

# Challenges to the Expansion of Newborn Screening Testing

2008 Policy Forum

Center for Genomics and Society

UNC Chapel Hill

May 5, 2008



# Decision Factors in Adding Tests to Newborn Screening

- Historically, a number of questions were considered prior to adding a test for a specific disorder.
  - Is a test available and suitable for screening large numbers of newborns?
  - Is the test sensitive and specific?
  - Is the test cost-prohibitive?
  - Is there an intervention for the disorder that significantly improves the health of the child?



# Screening Tests that Met the Criteria

- Phenylketonuria (PKU) *(and other amino acid, fatty acid and organic acid disorders)*
- Galactosemia
- Congenital Adrenal Hyperplasia (CAH)
- Hypothyroidism
- Biotinidase Deficiency
- Sickle Cell Anemia
- Cystic Fibrosis



# Range of Interventions

- Dietary Changes

- PKU, Cystic Fibrosis

- Dietary Supplements

- Biotinidase Deficiency

- Hormone Therapy

- Hypothyroidism

- Antibiotic Therapies

- Sickle Cell Anemia, Cystic Fibrosis





# Challenges

- Funding for new tests must go through several levels of approval, with final approval by the state legislature
- Availability of FDA approved methods is limited
- New laboratory and follow up program positions are required to support new methods
- New equipment is required for new methods
- Laboratory space is required for new equipment and new positions and space is currently limiting

## Challenges (cont'd)



- Effective interventions are less clear and/or more invasive for some disorders
- Ethical dilemmas are created by DNA testing (for example knowledge of carrier status or if no treatment is available)
- Recommendations by professional groups are perceived as “standard of care,” creating great political pressure on states to add tests to screening panel

# One Solution to Complex Decision-Making

- Active Newborn Screening Advisory Committee with broad member base
  - State Laboratory of Public Health staff
  - State Follow up Program staff
  - Medical Professionals
  - Researchers
  - Parents with affected children
  - Non-profit group members



# Contact Information

Dr. Leslie A. Wolf, Director

North Carolina State Laboratory of Public  
Health

306 N. Wilmington St.

Raleigh, NC 27601

919-733-7834

Website: <http://slph.state.nc.us>

Email: [Leslie.Wolf@ncmail.net](mailto:Leslie.Wolf@ncmail.net)

