# Implications of Newborn Screening for Nurses & Nursing Faculty

Jane M. DeLuca PhD RN
Clemson University School of Nursing
Alex R. Kemper MD MPH MS
Duke University School of Medicine

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# Webinar content will include:

- An overview of newborn screening (NBS) activities at state & national levels
- A description of NBS controversies and ethical considerations
- A description of nursing roles in NBS with suggestions for nursing education & research
- A discussion of new developments in NBS

# Brief history of NBS

- Dr. Folling & phenylketouria (PKU)
- Development of diet treatment for PKU
- PKU screening programs in 1960s & 70s
- New screening 1990s- ; tandem mass spectroscopy, DNA testing and other technologies

# State and national oversight of NBS

- Inclusion of disorders for NBS screening panels occurs at the level of the states
- Guidance provided by SACHDNC
  - Evidence based criteria
- Recommended uniform panel
  - 31 core conditions/ 26 secondary conditions
  - http://www.hrsa.gov/advisorycommittees/mchbadvisory/heritabledisorders/recommendedpanel/

# Recommended Screening Panel of Core Conditions

| | |
|---|---|
| *PROP* | Propionic acidemia |
| *MUT* | Methylmalonic acidemia (methylmalonyl-CoA mutase) |
| *Cbl A,B* | Methylmalonic acidemia (cobalamin disorders) |
| *IVA* | Isovaleric acidemia |
| *3-MCC* | 3-Methylcrotonyl-CoA carboxylase deficiency |
| *HMG* | 3-Hydroxy-3-methyglutaric aciduria |
| *MCD* | Holocarboxylase synthase def. |
| *ßKT* | ß-Ketothiolase deficiency |
| *GA1* | Glutaric acidemia type I |
| *CUD* | Carnitine uptake defect/carnitine transport defect |
| *MCAD* | Medium-chain acyl-CoA dehydrogenase deficiency |
| *VLCAD* | Very long-chain acyl-CoA dehydrogenase deficiency |
| *LCHAD* | Long-chain L-3 hydroxyacyl-CoA dehydrogenase deficiency |
| *TFP* | Trifunctional protein deficiency |
| *ASA* | Argininosuccinic aciduria |
| *CIT* | Citrullinemia, type I |
| *MSUD* | Maple syrup urine disease |
| *HCY* | Homocystinuria |
| *PKU* | Classic phenylketonuria |
| *TYR I* | Tyrosinemia, type I |
| *CH* | Primary congenital hypothyroidism |
| *CAH* | Congenital adrenal hyperplasia |
| *Hb SS* | S,S disease (Sickle cell anemia) |
| *Hb S/βTh* | S, βeta-thalassemia |
| Hb S/C | S,C disease |
| BIOT | Biotinidase deficiency |
| CCHD | Critical congenital heart disease |
| CF | Cystic fibrosis |
| GALT | Classic galactosemia |
| HEAR | Hearing loss |
| SCID | Severe combined immunodeficiences |

# Balancing benefits & harms of screening

- There are tensions in screening for disorders with limited or expensive treatments or poor outcomes
- Early identification of disorders may be beneficial for families
  - Avoidance of 'diagnostic odyssey'
  - Allows parents to prepare for expected outcomes
  - Allows for informed reproductive choices

# Potential harms of NBS

- Complications of identifying infants with
  - False positive results
  - Carrier results
  - Ambiguous or intermediate screening results

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# Controversies in biobanking

- Storage and use of residual dried blood spots
  - For quality assurance purposes
  - Forensics
  - New screening test development
  - Research

- Court challenges
- Issues of parental consent

# NBS in Nursing Practice

- Nurses and midwives are key providers of NBS education and the communication of screening information to parents throughout the NBS process

# NBS in Nursing Practice

- Preconception period
  - Persons may not receive information about newborn screening until planning a pregnancy or already pregnant
  - Parents prefer delivery of NBS education over time during the pregnancy
  - Brochures can be helpful
    - should describe how NBS results will be conveyed to parents and what to expect in the event of an abnormal NBS result

# NBS in Nursing Practice

- Perinatal period
  - NBS education around time of deliver can be lost
  - NBS is mandatory in most of U.S. operating as a routine procedure after delivery
    - Parents may lack information about screening refusal
  - NBS beyond the blood spot
    - critical congenital heart disease and hearing loss
  - Sufficient knowledge of the screening process and adequate communication skills are necessary for conversations with parents about abnormal screen results

# NBS in Nursing Practice

- Specialty care
  - Multidisciplinary teams of genetic providers, medical specialists, nurses, and nutritionists care for infants suspected and diagnosed with NBS disorders
  - Nurses can have a key role in coordination and communication during acute phase and chronic care of infants with metabolic and other disorders identified through NBS

# NBS in Nursing Practice

- Long-term follow up
  - High quality chronic disease management with condition specific treatment and age appropriate preventative care over the life span
    - LTF is crucial for understanding the natural history of rare disorders and innovating treatments
  - Surge in long-term tracking through voluntary national registries
  - Continuous quality improvement for advancing care and services

# Opportunities for Nurse Educators

- NBS as a model for teaching genetics and principles of public health to nurses in various stages of education from novice to advance practice
  - Wealth of education materials through state and national agencies
  - Opportunity for practicing communication skills for conveying complex information to parents and families

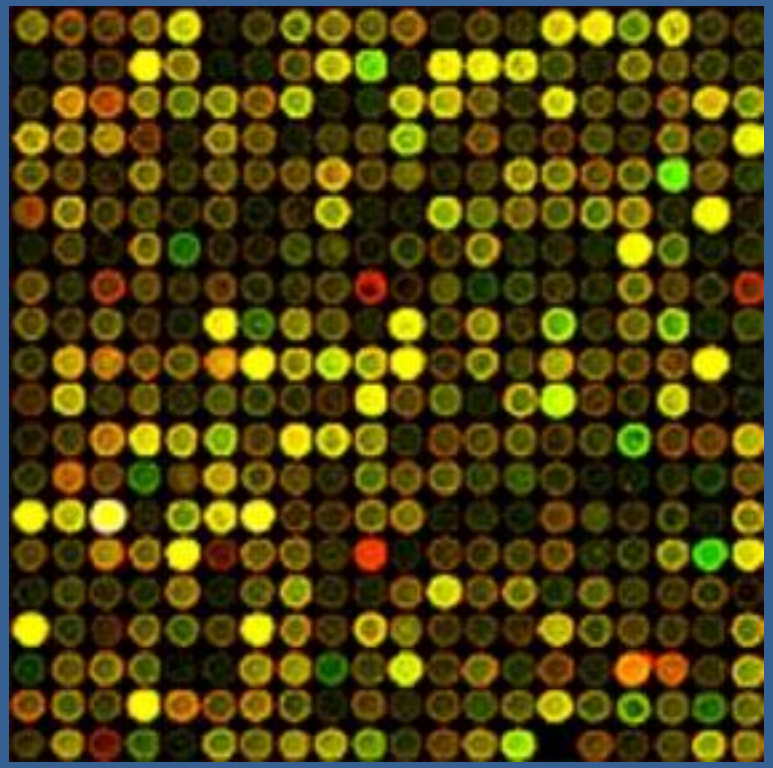
# Potential for Nursing Research in NBS

- Opportunities for conducting clinical and collaborative research in many areas of NBS
  - Biobanking
  - Informed consent
  - Disparities in screening services
  - Best practices in communicating NBS information
  - Cost-effectiveness of NBS

  – Foster the development of multidisciplinary research and clinical teams

# New screenings on the horizon

- Technological advances in screening continue:
  - Microarray
  - Sequencing entire genome
  - Personalized medicine
  - New disorders in the wings for screening
- Need for additional public education, research, medical and nursing education to meet the demands for newer screening processes



bio.cse.ohio-state.edu

# Websites for NBS information & education

- National Newborn Screening and Genetics Resource Center: http://www.nccrcg.org/
- National Newborn Screening & Genetics Resource Center: http://genes-r-us.uthscsa.edu
- Genetic Alliance: http://geneticalliance.org/
- March of Dimes: http://marchofdimes.com
- Babies First Test: http://www.babysfirsttest.org/
- Save Babies Through Screening Foundation: http://www.savebabies.org/