4050.0, "Genetics and Newborns Screening: Practice, Policy and Ethics"

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Using community based participatory approaches to provide genetics education

Community Based Participation

- The W.K. Kellogg Foundation's Community Health Scholars Program proposes that "community based participatory research in health is a collaborative approach to research that equitably involves all partners"

Community Based Participation

- "Participatory approaches are grounded in the assumption that knowledge that is connected to people's lived experience is more readily translated into action than knowledge that is disconnected from practice"

HRSA Initiatives for Families and General Public

- LSUHS - 7 **Things Parents Want to Know**
- March of Dimes - ***Community Genetics Education Network (CGEN)***
- Genetic Alliance - ***Family Health History***

Learning Objectives

- Recognize the ethical, legal, financial and social practice and public policy concerns regarding community engagement.
- Describe the concerns of families and other stakeholders in regards to lack of genetic education, services and input into policy development

Factors Limiting Participation

- Underserved and underrepresented families are coping with practical issues on a day to day basis.
- Practical issues often make it difficult for individuals and families to receive genetic services or participate in the development of educational materials.

NBS Communication Challenges

- New technology/rapidly changing environment
- State programs differ (disorders screened, info given, process of reporting results)
- Parents/ public lack basic knowledge
- Best practices yet to be identified

Hidden Barriers to Informing Parents about NBS

Patients/ providers/nurses/ state programs:

- Agendas/ communication styles/ knowledge level differ

Patients:

- Education/ Literacy/ Language
- Health Literacy:

  Capacity to

  - Obtain, process, understand basic health information and services
  - Make appropriate health care decisions (act on information)
  - Access/ navigate healthcare system

Is the layout user-friendly?

1. Is the layout user-friendly?

First impressions are important!

Does the pamphlet:

- Have ample white space?
- Limit paragraphs to 4 to 5 lines?
- Use bullets, boxes, indentation, bolding, vertical lists?
- Use bifold rather than trifold format?
- Use font that is 12 point or larger?
- Avoid use of ALL CAPS, *italics* and *specialty* fonts in large blocks of text?

Why does my baby need Newborn Screening tests?

Most babies are healthy when they are born.

We test all babies because a few babies look healthy but have a rare health problem.

If we find problems early, we can help prevent serious problems like mental retardation or death.

How will my baby be tested?

Before you leave the hospital, a nurse will take a few drops of blood from your baby's heel.

The hospital will send the blood sample to a newborn screening lab.

How will I get the results of the tests?

Parents are notified of test results if there is a problem.

Ask about results when you see your baby's doctor.

Do illustrations convey the message?

2. Do illustrations convey the message?

A picture may be worth a thousand words – but which thousand?

Are pictures and captions:

- Serving a purpose (they are not just decorative)?
- Clear and realistic?
- Familiar and likely to be understood?

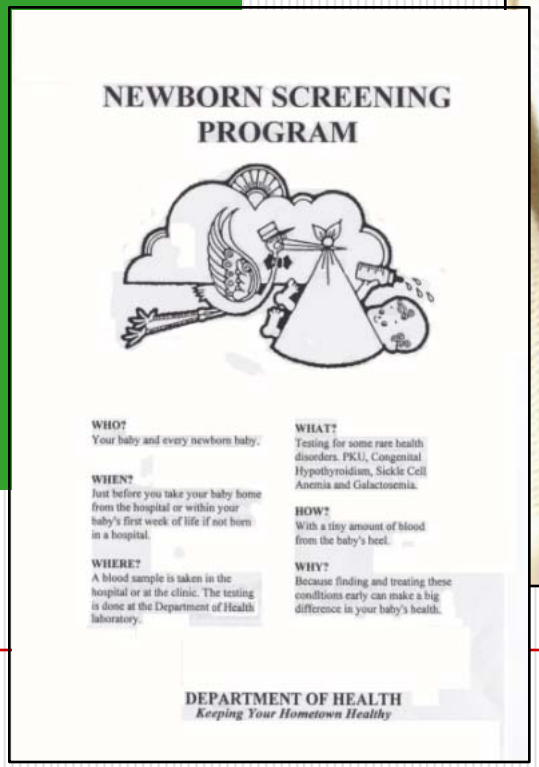
Is the message clear?

3. Is the message clear?

- Is the message obvious on the cover, title, and headings?
- Are key messages easy to pick out?
- Does pamphlet get to the point quickly?
- Does pamphlet easily inform the readers of what they need to know and do?

Does cover graphic:

- Target expectant parents?



NEWBORN SCREENING PROGRAM

WHO?
Your baby and every newborn baby.

WHEN?
Just before you take your baby home from the hospital or within your baby's first week of life if not born in a hospital.

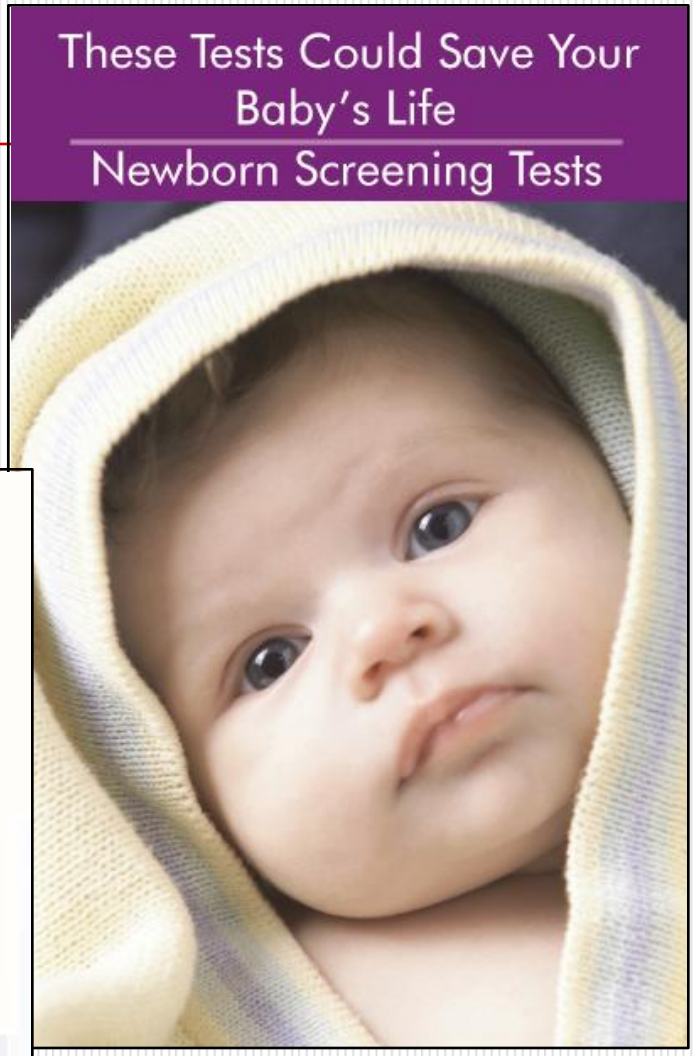
WHERE?
A blood sample is taken in the hospital or at the clinic. The testing is done at the Department of Health laboratory.

WHAT?
Testing for some rare health disorders. PKU, Congenital Hypothyroidism, Sickle Cell Anemia and Galactosemia.

HOW?
With a tiny amount of blood from the baby's heel.

WHY?
Because finding and treating these conditions early can make a big difference in your baby's health.

DEPARTMENT OF HEALTH
Keeping Your Hometown Healthy



Is the information manageable?

4. Is the information manageable?

Does the pamphlet:

- Focus on "need to know" rather than "nice to know"?
- Stick to a few key messages to avoid information overload?
- Limit the use of graphs and statistics?

Why does my baby need to be screened?

Routine newborn screening can determine if your baby has any of the following conditions: PKU (Phenylketonuria), Hypothyroidism, Galactosemia, Sickle Cell Disease or CAH (Congenital Adrenal Hyperplasia). These are rare, but serious conditions which can cause brain damage or even death if not treated. Even if your baby looks healthy, he or she may have one of these conditions. If any of these conditions go untreated, serious problems will arise. Therefore, (state) law requires that all newborn babies be tested. The blood tests will identify babies who need more testing, counseling and treatment. It is critical to detect these conditions as soon as possible. A few days or weeks could make the difference between life and death or disability.

(College Reading Level)

Why does my baby need Newborn Screening tests?

* Most babies are healthy when they are born.
* A few babies look healthy but have a rare health problem.
* Babies who are born with these diseases seem normal at birth.
* We test all babies to find the ones who may need treatment.
* If we find problems early, we can help prevent serious problems like mental retardation or death.

(7th Grade Reading Level)

"Meant for Me"

5. Does the pamphlet make the reader feel "this information is meant for me"?

Does the pamphlet:

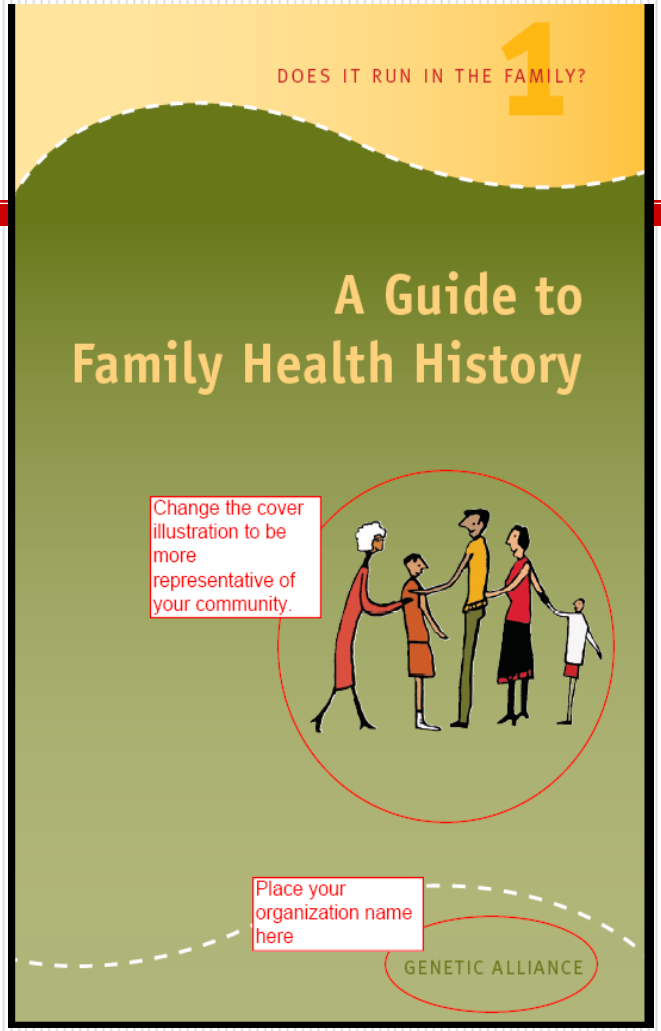
- Use a personal, conversational tone rather than a textbook or bureaucratic one?
- Foucs on parent rather than on the NBS program?
- Use familiar words, situations and pictures?
- Address the reader; personalize information ("*your baby*" not "*the baby*")?
- Show cultural sensitivity?

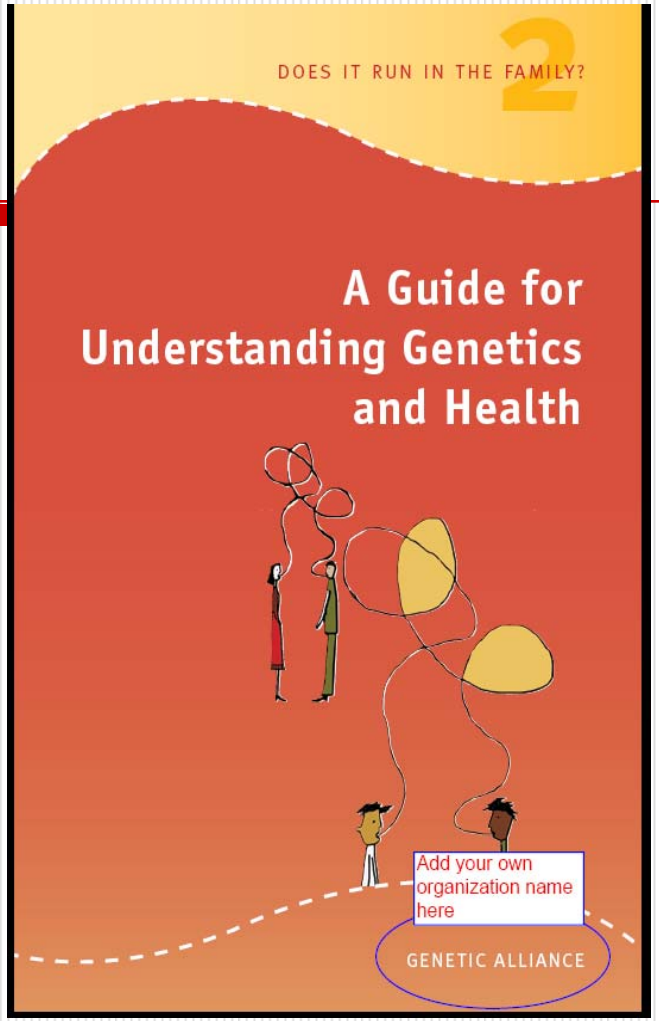
Newborn Screening is offered to families with new babies as a service through the Department of Health. The initial screening tests are performed by the Department of General Services, Division of Consolidated Laboratory Services (DCLS) which is located in (city). DCLS also performs repeat tests on infants up to six months of age. This service makes it possible to find out whether newborn babies might have disorders that may result in serious problems if treatment is not started soon after birth. Every infant in (state) is tested a few days after birth unless a parent or guardian objects on the grounds that the test conflicts with their religious practices.

How will my baby be tested?

* *Before you leave the hospital, a nurse will take a few drops of blood from your baby's heel.*
* *The hospital will send the blood sample to a newborn screening lab.*

Family Health History





Family Health History

Supplemental Materials

Healthcare Provider Card

Purpose: This card is for the individual to fill out and bring to their healthcare provider.

- One side of the card concentrates on concerns about family health history.
- On the other side, there is information for the provider on how to best use the family history to determine the patient's risk of getting a disease.
- Intermountain Health Care is currently conducting interviews with primary care providers to determine what types of information they want.

Healthcare Provider Card

Summary of family health history

I am concerned about my family history of: (check all that apply)

Health Concerns/Risk Factors

- ☐ Heart disease or heart attack
- ☐ Stroke
- ☐ Diabetes/sugar disease
- ☐ High blood pressure
- ☐ High cholesterol
- ☐ Asthma
- ☐ Hearing loss at young age
- ☐ Vision loss at young age
- ☐ Breast cancer
- ☐ Ovarian cancer
- ☐ Colon cancer
- ☐ Endometrial(uterine) cancer
- ☐ Other cancer:_____
- ☐ Mental health:_____
- ☐ Mental retardation/developmental delay
- ☐ Alzheimer disease/dementia
- ☐ Genetic conditions:_____

Prenatal Concerns

- ☐ Birth defects
- ☐ Miscarriage/stillbirth
- ☐ Genetic conditions:_____

Identify family members with each condition checked, including age of diagnosis, current age or age at death and cause of death (use extra sheets if needed)

Relationship	Condition	Age of onset	Current age	Age, cause at death
Example:				
Brother	*High Blood Pressure*	*35*	*45*	
Mother	*High Blood Pressure*	*40*		*65, Stroke*

Please include information about your children, your brothers and sisters, mother,(mother's side: aunts, uncles, grandparents), father,(father's side: aunts, uncles, grandparent)

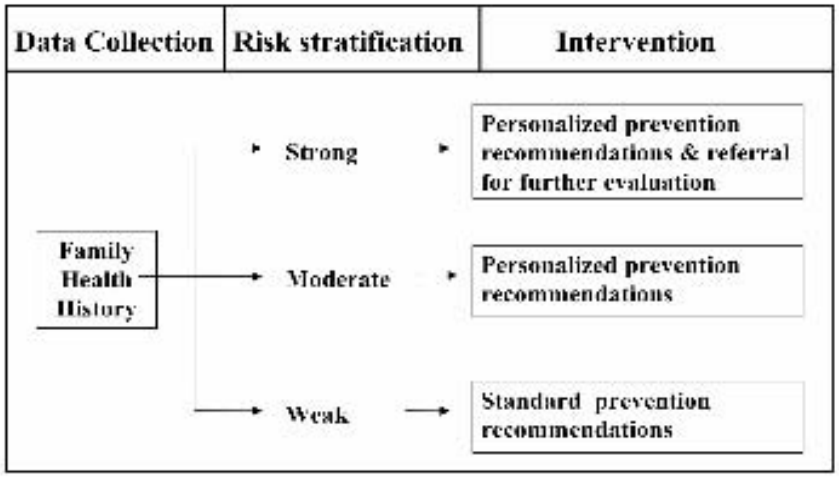
Funded in part by a grant (U33 MC06836) from the Maternal and Child Health Bureau, Health Resources and Services Administration.

DOES IT RUN IN THE FAMILY

Resources for the Provider

Recognizing Family Risk (Genetic Red Flags)

- Family history of known genetic disorder
- Multiple affected family members with same or related disorders
- Earlier age at onset of disease than expected
  - Breast, ovarian and endometrial cancer < 50 yrs (pre-menopausal)
  - Colon and prostate cancer < 50 yrs
  - Stroke and noninsulin-dependent diabetes < 50 yrs
  - Dementia < 60 yrs
  - Coronary artery disease < 55 yrs males and < 65 yrs in females
- Sudden cardiac death in a person who seemed healthy
- Multifocal or bilateral occurrence in paired organs
- Ethnic predisposition to certain genetic disorders



General Guidelines for Risk Stratification

High Risk

1. Premature disease in a 1st-degree relatives, (sibling, parent or child)
2. Premature disease in a 2nd-degree relative (CAD only)
3. Two affected 1st-degree relatives
4. One 1st-degree relative with late or unknown disease onset and an affected 2nd-degree relative with premature disease from the same lineage
5. Two 2nd-degree maternal or paternal relatives with at least one having premature onset of disease
6. Three or more affected maternal or paternal relatives
7. Presence of a "moderate risk" family history on both sides of the pedigree

Moderate risk:

1. One 1st-degree relative with late or unknown onset of disease
2. Two 2nd-degree relatives from the same lineage with late or unknown disease onset

Average risk:

1. No affected relatives
2. Only one affected 2nd-degree relative from one or both sides of the pedigree
3. No known family history
4. Adopted person with unknown family history

Scheuner et al., Am J Med Genet 1997; 71:315-324

Family History Website Resources

1. CDC – www.cdc.gov/genomics/famhistory/famhist.htm
2. AAFP Genomics CME – www.aafp.org
3. U.S. Surgeon General – www.hhs.gov/familyhistory/
4. Genetic Alliance – www.geneticalliance.org/familyhealthhistory
5. Recent Literature – www.geneticalliance.org/fhh.literature

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DOES IT RUN IN THE FAMILY?

Components for community engagement

- Trust
- Message resonates with community
- Constant communication
- Technical assistance- as needed
- Understanding of differences

Contact Information

- For questions please contact
- Penny Kyler Pkyler@hrsa.gov

- *Thank You*