to be done and the trust factor a person has in their licensed health care provider to do those things that need to be done.

Senator Tim Mathern explained what he was hearing – the House is willing to recede from their amendments if the House can recognize a procedure that is being done to assure that parents are really reading this or that it is actually being pointed out to them.

Rep. Damschen agreed that was pretty much an accurate reading – if they had some assurance that the parent is informed before the test. They are not trying to change the procedure.

Senator Spencer Berry responded that informed consent is vital to procedures but he has learned that to try to assure 100% is impossible. He believes that in life with rights come responsibilities. As it relates to having a child if the parents choose not to read the information in the packet they were given and instructed to read they can't be forced.

Rep. Paur stated that the committee in the House had very strong feelings that they assure the parents are notified. He spoke about New Jersey and suggested procedures of informing parents.

Senator Tim Mathern said it seemed to him that they were close to agreement. He thought they could ask the House to recede from their amendments and then take it to the practice level. The Department of Health and the providers are doing what is needed so people understand but it isn't to the level of assurance that the House members want. He didn't think it would need to be put into law because it is a practice issue.

Senator Judy Lee asked if there were any additional items to address.

Rep. Damschen was hesitant to recede from their amendments until they have something that would assure the House that parents are being informed. He couldn't imagine that it would be so difficult to present the opt out form with basic information to the parents.

Senator Judy Lee pointed out that they are getting the brochure and the opt out form now. She asked what wasn't adequate about that.

Rep. Damschen wasn't sure they are being informed of what they are opting out of.

Senator Judy Lee suggested they think about whether it has to be in statute, if it can be in policy, does it need to be in rule, and how much trust and confidence they have in the licensed health care professionals to do what they are obligated to do at this point.

The meeting was adjourned for the day and another meeting will be scheduled.

2011 SENATE STANDING COMMITTEE MINUTES

Senate Human Services Committee
Red River Room, State Capitol

SB 2067
4-11-2011
Job Number 16491

☒ Conference Committee

Committee Clerk Signature DMonson

Explanation or reason for introduction of bill/resolution:

Minutes: Attachment

Senator Judy Lee brought to order the conference committee on SB 2067. All members were present: Senator Judy Lee, Senator Spencer Berry, Senator Tim Mathern, Rep. Chuck Damschen, Rep. Gary Paur, and Rep. Tom Conklin.

Rep. Damschen provided amendments for review - .01002 – and also a copy of the amendment put in context.

Senator Judy Lee commented that with this amendment a reason for objecting to the testing does not need to be given.

Rep. Damschen confirmed that was correct. He went on to report that since the amendment was drafted he had received concerns that the parental informed consent was being taken out. He believed some of the information they were basing that on was incorrect. He didn't think this goes beyond a test and recording numbers. He asked if this records names and tracks if no disease is found.

Senator Judy Lee asked if the Department of Health could answer.

Barb Schweitzer, Department of Health, said that a child's reports that come in within normal limits is kept in security at the University of Iowa lab. A child's report with a presumptive positive disorder is forwarded on to the Health Dept. via the Iowa lab. The Department of Health contacts the local provider with recommendations of what they want done with confirmatory testing or repeat screening. Department of Health keeps registries of those tightly secured.

Senator Tim Mathern asked if it matters, in terms of how this actually works, if it's specific about the reason for objection or just that they object.

Ms. Schweitzer replied that at this time they opt out for religious practices but they do not have to document what their religious practices are. As long as there is an opt out it would be appropriate.

The actual practice and how this is implemented would be about the same whether they say religious tenet or don't give a reason. They can still opt out.

Senator Judy Lee asked Ms. Schweitzer if she saw anything about the proposed amendments that would cause concern as far as making sure the children get tested.

Ms. Schweitzer said her only concern was that on an annual basis they have provided a lot of education to providers and facilities and written information to parents about the nature of the proposed testing.

Senator Judy Lee reported that in her discussion with Rep. Weisz earlier he seemed comfortable with the information in the brochure and that it would be in law so it would be recognized by health care providers that they need to provide that information. She wanted to know if that was what the House Human Services Committee position would be.

Rep. Damschen said that would address their main concern. He also shared information from a lady who recently had a baby and said she did not have any information presented to her prior to the screening.

Rep. Paur had contacted several states and had a lot of background from the Academy of Pediatrics. All the Associations very strongly recommend informing the parents. Most recommend educating the parents. It is important to screen because if it comes back positive the turnaround for getting testing done is a lot shorter if the parents are aware and educated about the testing ahead of time.

Senator Judy Lee agreed and pointed out that a strong effort had been made by the health department to do it and those tending the moms are generally giving them a packet of information.

Senator Tim Mathern said it appeared to him that the amendments proposed by Rep. Damschen address the concerns and continue the process of an opt out procedure and puts in law what they believe is being done in terms of informing people but actually makes it part of the law.

Senator Judy Lee asked if the people from the health department feel this will strengthen their program and not create roadblocks.

Ms. Schweitzer said they can abide by this.

Rep. Damschen moved that the **House recede from their amendments and further amend** as contained in the version .01002.

Seconded by **Senator Tim Mathern.**

Clarification that the only thing that changes is in section 25-17-04.

Roll call vote 6-0-0. **Motion carried.**

Senate carrier is **Senator Judy Lee**.

House carrier is **Rep. Chuck Damschen**

11.8061.01002
Title.03000

Prepared by the Legislative Council staff for
Representative Damschen
April 8, 2011

4-11-11

PROPOSED AMENDMENTS TO SENATE BILL NO. 2067

That the House recede from its amendments as printed on page 900 of the Senate Journal and pages 1071 and 1072 of the House Journal and that Senate Bill No. 2067 be amended as follows:

Page 3, line 3, after "shall" insert "provide the parents with written information regarding the nature of the proposed testing and then"

Page 3, line 7, overstrike "on the grounds that testing for metabolic"

Page 3, line 7, remove "or"

Page 3, line 8, remove "genetic"

Page 3, line 8, overstrike "diseases"

Page 3, line 8, remove ", or both,"

Page 3, line 8, overstrike "conflicts with their religious tenets and practices"

Renumber accordingly

2011 SENATE CONFERENCE COMMITTEE ROLL CALL VOTES

Committee: Senate Human Services

Bill/Resolution No. 2067 as (re) engrossed

Date: 4-11-11

Roll Call Vote #: 1

Action Taken

- ☐ SENATE accede to House amendments
- ☐ SENATE accede to House amendments and further amend
- ☐ HOUSE recede from House amendments
- ☒ HOUSE recede from House amendments and amend as follows

Senate/House Amendments on SJ/HJ page(s) 900 --

- ☐ Unable to agree, recommends that the committee be discharged and a new committee be appointed

((Re) Engrossed) 2067 was placed on the Seventh order of business on the calendar

Motion Made by: Damschen Seconded by: Mathern

Senators	4-8	4-11		Yes	No	Representatives	4-8	4-11		Yes	No
Sen. Judy Lee	✓	✓		✓		Rep. Damschen	✓	✓		✓	
Sen. Berry	✓	✓		✓		Rep. Paur	✓	✓		✓	
Sen. Mathern	✓	✓		✓		Rep. Conklin	✓	✓		✓	

Vote Count: Yes 6 No 0 Absent 0

Senate Carrier Sen. J. Lee House Carrier Rep. Damschen

LC Number _____ . _____ of amendment

LC Number _____ . _____ of engrossment

Emergency clause added or deleted

Statement of purpose of amendment

REPORT OF CONFERENCE COMMITTEE

SB 2067: Your conference committee (Sens. J. Lee, Berry, Mathern and Reps. Damschen, Paur, Conklin) recommends that the **HOUSE RECEDE** from the House amendments as printed on SJ page 900, adopt amendments as follows, and place SB 2067 on the Seventh order:

That the House recede from its amendments as printed on page 900 of the Senate Journal and pages 1071 and 1072 of the House Journal and that Senate Bill No. 2067 be amended as follows:

Page 3, line 3, after "shall" insert "provide the parents with written information regarding the nature of the proposed testing and then"

Page 3, line 7, overstrike "on the grounds that testing for metabolic"

Page 3, line 7, remove "or"

Page 3, line 8, remove "genetic"

Page 3, line 8, overstrike "diseases"

Page 3, line 8, remove ", or both,"

Page 3, line 8, overstrike "conflicts with their religious tenets and practices"

Renumber accordingly

SB 2067 was placed on the Seventh order of business on the calendar.

2011 TESTIMONY

SB 2067

#1

Testimony
Senate Bill 2067
Human Services Committee
Wednesday, January 12, 2011; 11:00 a.m.
North Department of Health

Good morning, Madam Chair and members of the Senate Human Services committee. My name is Kim Mertz and I am the director of the Division of Family Health for the North Dakota Department of Health. I am here today to provide testimony in support of Senate Bill 2067, which amends NDCC 23-01-03.1 to include the words "and genetic" when referring to newborn metabolic disease screening tests.

Newborn screening is the practice of testing every newborn for certain harmful or potentially fatal disorders that aren't otherwise apparent at birth. With a simple blood test, doctors often can tell whether newborns have certain conditions that could eventually cause health problems. Even though these conditions are considered rare, early diagnosis and proper treatment can mean the difference between lifelong impairment and healthy development. Due to medical advancements, it will soon be common to screen infants for both metabolic and genetic disorders.

Metabolic disease occurs when abnormal chemical reactions in the body disrupt the process of breaking the food parts into sugars and acids, your body's fuel. Genetic diseases are caused by an abnormality in a person's genes.

The North Dakota Newborn Screening program started in 1964 screening for Pheylketonuria (PKU), a metabolic disorder. Since the development of the PKU test, researchers have developed blood tests that can screen newborns for additional disorders beyond metabolic diseases which, unless detected and treated early, can cause physical problems, developmental delay, and in some cases, even death. Today, North Dakota screens for 40 different disorders.

The term "metabolic" has been used nationally and in North Dakota for many years to describe newborn screening programs. Advances in research and technology have, and will no doubt continue to, identify additional disorders beyond metabolic that are recommended for testing. At the present time, the language in the law does not have the correct terminology for current or future testing. This change does not affect the process needed for adding new diseases to

be tested for, as new testing must be approved through the administrative rulemaking process.

This concludes my testimony. I am happy to answer any questions you may have.

#2

Wednesday, January 12, 2011

SB 2067: relating to newborn disease screening and research regarding metabolic and genetic diseases.

To: Senator J. Lee and members of the senate human services committee

My name is JoAnn Brager and I am the Vice President of Public Policy for the North Dakota Association for the Education of Young Children. NDAEYC represents approximately 400 members who work with or on behalf of children ages birth to eight years of age.

NDAEYC strongly supports the addition of genetic diseases to the North Dakota Century code. We know that early identification and timely intervention will lead to significant reduction in mortality, morbidity and associated disabilities. Those of us who work with young children address the whole child's well being as well as the needs and concerns of the people who care for our youngest North Dakotans. This includes parents, health care workers, teachers and the general public.

Thank you for your time today and I am happy to answer any questions you may have.

From: Jennifer Restemayer [mailto:jennmarie@bis.midco.net]
Sent: Wednesday, January 12, 2011 7:37 AM
To: Lee, Judy E.
Subject: SB 2067 ~ Sorry, I forgot the picture the first time :)

Senator Lee,

I am writing to you today in regard to Senate Bill 2067. I would like to let you know that I fully support the changes to the North
Dakota Century Code to include Genetic Diseases.

I am the mother of a child with a progressive and degenerative genetic disorder. My daughter Allison was diagnosed with
Mucopolysaccharidosis (MPS) type 1 just before her second birthday. I will never forget sitting in the Doctors office and being
handed a photocopy of the Physicians Desk Reference page on Hurler Syndrome (or MPS 1) and reading that she would be
bedridden by the age of 8 and dead by the age of 10.

We were fortunate that a treatment for her disorder was FDA approved shortly after Allison's diagnosis, and we were able to
start that treatment within weeks of approval. Allison is affected by her disorder, the damage that was done before she was able
to start treatment will never be undone, however, she has a much better quality of life and many complications of her disease
that have not yet touched her because she was able to start treatment at age 2. Allison will celebrate her 10th birthday on Feb 5, 2011.
I can only imagine the possibilities for the babies able to start treatment shortly after birth.

It is so important for us to remember that the earlier children with genetic and metabolic disorders are diagnosed and can start treatment,
the better their quality of life will be. Rare diseases and genetic disorders are becoming less and less rare, so unfortunately there will be
more children affected by them. Our children in the state of North Dakota deserve every chance we can give them to grow up healthy
and strong. With the changes to the North Dakota Century Code, we can ensure that as newborn screening tools become available for
more genetic disorders, we can request that our wonderful state start to screen for them. We can give this population of children the
best chance for a great life!

Thank you for your time.

Jennifer Restemayer
2217 E Capitol Ave
Bismarck, ND
(701) 471-8714

Testimony
Senate Bill 2067
House Human Services Committee
Wednesday, March 9, 2011; 9 a.m.
North Dakota Department of Health

#1

Good morning, Chairman Weisz and members of the House Human Services committee. My name is Barb Schweitzer and I am the director of the Newborn Screening Program for the North Dakota Department of Health. I am here today to provide testimony in support of Senate Bill 2067, which amends NDCC 23-01-03.1 to include the words "and genetic" when referring to newborn metabolic disease screening tests.

Newborn screening is the practice of testing every newborn for certain harmful or potentially fatal disorders that aren't otherwise apparent at birth. With a simple blood test, doctors often can tell whether newborns have certain conditions that could eventually cause health problems. Even though these conditions are considered rare, early diagnosis and proper treatment can mean the difference between lifelong impairment and healthy development. Due to medical advancements, it will soon be common to screen infants for both metabolic and genetic disorders.

Metabolic disease occurs when abnormal chemical reactions in the body disrupt the process of breaking the food parts into sugars and acids, your body's fuel. Genetic diseases are caused by an abnormality in a person's genes.

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The term "metabolic" has been used nationally and in North Dakota for many years to describe newborn screening programs. Advances in research and technology have, and will no doubt continue to, identify additional disorders beyond metabolic that are recommended for testing. At the present time, the language in the law does not have the correct terminology for current or future testing. This change does not affect the process needed for adding new diseases to be tested for, as new testing must be approved through the administrative rulemaking process.

This concludes my testimony. I am happy to answer any questions you may have.

Beth Nodland
905 N. Anderson Street
Bismarck, ND 58501
(701) 223-6306

Chairman Weisz and Committee Members, thank you for the opportunity to present testimony regarding **SB2067**. I appear before you as the parent of a healthy, active, almost three year old son, who happens to have a genetic condition, Down syndrome. I am also a member of the statewide network of parents of and individuals with Down syndrome, called Designer Genes. Three of our Board Officers recently returned from a national conference in Texas, where the issue of genetic registries got a bit of attention. This is going to be an issue to which we will start paying attention. Our group's President, Roxane Romanick and I have met with the State Department of Health to address my concerns and have begun to try to understand the Administrative Rules and programs already in place. I have agreed to volunteer to help address this with the State Department of Health. But I wanted to speak today, to ask for some consideration now.

Points of Concern with SB2~~076~~ 67

1. It is unclear what the point of the "research" is, (understanding that none as happened in ND to date,) and more importantly who will have access to potentially aggregated data. Is this "point-in-time" data or "longitudinal studies?" I have great concern with the State starting to track my child over time and aggregate his information. Is there data access by students, pharmaceutical, and insurance companies?

2. Ability for parents and individuals with genetic conditions to opt in or out. Currently the early screening heal prick happens with no informed consent or signed form by parents, the State "assumes consent." Parents and individuals with genetic conditions should have the informed ability to opt in or out of the genetic registry and research: 1) before the heal prick, 2) after the diagnosis, and 3) at any point during the course of life, for example when a person with a genetic condition reaches adulthood.

3. There is an important educational element, including outreach to doctors, nurses, and citizens. I think we all share the understanding of the importance of newborn screening, so I don't understand why there is no funding for either the newborn screening program nor for the educational outreach part?

4. I'm also concerned with the purpose of the genetic registry: the legislation simply says: "Maintain a registry of cases of metabolic and genetic diseases." I'd like the purpose defined.

5. As it stands now, the process of opting out is unclear. The language at the end of the bill, (and in the current code says that "parents of a newborn child [can] object to the testing on the grounds that testing for metabolic or genetic diseases, or both, conflicts with their religious tenets and practices." I ask that a "period" be placed after the first word "testing" or that the justification be expanded to include that parents can object because it conflicts with their ethical, or scientific tenets and practices, or simply because of their privacy wishes, tenets and practices.

Thank you for this opportunity.

March of Dimes Foundation
North Dakota Chapter

1330 Page Dr., Suite 102
Fargo, ND 58103
Telephone (701) 235-5530
1(800) 393-4637
Fax (701) 235-8725
Email:
ND407@marchofdimes.com

Karin Roseland
State Director

April 7, 2011

Re: SB 2067

Dear Conferees for SB 2067,

The mission of the March of Dimes is to improve maternal and child health by preventing birth defects, premature birth, and infant mortality.

As part of our work we have advocated for comprehensive and quality newborn screening (NBS) programs across the country. The March of Dimes' overriding interest is to safeguard the benefit to newborns, of early detection of life threatening conditions and initiation of treatment that is made possible by NBS. Early identification of these conditions is crucial, as timely intervention can lead to a significant reduction or morbidity, mortality and associated disabilities in affected infants.

The March of Dimes requests that you restore the original language in SB 2067, page 3, lines 7-8. The bill in its current amended form put infants at risk of death and disability.

Thank you for your attention to this matter.

On behalf of children in North Dakota,

Karin Roseland, State Director

march of dimes

NEWBORN SCREENING

Background

Newborn screening (NBS) is a public health program that provides early identification and follow-up for treatment of infants affected by certain genetic, metabolic, hormonal and/or functional conditions. Since the mid-1960s, the success of NBS programs has made screening routine for the over four million infants born in the United States each year and screened for congenital disorders. The CDC estimates that over 6,000 newborns[1] are diagnosed as having a treatable metabolic condition annually and another 12,000 a hearing impairment that requires follow up each year.[2] Except for hearing, screening tests are done using a few drops of blood from the newborn's heel, usually taken in the hospital 24 to 48 hours after birth.

Currently, each state or territory operates by law its own NBS program. Therefore, although all state programs now require screening for at least 26 conditions, individual programs vary in the number and types of conditions for which newborns are screened.[3] In 2000, the March of Dimes led the way in proposing a national standard for NBS which included a core list of 9 disorders, with provision for expanding the list as the science and technology evolved.

Recent advances in treatments and technology, such as tandem mass spectrometry, enable screening for many different disorders. In August 2004 the American College of Medical Genetics (ACMG) submitted a report to the federal Health Resources and Services Administration (HRSA), which included proposed nationwide standards for state NBS programs. The report listed 29 core treatable disorders that should be targeted by NBS programs and an additional 25 secondary target conditions for which test results should be reported.[4] These secondary target disorders are not actively sought by NBS, because they do not yet have documented treatments or there is limited knowledge of their natural history, but they are revealed in the course of screening for the core conditions. This recommendation was endorsed by the Secretary's Advisory Committee on Heritable Disorders in Newborns and Children (SACHDNC) as well as the March of Dimes. In 2010, another disorder, severe combined immunodeficiency (SCID) was added to SACHDNC's Recommended Uniform Screening Panel, making the new total 30 core conditions. Also in 2010, the Secretary of Health and Human Services adopted the panel of 30 disorders as the national standard for newborn screening as recommended by SACHDNC.[5]

NBS program practices vary by state and territory regarding retention of residual blood spots after newborn screening tests are completed, raising questions about both potential uses of the samples, and requirements for informed consent. Most NBS programs routinely use post screening residual samples for the purpose of laboratory quality assurance, i.e. comparing results of the tests from the screening laboratory with those of the reference laboratory, and for the development of new screening methods. In some states, these samples are also used for population based epidemiologic research and clinical studies.

March of Dimes Policy

NBS programs should offer comprehensive services, including high quality screening with state-of-the-art technology, trained personnel, and resources for timely follow-up and program evaluation. Moreover, state programs should ensure that every newborn is screened at birth for conditions/disorders that meet all of the following public health criteria: 1) There is documented medical benefit to the affected infant from early detection, and treatment; 2) There is a reliable screening test for the disorder; and 3) Early detection can be made from newborn blood spots, or other specific means (see separate Field Advisory on newborn hearing screening).[6] The core panel of 30 disorders (see attached appendix) meets these criteria and incorporates all of the conditions named in the Foundation's previous policy.

The March of Dimes also supports education of parents about newborn screening during the prenatal period as well as expansion of education programs for health professionals. Education should include information on treatable conditions as well as testing for disorders for which treatment is not yet available and on state policies for the storage, retention and use of blood spots. In addition to counseling by health professionals during prenatal visits, information about testing and associated costs can be obtained from multiple sources, including, genetic counselors, the American College of Medical Genetics, and the U.S. Department of Health and Human Services Web site which is operated under contract by the University of Texas Health Science Center (genes-r-us.uthscsa.edu).

The March of Dimes recognizes both the value of these samples for research uses by the scientific community and the importance of appropriate parental involvement in decisions regarding storage and

use of post-screening residual samples. The Foundation's overriding interest is to safeguard the benefit to newborns, of early detection of life threatening conditions and initiation of treatment that is made possible by NBS. The March of Dimes encourages policy makers to ensure that state practices for post screening storage and use of residual blood spots work in concert with—and do not jeopardize—longstanding and highly effective newborn screening public health programs that now operate in all states.

Regarding the issue of parental involvement in the approaches to use and storage of residual samples, March of Dimes recommends the following: 1) Research using residual specimens should be undertaken only after review and approval by an Institutional Review Board whose policies are consistent with federal and state regulations concerning informed consent; 2) If some form of parental consent such as the opportunity to opt out relating to storage and use becomes or is required by law or regulation, it should be obtained only after the newborn has been screened; 3) Where a newborn screening program would otherwise be jeopardized, March of Dimes will not object to offering parents the opportunity to "opt out" of storage and use of residual samples.

March of Dimes Practice

March of Dimes grantees developed the first screening tests for phenylketonuria, biotinidase deficiency and congenital adrenal hyperplasia, and contributed to development of screening for hypothyroidism. Since the 1970s, March of Dimes state chapters and volunteers have worked closely with governors, state legislators, health departments, and other organizations to expand and improve state NBS programs. In addition, the March of Dimes has been instrumental in developing legislation and obtaining federal resources to support state and national NBS activities. March of Dimes representatives participated in the ACMG project to develop recommendations for NBS standards and a March of Dimes representative serves as a liaison member of the U.S. Dept. of Health and Human Services SACHDNC.

In deliberations over proposals to modify the scope of state screening panels, the March of Dimes will support the addition of conditions only when they meet all three public health criteria described in the opening paragraph of the Policy section above. Currently, the March of Dimes advocates in support of screening newborns for at least the 30 core disorders as recommended by the Secretary and to ensure reporting of the additional secondary target conditions which are revealed in the course of screening for the core disorders. The March of Dimes also urges states to provide comprehensive educational information to parents, including guidance on testing for disorders where treatment is not yet available. In deliberations over proposals to modify state screening programs by adding provisions for storage and use of residual blood spots, March of Dimes will support appropriate parental involvement for all post screening uses, as recommended above. In addition, the Foundation supports providing health professionals with information about the availability and benefits of NBS and encourages professional societies and government agencies to work toward strengthening education of clinicians and other health care providers about newborn screening.

The March of Dimes also supports ongoing improvement of policies, standards, procedures and quality assurance systems for state programs and urges public and private entities to work together to strengthen such programs so that they include research, validation of methods to detect and treat disorders, and provide follow-up and counseling for affected families.

References

[1] Centers for Disease Control and Prevention (CDC). Impact of expanded newborn screening--United States, 2006. MMWR Morb Mortal Wkly Rep. 2008;57:1012-5.

[2] National Center for Hearing Assessment & Management, Early Hearing Detection and Intervention/Universal Newborn Hearing Screening. Fact Sheet, http://www.infanthearing.org/screening/nhsresources.html.

[3] National Newborn Screening and Genetics Resource Center. National Newborn Screening Status Report. http://genes-r-us.uthscsa.edu/nbsdisorders.pdf.

[4] American College of Medical Genetics. Newborn Screening: Towards a Uniform Screening Panel and System. March 8, 2005. http://mchb.hrsa.gov/screening/.

[5] Letter from the Secretary of Health and Human Services to the SACHDNC. May 21, 2010. Available at: http://www.hrsa.gov/heritabledisorderscommittee/correspondence

[6] March of Dimes. Newborn Hearing Screening. White Plains, NY, 1999.

NEWBORN SCREENING FIELD ADVISORY

APPENDIX
National Standard for Newborn Screening
Recommended Panel

30 conditions, organized into 5 categories.

MS/MS = tandem mass spectrometry.

Organic Acid Metabolism Disorders – test by MS/MS
IVA: Isovaleric acidemia
GA-I: Glutaric acidemia type I
HMG: Hydroxymethylglutaric aciduria, also called 3-OH 3-CH3 glutaric aciduria
MCD: Multiple carboxylase deficiency
MUT: Methylmalonic acidemia, mutase deficiency form
3MCC: 3-Methylcrotonyl-CoA carboxylase deficiency
Cbl A,B: Methylmalonic acidemia, Cbl A and Cbl B forms
PROP: Propionic acidemia
BKT: Beta-ketothiolase deficiency

Fatty Acid Oxidation Disorders – test by MS/MS
MCAD: Medium-chain acyl-CoA dehydrogenase deficiency
VLCAD: Very-long-chain acyl-CoA dehydrogenase deficiency
LCHAD: Long-chain hydroxyacyl-CoA dehydrogenase deficiency
TFP: Trifunctional protein deficiency
CUD: Carnitine uptake defect

Amino Acid Metabolism Disorders – test by MS/MS
PKU: Phenylketonuria
MSUD: Maple syrup urine disease
HCY: Homocystinuria
CIT: Citrullinemia
ASA: Argininosuccinic acidemia
TYR I: Tyrosinemia type I

Hemoglobinopathies (Hemoglobin Disorders)
SCA: Sickle cell anemia
Hb S/Th : Hb S/Beta-thalassemia
Hb S/C: Hb S/C disease

Others
HYPOTH: Congenital hypothyroidism
BIOT: Biotinidase deficiency
CAH: Congenital adrenal hyperplasia
GALT: Galactosemia
HEAR: Hearing deficiency
CF: Cystic fibrosis
SCID: Severe combined immunodeficiency

The North Dakota Newborn Screening Program in-services are designed to educate healthcare facilities and healthcare professionals regarding the rapid changes with newborn screening.

Newborn Screening Guidelines

Purpose

To provide education to healthcare staff regarding newborn screening process.

WHERE?

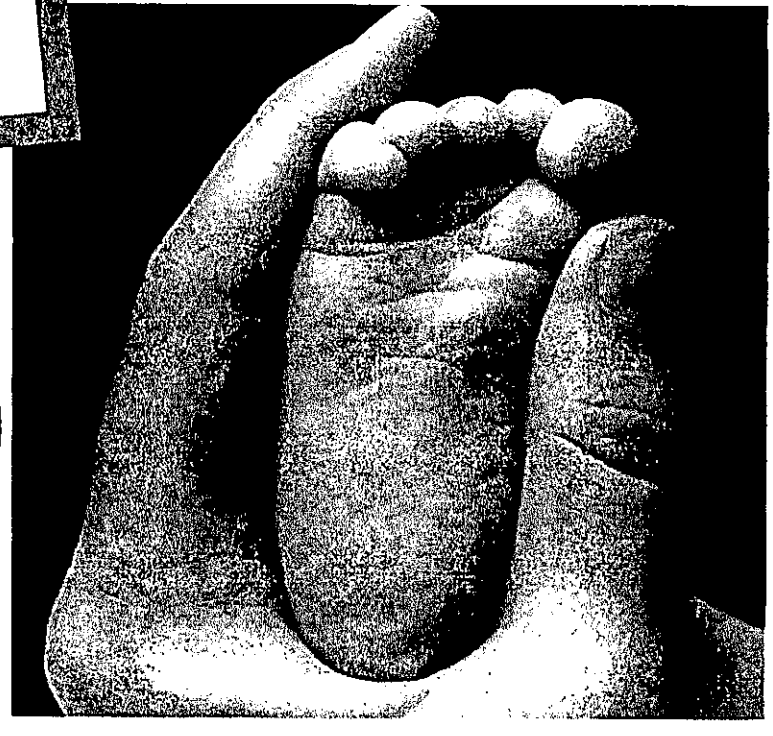
WHEN?

WHO?

NORTH DAKOTA DEPARTMENT of HEALTH

NEWBORN SCREENING STAFF

BARB SCHWEITZER, RN

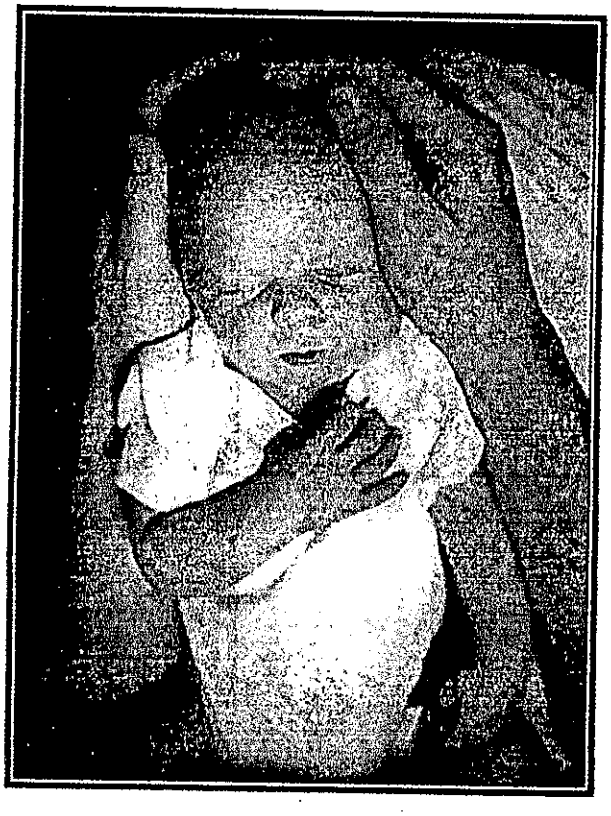


Earn 1 Contact Hour

Contact hours will be awarded. Community Health Section, North Dakota Department of Health is an approved provider of continuing nursing education by CNE-Net, the education division of the North Dakota Nurses Association, an accredited approver by the American Nurses Credentialing Center's Commission on Accreditation.

NORTH DAKOTA DEPARTMENT OF HEALTH

NEWBORN SCREENING PROGRAM



GUIDELINES FOR HEALTH-CARE PROVIDERS

NORTH DAKOTA
DEPARTMENT *of* HEALTH

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Appendices

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North Dakota Newborn Screening Program
Refusal of Newborn Screening Tests

Infant's Name: _____ Medical Record # _____

Date of Birth: _____

Attending Physician or Practitioner: _____

Place of Birth: _____

I have received and read the parent informational brochure entitled "Screening for a Healthy Baby," describing newborn screening in North Dakota. I understand that these disorders are easily detected by testing a blood sample from my baby's heel.

I have been informed and I understand that it is the Law of the State of North Dakota that all newborns shall be screened for these disorders, unless the parents/guardians refuse "on the grounds that testing for metabolic diseases conflicts with their religious tenets and practices".

I have been informed and I understand that, if untreated, these conditions may cause permanent damage to my child, including serious mental retardation, growth failure, and even death.

I have discussed this screening with _____ and I understand the risks to my child if this screening is not completed.
(Physician or practitioner)

I do not want to have _____ screened for these disorders.
(Name of Infant)

My decision was made freely and I accept the legal responsibility for the consequences of this decision.

_____ _____
Parent or legal guardian printed name Date

_____ _____
Parent or legal guardian signature Date

_____ _____
Witness signature Date

Original: Infant's medical record
Copy: North Dakota Newborn Screening Office
Family Health Division, ND Department of Health
600 E. Boulevard Ave., Dept 301
Bismarck, ND 58505-0200
Copy: Parent/Guardian

10/24/06

Newborn Screening Fact Sheet

Index

25. CH (Congenital Hypothyroidism)
26. Galactosemia
27. Hemoglobin E Disease
28. MSUD (Maple Syrup Urine Disease)
29. PKU (Pheylketonuria)
30. Sickle Cell Disease
31. Cystic Fibrosis

History of North Dakota Newborn Screening

1964
Screening for Pheylketonuria began

1977
Screening for Hypothyroidism began

1991
Screening for Galactosemia began

1992
North Dakota lab was no longer able to do the newborn screening lab tests. North Dakota entered into an agreement with the Iowa Metabolic Screening Program for the lab testing

1992
Screening began for Congenital Adrenal Hyperplasia (CAH) and Maple Syrup Urine Disease (MSUD)

November 2002
Participated in pilot project with the Iowa lab for testing of an expanded panel of disorders detectable by tandem mass spectrometry

April 2003
Screening for MCAD, Biotinidase, Sickle Cell and other hemoglobinopathies began

August 2004
Screening for disorders detected by tandem mass spectrometry were officially part of newborn screening

January 2006
Newborn Screening for Cystic Fibrosis started

June 1, 2008
Implemented private courier service

Newborn Screening Saves Lives and Money
Barb Schweitzer

- Universal newborn screening for phenylketonuria (PKU) was first introduced in the 1960s for the purpose of preventing mental retardation, or intellectual disability, by allowing for timely introduction of a special diet to prevent brain damage. At the time, it was calculated that the economic benefits to states from the reduced costs of lifetime care for individuals with intellectual disability greatly exceeded the cost of screening, not even taking into account the tremendous benefits in terms of health and realized human potential.

- In the 1980s, states began screening all babies for congenital hypothyroidism (CH) for the same reason. Screening for other disorders, such as galactosemia, congenital adrenal hyperplasia, and sickle cell disease, was introduced in order to save lives through prompt diagnosis and treatment.

- More than 4 million babies are born each year in the United States. Medical professionals estimate that thousands of babies will have a disorder detectable through universal comprehensive newborn screening. Even though every baby born in the United States receives a newborn screen, the number of diseases screened for is determined by the state in which that baby is born. Without proper screening, affected babies will likely suffer mental retardation, physical disability or even death. Most affected babies can lead normal, healthy lives when diagnosed early and started on treatment shortly after birth.

- Since 1964, North Dakota law requires all babies to be screened at birth. Today, North Dakota screens for more than 40 diseases and disorders.

- CDC published an estimate in 2004 that each child born with an intellectual disability incurs an average lifetime cost of roughly $1 million. That was in 2003 dollars and is even more costly in today's dollars. That figure includes both direct (medical, educational and developmental services) and indirect (lost productivity) costs. It does not include the costs to families of caring for a family member with a disability, in particular the lost earning potential of family members who provide unpaid care. Nor does it include the costs of long-term residential care for those whose families are not able to care for them in their own homes. Therefore, the $1 million per child estimate should be regarded as a lower bound estimate.

- North Dakota's newborn screening charge is $60.00 per screen. South Dakota's charge is $60.00, Minnesota is $101.00 and Montana is $91.70.

North Dakota Newborn Screening Program
List of Metabolic Disorders

AMINO ACIDEMIAS AND UREA CYCLE DISORDERS (AA)

(ASA) Argininosuccinate acidemia
(CIT 1) Citrullinemia or ASA Synthetase Deficiency
(HCY) Homocystinuria (cystathionine beta synthetase)
(MSUD) Maple Syrup Urine Disease
(PKU) Phenylketonuria
(TYR-1) Tyrosinemia Type 1
(ARG) Arginiemia
(BIOPT-BS) Defects of biopterin cofactor biosynthesis
(CIT-II) Citrullinemia type II
(BIOPT-RG) Defects of biopterin cofactor regeneration
(H-PHE) Benign hyperphenylalaninemia
(MET) Hypermethioninemia
(TYR II) Tyrosinemia type II
(TRY III) Tyrosinemia type III

ORGANIC ACIDEMIAS (OA)

(GA-1) Glutaric acidemia type 1
(HMG) 3-Hydroxy 3-methylglutaric aciduria
(IVA) Isovaleric acidemia
(3-MCC) 3-Methylcrotonyl-CoA carboxylase
(Cbl-A,B) Methylmalonic acidemia (vitamin B12 disorders)
(BKT) Beta Ketothiolase
(MUT) Methylmalonic Acidemia (methylmalonyl-CoA mutase)
(PROP) Propionic acidemia
(MCD) Multiple carboxylase
(2M3HBA) 2-Methyl-3-hydroxybutyric aciduria
(2MGB) 2-Methylbutyrl-CoA dehydrogenase
(3MGA) 3-Methylglutaconic aciduria
(Cbl-C, D) Methylmalonic acidemia
(IBG) Isobutyrl-CoA dehydrogenase
(MAL) Malonic acidemia

FATTY ACID OXIDATION DISORDERS (FAO)

(CUD) Carnitine uptake defect (Carnitine transport defect)
(LCHAD) Long-chain L-3 hydroxyacyl-CoA dehydrogenase
(MCAD) Medium chain acyl-CoA dehydrogenase
(TRP) Trifunctional protein deficiency
(VLCAD) Very long-chain acyl-CoA dehydrogenase
(CACT) Carnitine acylcarnitine translocase
(CPT-Ia) Carnitine palmitoyltransferase I
(CPT-II) Carnitine palmitoyltransferase II
(GA-II) Glutaric acidemia Type II
(MCKAT) Medium-chain ketoacyl-CoA thiolase
(M/SCHAD) Medium/Short chain L-3-hydroxy acyl-CoA dehydrogenase
(SCAD) Short-chain acyl-CoA dehydrogenase

North Dakota Newborn Screening Program

METABOLIC

Biotinidase deficiency
Congenital adrenal hyperplasia (CAH)
Congenital hypothyroidism (CH)
Galactosemia

GENETIC

Cystic Fibrosis
Sickle cell disease and other hemoglobin disorders

North Dakota Newb[redacted]creening Numbers

Year	91	92	93	94	95	96	97	98	99	00	01	02	03	04	05	06	07	08	09	10	11	12	13	14	GT
Disorder																									
Biotinidase	--	--	--	--	--	--	--	--	--	--	--	--	1	--	--	3	1	1	2	1					9
CAH	--	2	--	1	1	--	1	1	1	1	1	1	--	1	3	--	0	4	1	1					20
Galactosemia	--	2	1	1	--	--	1	3	7	8	4	3	6	2	3	2	3	4	3	1					54
MCAD	--	--	--	--	--	--	--	--	--	--	--	--	1	2	1	--	1	0	0	1					6
LCHAD	--	--	--	--	--	--	--	--	--	--	1	--	--	1	--	--	0	1	0	0					3
SCAD	--	--	--	--	--	--	--	--	--	--	--	--	--	1	1	--	0	0	2	0					4
3MCC																		1	0	1					1
VLCAD																	1	2	1						2
MSUD	1	--	--	--	--	--	--	--	--	--	--	--	--	--	--	--	0	0	0						1
CPT-1	--	--	--	--	--	--	--	--	--	--	--	--	--	--	--	1	0	0	0	1					2
CUD	--	--	--	--	--	--	--	--	--	--	--	--	--	--	--	1	0	0	0						1
CPT 2																			1	1					1
TYR 2																			1	0					1
Isovaleric Acidemia	--	--	--	--	--	--	--	--	--	--	--	--	--	--	--	1	0	0	0	0					1
PKU	1	1	1	1	2	2	1	2	--	--	1	2	1	--	--	--	2	0	0	3					20
TSH (CH)	2	2	3	7	--	6	2	4	2	3	5	6	4	1	4	9	6	6	7	6					85
Cystic Fibrosis	--	--	--	--	--	--	--	--	--	--	--	--	--	--	--	4	1	3	2	2					12
Sickle Cell	--	--	--	--	--	--	--	--	--	--	--	--	--	--	--	--	0	1	0						1
Hemoglobin:																									
FF	--	--	--	--	--	--	--	--	--	--	--	--	--	1	--	--	--	0	0						1
FAE	--	--	--	--	--	--	--	--	--	--	--	--	1	5	3	--	3	6	2						20
FAD	--	--	--	--	--	--	--	--	--	--	--	--	1	1	1	1	1	2	0						7
FAC	--	--	--	--	--	--	--	--	--	--	--	--	7	4	6	4	6	7	10						44
S Beta thal.																			1	2					1
FAV	--	--	--	--	--	--	--	--	--	--	--	--	12	10	10	6	20	11	11						80
FAS	--	--	--	--	--	--	--	--	--	--	--	--	8	18	9	7	16	20	24						102
FA + Barts	--	--	--	--	--	--	--	--	--	--	--	--	1	4	3	7	3	8	9						35
Total													30	43	32	25	48	50	57						235

Year	62	63	65	66	68	69	70	72	73	74	75	76	77	78	80	81	82	83	84	85	86	87	88	89	90
Disorder																									
PKU	1	1	1	1	2	4	1	1	1	--	1	4	2	1	3	2	--	1	1	1	2	1	2	--	--
TSH (CH)	--	--	--	--	--	--	--	--	--	--	--	--	--	--	1	1	2	--	1	1	3	2	4	2	3

* GT = Grand Total

4/7/2011

NEWBORN SCREENING EDUCATION IN NORTH DAKOTA

In complying with the North Dakota statue of Chapter 25-17-01 in part "the state department of health shall: Develop and implement a metabolic disease educational program.....This educational program must include information about the diseases so that proper measures may be taken to reduce mortality, morbidity, and associated disabilities."
As a result of this directive, ongoing educational efforts are a priority.

1. Developed and distributed "Guidelines for Health Care Providers" to all health care birthing facilities and clinics. The guidelines include the state law, rules, as well as individual comprehensive protocols for the screening disorders.
2. Distributed Clinical and lab standards institute (CLSI) guidelines for newborn screening collection that includes DVR educational tool for staff use training as well as procedures for blood collection and newborn screening follow up.
3. Developed and distributed disorder specific "Parent Fact Sheets" to health care facilities and health care providers.
4. Distributed CD's "A Parent's guide to Newborn Screening" to health care providers.
5. Update newborn screening website which includes the North Dakota laws, disorders for which babies are screened, parent fact sheets, the screening for a healthy baby brochure.
6. The North Dakota Newborn Screening Director has conducted educational site visits to healthcare facilities and health care providers every year since 2002.
7. Additional education is provided to facilities/providers as requested and needed.
8. Dr. Sara Copeland, Iowa's metabolic consultant to North Dakota presented to the North Dakota American Academy of Pediatrics annual meetings in April 2005, April 2007, and September 2008.

Metabolic Screening

Who?

Every baby born in North Dakota.

What?

Screening for genetic disorders.

When?

Between one and two days of age is the best time for screening.

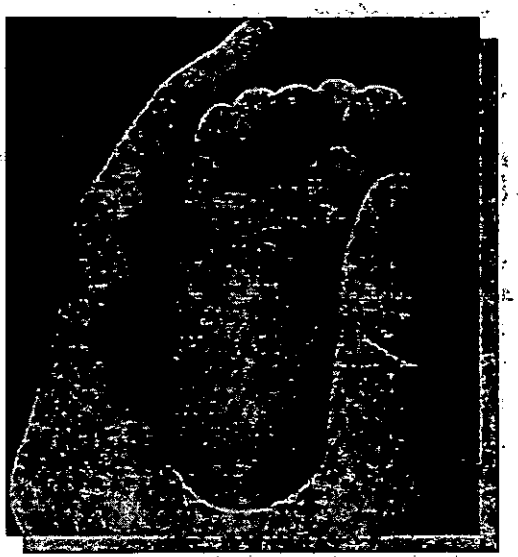
How?

Several small drops of blood, usually from one poke of your baby's heel, are allowed to dry on a special paper and then sent to a laboratory for testing.

Where?

The sample is sent to the University of Iowa Hygienic Laboratory in Ankeny, Iowa.

Ask your health-care provider for more details about North Dakota's Newborn Screening Program, or visit this website:
www.ndhealth.gov/familyhealth



North Dakota Department of Health
Division of Family Health
600 E. Boulevard Ave., Dept. 301
Bismarck, ND 58505-0200
800.472.2286 or 701.328.2493

Revised 08/06

Screening for a

Healthy Baby

North Dakota
Department of Health
Newborn Screening Program

What Is Newborn Screening?

Newborn screening is a blood test for certain metabolic and inherited disorders. The test is performed shortly after a baby's birth. North Dakota law requires all newborns and infants born in the state to be screened.

The North Dakota Newborn Screening Program identifies babies who may have one of these disorders and alerts the baby's health-care provider to the need for further testing and special care. With early diagnosis and treatment, complications from these serious disorders usually can be prevented.

Why Should My Baby Be Tested?

Babies with certain congenital disorders, often called "hidden birth defects," appear normal at birth. If untreated, these conditions may affect the baby's brain or physical development or cause other medical problems. These conditions can begin to affect the baby in the first days or weeks of life.

These hidden disorders are problems in the body's ability to make and use hormones, proteins, sugars or blood cells. These defects may be found in the blood long before symptoms appear or before they can cause serious damage.

By testing **all** newborns, many of these disorders can be found early. The earlier the physician diagnoses the disorder, the sooner treatment can begin. Earlier treatment gives the baby a better chance for normal growth and development, and can prevent many of the medical problems associated with these conditions.

How Is My Baby Tested?

All of the tests are performed from a few drops of blood obtained by pricking baby's heel at least 24 hours after birth or just before the baby is discharged from the hospital. The blood is collected on an absorbent paper collection form, which is then sent to the University of Iowa Laboratory.

IMPORTANT!

Babies born outside of hospitals also should be tested, preferably at about 24 hours to five days after birth. Parents can arrange the screening with their health-care provider.

Why Are Tests Needed When My Baby Seems Very Healthy?

All infants, even those who seem healthy, need to be tested because most infants with one of these disorders show no signs immediately after birth. When a disorder is found through early testing, the doctor can give the baby special care before the disorder begins to affect the child.

How Do I Receive the Results?

Parents are usually **not** notified if the screen was in a normal range. Your health-care provider will be informed when the tests are completed and you may ask them about the results. Generally, parents are notified only if retesting or further testing is needed.

If your health-care provider asks you to bring your baby in for retesting, do so as soon as possible. Retesting does not necessarily mean there is anything wrong with your baby. It may simply mean that another sample must be obtained.

How Can I Help the Doctor To Help My Baby?

Make sure the hospital knows how to contact you when you leave the hospital. If you don't have a telephone, leave the phone number of a friend, relative or neighbor. Let your doctor know immediately if you move soon after the baby is born. Then, if your baby needs retesting, the doctor can reach you.

What If I Don't Want My Baby Screened?

North Dakota law requires that all babies be tested, unless the parents object for religious reasons. Discuss refusal procedures with your health-care provider.

If you are asked to bring your baby in for re-screening, do so as soon as possible!

Regional Newborn Screening System Harmonization and Quality Improvement

May 4, 2010

Carol Johnson, Project Director
Kim Turner, RN, BSN, Nurse Clinician Specialist-Supervisor
University of Iowa Children's Hospital
Barb Schweitzer, RN, BSN
NDDoH Newborn Screening Director,
Regional Newborn Screening Coordinator

History Overview

- 1966 - The Iowa University Hygienic Lab began providing PKU testing
- 1992 - Lab contract with North Dakota to perform all newborn screening
- Fall of 2005 - Hurricane Katrina ~ Overnight the Iowa lab saw their newborn screening workload triple

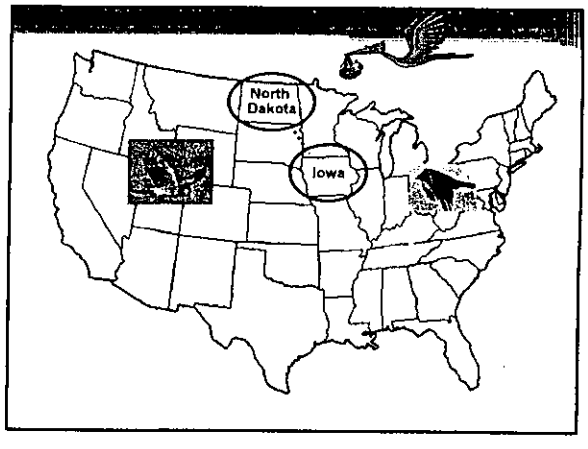
History Overview *con't*

- 2006 - Courier service started; the lab now receives specimens on the same day or next day after collection
- 2006 - Night shift in lab implemented
- 2007 - Contract with South Dakota to perform newborn screening lab testing and initial notification of abnormal results

History Overview *con't*

- 2004 to 2009 - Dr. Sara Copeland was Medical Director of Newborn Screening
- 2009 - Implemented Regional Coordinator position for Iowa and North Dakota

2010 - Recruiting a Newborn Screening Medical Director



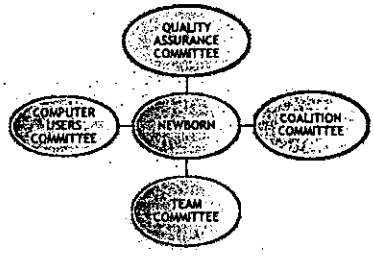
Need for Regional Coordinator:

- Both systems with similar needs
  - Education, quality improvement, follow-up
- Realization that duplications of efforts existed between Iowa and North Dakota
  - People doing the same job in both states, partially employed and partially dedicated to other programs- unable to delineate clearly where one job ends and the other begins
- Maintain cost effectiveness and sustainability
  - No change in number of staff, but more streamlined jobs
- Bring new perspective and ideas for systems improvement- avoid stagnation

- **Strengths Identified between systems:**
  - Caring, dedicated staff
  - State-of-the-art lab equipment
  - Weekend/holiday coverage
  - Rapid turn-around time
  - Strong collaboration with IDPH and NDDoH
- **Weaknesses Identified**
  - Lack of understanding and knowledge of each other's role
  - Division of responsibility for lab, short- term follow-up staff, long-term follow-up staff, genetics, information technology and medical consultants

SHARED PURPOSE - MOVING AS ONE

A shared purpose builds communication among the organization and individuals who share the same purpose. A Newborn Screening Committee is a collaborative between individuals and groups which join together for the common cause of newborn screening.



Before	After
1) No method to evaluate all aspects of newborn screening • Each state trying to do own thing, no communication or identification of similar problems; lack of communication between lab and follow-up	1) Bi-monthly Quality Assurance Committee • Developed method to discuss issues/concerns through event reports • Identified processes needing change

Before	After
2) No communication among stakeholders • Physicians would communicate with their state, but wouldn't necessarily be transmitted through program; • families with similar problems, but solutions were not communicated ; • 2 separate NBS advisory committees, with little connection between although utilizing same resources	2) Bi-monthly all-team meetings established to discuss: • Cut-off values • Program updates • Administrative issues

Before	After
3) Disconnect among newborn screening team members • Lab in one city, follow-up in another city or state	3) Lab and short-term follow up visit each other's worksite • As a result of QA event reports, guidelines, protocols and policies have been implemented • Roles clearly defined

Before	After
4) Lack of knowledge and understanding of database system • New electronic database, not a good fit for either lab or follow-up, both had needs that not met- better than previous system, but unwieldy	4) Quarterly database meetings implemented • Database changes made to meet needs now • Input shared for design of new system

Adapting to the Day-to-Day Multi-State Operations

Iowa short-term follow-up (STFU) staff:

- Clerk IV
- LPN
- RNs - 2
- Nurse Clinician Supervisor - RN

Daily Case Manager Staffing:

Including Weekends/ Holidays On Call

- Only nurses rotate on call
- Cross-training for all nursing staff; limited cross-training for Clerk IV
- Roles shifted to cover all states (IA, SD, ND)
- No increase in cost to either state or program
- Allows Regional Coordinator to focus on program harmonization

Approximate Births Per Year:

- Iowa - 40,000
- North Dakota - 10,000

Protocol Development:

- Involves case manager and lab staff input
- Nurse clinician submits proposed revisions to physician/regional coordinator for review/ approval
- Review and approval by medical consultants for different subspecialties and protocol implemented
- Annual review and revisions, as needed

- Increasing Awareness of Roles and Responsibilities- cross-training and contextual understanding of roles

Worksite Visits:

- STFU to lab
- Lab to STFU
- Regional Coordinator periodic visit to Iowa
- Lab staff periodic visit to North Dakota

Communication Issue **Identified and Addressed to Date:**

- Concern Identified: PCP/facility omitting necessary demographic information on blood spot card
- No clear process to manage/address
- Lack of information causes inaccurate database information and potential compromised health of infant

Solution

- Process and policy implemented February 1, 2010
- STFU and UHL buy in
- Accurate, timely information provided to all stakeholders

Has Regional Harmonization Been Achieved?

- 3-year strategic work plan developed
- Secure website
- Joint policy book
- The Regional Coordinator has provided leadership through communication and collaboration

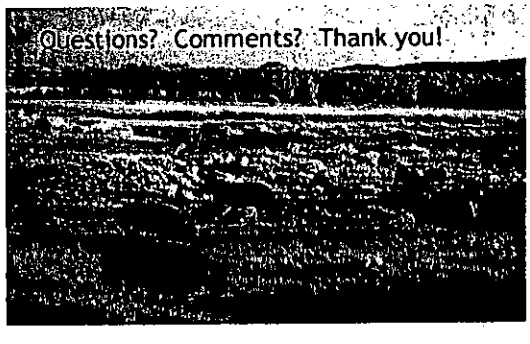
Acknowledgements

Sara Copeland, MD
Deputy Branch Chief, Genetic Services Branch
Division of Services for Children with Special Health Needs, Maternal and Child Health Bureau

Stan Berberich, PhD
Program Manager, Maternal and Newborn Screening University of Iowa Hygienic Lab

Kim Piper, RNC, BS.CPH, CPHG
State Genetics Coordinator
Iowa Department of Public Health Center for Congenital and Inherited Disorders

North Dakota Department of Health



Newborn Screening Guidelines

Barb Schweitzer, RN
Director
800-472-2286 (Press 1)
Phone: 701-328-4538
Email: bschweit@nd.gov
Cell Phone: 701-220-0366
Fax: 701-328-1412

NORTH DAKOTA DEPARTMENT of HEALTH

Objectives

- Describe the Newborn Screening process in North Dakota
- Identify protocol in performing a heel stick blood spot collection and the completion of the specimen card

NORTH DAKOTA DEPARTMENT of HEALTH

The North Dakota **Law**

- 25-17-04. Testing and reporting requirements.
  - The physician attending a newborn child, or the birth attendant in the case of an out-of-hospital birth, shall cause that newborn child to be subjected to testing for metabolic diseases, in the manner prescribed by the state department of health. A physician attending a patient with a metabolic disease shall report the case to the state department of health. The testing requirements of this section do not apply if the parents of a newborn child object to the testing on the grounds that testing for metabolic diseases conflicts with their religious tenets and practices.

NORTH DAKOTA DEPARTMENT of HEALTH

The North Dakota Administrative **Rules**

- 33-06-16-02. Testing of Newborns.
  - Under the newborn screening system, except as authorized by section 33-06-16-04, each newborn infant born in this state shall be tested for the metabolic diseases, **cystic fibrosis**, hypothyroidism, galactosemia, congenital adrenal hyperplasia, biotinidase deficiency, sickle cell disease, and other hemoglobinopathies, and any other disease that can be identified through tandem mass spectrometry that is designated on the department's test schedule with a designated laboratory engaged to perform this testing on behalf of the program.

NORTH DAKOTA DEPARTMENT of HEALTH

The North Dakota Administrative **Rules** (continued)

- 33-06-16-03. Physician responsibility.
  - The physician or other birth attendant shall order that:
    - A specimen of blood be collected from a newborn in accordance with directions supplied by the laboratory designated by the state department of health and the program; and
    - The specimen be sent to that laboratory.

NORTH DAKOTA DEPARTMENT of HEALTH

The North Dakota Administrative **Rules** (continued)

- 33-06-16-04. Refusal of testing.
  - If the parents or guardians refuse to have their infant receive newborn screening testing as authorized by North Dakota Century Code section 25-17-04, that refusal shall be documented by a written statement signed by the parents or guardians.
  - **The original refusal statement shall become a part of the infant's medical record and a copy of the statement shall be submitted to the program.**

NORTH DAKOTA DEPARTMENT of HEALTH

Newborn Screening Purpose

- The purpose of newborn screening for metabolic and genetic disorders is the **early identification and treatment** of affected individuals in order to avoid adverse health consequences.

Disorders/Conditions ND Screens for:

- Amino Acidemias and Urea Cycle Disorders
  - 5-Oxoprolinuria
  - Agininemia (ARG)
  - Arginiosuccinic Aciduria (ASA)
  - Citrullinemia Type 1 (CTLN1)
  - Citrullinemia Type 2 (CTLN2)
  - Hypermethioninemia
  - Tyrosinemia Type 1, 2 & 3 (TYR)

Disorders/Conditions ND Screens for: (continued)

- Organic Acidemias
  - 2-Methyl Butyryl-CoA Dehydrogenase Deficiency (2MBDH Deficiency)
  - 3-Methylcrontonyl-CoA Carboxylase Deficiency (3-MCC)
  - 3-Methylglutaconyl-CoA Hydratase (3MGH Deficiency)
  - Beta-Ketothiolase Deficiency (Ketone Utilization Disorder) (BKT)
  - Glutaric Acidemia Type 1/Glutaryl-CoA Dehydrogenase Deficiency (GA1)
  - Glutaric Acidemia Type 2 (GA2)

Disorders/Conditions ND Screens for: (continued)

- Organic Acidemias (continued)
  - Isobutyryl-CoA Dehydrogenase Deficiency (IBD)
  - * Isovaleric Acidemia/Isovaleryl-CoA Dehydrogenase Deficiency (IVA)
  - Methylmalonic Acidemia, Vitamin B12 Non-Responsive (MMA)
  - Methylmalonic Acidemia, Vitamin B12 Responsive (MMAA)
  - Multiple CoA Carboxylase Deficiency
  - Proprionic Acidemia/Propionyl-CoA Carboxylase Deficiency (PA)
  - * Carnitine Uptake Defect (CUD)

* Identified in North Dakota

Disorders/Conditions ND Screens for: (continued)

- Fatty Acid Oxidation Disorders
  - 2,4 Dienoyl-CoA Reductase Deficiency
  - 3-Hydroxy 3-Methylglutaryl-CoA Lyase Deficiency (HMG)
  - Carnitine/Acylcarnitine Translocase Deficiency (CACT)
  - * Carnitine Palmitoyl Transferase Deficiency Type 1 (CPT1)
  - Carnitine Palmitoyl Transferase Deficiency Type 2 (CPT2)
  - * Long-chain Acyl-CoA Dehydrogenase Deficiency (LCADD)

* Identified in North Dakota

Disorders/Conditions ND Screens for: (continued)

- Fatty Acid Oxidation Disorders (continued)
  - Long-chain Hydroxy Acyl-CoA Dehydrogenase Deficiency/3-Hydroxyacyl CoA Dehydrogenase Deficiency (LCHADD)
  - * Medium-chain Acyl-CoA Dehydrogenase Deficiency (MCADD)
  - * Short-chain Acyl-CoA Dehydrogenase Deficiency (SCADD)
  - Trifunctional Protein Deficiency (TFP)
  - * Very Long-chain Acyl-CoA Dehydrogenase Deficiency (VLCADD)

* Identified in North Dakota

Disorders/Conditions ND Screens for: (continued)

- Others
  - * Biotinidase Deficiency (BT)
  - * Congenital Adrenal Hyperplasia (CAH)
  - * Congenital Hypothyroidism (CH)
  - * Galactosemia (GALT)
  - * Hemoglobinopathies (HGB)
  - * Maple Syrup Urine Disease (MSUD)
  - * Phenylketonuria (PKU)
  - * Sickle Cell Disease
  - * Cystic Fibrosis

* Identified in North Dakota

NORTH DAKOTA DEPARTMENT of HEALTH

North Dakota Resources

Dr. John Martsolf, Medical Genetics
University of North Dakota
Grand Forks, ND

Mary Riske, Clinical Nurse Geneticist
University of North Dakota
Grand Forks, ND

NORTH DAKOTA DEPARTMENT of HEALTH

Resources (continued)

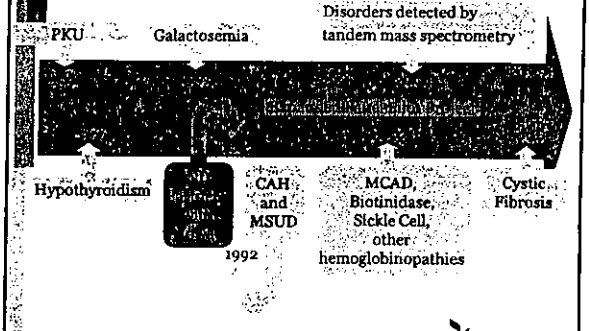
Dr. Alan Kenien, Fargo, ND
Pediatric Endocrinologist
Conducts Metabolic Clinics 4 times a year funded by Children's Special Health Services

Dr. Sara Copeland, Iowa
Assistant Professor in Pediatrics
Medical Director of Newborn Screening for Iowa

Dr. Nathan Kobrinsky, Fargo, ND
Pediatric Hematology

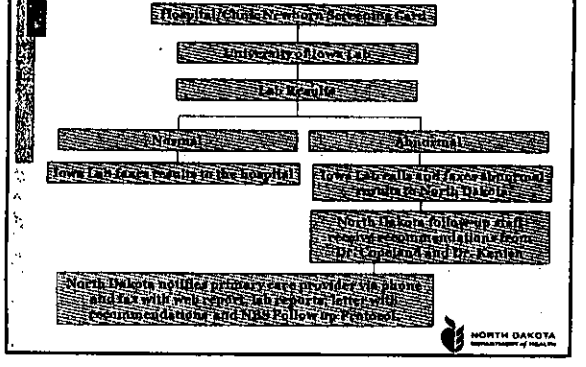
NORTH DAKOTA DEPARTMENT of HEALTH

History of North Dakota Newborn Screening



NORTH DAKOTA DEPARTMENT of HEALTH

Coordination



NORTH DAKOTA DEPARTMENT of HEALTH

Follow-up

- State newborn screening programs have evolved with the goal of providing safe screening tests and appropriatefollow-up to every newborn.
- State NBS follow-up staff follow the baby until lab result is **normal or confirmed diagnosis**.
- A total of 917 cases were followed up by the State NBS staff in 2007.

NORTH DAKOTA DEPARTMENT of HEALTH

Follow-up

- Depending on the pattern, may just repeat the specimen or go directly to confirmatory lab testing.
- Generally confirmatory labs will be either blood or urine tests depending on the pattern of elevations and the possible disorder.

Practitioner Follow-Up Responsibilities

- Prompt action on unsatisfactory or abnormal NBS result
- Verify results for every infant
- Provide education to parents on NBS disorder
- Keep NBS Program staff informed of further testing and results

Variables that can affect newborn screening results—MAY 2008
NORTH DAKOTA NEWBORN SCREENING.
REVIEWED BY DR. SARA COPELAND

See NBS Variables Table in handout.

To Do List for North Dakota Newborn Screening

1) Provide Patient/Family Education
2) Maintain and expand Quality Management Program
3) Prepare Facility Report Card
4) Provide annual Report for all of North Dakota

Dr. Alan Kenien
D

Dr. Sara Copeland

Stan Berberich, PhD., Lab Manager

Acknowledgements

- Terry Dwelle, MD, State Health Officer
  ND Department of Health
- Kim Senn, RN, North Dakota Title V Director
- Pat Welle, MD, Chair, Newborn Screening Advisory Committee
- Alan Kenien, MD, Pediatric Endocrinologist
- Sara Copeland, MD, Metabolic Consultant to North Dakota

Newborn Screening Guidelines

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Fax: 701-328-1412

NORTH DAKOTA DEPARTMENT of HEALTH

Objectives

- Describe the Newborn Screening process in North Dakota
- Identify protocol in performing a heel stick blood spot collection and the completion of the specimen card

NORTH DAKOTA DEPARTMENT of HEALTH

The North Dakota **Law**

- 25-17-04. Testing and reporting requirements.
  - The physician attending a newborn child, or the birth attendant in the case of an out-of-hospital birth, shall cause that newborn child to be subjected to testing for metabolic diseases, in the manner prescribed by the state department of health. A physician attending a patient with a metabolic disease shall report the case to the state department of health. The testing requirements of this section do not apply if the parents of a newborn child object to the testiug on the grounds that testing for metabolic diseases conflicts with their religious tenets and practices.

NORTH DAKOTA DEPARTMENT of HEALTH

The North Dakota Administrative **Rules**

- 33-06-16-02. Testing of Newborns.
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NORTH DAKOTA DEPARTMENT of HEALTH

The North Dakota Administrative **Rules** (continued)

- 33-06-16-03. Physician responsibility.
  - The physician or other birth attendant shall order that:
    - A specimen of blood be collected from a newborn in accordance with directions supplied by the laboratory designated by the state department of health and the program; and
    - The specimen be sent to that laboratory.

NORTH DAKOTA DEPARTMENT of HEALTH

The North Dakota Administrative **Rules** (continued)

- 33-06-16-04. Refusal of testing.
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  - **The original refusal statement shall become a part of the infant's medical record and a copy of the statement shall be submitted to the program.**

NORTH DAKOTA DEPARTMENT of HEALTH

Disorders/Conditions ND Screens for: (continued)

- Others
  - Biotinidase Deficiency (BT)
  - Congenital Adrenal Hyperplasia (CAH)
  - Congenital Hypothyroidism (CH)
  - Galactosemia (GALT)
  - Hemoglobinopathies (HGB)
  - Maple Syrup Urine Disease (MSUD)
  - Phenylketonuria (PKU)
  - Sickle Cell Disease
  - Cystic Fibrosis

* Identified in North Dakota

NORTH DAKOTA DEPARTMENT of HEALTH

North Dakota Resources

Dr. John Martsolf, Medical Genetics
University of North Dakota
Grand Forks, ND

Mary Riske, Clinical Nurse Geneticist
University of North Dakota
Grand Forks, ND

NORTH DAKOTA DEPARTMENT of HEALTH

Resources (continued)

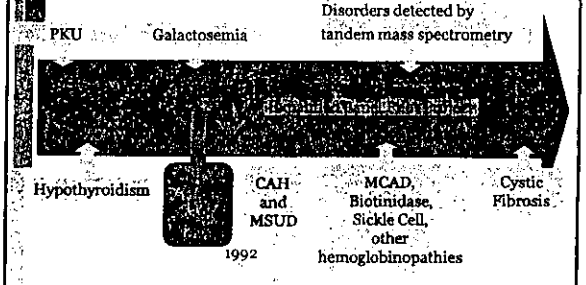
Dr. Alan Kenien, Fargo, ND
Pediatric Endocrinologist
Conducts Metabolic Clinics 4 times a year funded by Children's Special Health Services

Dr. Sara Copeland, Iowa
Assistant Professor in Pediatrics
Medical Director of Newborn Screening for Iowa

Dr. Nathan Kobrinsky, Fargo, ND
Pediatric Hematology

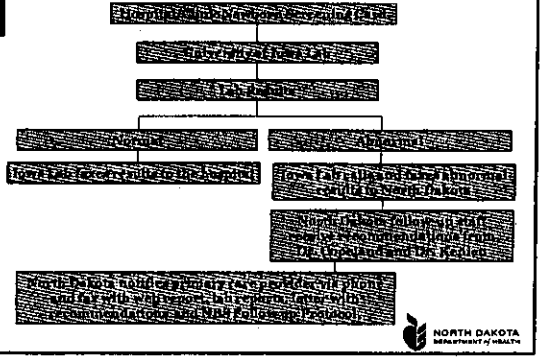
NORTH DAKOTA DEPARTMENT of HEALTH

History of North Dakota Newborn Screening



NORTH DAKOTA DEPARTMENT of HEALTH

Coordination



NORTH DAKOTA DEPARTMENT of HEALTH

Follow-up

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NORTH DAKOTA DEPARTMENT of HEALTH

Stan Berberich, PhD., Lab Manager

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- Alan Kenien, MD, Pediatric Endocrinologist
- Sara Copeland, MD, Metabolic Consultant to North Dakota

State NBS Program

- North Dakota Department of Health
  Community Health Section
  Family Health Division
  600 East Boulevard Ave, Dept. 301
  Bismarck, North Dakota 58505

North Dakota State Capitol

How is NBS program funded?

- The newborn screening is a fee for service
- The purchase for the screening specimen cards and all follow-up activities are funded through the MCH Title V Block Grant

State NBS Statistics

- Population = 635,867
- Birthrate = 9,877
- Average yearly initial screens = 9,587
- Average yearly repeat screens = 620
- Combined NBS disorder detection rate=18

Disorders Screened

(Year Started)

- PKU.....1964
- CH.....1977
- GALT.....1991
- Hb.....2003
- CAH.....1992
- MS/MS Pilot.....2002
  MS/MS full implementation.....2004
- Cystic Fibrosis.....January 2006

NBS Program Staff

- .7 FTE Program Director, Follow-up RN
- .3 FTE Follow-up RN (Vacant)
- .5 Clerical

Barb Schweitzer & Courtney Smith

What's new with your program?

- Working on developing Parent Fact Sheets
- Investigating the possibility of the Iowa follow-up staff in assisting North Dakota with weekend/holiday coverage

What makes you most proud of your program?

- Being on the cutting edge with Iowa in the implementation of Tandem Mass and Cystic Fibrosis
- All in all, being associated with the University of Iowa lab, follow-up staff as well as the IT staff

Most challenging past experience?

- Regaining the trust of the State's Pediatricians with the expansion of Tandem Mass in 2004 and the addition of Cystic Fibrosis in 2006
- Both of these required administrative rule changes

Most memorable or rewarding past experience?

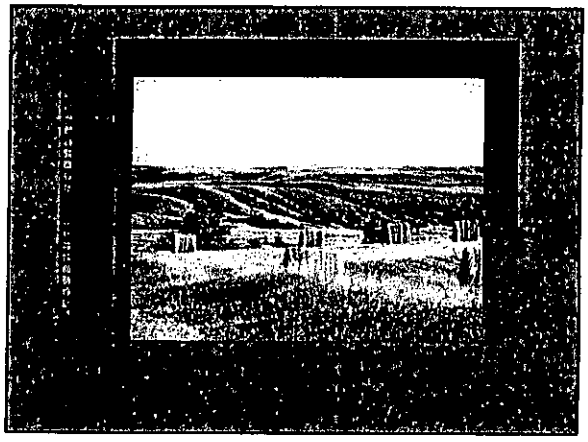
- Identifying a C.U.D. case in April 2006 and began treatment at 4 days of age

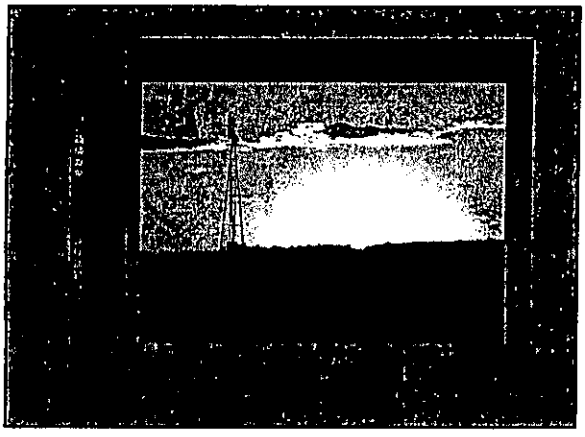
What is biggest current challenge?

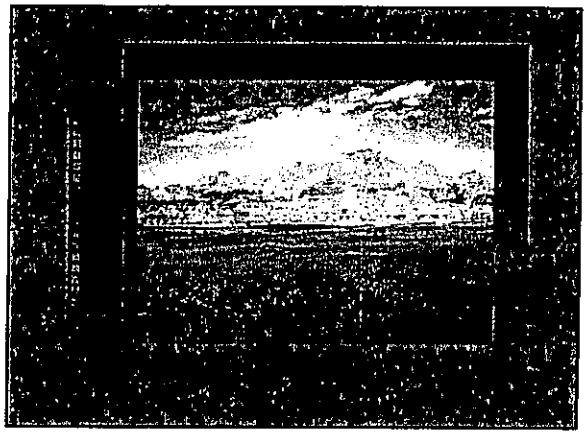
- Lack of medical specialists in North Dakota
- Lack of trained follow-up RN staff

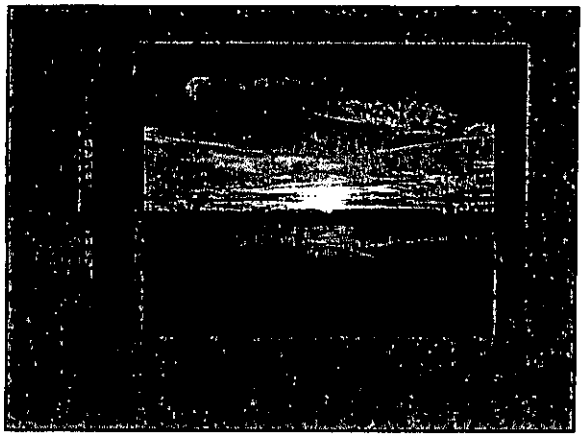
Some good lessons learned:

- Engage the State's Pediatricians as changes occur
- Maintain the Newborn Screening Advisory group









Hearing Hoofbeats and Thinking Zebras

April 23-24, 2007

Newborn Screening in North Dakota

Barb Schweitzer, RN
Director
N.D. 800.472.2286 (Press 1)
Phone: 701.328.4538
Email: bschweit@nd.gov
On Call Phone: 701.220.0366

NORTH DAKOTA DEPARTMENT OF HEALTH

- North Dakota Department of Health
  Community Health Section
  Family Health Division
  Bismarck, North Dakota 58505
- Contract with University of Iowa Lab for lab services for all ND babies since 1992

NORTH DAKOTA DEPARTMENT OF HEALTH

The North Dakota **Law**

- 25-17-04. Testing and reporting requirements.
  - The physician attending a newborn child, or the birth attendant in the case of an out-of-hospital birth, shall cause that newborn child to be subjected to testing for metabolic diseases, in the manner prescribed by the state department of health. A physician attending a patient with a metabolic disease shall report the case to the state department of health. The testing requirements of this section do not apply if the parents of a newborn child object to the testing on the grounds that testing for metabolic diseases conflicts with their religious tenets and practices.

NORTH DAKOTA DEPARTMENT OF HEALTH

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NORTH DAKOTA DEPARTMENT OF HEALTH

The North Dakota Administrative **Rules** (continued)

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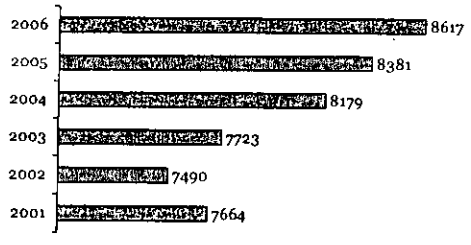
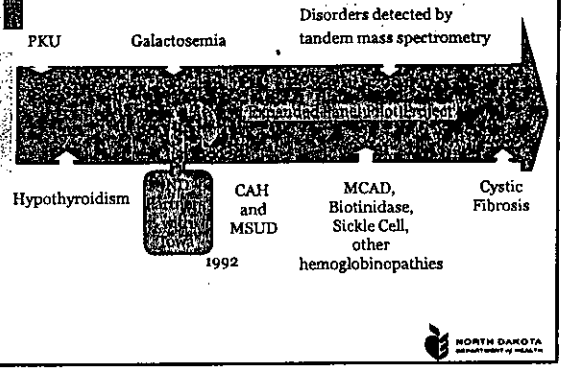
NORTH DAKOTA DEPARTMENT OF HEALTH

North Dakota Births

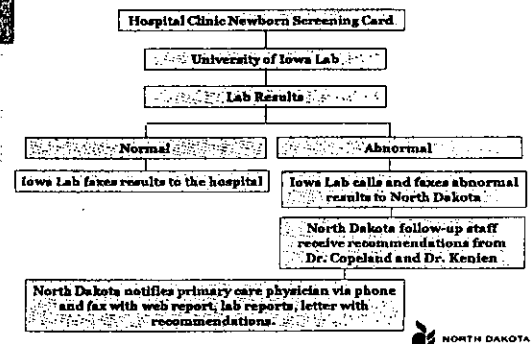


NORTH DAKOTA DEPARTMENT OF HEALTH

History of North Dakota Newborn Screening



Coordination



Disorders/Conditions ND Screens for:

- Amino Acidemias and Urea Cycle Disorders
  - 5-Oxoprolinuria
  - Agininemia (ARG)
  - Arginiosuccinic Aciduria (ASA)
  - Citrullinemia Type 1 (CTLN1)
  - Citrullinemia Type 2 (CTLN2)
  - Hypermethioninemia
  - Tyrosinemia Type 1, 2 & 3 (TYR)

Disorders/Conditions ND Screens for: (continued)

- Organic Acidemias
  - 2-Methyl Butyryl-CoA Dehydrogenase Deficiency (2MBDH Deficiency)
  - 3-Methylcrontonyl-CoA Carboxylase Deficiency (3-MCC)
  - 3-Methylglutaconyl-CoA Hydratase (3MGH Deficiency)
  - Beta-Ketothiolase Deficiency (Ketone Utilization Disorder) (BKT)
  - Glutaric Acidemia Type 1/Glutaryl-CoA Dehydrogenase Deficiency (GA1)
  - Glutaric Acidemia Type 2 (GA2)

Disorders/Conditions ND Screens for: (continued)

- Organic Acidemias (continued)
  - Isobutyryl-CoA Dehydrogenase Deficiency (IBD)
  - * Isovaleric Acidemia/Isovaleryl-CoA Dehydrogenase Deficiency (IVA)
  - Methylmalonic Acidemia, Vitamin B12 Non-Responsive (MMA)
  - Methylmalonic Acidemia, Vitamin B12 Responsive (MMAA)
  - Multiple CoA Carboxylase Deficiency
  - Proprionic Acidemia/Propionyl-CoA Carboxylase Deficiency (PA)

* Identified in North Dakota

Disorders/Conditions ND Screens for: (continued)

- Fatty Acid Oxidation Disorders
  - 2,4 Dienoyl-CoA Reductase Deficiency
  - 3-Hydroxy 3-Methylglutaryl-CoA Lyase Deficiency (HMG)
  - Carnitine/Acylcarnitine Translocase Deficiency (CACT)
  - Carnitine Palmitoyl Transferase Deficiency Type 1 (CPT1)
  - Carnitine Palmitoyl Transferase Deficiency Type 2 (CPT2)
  - * Long-chain Acyl-CoA Dehydrogenase Deficiency (LCADD)

* Identified in North Dakota

Disorders/Conditions ND Screens for: (continued)

- Fatty Acid Oxidation Disorders (continued)
  - Long-chain Hydroxy Acyl-CoA Dehydrogenase Deficiency/3-Hydroxyacyl CoA Dehydrogenase Deficiency (LCHADD)
  - * Medium-chain Acyl-CoA Dehydrogenase Deficiency (MCADD)
  - * Short-chain Acyl-CoA Dehydrogenase Deficiency (SCADD)
  - Trifunctional Protein Deficiency (TFP)
  - * Very Long-chain Acyl-CoA Dehydrogenase Deficiency (VLCADD)

* Identified in North Dakota

NORTH DAKOTA DEPARTMENT OF HEALTH

Disorders/Conditions ND Screens for: (continued)

- Others
  - Biotinidase Deficiency (BT)
  - * Congenital Adrenal Hyperplasia (CAH)
  - * Congenital Hypothyroidism (CH)
  - * Galactosemia (GALT)
  - * Hemoglobinopathies (HGB)
  - Maple Syrup Urine Disease (MSUD)
  - * Phenylketonuria (PKU)
  - * Sickle Cell Disease
  - Cystic Fibrosis

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NORTH DAKOTA DEPARTMENT OF HEALTH

Follow-up

- State newborn screening programs have evolved with the goal of providing safe screening tests and appropriatefollow-up to every newborn.
- State NBS follow-up staff follow the baby until lab result is **normal or confirmed diagnosis**.
- A total of 315 cases were followed up by the State NBS staff in 2005 and 457 cases were followed up in 2006.

NORTH DAKOTA DEPARTMENT OF HEALTH

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Newborn Screening in North Dakota

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Becky Bailey, RN
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On Call Phone: 701-220-0366

NORTH DAKOTA DEPARTMENT of HEALTH

Objectives

- Understand the North Dakota Newborn Screening Program.
- Discuss the role of physician and/or birth attendant.
- Review the disorders that have been identified in North Dakota.

NORTH DAKOTA DEPARTMENT of HEALTH

Newborn Screening in North Dakota

- North Dakota Department of Health
- Division of Family Health
- Title V (MCH) funds coordination and short-term follow-up.
- Laboratory Testing: University of Iowa Lab
- Newborn Screening – Fee for Service

NORTH DAKOTA DEPARTMENT of HEALTH

The North Dakota Law

- 25-17-04. Testing and reporting requirements.
  - The physician attending a newborn child, or the birth attendant in the case of an out-of-hospital birth, shall cause that newborn child to be subjected to testing for metabolic diseases, in the manner prescribed by the state department of health. A physician attending a patient with a metabolic disease shall report the case to the state department of health. The testing requirements of this section do not apply if the parents of a newborn child object to the testing on the grounds that testing for metabolic diseases conflicts with their religious tenets and practices.

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NORTH DAKOTA DEPARTMENT of HEALTH

The North Dakota Administrative Rules (continued)

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  - If the parents or guardians refuse to have their infant receive newborn screening testing as authorized by North Dakota Century Code section 25-17-04, that refusal shall be documented by a written statement signed by the parents or guardians.
  - **The original refusal statement shall become a part of the infant's medical record and a copy of the statement shall be submitted to the program.**

NORTH DAKOTA DEPARTMENT of HEALTH

Collaboration with Iowa and North Dakota Follow-up Staff

- Participate with quarterly meetings via conference calls with the Iowa follow-up staff.
- Participate in quarterly conference calls with Iowa Lab.
- Participate via conference calls with the redesign of the web reports.

Follow-up (continued)

- State newborn screening programs have evolved with the goal of providing safe screening tests and appropriatefollow-up to every newborn.
- State follow-up staff follow the baby until lab result is **normal or confirmed diagnosis**.
- A total of 315 cases were followed up by the state staff in 2005.

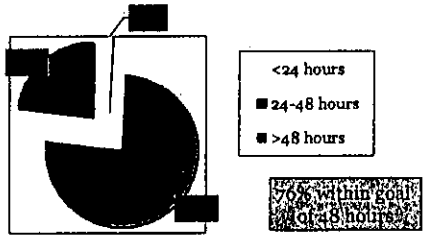
Follow-up (continued)

- Depending on the pattern, may just repeat the specimen or go directly to confirmatory studies
- Generally confirmatory studies will be either blood or urine tests depending on the pattern of elevations and the possible disorder.

Practitioner Follow-Up Responsibilities

- Contact Iowa Lab and/or ND Department of Health if results are not received in two weeks
- Prompt action on unsatisfactoryor abnormal result
- Communication with consultants regarding infants with abnormal results
- Verify results for every infant

Infant's Age at Time of Initial Testing



Total 14,585 Specimens from 1/1/04 to 6/30/05

Timing - When to Test

- In a perfect world – collect at 24-48 hours
- ALWAYS collect prior to blood transfusion
- OK to collect prior to 24 hours if being discharged home
  - Better to collect early than lose to follow-up
- Repeat screening will not hurt
  - May make for more accurate results if tested early

What Are Our Neighbors Doing? (continued)

Montana

- Mandatory Tests
  - PKU = $10.19
  - Galactosemia = $11.10
  - CH-TSH = $8.71
  - T4 = $9.88
  - Hemoglobinopathies, i.e., mainly Hemoglobin S, Hemoglobin C, and beta-thalassemia = $8.17
  - Total for mandatory tests = $48.05
- Optional Tests
  - CF = $9.88
  - CAH and Biotinidase = $6.50
  - Acylcarnitine profile = $10.75
  - Aminoacidopathies = $4.25
  - Total for optional tests = $31.38
  - Total for mandatory + optional testing = $79.43

What Are Our Neighbors Doing? (continued)

Montana (continued)

- The optional tests are referred to the Wisconsin Newborn Screening Lab
- Optional tests are ordered separately on the NBS specimen collection card

North Dakota

- Screenings performed as previously noted
- Have used the University of Iowa Lab since 1992
- Charge is $42.50

Long Term Follow-up

- Long term follow-up begins with the confirmation of a diagnosis and initiation of treatment (if necessary and appropriate) and ends with the transfer of treatment or care to another health care provider outside of a designated catchment area or the death of the patient. In an ideal long-term follow-up program, contact is maintained on a regular, protocol-based schedule despite changes in state of residence, care provider or insurance carrier.

Establishing the Need for LTFU

- To facilitate evaluations of NBS programs, short-term and long-term performance measures should be collected.
- Essential long-term measures should assess whether infants with a diagnosed disorder have developmental disabilities, mental retardation, and premature mortality and should identify adverse health outcomes associated with each disorder beyond the newborn period.

Why do it?

- Continuing communication with and education of physicians seeing the children
- Documented outcomes of patients over life span
- Proof of utility of NBS-documentation of health of ND children ID'd on NBS.

Our To Do List for North Dakota Newborn Screening for 2006

1) Develop Patient/Family Education
2) Develop Quality Management Program
3) Set-up Facility Report Card
4) Do Annual Report for all of North Dakota

The North Dakota Department of Health Newborn Screening Program

Barb Schweitzer, Director
800-472-2286 (Press 1 for MCH)
701-328-4538 (Direct Line)
Email: bschweit@state.nd.us

The North Dakota Law

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The North Dakota Administrative Rules

- 33-06-16-01. Definitions. As used in this chapter:
  - "Diagnostic test" means a test that is used to establish a definitive diagnosis of some condition in an affected newborn.
  - "Newborn screening system" means the routine testing of newborn infants for congenital conditions by analysis of a dried blood specimen through laboratory procedures that identify infants with an increased risk for specified diseases and conditions, and that justify follow-up actions and diagnostic tests or procedures.

The North Dakota Administrative Rules (continued)

- 33-06-16-01. Definitions. As used in this chapter:
  - "Program" means the North Dakota newborn screening program in the division of maternal and child health in the state department of health.
  - "Protected health information" has the meaning set forth in North Dakota Century Code section 23-01.3-01.
  - "Tandem mass spectrometry" is a laboratory technology that uses a machine consisting of two mass spectrometers joined by a fragmentation chamber. Tandem mass spectrometry technology allows the identification of an array of metabolic conditions, such as amino acid, fatty acid, and organic acid disorders, from a single dried blood spot.

The North Dakota Administrative Rules (continued)

- 33-06-16-02. Testing of Newborns.
  - Under the newborn screening system, except as authorized by section 33-06-16-04, each newborn infant born in this state shall be tested for the metabolic diseases phenylketonuria, hypothyroidism, galactosemia, congenital adrenal hyperplasia, biotinidase deficiency, sickle cell disease, and other hemoglobinopathies, and any other disease that can be identified through tandem mass spectrometry that is designated on the department's test schedule with a designated laboratory engaged to perform this testing on behalf of the program.

The North Dakota Administrative Rules (continued)

- 33-06-16-03. Physician responsibility.
  - The physician or other birth attendant shall order that:
    - A specimen of blood be collected from a newborn in accordance with directions supplied by the laboratory designated by the state department of health and the program; and
    - The specimen be sent to that laboratory.

The North Dakota Administrative Rules (continued)

- 33-06-16-03. Physician responsibility.
  - If a patient, who has a condition for which the program conducts a screening test, but which has been detected by another mechanism or by an out-of-state screening program, the patient's physician shall, within thirty days of becoming aware of the patient's condition, notify the program of the patient's name, parent's name, parent's name if the patient is under eighteen years of age, date of birth, address, and condition.

The North Dakota Administrative Rules (continued)

- 33-06-16-04. Refusal of testing.
  - If the parents or guardians refuse to have their infant receive newborn screening testing as authorized by North Dakota Century Code section 25-17-04, that refusal shall be documented by a written statement signed by the parents or guardians.
  - **The original refusal statement shall become a part of the infant's medical record and a copy of the statement shall be submitted to the program.**

Newborn Screening Purpose

- The purpose of newborn screening for metabolic and genetic disorders is the **early identification and treatment** of affected individuals in order to avoid adverse health consequences.

Disorders/Conditions ND Screens for:

- Amino Acidemias and Urea Cycle Disorders
  - 5-Oxoprolinuria
  - Agininemia (ARG)
  - Arginiosuccinic Aciduria (ASA)
  - Citrullinemia Type 1 (CTLN1)
  - Citrullinemia Type 2 (CTLN2)
  - Hypermethioninemia
  - Tyrosinemia Type 1, 2 & 3 (TYR)

Disorders/Conditions ND Screens for: (continued)

- Organic Acidemias
  - 2-Methyl Butyryl-CoA Dehydrogenase Deficiency (2MBDH Deficiency)
  - 3-Methylcrontonyl-CoA Carboxylase Deficiency (3-MCC)
  - 3-Methylglutaconyl-CoA Hydratase (3MGH Deficiency)
  - Beta-Ketothiolase Deficiency (Ketone Utilization Disorder) (BKT)
  - Glutaric Acidemia Type 1/Glutaryl-CoA Dehydrogenase Deficiency (GA1)
  - Glutaric Acidemia Type 2 (GA2)

Disorders/Conditions ND Screens for: (continued)

- Organic Acidemias (continued)
  - Isobutyryl-CoA Dehydrogenase Deficiency (IBD)
  - Isovaleric Acidemia/Isovaleryl-CoA Dehydrogenase Deficiency (IVA)
  - Methylmalonic Acidemia, Vitamin B12 Non-Responsive (MMA)
  - Methylmalonic Acidemia, Vitamin B12 Responsive (MMAA)
  - Multiple CoA Carboxylase Deficiency
  - * Proprionic Acidemia/Propionyl-CoA Carboxylase Deficiency (PA)

* Identified in North Dakota

Disorders/Conditions ND Screens for: (continued)

- Fatty Acid Oxidation Disorders
  - 2,4 Dienoyl-CoA Reductase Deficiency
  - 3-Hydroxy 3-Methylglutaryl-CoA Lyase Deficiency (HMG)
  - Carnitine/Acylcarnitine Translocase Deficiency (CACT)
  - Carnitine Palmitoyl Transferase Deficiency Type 1 (CPT1)
  - Carnitine Palmitoyl Transferase Deficiency Type 2 (CPT2)
  - * Long-chain Acyl-CoA Dehydrogenase Deficiency (LCADD)

* Identified in North Dakota

NORTH DAKOTA DEPARTMENT of HEALTH

Disorders/Conditions ND Screens for: (continued)

- Fatty Acid Oxidation Disorders (continued)
  - Long-chain Hydroxy Acyl-CoA Dehydrogenase Deficiency/3-Hydroxyacyl CoA Dehydrogenase Deficiency (LCHADD)
  - * Medium-chain Acyl-CoA Dehydrogenase Deficiency (MCADD)
  - * Short-chain Acyl-CoA Dehydrogenase Deficiency (SCADD)
  - Trifunctional Protein Deficiency (TFP)
  - * Very Long-chain Acyl-CoA Dehydrogenase Deficiency (VLCADD)

* Identified in North Dakota

NORTH DAKOTA DEPARTMENT of HEALTH

Disorders/Conditions ND Screens for: (continued)

- Others
  - * Biotinidase Deficiency (BT)
  - * Congenital Adrenal Hyperplasia (CAH)
  - * Congenital Hypothyroidism (CH)
  - * Galactosemia (GALT)
  - Hemoglobinopathies (HGB)
  - * Maple Syrup Urine Disease (MSUD)
  - * Phenylketonuria (PKU)
  - Sickle Cell Disease

* Identified in North Dakota

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Not a PKU Test!!!!!!!!!

- NEWBORN SCREENING TEST
- NEWBORN SCREENING TEST
- NEWBORN SCREENING TEST
- NEWBORN SCREENING TEST

NORTH DAKOTA DEPARTMENT of HEALTH

Difference Between Treated and Untreated

- Untreated PKU

- Treated PKU

NORTH DAKOTA DEPARTMENT of HEALTH

Difference Between Treated and Untreated

- Untreated Fatty Acid Oxidation Defect

- Treated Fatty Acid Oxidation Defect

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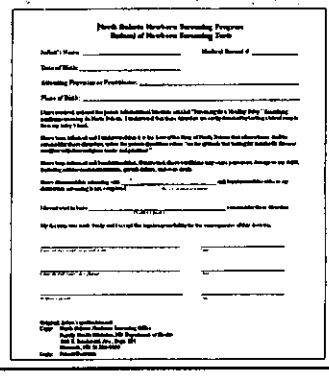
Purpose of the Guidelines Book

- Insert Table of Contents here once updated....

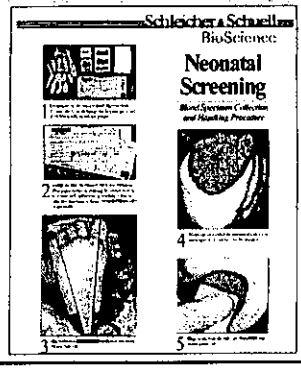
Newborn Screening Refusal Form



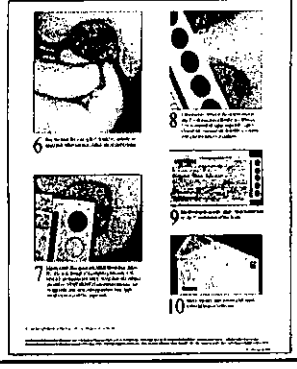
Important Screening Considerations

- Collection Form Information
- Collection Times
- Specimen Collection
- Premature or Ill Infants
- Transferred Infants
- Transfusions
- Treatment

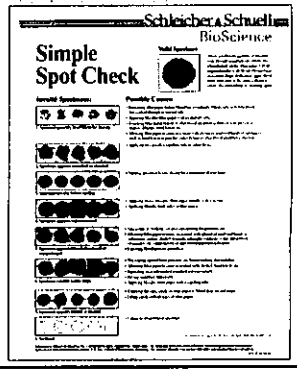
Collection and Handling Procedures



Collection and Handling Procedures (continued)



Simple Spot Check



Newborn Screening Protocols

NORTH DAKOTA DEPARTMENT OF HEALTH
NEWBORN SCREENING FOLLOW-UP PROTOCOLS

Galactosemia (GALT)

NORTH DAKOTA DEPARTMENT of HEALTH

Newborn Screening Protocols (continued)

NORTH DAKOTA DEPARTMENT of HEALTH

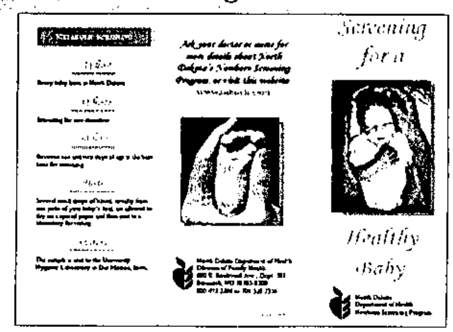
Parent Fact Sheets

North Dakota Department of Health
Parent Fact Sheet

Galactosemia

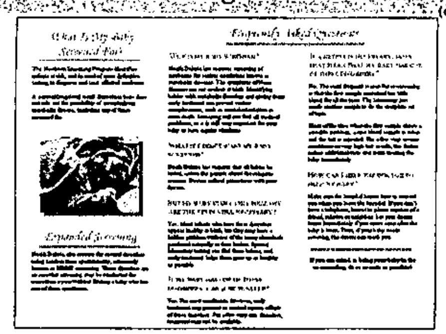
NORTH DAKOTA DEPARTMENT of HEALTH

Newborn Screening Brochure



NORTH DAKOTA DEPARTMENT of HEALTH

Newborn Screening Brochure (continued)



NORTH DAKOTA DEPARTMENT of HEALTH

Contact Information

Barb Schweitzer, Director
800-472-2286 (Press 1 for MCH)
701-328-4538 (Direct Line)
701-328-1412 (Fax)
Email: bschweit@state.nd.us

Dr. Alan Kenien
Pediatric Endocrinologist
MeritCare Medical Center
701-234-2431

Sara Copeland, MD
Assistant Professor in Pediatrics
Division of Medical Genetics
University of Iowa Hospitals and Clinics
Consultant to North Dakota
319-356-4783 (Office phone)
515-725-1630 #2 (Iowa Lab phone number)
515-725-1650 (Fax)
E-mail: sara-copeland@uiowa.edu

NORTH DAKOTA DEPARTMENT of HEALTH

Metabolic Disorders and Newborn Screening

Sara Copeland, MD
Assistant Professor University of Iowa
Department of Pediatrics, Division of Medical Genetics
Medical Consultant for Newborn Screening
4/29/05

Objectives

- Recognize signs and symptoms of metabolic disease
- Understand the strengths and weaknesses of newborn screening
- Know what labs are best for work-up of possible metabolic disorders

How to Break Metabolic Defects Down

- Problems of intoxication
  - Acute or progressive
  - Accumulation of toxic compounds
- Problems of energy metabolism
  - Symptoms from deficiency in energy production or utilization
- Problems of complex molecules
  - Disturbed synthesis or catabolism
  - Symptoms are permanent and progressive

Types of Metabolic Disorders Covered by Newborn Screening

- Intoxications
  - Amino acid disorders (some)
  - Organic acidurias
  - Some urea cycle defects
- Energy Metabolism Defects
  - Fatty acid oxidation defects
- NOT Identified on screening
  - Complex molecule defects
  - Glycogen metabolism defects
  - Congenital lactic acidosis
  - Mitochondrial defects

NOT "PKU" TEST!!!!!!

NEWBORN SCREENING TEST
NEWBORN SCREENING TEST
NEWBORN SCREENING TEST
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NEWBORN SCREENING TEST
NEWBORN SCREENING TEST
NEWBORN SCREENING TEST

NOT "PKU" TEST!!!!!!

Arginase def, argininosuccinic aciduria, citrullinemia, homocystinuria, hyperphenylalaninemias, tyrosinemia, beta-ketothiolase def, glutaric acidemia 1 & 2, isovaleric acidemia, malonic acidemia, MMA, PA, 2-methyl-3-hydroxy coA dehydrogenase def, 2-methylbutyryl CoA dehydrogenase def, HMG CoA lyase def, 3-methylcrotonyl CoA carboxylase deficiency, 3-methylglutaconyl CoA hydratase def, SCAD, MCAD, LCHAD, VLCAD, carnitine transport disorders CPT 1 & 2, hemoglobinopathies, congenital hypothyroidism, and congenital adrenal hyperplasia

Testing Performed in North Dakota

- Enzyme analysis for galactosemia and biotinidase deficiency
- Hemoglobinopathies
- Endocrine disorders
- Amino acid quanitation
- Acylcarnitine quanitation

- Coming soon-Cystic Fibrosis!!

Guthrie Bacterial Assay

- One Disease = One Blood Spot
- Run Time:
  - Prep
  - Overnight incubation
  - Read
- High False Positive Rate
- Antibiotic Interference
- COST: $1/specimen

Tandem Mass Spectrometry

- 25-30 Disorders: 1 blood spot
- Run Time: 1 – 2 minutes/specimen
- Said to be sensitive and specific
- Cost: $10/sample
- Tests for:
  - Amino acids
  - Acylcarnitines
- Interpretation is based on pattern analysis

Disorders Detected

- Acylcarnitines
  - Fatty Acid Oxidation Defects
  - Organic Acidurias
    - Elevations of various odd chain acylcarnitine reflect the ketoacid buildup.
- Amino Acids
  - Amino acidopathies
  - Urea cycle defects

Intoxications

- Aminoacidopathies
- Organic acidemias
- Urea cycle defects
- Sugar intolerances

Disorders with Build-up of Toxic Metabolites (Intoxications)

- Generally the enzyme block is somewhere in protein degradation and disposal
- Get build-up of ketoacids or nitrogen (ammonia)
  - These are the metabolites that cause the clinical problems.
- Catabolism or increased protein intake makes worse
- Often patients will self-select low protein foods once old enough to choose diet.

Energy Metabolism Defects

- Defects of making or breaking down glycogen
- Fatty acid oxidation defects*
- Krebs's cycle/ Mitochondrial respiratory-chain disorders
  - Congenital lactic acidemias

* Screened for on NBS

Defects of Energy Metabolism

- Unable to utilize normal resources of stored energy
- The problem can be anywhere in the ATP cycle
  - Fatty acid metabolism
  - Glucose metabolism
  - Protein metabolism
- Fasting or illness with increased energy needs can exacerbate disorder and bring on decompensation
- Hypoglycemia is usually the last sign, often will be lethargic prior to complete collapse.

Complex Molecule Defects

- **Lysosomal storage disorders**
- **Peroxisomal storage disorders**
- **Intracellular trafficking and processing defects**
  - Alpha-1-antitrypsin deficiency
  - Wilson's Disease
- **Congenital disorders of glycosylation**
- **Inborn errors of cholesterol synthesis**

Inborn Errors Of Metabolism

- Affect Approximately 1% of the Population
- Often Treatable But Outcome Depends on Early Diagnosis
- IEM Frequently Mimic Signs and Symptoms of Sepsis in Neonates
- Severe Forms Present in the First Few Days of Life

Follow-up

- Depending on the pattern, may just repeat the specimen or go directly to confirmatory studies
- Generally confirmatory studies will be either blood or urine tests depending on the pattern of elevations and the possible disorder.

Apocryphal Stories

- Parents look "OK"-testing unnecessary
- Mother's blood used to avoid 'hurting' baby
- Multiple birth: only need to test one baby
- Blood soaked on paper towels/tolet paper
- Several babys' blood applied to one filter paper
- Circles "filled" with red ink
- Test at discharge: baby 6 months old
- Specimens "dried" in microwave
- Specimen "FAXed" to laboratory

"THERE'S NO SUCH THING AS A 'FREE' SCREENING TEST"

Dr. Ed McCabe

Parent's Views

- Early diagnosis decreases anxiety
- Early diagnosis decreases morbidity and costs of seeking diagnosis
- Child's life is valuable even if treatment imperfect or child has disabilities
- Need to know diagnosis for family planning
- Parents want "EVERYTHING" screened for

NBS Infrastructure For a Successful Program

- Sub specialist Consultants
- Lab Technology Consultant
- Back-up Instruments
- Good Screening Practices
- Excellent Lab and Clinical Follow-up Practices
- **REASONABLE POLITICAL CLIMATE**

Lessons From Newborn Screening

- Death and mental retardation can be prevented
- Treatment for most disorders has improved
- Benefits of screening are often unknown in the beginning
- Long term outcome uncertain for years

Lesson From Newborn Screening

- Not a "simple" blood test
- Infrastructure must exist for early dx and long term treatment and evaluation
- "Benefits" of screening may not necessarily accrue to the individual
- Political agendas have a major impact on the quality and quantity of NBS

NBS Dilemmas

- We Expect to Find Every Affected Baby (perfect sensitivity) and Have Acceptable Specificity
- Sensitivity and Specificity is Still Unknown for Many of the Disorders (need time and cases)
- Great Need For Comparing Data, Identifying Problems, Establishing Incidence, and Long Term Outcome Measures

NBS Dilemmas

- Fatty acid oxidation disorders: hypoglycemia, liver disease, cardiomyopathy, SIDS
- Organic acidemias: acidosis, stroke, death
- Urea cycle defects: increased NH^3, cerebral edema, death

ALL CAN KILL OR MAIM IN THE FIRST WEEK OF LIFE

Early testing and rapid transit are critical!!!

US Recommendations

Current	Future
• Collect 24 HRS TO 7 days	• Collect 24-48 HRS
• Mail within 24 HRS	• Mail within 6-12 HRS
• Transit Time 24 HRS-10 days	• Transit Time =/< 24 HRS
• Testing 1-4 DAYS	• Testing 24 HRS
Age at diagnosis : 5 - 22 days	Age at diagnosis: 2.8 - 4.5 days

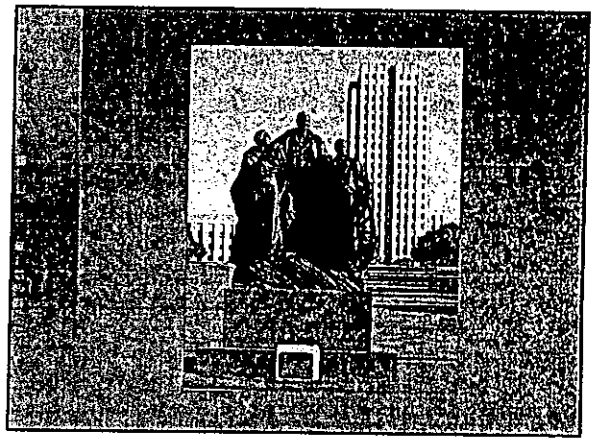
Practitioner Follow-Up Responsibilities

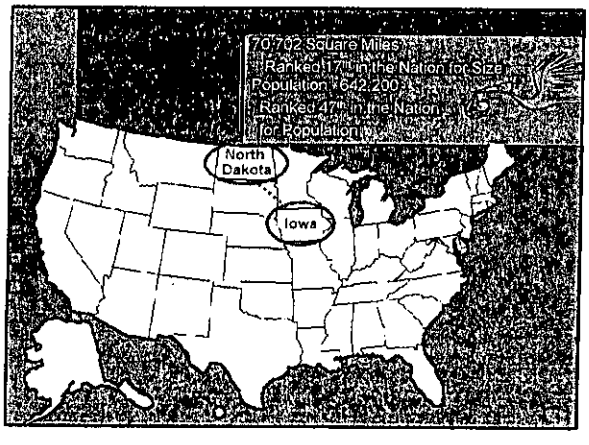
- Contact State Lab (515-243-0141) if results are not received in two weeks
- Prompt action on unsatisfactory or abnormal result
- Communication with consultants regarding infants with abnormal results
- Verify results for every infant

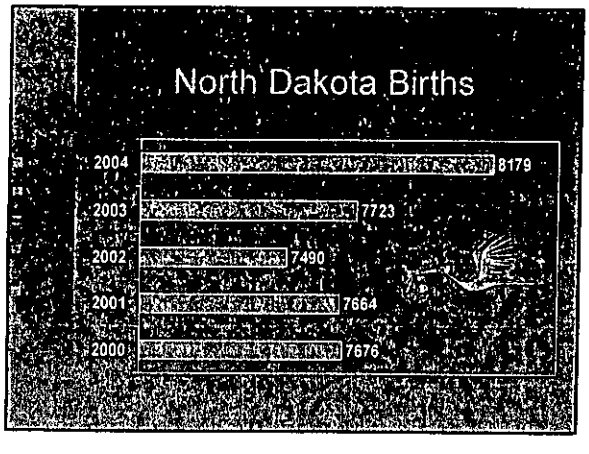
Contacts

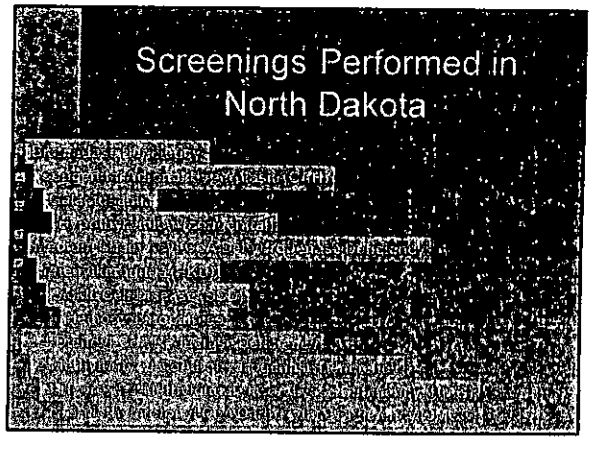
- E-mail: sara-copeland@uiowa.edu
  - Office phone: 319-356-4783
- NBS Lab phone number: 515-243-0141 #2
- NBS Coordinator- Barb Schweitzer
- Coordinator Office: 701-328-4538
- Other Team Members:
  - Cathy Pearce

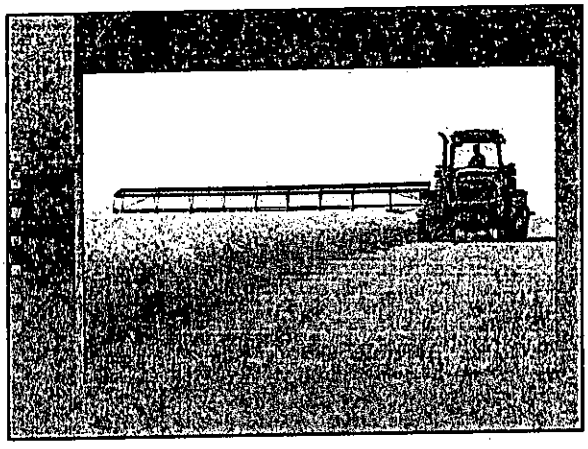
✲Assessing Coordination with the Iowa lab and ND Follow-up Staff
October 17, 2005
✲National NBS conference – Portland OR

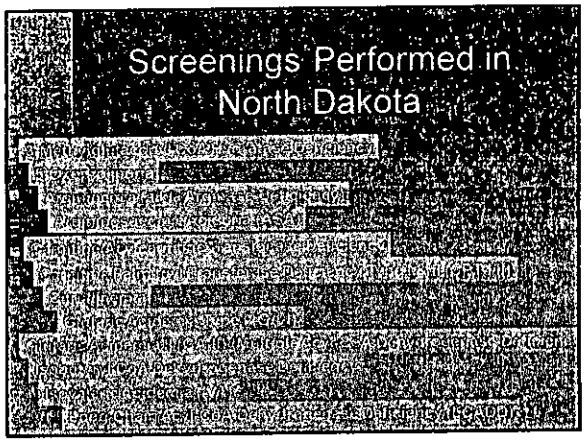


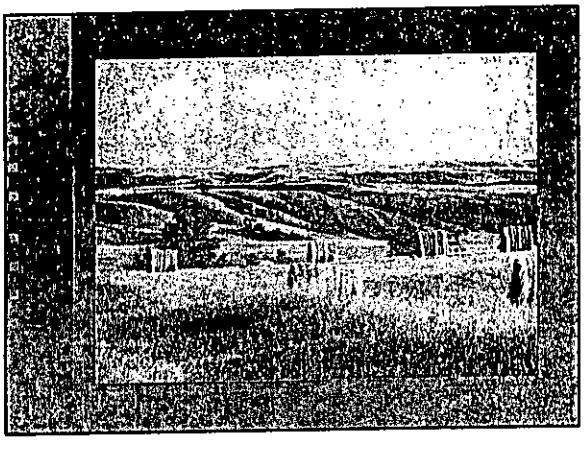


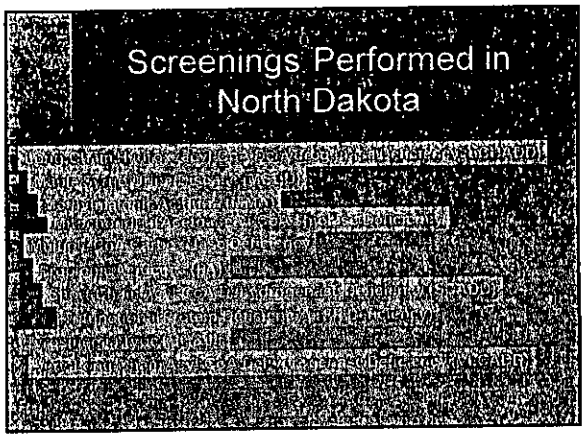


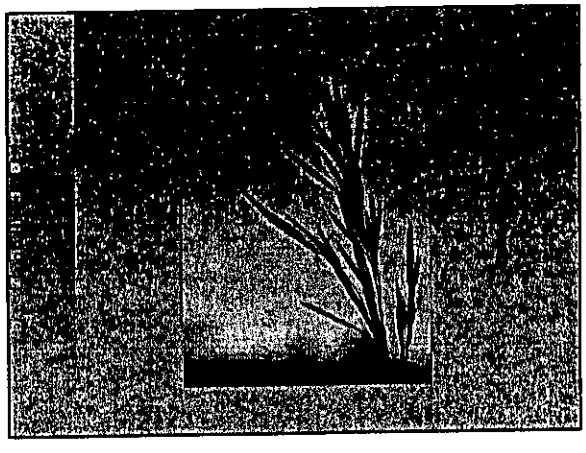


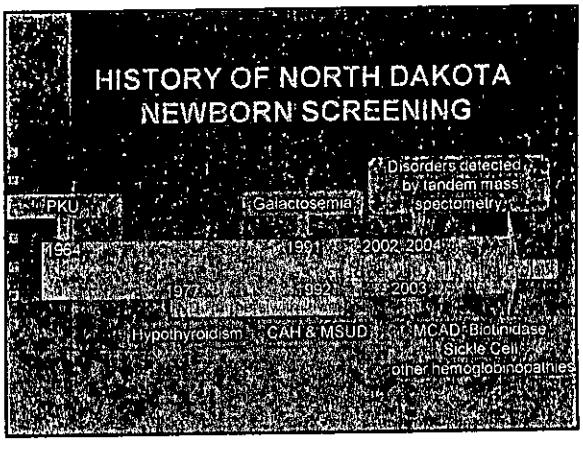


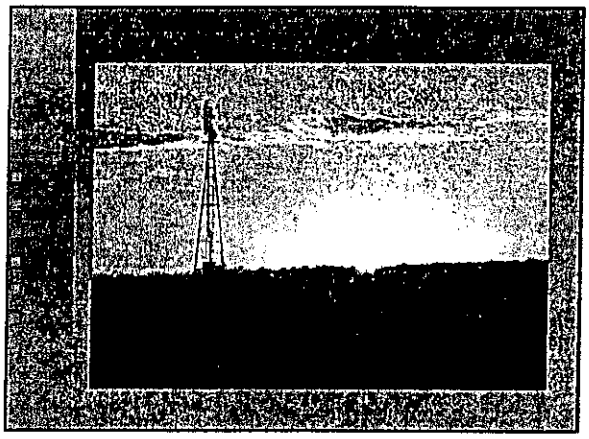

Screenings Performed in North Dakota




Screenings Performed in North Dakota

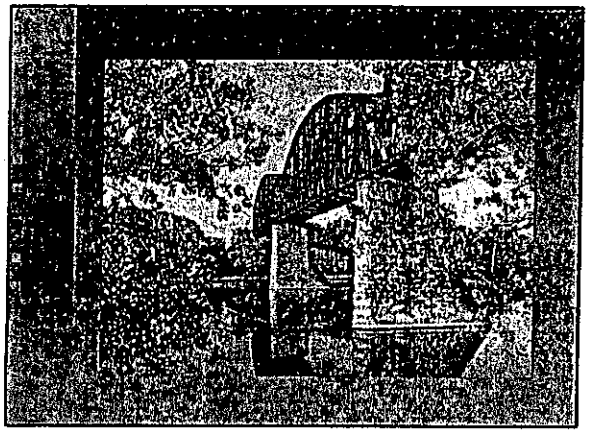



HISTORY OF NORTH DAKOTA NEWBORN SCREENING
Disorders detected by tandem mass spectometry:
PKU
Galactosemia
1954
1991
2002-2004
1977
1992
2003
Hypothyroidism
CAH & MSUD
MCAD, Biotinidase
Sickle Cell
other hemoglobinopathies



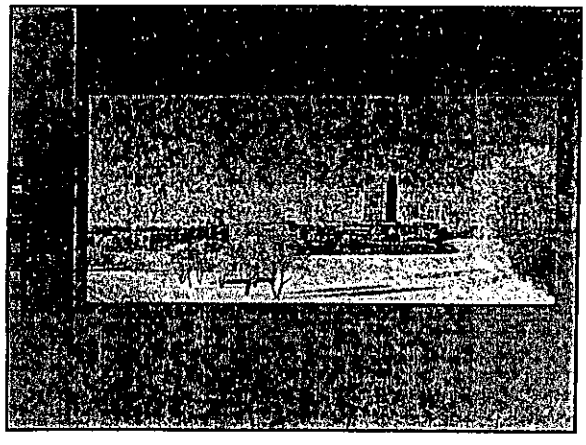
Resources

- Dr. John Martsoff, Medical Genetics
  University of North Dakota
  Grand Forks, ND
- Mary Riske, Clinical Nurse Geneticist
  University of North Dakota
  Grand Forks, ND
- Dr. William Rhead, Wisconsin
  November 2002 – August 2004

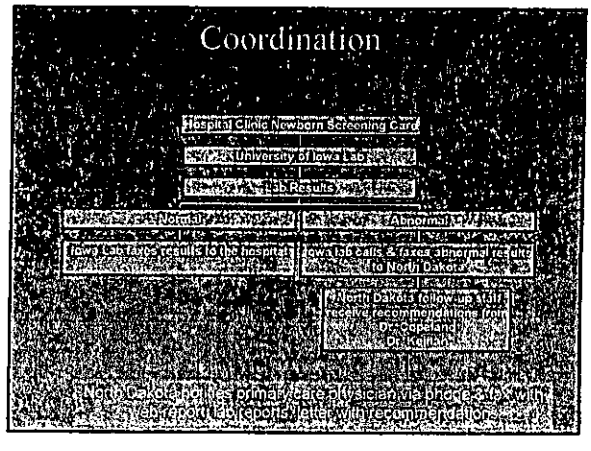


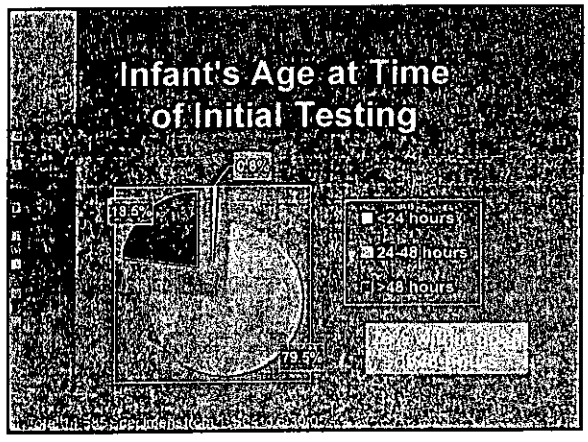
Resources

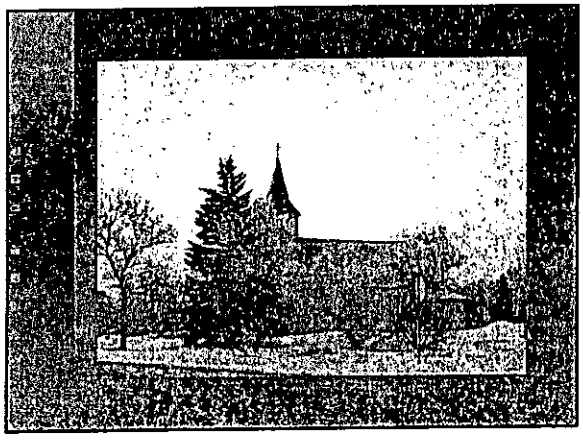
- Dr. Alan Kenien, Fargo, ND
  Pediatric Endocrinologist
  Conducts Metabolic Clinics 4 times a year, funded by Children's Special Health Services
- Dr. Sara Copeland, Iowa
  Assistant Professor in Pediatrics
  Medical Director of Newborn Screening for Iowa

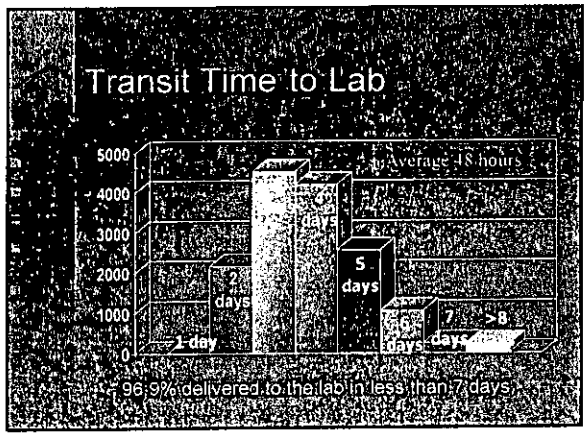


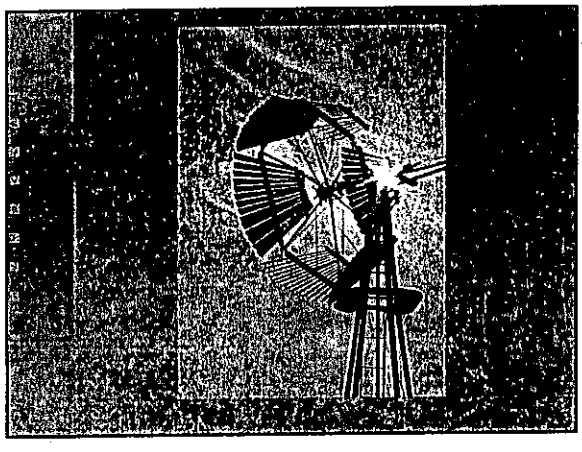
Coordination

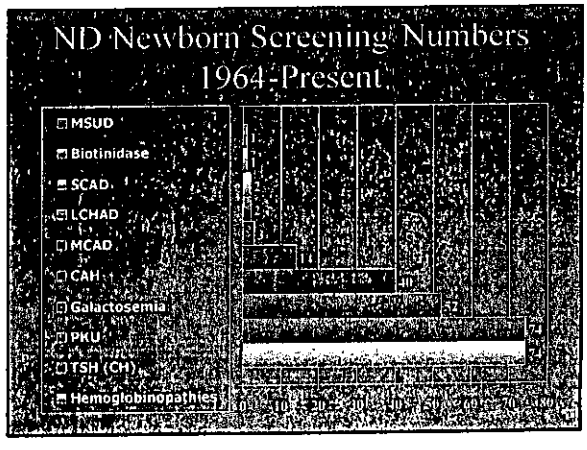


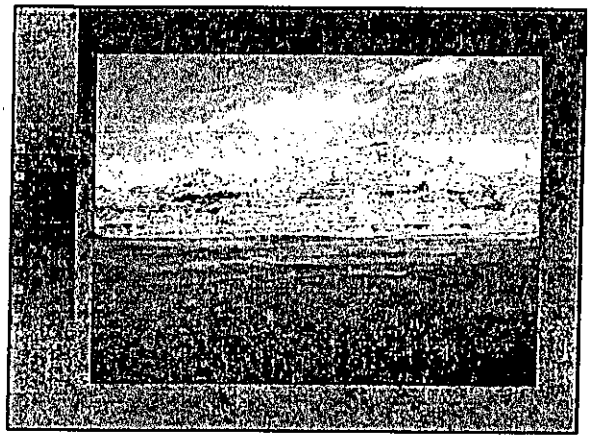

Infant's Age at Time of Initial Testing
19.5%
79.5%
24 hours
24-48 hours
>48 hours

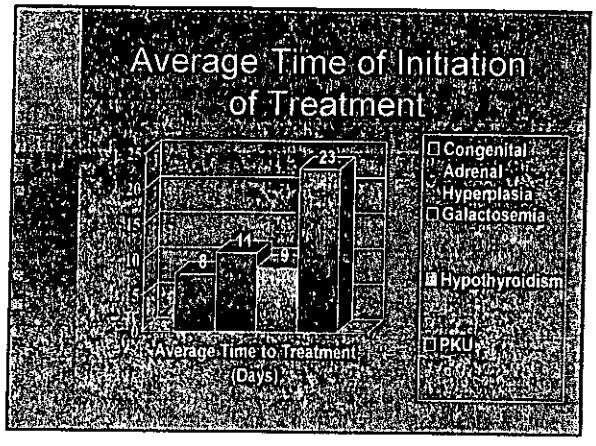



Transit Time to Lab
5000
4000
3000
2000
1000
0
Average 18 hours
2 days
5 days
7 days
>8 days
1 day
96.9% delivered to the lab in less than 7 days




ND Newborn Screening Numbers
1964-Present
MSUD
Biotinidase
SCAD
LCHAD
MCAD
CAH
Galactosemia
PKU
TSH (CH)
Hemoglobinopathies





Conclusions

The increased demand for expanded newborn screening suggests collaboration between states can serve as a model for other states.