The Role of Birth Defects State Programs in Screening for Critical Congenital Heart Disease

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Outline

- Federal role in CCHD implementation
- NBDPN survey of CCHD implementation
- POC Newborn Screening
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Newborn Screening Saves Lives Act, 2007

- Two oversight committees to assure equity in NBS in U.S.
  - Secretary’s Advisory Committee on Heritable Disorders in Newborns and Children
    - Federal Advisory Committee
    - Recommended Newborn Screening Panel
  - Interagency Coordinating Committee on Newborn and Child Screening
    - CDC and HRSA – co-leads
    - Guide implementation of SACHDNC recommendations
CCHD is added as a condition to the SACHDNC Recommended Newborn Screening Panel, September 2011

THE SECRETARY OF HEALTH AND HUMAN SERVICES
WASHINGTON, D.C. 20201

September 21, 2011

R. Rodney Howell, M.D.
Committee Chairperson
Secretary’s Advisory Committee on Heritable Disorders in Newborns and Children
5600 Fishers Lane, Room 18A19
Rockville, MD 20857

Dear Dr. Howell:

As indicated in my letter to you on April 20, 2011, I determined that the Secretary’s Advisory Committee on Heritable Disorders in Newborns and Children’s (SACHDNC) recommendations pertaining to the addition of Critical Congenital Heart Disease (CCHD) screening to the Recommended Uniform Screening Panel (RUSP) were not yet ready for adoption.
ICC on Newborn and Child Screening

“Screening for Critical Congenital Heart Disease: A Federal Agency Plan of Action”

- **Screening Standards and Infrastructure**: HRSA to guide the development of screening standards/infrastructure
- **Education and Training**: HRSA to fund the development of appropriate educ/training materials
- **Research**: NIH to determine the screening technology, diagnostic processes, care provided and health outcomes
- **Surveillance**: CDC to monitor infant mortality and other health outcomes *(clinical utility and program evaluation)*
CDC

- Evaluate state surveillance and tracking
  - National Birth Defects Prevention Network survey of states
  - EpiAid in NJ
- Conduct a cost-effectiveness analysis of newborn screening
  - NJ EcoAid
  - cost-effectiveness model of newborn screening for CCHD
- Leverage electronic health records
  - CDC created an EHR workgroup for BD; working with external partners
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  - Role of public health
NBDPN Survey of BD Surveillance Programs

- Potential role of state birth defects surveillance programs in assuring screening for CCHD
- First survey in Oct 2010 and revised in Nov 2011 following the addition of CCHD to RUSP
Is your state engaged in pulse oximetry screening for CCHD?

- Yes: 1 (2010), 10 (2011)
- No: 30 (2010), 21 (2011)
- Don't know: 12 (2010), 3 (2011)
November 2011: If your State adopts NBS for CCHD, how could the birth defects surveillance program assist with the confirmed cases of CCHD?

- Link children identified by screening to support services: 16
- Report on health care utilization by affected children: 13
- Report on support services utilization by affected children: 7
- Report on enrollment of affected children in special education services: 1
- Other (please specify): 8
### Nov 2011: How could the birth defects surveillance program assist with evaluation of CCHD newborn screening?

<table>
<thead>
<tr>
<th>Possible Options for Assistance</th>
<th>Number of States</th>
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<tbody>
<tr>
<td>Evaluate mortality associated with CCHD</td>
<td>28</td>
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<tr>
<td>Evaluate morbidities associated with CCHD</td>
<td>16</td>
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<tr>
<td>Evaluate interventions associated with CCHD</td>
<td>11</td>
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<tr>
<td>Compare outcomes of children with CCHD</td>
<td>14</td>
</tr>
<tr>
<td>Evaluate all true and false positive screens</td>
<td>13</td>
</tr>
<tr>
<td>Evaluate false negative screens</td>
<td>13</td>
</tr>
<tr>
<td>Assist with economic evaluation of screening</td>
<td>8</td>
</tr>
<tr>
<td>Other</td>
<td>8</td>
</tr>
</tbody>
</table>
What is the working relationship between your state’s birth defects surveillance program and newborn screening program?

- Organizationally located together: 11
- Contained within the same program/bureau: 15
- Physically located in the same building: 14
- Currently share a common database/data system: 8
- None/no working relationship between birth defects surveillance program and newborn screening program: 5
- Other: 10
Nov 2011: What is the average time lag for collection of complete data (≥ 95%) for all major birth defects under surveillance in your state?
November 2011: What are the likely barriers in your state to your program’s involvement with newborn screening for CCHD?

- Lack of legislative/regulatory language/authority: 19
- Inadequate staffing: 29
- Insufficient funds: 27
- Information technology/data linkage needs: 19
- Other (please specify): 8
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Secretary’s Advisory Committee on Heritable Disorders in Newborns and Children
Subcommittee on Treatment and Follow up

Kemper AK, et al “Implementing Point-of-Care Newborn Screening” (under review -- Genet Med)
What is Point-of-Care Screening?

- POC Testing - defined as: “[Screening] at or near the site of patient care.
- The driving notion behind POC is to bring the test conveniently and immediately to the patient.
- This increases the likelihood that the patient will receive the results in a timely manner.”

Traditional Newborn Screening

- Provided under public health authority as *an essential public health activity*
- Public agencies provide direct oversight to ensure timeliness and uniform quality
Criteria for Inclusion in POC NBS

Shared imperatives of Traditional NBS-
PLUS:
• Urgent treatment is required earlier than the feasible turnaround time for a centralized laboratory;

OR
• When screening is based on physiologic testing requiring presence of the newborn
Public Health Role in POC NBS

Assurance

– Implementation and follow-up
  – Evaluation of quality of the implementation
  – Especially if screening requires special equipment or staff training

– Availability of confirmatory diagnostic testing or treatment (e.g. need to transfer care)

– Surveillance and tracking

No Single Right Way or Directive
Depends on the Condition, the State, Other Factors
Summary -- Opportunity

- Many BD state surveillance programs have the data needed to evaluate CCHD NBS implementation
- Important opportunity to provide the needed ‘assurance’ function
- Engage with NBS programs early in the process
  - NJ, IN, MD