NBS Candidate Condition
ELSI Framework

Your Participation Needed

Jeffrey P. Brosco MD PhD
Mailman Center for Child Development
Disclosures

• No conflicts of interest to disclose
• This work represents my opinions and not necessarily those of the SACHDNC
Outline

• ELSI at the beginning of NBS
• Framework
  – How to ensure that ELSI issues are addressed as a new condition is considered for the RUSP
  – What questions might be answered during an NBS pilot study
• Audience participation
Carol Buck

Pearl S. Buck
“The Girl Who Never Grew”
*Ladies Home Journal*, 1950
NBS in the 1960s
What do you think were the key ELSI as NBS began in the 1960s?
What do you think were the key ELSI as NBS began in the 1960s?

- False negatives
  - Move from hospital-based to state-legislated

- Few/No concerns about parental consent
- Few/No concerns about “genetics”
What do you think were the key ELSI as NBS by 1968, after 1 million children screened for PKU?
Key ELSI in 1968

- Indeterminant values (who to treat)
- How to treat (what level of Phe ok)
- When to stop treatment
- False positives (few physical harms)
Preventing Intellectual Disability
Prevalence ID per 1 million births
Brosco, Arch. Peds., 2006

- CS
- Rh
- Measles
- H.flu
- CH
- PKU
- CRS
What are the key NBS ELSI today?

(Focus on conditions that are or might be considered for addition to the RUSP)
NBS Candidate Condition ELSI Framework

• Goals
  – Provide a logical way to approach the range of potential ELSI for a candidate condition
  – Allow anyone interested in nominating a new condition to systematically identify and address specific ELSI
  – Encourage scholars to include ELSI research questions in pilot studies
<table>
<thead>
<tr>
<th>Stakeholder Group</th>
<th>Non-Result Related ELSI Issues</th>
<th>Result Related ELSI Issues</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Individual Newborn</strong></td>
<td>1. What are the potential harms of screening to a newborn?</td>
<td>1. What are the potential harms of the diagnostic procedures associated with a condition?</td>
</tr>
<tr>
<td></td>
<td>2. What are the potential harms associated with the treatment of a condition?</td>
<td>2. Are there any potential physical impacts of heterozygous status?</td>
</tr>
<tr>
<td></td>
<td>3. Potential stigmatization of a true positive result.</td>
<td>3. What may be the reproductive impact of carrier information for the newborn?</td>
</tr>
<tr>
<td><strong>Family</strong></td>
<td>1. Are there any potential harms to families when screening for a new condition?</td>
<td>1. What are the potential financial costs and psychosocial harms of the diagnostic procedures associated with a condition?</td>
</tr>
<tr>
<td></td>
<td>2. What are the potential psychosocial and financial costs impacts associated with the treatment of a condition?</td>
<td>2. What are the potential psychosocial impacts of a false positive?</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td><strong>NBS State Program</strong></td>
<td>1. What are the costs and resources needed to implement a new NBS condition?</td>
<td>1. What are the costs and resources needed for short-term and long-term follow-up?</td>
</tr>
<tr>
<td></td>
<td>2. What is the impact of state services (e.g. Medicaid, public school system)?</td>
<td>2. What is the impact of state services on reproductive health?</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td><strong>Population</strong></td>
<td>1. What are the opportunity costs to society of implementing a new NBS condition?</td>
<td>1. Do excessive false positive results undermine faith in NBS, science, or government?</td>
</tr>
</tbody>
</table>
ELSI Framework

- 4 “stakeholder” groups
  - Individual/newborn
  - Family
  - State NBS program (health department)
  - Population
ELSI Framework

• Non-result category
• Result category
  – True positive
  – False positive
  – Carrier
  – Unclear result (e.g. late onset, intermediate value, natural history unknown)
“Life Course” of a Condition

- Pre-nomination
  - Identify ELSI issues; conduct pilot studies; make specific recommendations
- Evidence review
  - Identify/review evidence; note research gaps
- SACHDNC review
  - Identify issues; make recommendations
- Implementation
  - Identify issues; study ELSI outcomes; make rec’s
What should we change?

- Open discussion
- Written comments
- Follow-up informally