Newborn Screening and Ethical Underpinnings

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November 17, 2010
History of Newborn Screening

- **Development:**
  - State-based public health service
  - Wilson and Jungner Criteria

- **Purpose:**
  - Early treatment can prevent serious complications
  - From healthy families and appear healthy at birth
  - “silent carriers”
1. The condition being screened for should be an important health problem
2. The natural history of the condition should be well understood
3. There should be a detectable early stage
4. Treatment at an early stage should be of more benefit than at a later stage
5. A suitable test should be devised for the early stage
6. The test should be acceptable to the population
7. Intervals for repeating the test should be determined
8. Adequate health service provision should be made for the extra clinical workload resulting from screening
9. The risks, both physical and psychological, should be less than the benefits
10. The costs should be balanced against the benefits

(World Health Organization 1968)
Newborn Screening in Texas

- The Newborn Screening Laboratory (NBS)
- Texas Newborn Screening Program
- 24-48 Hours after birth or hospital discharge
- One to two weeks of age
- An abnormal screen
- Follow-up with a health care provider for confirmatory testing
Texas NBS

- Texas currently screens for all 29 core panel disorders recommended by the American College of Medical Genetics (ACMG)
- The program expanded from 7 to 27 disorders with passage of HB 790 in 2005
- Required expansion of NBS program using the ACMG recommended panel as funds allowed
- Expanded screening began in December 2006
- Babies born in hospitals or birthing centers receive a hearing screen (or are referred for screening)
- ACMG Core Panel includes hearing screen
- Screening for Cystic Fibrosis was funded by the 81st Legislature
- Screening began December 1, 2009
Texas NBS Statistics

- Approximately 400,000 births annually
- DSHS screens approximately 800,000 newborn specimens annually
- In FY09, the NBS program followed-up on approximately 27,000 abnormal screens a year
- 700 disorders diagnosed each year
Disorders Included in the DSHS Newborn Screening Panel December 1, 2009

- **AMINO ACID DISORDERS:**
  - Argininosuccinic Acidemia (ASA)
  - Citrullinemia (CIT)
  - Homocystinuria (HCY)
  - Maple Syrup Urine Disease (MSUD)
  - Phenylketonuria (PKU)
  - Tyrosinemia Type I (TYRI)

- **FATTY ACID DISORDERS:**
  - Medium Chain Acyl-CoA Dehydrogenase Deficiency (MCAD)
  - Very Long Chain Acyl-CoA Dehydrogenase Deficiency (VLCAD)
  - Long Chain Hydroxyacyl-CoA Dehydrogenase Deficiency (LCHAD)
  - Trifunctional Protein Deficiency (TFP)
  - Carnitine Uptake Deficiency (CUD)
  - Carnitine Palmitoyl Transferase Deficiency1 (CPT1)

- **ORGANIC ACID DISORDERS:**
  - Glutaric Acidemia I (GA-I)
  - 3-OH 3-Methyl Glutaric Aciduria (HMG)
  - Isovaleric Acidemia (IVA)
  - Multiple Carboxylase Deficiency (MCD)
  - 3-Methylcrotonyl-CoA Carboxylase Deficiency (3-MCC)
  - Methylmalonic Acidemia (MMA)
  - Propionic Acidemia (PA)
  - Beta-Ketothiolase Deficiency (BKT)

- **CYSTIC FIBROSIS**

- **GALACTOSEMIA**

- **BIOTINIDASE DEFICIENCY**

- **ENDOCRINE DISORDERS:**
  - Congenital Hypothyroidism (CH)
  - Congenital Adrenal Hyperplasia (CAH)

- **HEMOGLOBINOPATHIES including:**
  - Hb S/S
  - Hb S/C
  - Hb S-Beta thalassemia
Ethical and Public Health Considerations

- Tandem mass spectrometry (MS/MS) to increase the number of disorders screened
- As many as 34 states are now instituting or preparing to institute expanded newborn screening
- Increasing complexity of technology and the increasing scope of disorders identified
- Even more important to maintain the public health perspective in these programs
Ethical Concerns

- Using Blood Spots for Research
  - Prior to 2009, a small percentage of newborn screening blood spots were used in research projects
  - A procedure was in place to review the significance, scientific merit and ethical integrity of all requests for blood spots for research
  - As appropriate, the researcher would then submit the study to the DSHS Institutional Review Board (IRB)
    - In the past, de-identified blood spots were exempt from IRB review
    - The main purpose of the DSHS IRB review is to ensure the safety, rights and welfare of human participants involved in the proposed research
    - If the study is approved, the researcher may then receive and use de-identified blood spots only as provided in the final IRB approval of the study
    - No identifying information has been released outside of the department to a researcher except when advance parental consent was obtained
HB 1672 made five changes to Chapter 33, Texas Health and Safety Code:

- Requires disclosure to parents, managing conservators or guardians that newborn screening specimens will be retained and may be used for other purposes

- Allows parents or adults to request destruction of their child’s or their own specimen(s)

- Provides confidentiality provisions for NBS data and specimens and defines acceptable uses

- Requires an interim legislative study of newborn screening in Texas

- Adds requirement to screen for sickle cell trait
Ethical Concerns: Potential Harms

- Unnecessary worry
- Children labeled as having serious health problems
- Insurability and employability
- Risk-benefit evaluation is inadequate
- Provide no demonstrable health benefit to the children while spending scarce resources
Informed Consent

- Ensure that parents understand the potential risk of refusing participation
- Improve rate of follow-up for children an abnormal screen
- Minimize the harm of false-positive results
- Allow parent the autonomy
- Documented informed consent has been controversial
2009 Texas Legislative Requirements

- 2009 Legislative requirements for Providers Concerning Newborn Screening
- Disclosure/Destruction Directive Form
- parents, legal guardians, or managing conservators
- Healthcare providers are **ONLY** required to provide the information by giving the form to the parent, legal guardian, or managing conservator
- Does **NOT** allow the parent, legal guardian, or managing conservator to choose to **NOT** have their child receive the newborn screen test
- Conflicts with their religious beliefs stemming from an established religion
Use and Storage of Newborn Screening Blood Spot Cards

What is newborn screening? The Texas Newborn Screening Program checks Texas babies for a list of serious medical conditions. These conditions can cause death or severe disability. Finding a medical problem during newborn screening can help prevent problems and may save your baby’s life.

How does your baby get screened? A small amount of your baby’s blood is placed on a special blood spot card. The blood spot card is sent to the state laboratory and tested.

What happens after the blood is tested? After testing, blood spot cards are safely stored by the Texas Department of State Health Services (DSHS) because they still have important public health uses. The main uses are 1) quality assurance/quality control, such as making sure that testing equipment continues to produce accurate newborn screening test results for Texas babies and 2) medical research [see Texas Health and Safety Code Sec. 35.91(a-4)] for a complete list of uses allowed by law. Specific information that could identify your child and connect him/her to a particular blood spot card is not allowed outside of DSHS without permission from the child’s parent, managing conservator, or legal guardian unless otherwise provided by law.

You can have your baby’s blood sample destroyed if you do not want it to be used after the newborn screening tests are completed. If you are okay with the sample being stored and used, as described above, there are no further steps for you to take.

If you want your baby’s blood sample to be destroyed, you must fill out ALL of the information on this form and send it back to DSHS at the address given below. DSHS will also accept the form from your healthcare provider, once you have completed and signed the form. If the newborn screening blood spot card is destroyed, the blood sample will not be available for any future needs you may have for the sample.

Directive to Destroy Newborn Screening Blood Spot Card Following Testing

To request to have your child’s blood spot(s) destroyed:
1. Fill out the form below ONLY if you want your baby’s blood sample destroyed after newborn testing is complete
2. Fill out the same attached form. Do not leave any fields blank.
3. Mail original to: Texas Department of State Health Services
   Newborn Screening Laboratory, MC 1647
   300 Box 14943-47
   Austin, Texas 78714-9437
4. For additional information, call (512) 453-7111 ext. 7332 or visit web site: http://www.dshs.state.tx.us/lab/newbornscreening.htm

I, _______________ (please print full name) hereby certify that I am the (check one) = parent, managing conservator, or legal guardian of the child named below, and I further certify that there is no court order in effect which restricts my legal ability to make this request.

As parent, managing conservator, or legal guardian, I am telling DSHS to destroy my child’s blood spot card(s) after the newborn screening testing is finished.

Full Name of Child: __________________________
Child’s Date of Birth: ________________________
Full Name of Mother: _________________________
Contact Information: _________________________
(Telephone number, e-mail address)

(Mailing address) ____________________________
(City, State, Zip) ____________________________

Check here □ to instruct DSHS to destroy NSB specimens from the child named above.

(Signature) ____________________________ (Date) ____________

I hereby certify under penalty of law that all the information I have provided herein is true and accurate. I understand that providing false information on this form constitutes a crime in Texas under Penal Code Sec. 37.10.

DSHS Laboratory Services Section
NBS Form EP14-12230S – May 2010
Use and Storage of Newborn Screening Blood Spot Cards

PARENT / MANAGING CONSERVATOR / LEGAL GUARDIAN
PLEASE READ CAREFULLY

What is newborn screening? The Texas Newborn Screening Program checks Texas babies for a list of serious medical conditions. These conditions can cause death or severe disability. Finding a medical problem during newborn screening can help prevent problems and may save your baby's life.

How does your baby get screened? A small amount of your baby's blood is placed on a special blood spot card. The blood spot card is sent to the state laboratory and tested.

What happens after the blood is tested? After testing, blood spot cards are safely stored by the Texas Department of State Health Services (DSHS), because they still have important public health uses. The main uses are: 1) quality assurance/quality control, such as making sure that testing equipment continues to produce accurate newborn screening test results for Texas babies, and 2) medical research (see Texas Health and Safety Code Sec. 33.017(b)-(d) for a complete list of uses allowed by law). Specific information that could identify your child and connect him/her to a particular blood spot card is not allowed outside of DSHS without permission from the child's parent, managing conservator, or legal guardian unless otherwise provided by law.

You can have your baby's blood sample destroyed if you do not want it to be used after the newborn screening tests are completed. If you are okay with the sample being stored and used, as described above, then there are no further steps for you to take.

If you want your baby's blood sample to be destroyed, YOU must fill out ALL of the information on this form and send it back to DSHS at the address given below. DSHS will also accept the form from your healthcare provider, once you have completed and signed the form. If the newborn screening blood spot card is destroyed, the blood sample will not be available for any future needs you may have for the sample.

To Request to have your child's blood spot(s) destroyed:

1. Fill out the entire attached form. Do not leave any fields blank.
2. Mail original to: Texas Department of State Health Services, Newborn Screening Laboratory, MC 14947, PO Box 149347, Austin, Texas 78714-9347
3. For additional information, call (512) 453-7111 ext. 7333 or visit web site: http://www.dshs.state.tx.us/lab/newborn/screening.shtml

DSHS Laboratory Services Section - NBS Form F14-15230 - February 2010

Directive to Destroy Newborn Screening Blood Spot Card Following Testing

Fill out this form ONLY if YOU WANT your baby's blood sample destroyed after newborn testing is complete.

I, _____________________________________________________________________________________(please print full name) hereby certify that I am the (check one) □ parent, □ managing conservator, or □ legal guardian of the child named below, and I further certify that there is no court order in effect which restricts my legal ability to make this request.

As parent, managing conservator, or legal guardian, I am telling DSHS to destroy my child's blood spot card(s) after the newborn screen testing is finished.

Full Name of Child: ____________________________
Child’s Date of Birth: ____________________________
Full Name of Mother: ____________________________
Contact Information: ____________________________
(Telephone number, e-mail address)
(Mailing address)
(City, State, Zip)
Check here □ to instruct DSHS to destroy NBS specimens from the child named above.

(Signature) ______________________ (Date) __________

I hereby certify under penalty of law that all the information I have provided herein is true and accurate. I understand that providing false information on this form constitutes a crime in Texas under Penal Code Sec. 37.09.
Texas NBS Lawsuit

- DSHS and Texas A&M University were sued in federal court regarding the State of Texas Newborn Screening Program
- Constitutionality of how the program operated under Texas law
- Storage and use of the newborn screening blood spots after newborn screening was completed
- Lawsuit against the Newborn Screening program was settled
- DSHS believes settling the lawsuit was in the best interest of the State of Texas
  - Preserves the NBS program’s core mission to screen all newborn babies for life-threatening disorders
  - Avoids cost of class action lawsuit
  - DSHS destroyed blood spot cards received by the department prior to May 27, 2009
  - HB 1672 was signed into law on this date
  - Blood spot destruction was completed on March 2, 2010
  - 5.3 million samples were destroyed
Question?

- Should specific consent be required for any use of blood spots in research (external, internal, etc.)?
Decision-Making

- Does not necessarily demonstrate informed decision making

- Few states have required informed consent for newborn screening

- Removing parents from the decision-making process for expanded newborn screening
Case Study

“My family lived the consequences of a false-positive newborn screening test result after the birth of our son in 2008”

- Baby Tested positive for glutaric acidemia type I, an organic acid disorder
- Baby placed on a strict feeding regimen; feeding every four hours of primarily breast milk or a special, low protein formula to prevent sudden and irreversible brain damage
- Preliminary negative results about five months later
- Advised to avoid daycare
- Follow-up tests cost without insurance discount, more than $9,500
- False Positive
- Neglectful parents?
- Parental rights
The End


References


