Implications of Newborn Screening for Nurses & Nursing Faculty

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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
# Recommended Screening Panel of Core Conditions

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

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