



**MSGRCC**  
**NBS Registry**  
**Project**  
**Overview**

**Jeffrey R. Botkin, MD**

University of Utah

# NBS Registry

- Regional activities funded by Mountain States through HRSA funds
- Background
  - ❖ Rapid expansion in NBS services
  - ❖ Assessment of longer-term outcomes necessary to evaluate programs and better understand the conditions
  - ❖ State programs provide limited long-term follow-up
  - ❖ Limited data on long-term outcomes of
    - affected children and their families
    - children with conditions of uncertain clinical significance
  - ❖ Many conditions targeted are uncommon
    - Pooled data essential to evaluate rare conditions

# NBS Registry

## ➤ Background (cont)

❖ General consensus that data on long-term outcomes is necessary to:

- Assess clinical outcomes of affected children
- Assess developmental outcomes of affected children
- Assess family and social impacts
- Establish a resource and infrastructure for research to:
  - Assess test result/phenotype correlations
  - Assess genotype/phenotype correlations
  - Assess treatment and environmental variables associated with better outcomes
- Establish a mechanism to communicate with children, families, and medical homes

# Registry Planning Project

- Objective 1: Team building of regional and national collaborators and system planning for the registry to include data in several domains:
  - ❖ clinical status,
  - ❖ developmental status,
  - ❖ economic impact,
  - ❖ school performance,
  - ❖ family impact.

# Registry Planning Project

- Objective 2: Collaborate with the Nicola Longo's project entailing pilot testing of a database software tool in metabolic clinic to identify strengths, weaknesses, and opportunities.

# Registry Planning Project

- ❖ Objective 3: Collaborate with Janet Thomas's Colorado project of disease specific care plans with defined outcome measures for all metabolic disorders diagnosed by newborn screening. Outcome measures for metabolic conditions will be established and used to model process and outcome measures for other NBS conditions, including endocrinopathies and hemoglobinopathies.

# Registry Planning Project

- Initial plan will be ambitious
  - ❖ All conditions (virtually all)
  - ❖ All affected children
  - ❖ Multiple outcome domains
  - ❖ Multiple data sources
  - ❖ DNA repository (?)
- Regional model beginning with UT and CO
- Potential model for multi-regional or national system

# Data Sources

- Newborn screening program
- Subspecialty clinics
- Medical Home
- Families
- Schools

# Collaborators

- MSGRCC
- University of Utah
  - ❖ GeneSiS Center/ Pediatrics
  - ❖ Clinical Genetics
  - ❖ Medical Informatics
  - ❖ College of Nursing
  - ❖ Division of Medical Ethics
- Utah Department of Health
- IHC Clinical Genetics Institute
- University of Colorado
- Dartmouth University
- Utah State University
- Boston Children's Hospital
- Oregon Health and Science University
- University of Iowa

# Questions/Discussion

