Newborn Screening for Critical Congenital Heart Disease: An Update on CDC Activities

Cindy Hinton, PhD, MS, MPH
Health Scientist, Pediatric Genetics Team
DBDDDD/NCBDDDD/CDC

30th Meeting of the Secretary’s Advisory Committee on Heritable Disorders in Newborns and Children: Webinar
April 19, 2013
CDC Role: Newborn Screening for Critical Congenital Heart Disease (CCHD)

- Recommendation was endorsed by Secretary Kathleen Sebelius on September 21, 2011. CDC was assigned three tasks:
  1. Evaluate state surveillance and tracking to monitor the effectiveness of CCHD newborn screening programs
  2. Conduct a cost-effectiveness analysis of newborn screening for the early identification of CCHD
  3. Leverage an electronic health record framework for congenital heart defects, including CCHD
1. SURVEILLANCE, PUBLIC HEALTH PRACTICE, & APPLIED RESEARCH
CDC Supports CCHD Surveillance and Research

- **Assess state readiness**
  - Survey and field investigations in NJ and GA

- **Support birth defects surveillance programs**
  - Direct support to states and National Birth Defects Prevention Network (NBDPN)
  - Funding Association of Maternal and Child Programs (AMCHP) for joint newborn screening (NBS) and birth defects program meeting
  - Congenital heart defects (CHD) surveillance in metropolitan Atlanta

- **Support public health research**
  - National Birth Defects Prevention Study (NBDPS)
  - Collaboration with FL Birth Defects Registry and March of Dimes
Public Health Cycle

- Monitoring
- Research
- Prevention
Survey of State Birth Defects Surveillance Programs

- To assess the potential role of state birth defects surveillance programs with screening for CCHD

- Distributed in October 2010 by National Birth Defects Prevention Network and re-sent in November 2011 after addition of CCHD to uniform newborn screening panel
  - States were asked to confirm or change responses from 2010

What are the likely barriers in your state to your program’s involvement with newborn screening for CCHD?

Summary of Survey Findings

- Involvement of state birth defects surveillance programs in surveillance and evaluation of CCHD screening implementation has potential to be hindered by:
  - Limited relationship between the state birth defects and newborn screening programs
  - Inadequate staffing and insufficient funds

- Recommendation: States should evaluate infrastructure and resource needs prior to adoption of screening for CCHD
Defining a Role for Birth Defects Surveillance Programs

Evaluation Questions for Birth Defects Surveillance Programs:

- Health outcomes after newborn screening among affected children
- Missed primary targets of screening (i.e., affected children who were not screened or had false-negative screens)
- Burden and screening accuracy for secondary targets
- Role of altitude, sociodemographic characteristics, and other special circumstances
- Contribution of prenatal and clinical diagnoses before newborn screening
- Costs and service utilization

Olney RS and Botto LD. Newborn screening for critical congenital heart disease: essential public health roles for birth defects monitoring programs. BDRA 2012;94(12):965-969.
Challenges for Birth Defects Surveillance Programs

- Data sources and quality
- Timeliness of data collection
- Long-term follow-up for comprehensive outcomes
- Standardization of reporting
- State and national program coordination

Olney RS and Botto LD. Newborn screening for critical congenital heart disease: essential public health roles for birth defects monitoring programs. BDRA 2012:94(12);965-969.
New Jersey Field Investigation (January 2012)

- Conduct assessment of:
  - Screening data flow and tracking at each facility
  - Electronic health records (EHR) capabilities at each facility
  - Process of communicating screening data to the NJ Birth Defects Registry

- Provide technical assistance to the NJ Department of Health for development and pilot of a questionnaire for follow-up of all infants that do not pass screening

- Describe epidemiology of CCHD cases detected during the first three months of screening
Georgia Field Investigation (June–September 2012)

- **Conduct assessment of:**
  - Screening data flow and tracking at each facility
  - Electronic health records (EHR) capabilities at each facility
  - Process of communicating screening data to the Georgia Department of Health

- **Assess the extent to which Georgia birthing hospitals are currently conducting or are planning to conduct universal newborn screening for CCHD**

- **Describe barriers and/or challenges Georgia hospitals have encountered to implementing screening**
Publications from Field Investigations

- **MMWR**
  - Release date: April 18, 2013
  - “Rapid Implementation of Statewide Mandate for Pulse Oximetry Newborn Screening to Detect Critical Congenital Heart Defects — New Jersey, 2011.”

- **Manuscript for the peer-review literature**
  - New Jersey state health department lead author
  - More in depth about the case ascertainment
  - Manuscript under peer review
CDC Activities with State Birth Defects Surveillance Programs

- Currently fund 14 state programs
- Provide technical assistance with development and enhancement of surveillance systems
- Collaborate on and assist with epidemiological analyses of pooled surveillance data – including analyses of prevalence and survival
- Facilitate exchange of information between states
- Publish state-specific surveillance data annually
- Assist with cluster investigations
Current CDC Cooperative Agreements for Birth Defects

State Birth Defects Surveillance and Data Utilization

- Arizona
- Colorado
- Florida
- Illinois
- Kentucky
- Louisiana
- Michigan
- Minnesota
- New Hampshire
- New Jersey
- Ohio
- Oklahoma
- Puerto Rico
- Rhode Island
- Utah
- Arkansas
- California
- Iowa
- Georgia (CDC)
- Massachusetts
- New York
- North Carolina
- Texas
- Alaska
- Hawaii

Centers for Birth Defects Research and Prevention (National Birth Defects Prevention Study)
Pooling Data Across Surveillance Sites

- NBDPN publishes an annual report on 41 major birth defects
- 2012 report focused on CCHD
  - Prevalence of CCHD reported per 10,000 live births by state and by type of surveillance system
  - Active and passive surveillance systems
  - Pooled summary estimates reported

Table 1
Critical Congenital Heart Defects Targeted for Newborn Screening: Counts and Prevalence among Live Births, 2005–2009 (Prevalence per 10,000 Live Births)

<table>
<thead>
<tr>
<th>State</th>
<th>Common Truncus</th>
<th>Hypoplastic Left Heart Syndrome</th>
<th>Pulmonary Valve Atresia and Stenosis</th>
<th>Pulmonary Valve Atresia</th>
<th>Tetralogy of Fallot</th>
<th>TAPVR</th>
<th>All TGA</th>
<th>d-TGA</th>
<th>Tricuspid Valve Atresia and Stenosis</th>
<th>Tricuspid Valve Atresia</th>
<th>Notes</th>
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<tbody>
<tr>
<td>Alaska</td>
<td>12</td>
<td>2.2</td>
<td>80</td>
<td>30</td>
<td>11</td>
<td>24</td>
<td>9</td>
<td>1.6</td>
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<tr>
<td>Arkansas</td>
<td>0.6</td>
<td>6</td>
<td>136</td>
<td>14</td>
<td>86</td>
<td>22</td>
<td>84</td>
<td>74</td>
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<td>Arizona</td>
<td>25</td>
<td>130</td>
<td>228</td>
<td>105</td>
<td>210</td>
<td>107</td>
<td>96</td>
<td></td>
<td>1.0</td>
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<tr>
<td>Colorado</td>
<td>0.5</td>
<td>83</td>
<td>98</td>
<td>75</td>
<td>144</td>
<td>38</td>
<td>100</td>
<td>61</td>
<td>1.5</td>
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<td>Connecticut</td>
<td>7</td>
<td>1.8</td>
<td>66</td>
<td>5</td>
<td>72</td>
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<td>9</td>
<td>2</td>
<td>0.5</td>
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<tr>
<td>Delaware</td>
<td>0.0</td>
<td>10</td>
<td>36</td>
<td>3</td>
<td>11</td>
<td>2</td>
<td>7</td>
<td>7</td>
<td>3</td>
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<tr>
<td>Florida</td>
<td>92</td>
<td>351</td>
<td>1181</td>
<td>172</td>
<td>566</td>
<td>98</td>
<td>551</td>
<td>290</td>
<td>141</td>
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<tr>
<td>Georgia / CDC</td>
<td>0.8</td>
<td>3.1</td>
<td>10.3</td>
<td>1.5</td>
<td>4.9</td>
<td>0.9</td>
<td>4.8</td>
<td>2.5</td>
<td>1.2</td>
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<tr>
<td>Hawaii</td>
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<td>44</td>
<td>3</td>
<td>2</td>
<td>5</td>
<td>0</td>
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<tr>
<td>Iowa</td>
<td>1.1</td>
<td>1.1</td>
<td>24.6</td>
<td>1.7</td>
<td>1.1</td>
<td>2.8</td>
<td>0.0</td>
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<td>23</td>
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AMCHP Meeting: State NBS and Birth Defects Program Roles

- CDC supported AMCHP to convene a one day meeting to discuss interactions between birth defects surveillance and NBS programs
- Atlanta, February 28, 2013
- Brought together 12 states (9 HRSA funded plus 3 others), birth defects programs, newborn screening programs, Title V directors, NEWSteps
- Shared programs, facilitated discussion on how to build collaboration
- AMCHP writing an issue brief for stakeholders
CHD Surveillance in Metropolitan Atlanta

- Metropolitan Atlanta Congenital Defects Program (MACDP) has been conducting active, population-based birth defects surveillance since 1968

- MACDP case definition:
  - Residency in metropolitan Atlanta (3 counties as of January 2012)
  - Infant, fetus or child has major structural or chromosomal anomaly present at delivery
  - Infant, fetus or child must have been of at least 20 completed weeks of gestation at the time of delivery
  - If it’s a live birth, birth defect must have been diagnosed before the child’s 6th birthday

- CHDs are among the birth defects ascertained by MACDP
CHD Surveillance in Metropolitan Atlanta
1998–2005

Table II. Congenital heart defect prevalence per 10,000 live births: Comparison of 1998–2005 Atlanta estimates with previous estimates

<table>
<thead>
<tr>
<th>Congenital Heart Defects</th>
<th>1998-2003 Atlanta</th>
<th>Hoffman and Kaplan*</th>
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</thead>
<tbody>
<tr>
<td>Left-to-right shunts</td>
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<tr>
<td>Ventricular septal defect</td>
<td>1665</td>
<td>17.6-44.8</td>
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<tr>
<td>Perimembranous ventricular septal defect</td>
<td>423</td>
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<tr>
<td>Muscular ventricular septal defect</td>
<td>1096</td>
<td>—</td>
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<tr>
<td>Subarterial ventricular septal defect</td>
<td>20</td>
<td>—</td>
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<tr>
<td>Ventricular septal defect NOS†</td>
<td>126</td>
<td>—</td>
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<tr>
<td>Atrial septal defect</td>
<td>523</td>
<td>3.7-10.6</td>
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<td>Secundum atrial septal defect</td>
<td>411</td>
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<tr>
<td>Sinus venosus atrial septal defect</td>
<td>15</td>
<td>—</td>
</tr>
<tr>
<td>Atrial septal defect NOS†</td>
<td>97</td>
<td>—</td>
</tr>
<tr>
<td>Atroventricular septal defect</td>
<td>163</td>
<td>2.4-4.0</td>
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<tr>
<td>Complete atroventricular septal defect</td>
<td>88</td>
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<tr>
<td>Patent ductus arteriosus</td>
<td>114</td>
<td>3.2-7.8</td>
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<td>Cyanotic congenital heart defects</td>
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<tr>
<td>Tetralogy of Fallot</td>
<td>189</td>
<td>2.9-5.8</td>
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<tr>
<td>Transposition of the great arteries</td>
<td>90</td>
<td>2.3-3.9</td>
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<tr>
<td>Discordant atroventricular connections</td>
<td>10</td>
<td>—</td>
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<tr>
<td>Truncus arteriosus</td>
<td>24</td>
<td>0.6-1.4</td>
</tr>
<tr>
<td>Total anomalous pulmonary venous return</td>
<td>31</td>
<td>0.6-1.2</td>
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<tr>
<td>Tricuspid atresia</td>
<td>19</td>
<td>0.2-1.2</td>
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<tr>
<td>Ebstein’s anomaly</td>
<td>24</td>
<td>0.4-1.6</td>
</tr>
<tr>
<td>Singleventricle complex</td>
<td>41</td>
<td>0.5-1.4</td>
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<tr>
<td>Heterotaxy syndrome</td>
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<td>Left heart obstructive defects</td>
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<tr>
<td>Coarctation of the aorta</td>
<td>177</td>
<td>2.9-4.9</td>
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<td>Valvar aortic stenosis</td>
<td>45</td>
<td>1.6-3.9</td>
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<tr>
<td>Interrupted aortic arch type B</td>
<td>15</td>
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<tr>
<td>Hypoplastic left heart syndrome</td>
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<td>1.5-2.8</td>
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<td>Right heart obstructive defects</td>
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<td>Valvar pulmonic stenosis</td>
<td>220</td>
<td>3.6-8.4</td>
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<tr>
<td>Pulmonary atresia</td>
<td>17</td>
<td>0.8-1.5</td>
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<tr>
<td>Critical CHD†</td>
<td>621</td>
<td>10.8-15.3</td>
</tr>
<tr>
<td>All CHD†</td>
<td>3240</td>
<td>60.2-105.7</td>
</tr>
</tbody>
</table>

†Not otherwise specified.
‡Prevalence for critical CHD and all CHD is based on the number of infants and fetuses, not on the number of defects.

MACDP CCHD Survival Study


- Over 1 million births during 1979–2005 in metropolitan Atlanta
  - Approximately 7,000 were born with a congenital heart defect
  - Nearly 2,000 had a CCHD

- Analysis looks at survival trends by time period, clinical and maternal demographic factors
APPLIED RESEARCH
National Birth Defects Prevention Study (NBDPS) (1997–Present)

- Study centers in 10 US states
- On-going population-based case-control study
- Case definition
  - Live births, stillbirths or terminations of pregnancy
  - Chromosomal anomalies and single-gene disorders excluded
  - CHD cases classified by clinicians with expertise in pediatric cardiology
  - CHD cases must be confirmed by echocardiography, catheterization, surgery or autopsy
- Extensive clinical data ascertained from medical records by participating surveillance systems
National Birth Defects Prevention Study (NBDPS)  
(1997–Present)

- Controls
  - Live births without major birth defects
  - Selected from hospital data or vital records

- Extensive maternal interview conducted via telephone between 6 weeks and 24 months after estimated date of delivery in English or Spanish
NBDPS CCHD Analysis (On-going)

- **Research Question:** What proportion of cases of CCHD might benefit from the new U.S. recommendations for routine newborn CCHD screening?

- **Operationalized as**
  - Estimate the proportion of live-born infants in NBDPS with CCHD whose condition was detected late
  - Investigate clinical and demographic factors associated with late detection
Florida Birth Defects Registry

- Using linked, longitudinal birth defects registry and hospital discharge data from Florida
  - Assessment of mortality and hospital resource utilization among infants with timely vs. late detection of CCHD (timely=before birth hospital discharge)
  - Examination of factors associated with timely vs. late detection of infants with CCHD
2. HEALTH ECONOMICS AND SERVICE UTILIZATION FOR CHILDREN AND ADULTS WITH CCHD
Health Economics Studies

- **New Jersey cost study**
  - Time-motion studies and resource utilization questionnaire to assess hospital cost burden
  - Manuscript under peer review

- **Florida service utilization and costs for late diagnosis of CCHDs**
  - Manuscript under peer review

- **Cost–effectiveness analysis for routine CCHD newborn screening**
  - Manuscript under peer review
Health Care Utilization for Children and Adults with CHDs

- The Healthcare Cost and Utilization Project (HCUP), which is maintained by the Agency for Healthcare Research and Quality (AHRQ), collects discharge-level hospital administrative billing data from participating hospitals across the United States. Data collected include:
  - Principal and secondary diagnoses
  - Procedures
  - Hospital charges
  - Hospital length of stay
  - Expected primary and secondary payer
HCUP: KID and NIS

- **Kids’ Inpatient Database (KID) and Nationwide Inpatient Sample (NIS)**
  - Stratified random samples
  - Weighted to obtain results interpreted as national estimates

- **Research questions:**
  - What is the healthcare resource utilization of pediatric and/or adult congenital heart defect hospital discharges at different ages?
  - How do discharges with critical congenital heart defects differ in their healthcare utilization from discharges with non-critical congenital heart defects?
  - What factors (age, procedure type, insurance status, discharge disposition, etc.) impact the healthcare resource utilization of discharges with congenital heart defects?
3. LEVERAGE ELECTRONIC HEALTH RECORD
Cross-Agency Collaboration on CCHD Coding

- Working towards case definitions
- CDC is collaborating with the National Library of Medicine and the National Heart Lung Blood Institute
- Mapping CCHD conditions to various coding systems
  - Highlight similarities and differences between codes
- Goal: Facilitate meaningful data exchange between stakeholders
- Dr. Alan Zuckerman to present abstract at May Newborn Screening meeting in Atlanta
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For more information please contact Centers for Disease Control and Prevention

1600 Clifton Road NE, Atlanta, GA 30333
Telephone, 1-800-CDC-INFO (232-4636)/TTY: 1-888-232-6348
E-mail: cdcinfo@cdc.gov Web: www.cdc.gov

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