



# Advisory Committee on Heritable Disorders and Genetic Diseases in Newborns and Children

September 2007

Health Resources And Services Administration  
Maternal And Child Health Bureau

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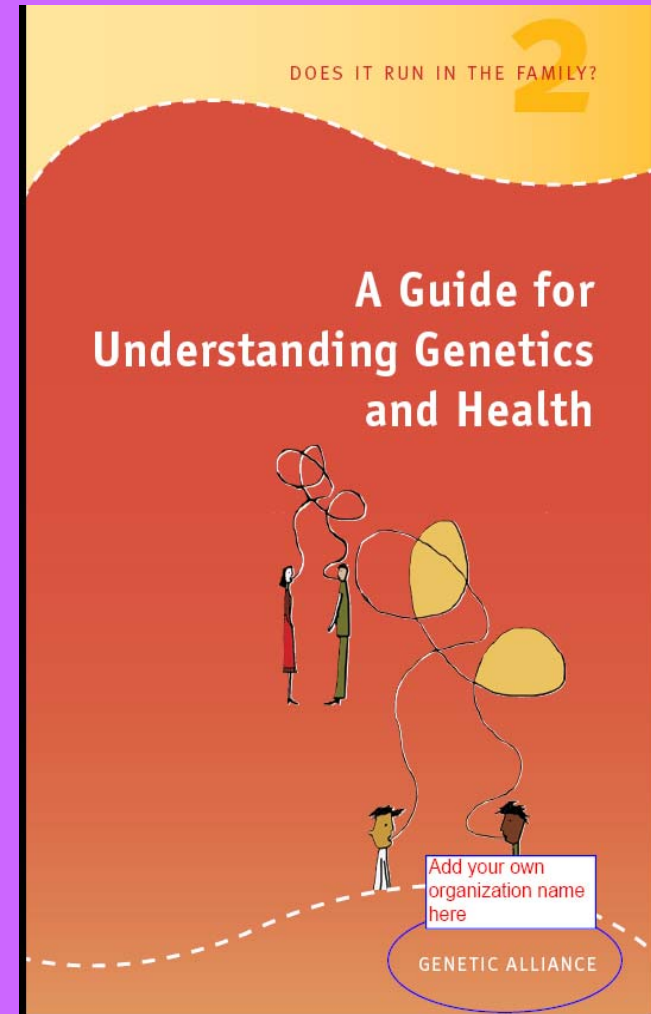
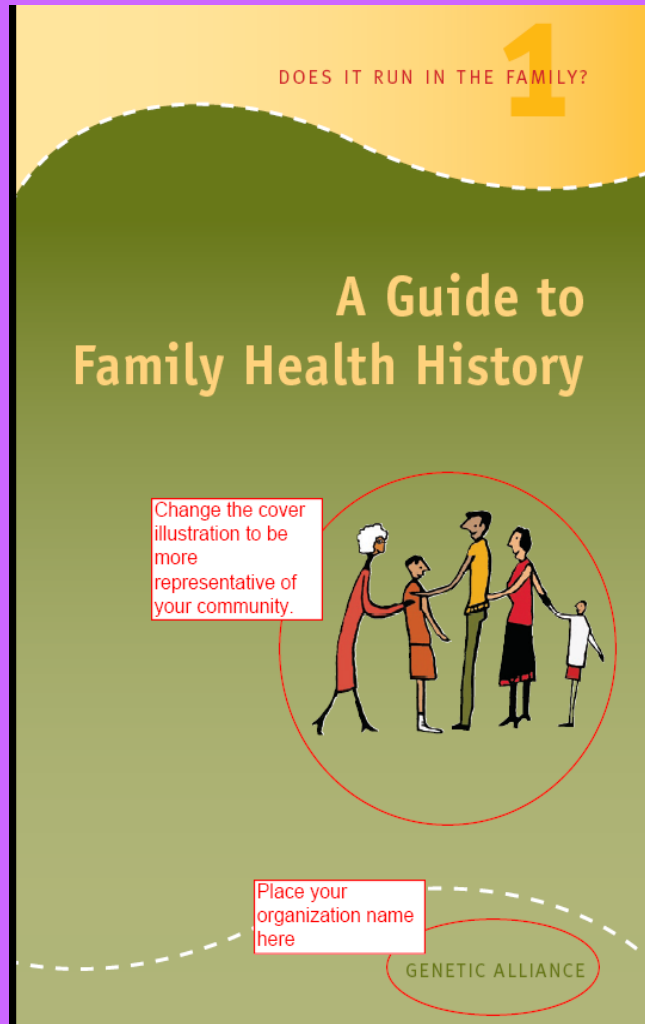


## Present Initiatives for Families and General Public

- Genetic Alliance - *Family Health History*
- March of Dimes - *Community Genetics Education Network (CGEN)*
- Genetic Alliance/ University of Maryland and Genetics & Public Policy, Iowa Department of Health/ University of Iowa and Hawaii Health Department - *Screening for Heritable Disorders in Children: Efficacy from a Family/Consumer Perspective*
- MCHB - **Family to Family Health Information Centers**



# 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

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

Prenatal Concerns

- |  |   |
|--|---|
| <input type="checkbox"/> Birth defects             | <input type="checkbox"/> Miscarriage/stillbirth |
| <input type="checkbox"/> 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
<i>Example:</i>				
<i>Brother</i>	<i>High Blood Pressure</i>	<i>35</i>	<i>45</i>	
<i>Mother</i>	<i>High Blood Pressure</i>	<i>40</i>		<i>65, Stroke</i>

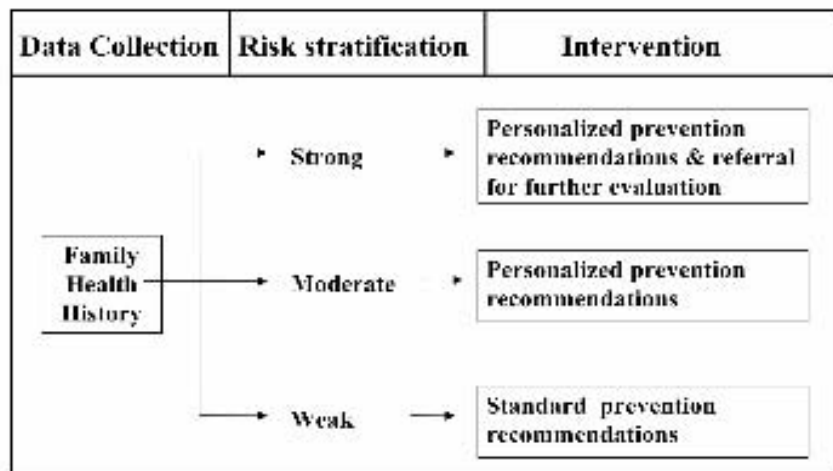
*Please include information about your children, your brothers and sisters, mother, (mother's side: aunts, uncles, grandparents), father, (father's side: aunts, uncles, grandparent)*

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

# 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 (premenopausal)
  - 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 1<sup>st</sup>-degree relatives, (sibling, parent or child)
2. Premature disease in a 2<sup>nd</sup>-degree relative (CAD only)
3. Two affected 1<sup>st</sup>-degree relatives
4. One 1<sup>st</sup>-degree relative with late or unknown disease onset and an affected 2<sup>nd</sup>-degree relative with premature disease from the same lineage
5. Two 2<sup>nd</sup>-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 1<sup>st</sup>-degree relative with late or unknown onset of disease
2. Two 2<sup>nd</sup>-degree relatives from the same lineage with late or unknown disease onset

### Average risk:

1. No affected relatives
2. Only one affected 2<sup>nd</sup>-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](http://www.cdc.gov/genomics/famhistory/famhist.htm)
2. AAFP Genomics CME – [www.aafp.org](http://www.aafp.org)
3. U.S. Surgeon General – [www.hhs.gov/familyhistory/](http://www.hhs.gov/familyhistory/)
4. Genetic Alliance – [www.geneticalliance.org/familyhealthhistory/](http://www.geneticalliance.org/familyhealthhistory/)
5. Recent Literature – [www.geneticalliance.org/fhh.literature](http://www.geneticalliance.org/fhh.literature)

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# Community Genetics Educational Network (CGEN)

- Working with 4 communities on projects to increase genetics knowledge.
- The communities are in New York, Utah and Washington DC and include individuals representing:
  - Asian-American including Chinese, Korean and Vietnamese
  - African-American and West African immigrant communities
  - Latin American community including Dominican, Puerto Rican, South American and Afro-Caribbean
  - Mexican American and Native American



# Community Genetics Educational Network (CGEN)

Charles B. Wang CHC has produced 4 bilingual brochures:

- *The Benefits of Genetic Testing & Counseling For You & Your Family*
- *Amniocentesis: How It Can Help You & Your Baby*
- *Understanding Thalassemia: How It Affects You & Your Baby*
- *Maternal Serum-Triple Screen: A Test That Lets You Know About the Risks Your Baby May Face*





# Community Genetics Educational Network (CGEN)

## **Family Genetics Education Through School and Community Partnerships in Utah**

Hispanic/Latino Community Advisory Committee

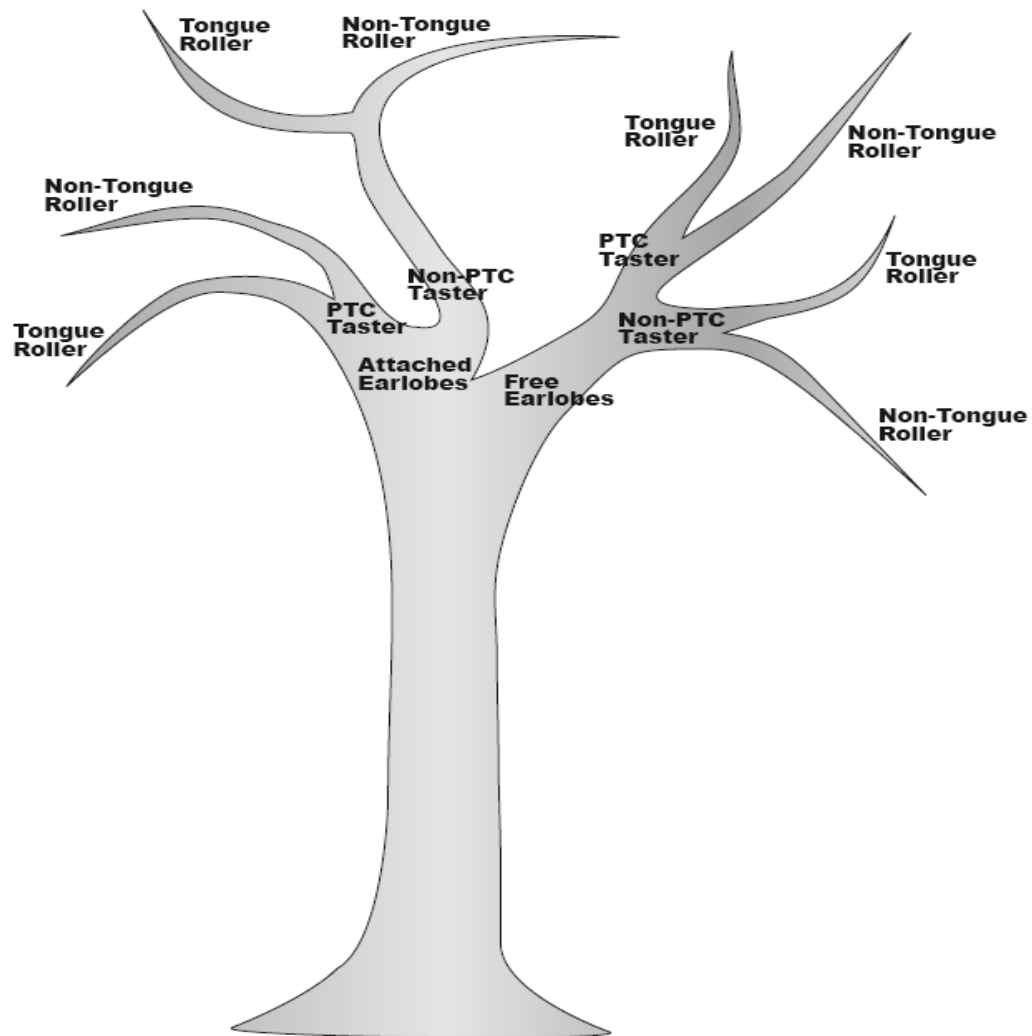
### **Project Goals**

1. To teach students in grades 5-10 and their families about genetics. And to teach them about the role of genetics in diseases such as diabetes, cancer and heart disease.
2. To encourage families to fill out a family health history. This will help them learn their family's risk of getting these diseases. They also will learn what they can do to reduce their risk.



## Teacher References: A Tree of Genetic Traits

### B. Example Trait Tree



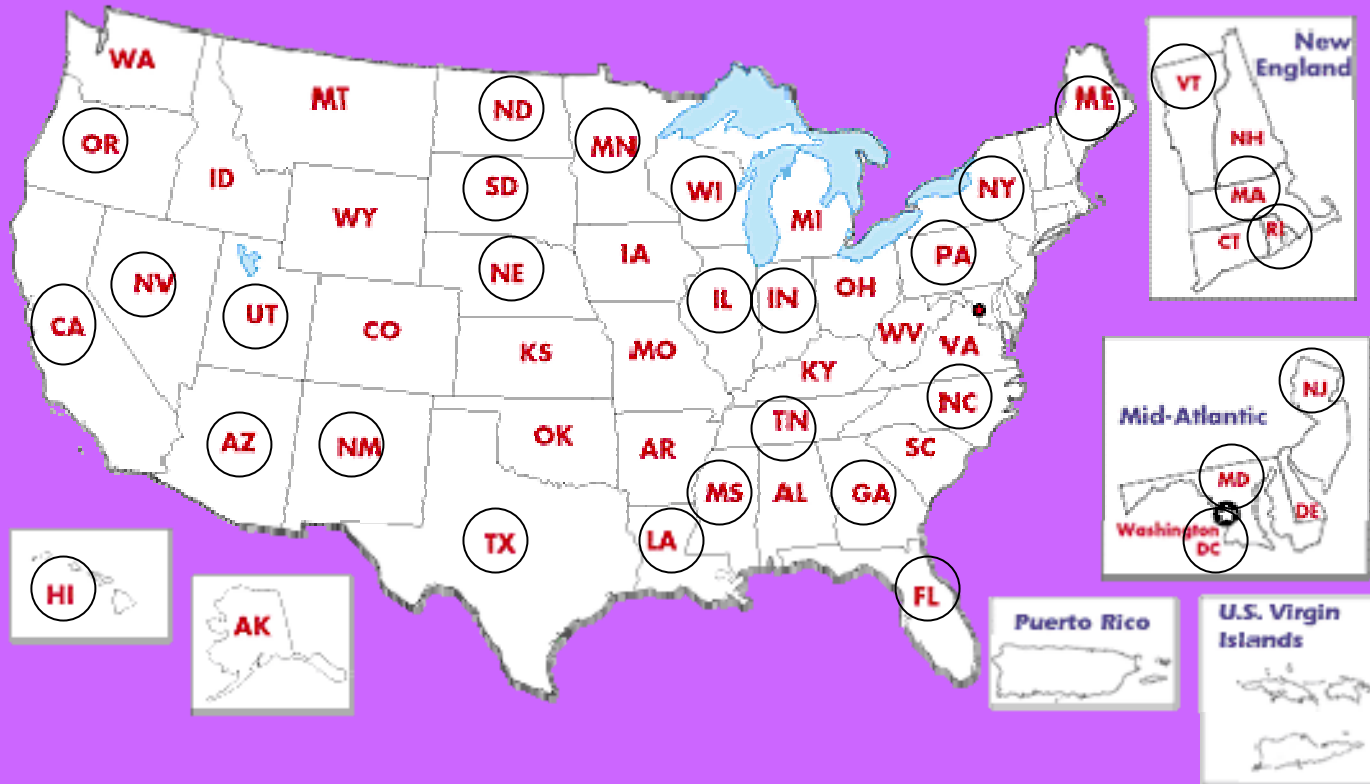
Adapted from "State Your Traits - Genetic Traits Tree", the GENETICS Project, University of Washington (2001).



# Family to Family Health Information Centers

- **Family-to-Family Health Information Centers:**
  - Provide information to families and providers regarding the health care needs of and resources available for CSHCN;
  - Assist families of children with special health care needs (CSHCN) make informed choices;
  - Develop partnerships with providers, managed care organizations, health care purchasers, and appropriate State agencies – models;
  - Provide training and guidance regarding the care of CSHCN; and
  - Are staffed by families with CSHCN.

# Currently Funded Centers



**30 Centers are funded through 2010. In all 50 states and District of Columbia by 2009**



# National Newborn Screening and Genetics Resource Center

## Some of the NNSGRC Activities:

- A point of contact for newborn screening (NBS) accessible to all (telephone, website, listservs)
- Provide expert consultative services to NBS programs who might request it (telephone consultation, expert review teams, reports and recommendations)
- Collect and report national NBS data for program evaluation (National Newborn Screening Information System – NNSIS: cases detected, births, presumptive positive tests, unsatisfactory specimens, etc.)
- Provide input into issues of national and regional importance (meetings of experts, white papers, funds for small projects, etc.)



# Regional Genetics and Newborn Screening Service Collaboratives

## 5 Year Grant Awards

- Heritable Disorders Program – the grant program
  - 1 National Coordinating Center (NCC)
  - 7 Regional Collaboratives (RCs)
- Project Period: 06/01/07 – 5/31/12
- RC Base Funding of \$500,000
- RC Additional Priority Activities (\$250,000/priority project)
  - Laboratory Performance Priority Activity 1
  - LTFU Priority Activity 2
- Cooperative Agreement
- Visit the Website: <http://www.nccrcg.org>.



# Regional Genetics and Newborn Screening Service Collaboratives

## National Coordinating Center (NCC)\*

1. Facilitate, coordinate and evaluate the implementation of activities carried out by the Regional Genetic and Newborn Screening Service Collaboratives (RCs).
2. Serve as the bridge between the RCs and MCHB; consumer organizations; health care professionals and professional organizations; researchers and research entities (including the NIH] and private entities); State, regional, and national public health organizations; and policy makers.
3. Work with the RCs and other partners to identify, prioritize, and address issues of importance regarding access to and utilization of genetic services at the national, State, and community levels.

\*Housed at the American College of Medical Genetics

## RC Primary Goal

- Ensure that children with heritable disorders and their families have access to quality care and appropriate genetic expertise and information in the context of a medical home that provides accessible, family-centered, continuous, comprehensive, coordinated, compassionate, and culturally effective care.



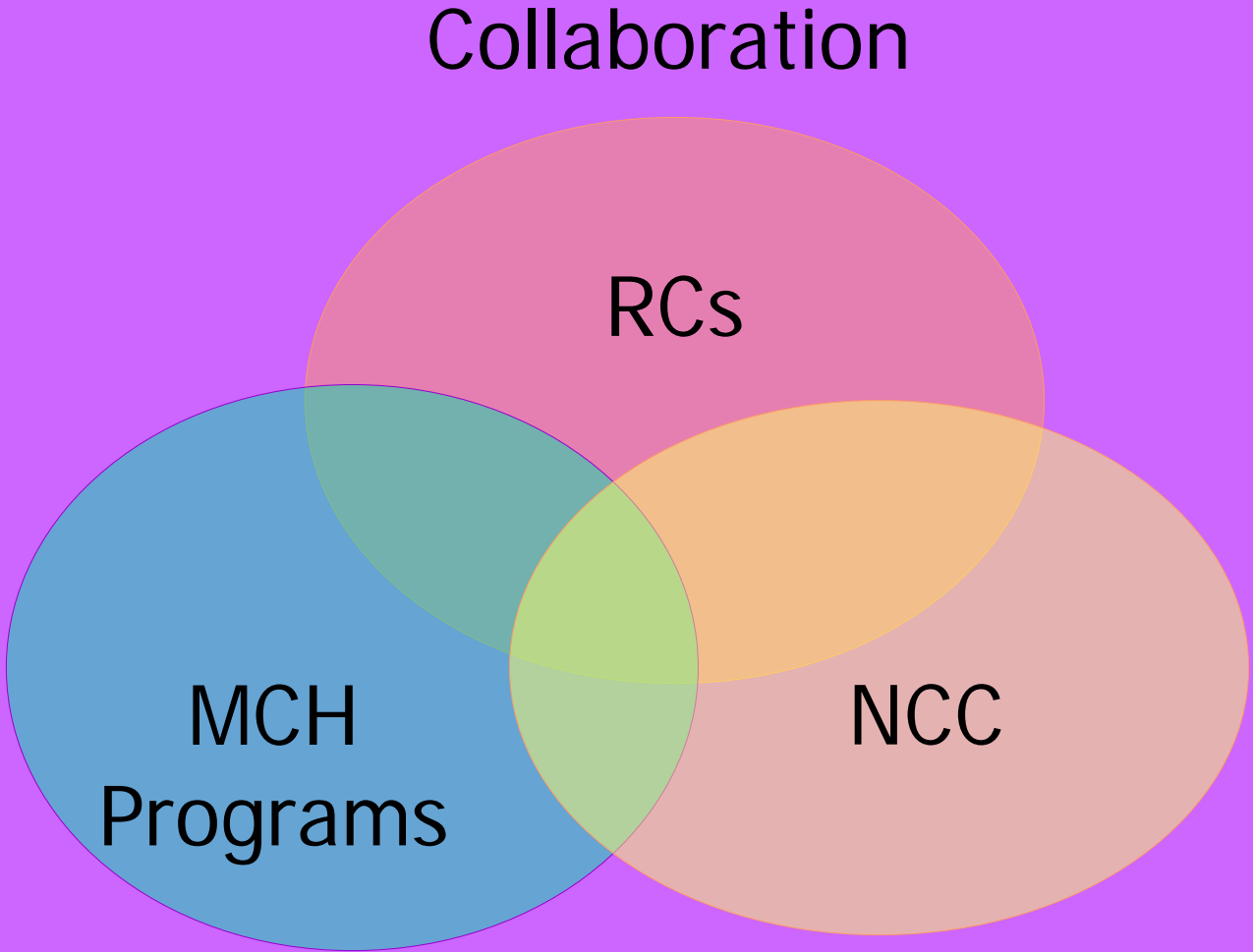


# Regional Genetics and Newborn Screening Service Collaboratives

## RC Two Objectives

- To strengthen communication and collaboration among public health, individuals, families, primary care providers, and genetic medicine and other subspecialty providers.
- To quantitatively and qualitatively evaluate outcomes of projects undertaken to accomplish their goals.

# Regional Genetics and Newborn Screening Service Collaboratives



Collaboration

RCs

MCH  
Programs

NCC



# Regional Genetics and Newborn Screening Service Collaboratives

## RC Activities

- Developing:
  - partnerships with primary care providers, individuals and families, and public health programs.
  - practice models to facilitate coordination between genetic services providers and nongenetic health services professionals.
  - telemedicine capacity and use of other distance learning technologies.
  - education, communication, marketing and dissemination activities
- Working toward regional NBS State panel standardization and expansion, and testing harmonization.
- Establishing NBS Emergency Preparedness Plans.



# Regional Genetics and Newborn Screening Service Collaboratives

## Laboratory Quality Assurance Priority Activity 1

- Undertake specific NBS public health laboratory quality-improvement projects such as enhancing NBS analytical laboratory test performance across the country and focusing on existing technologies currently in use in State NBS programs such as tandem mass spectrometry.
- Primary outcomes should be - harmonization of case definitions of disorders screened in NBS programs, NBS panels, and testing methodologies and decreasing the number of false positives.
- RC's funded:
  - Region 4 Great Lakes RC
  - Southeastern RC
  - New England RC
  - Mountain States RC



# Regional Genetics and Newborn Screening Service Collaboratives

## Follow-up Priority Activity 2

- Participate in collaborative study and health information technology and information exchange activities including the creation and use of regional and national information systems designed to: monitor health outcomes of infants and children identified with heritable disorders in NBS programs; evaluate NBS performance; and evaluate treatment protocols.
- Reflect collaborative activities between the public health NBS program and the service delivery system. Build on existing child health information systems activities in the region. Address issues of informed consent and family acceptance of screening and treatment.
- RCs funded:
  - Region 4 Great Lakes RC
  - Southeastern RC
  - New England RC



# Regional Genetics and Newborn Screening Service Collaboratives

## VISION – For the Next 5 Years!

- All Health care and public health professionals will have genetic resources readily accessible and will understand what it means to “think genetically”.
- Healthcare and public health professionals will know that their regional collaborative is the “go to place” for information about genetic resources and services within the region.



# Medical Home: A Core Component of Care

Growing interest in the medical home concept for comprehensive, coordinated care.

- *Commonwealth Fund Health Care Quality Survey* When adults have health insurance coverage and a medical home defined as a health care setting that provides patients with timely, well-organized care, and enhanced access to providers, racial and ethnic disparities in access and quality are reduced or even eliminated (2006).
- *Physician Practice Connections (PPC), Patient-Centered Medical Home (PCMH) Standards*. Being developed by NCQA, with collaboration with ACP, AAFP, AAP, and AOA, to assess PCMH. (2007).



# Medical Home: A Core Component of Care

- *Joint Principles of the Patient-Centered Medical Home* (PCMH). The Patient-Centered Medical Home is an approach to providing comprehensive primary care for children, youth, and adults. It is a health care setting that facilitates partnerships between individual patients and their personal physicians, and when appropriate, the patient's family ( AAFP, AAP, ACP, AOA, 2007)
- Medicare Medical Home Demonstration (*Tax Relief and Health Care Act of 2006—Sec. 204*)  
...redesign the health care delivery system to provide targeted, accessible, continuous and coordinated, family-centered care to high-need populations.





# Contact Information

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