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

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National Center on Birth Defects and Developmental Disabilities
Division of Birth Defects and Developmental Disabilities

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

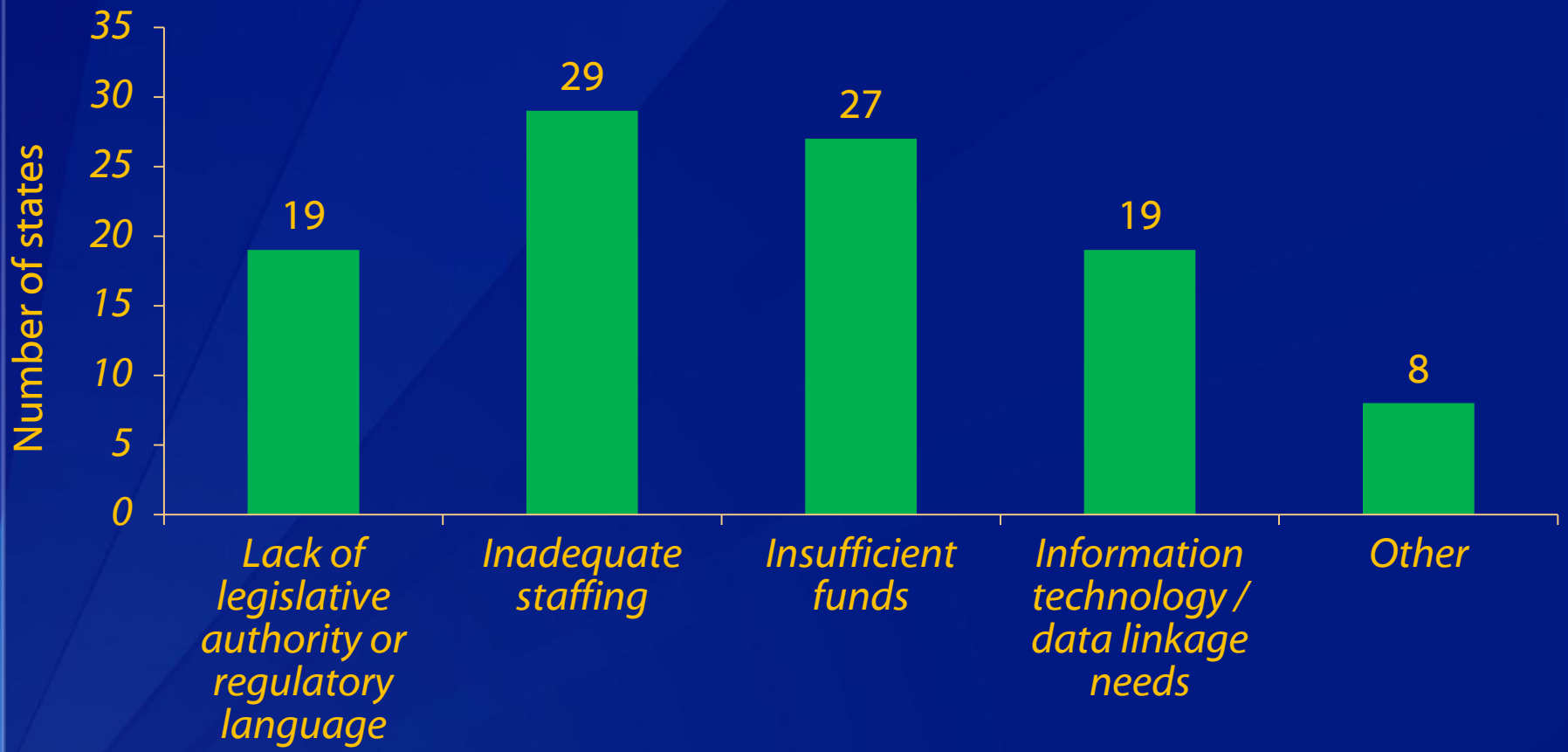
- **Assess state readiness**
  - Survey and field investigations in NJ and GA
- **Support birth defects surveillance programs**
  - Direct support to states and National Birth Defects Prevention Network (NBDPN)
  - Funding Association of Maternal and Child Programs (AMCHP) for joint newborn screening (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

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
- **"Newborn Screening for Critical Congenital Heart Disease: Potential Roles of Birth Defects Surveillance Programs—United States, 2010-2011." MMWR 2012; 61: 849-853.**

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



Centers for Disease Control and Prevention. "Newborn Screening for Critical Congenital Heart Disease: Potential Roles of Birth Defects Surveillance Programs—United States, 2010-2011." MMWR 2012; 61: 849-853.

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

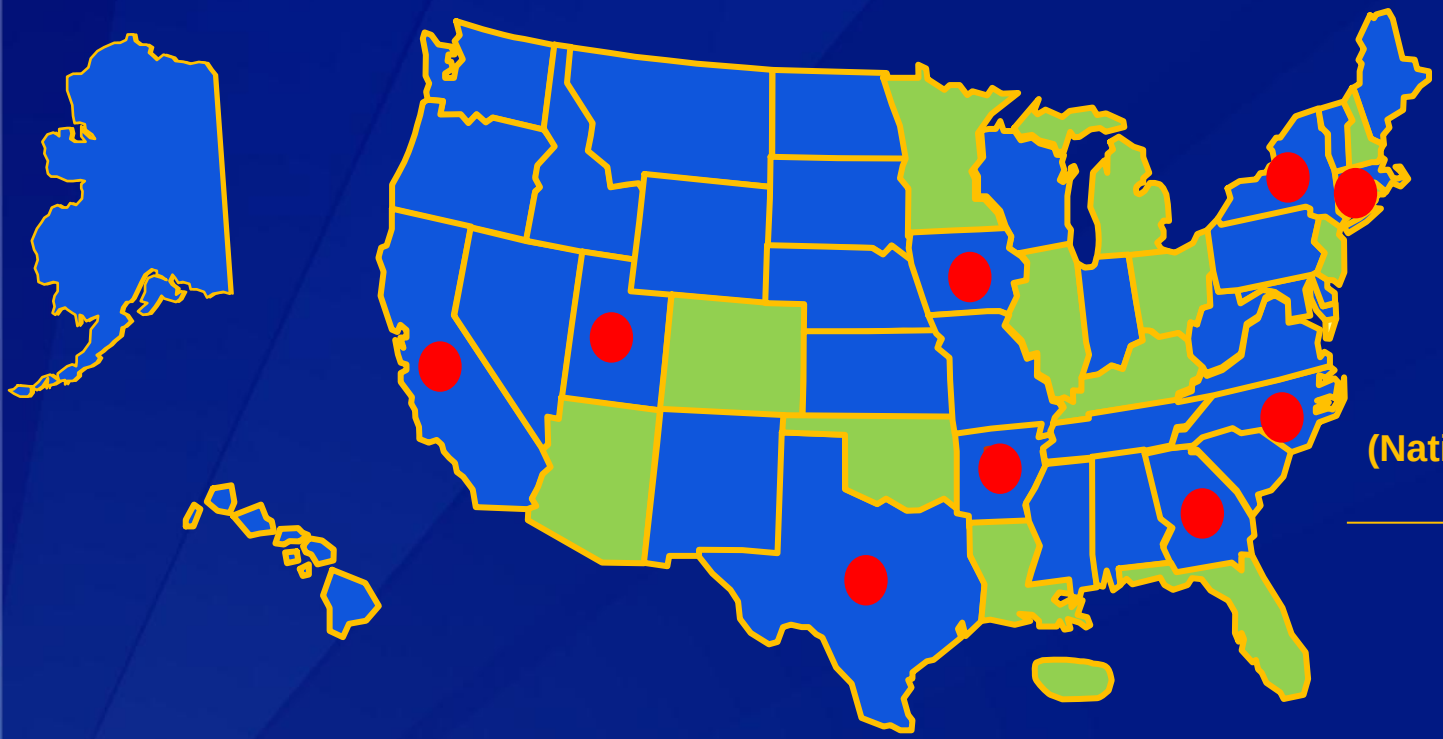
- **MMWR**
  - Release date: April 18, 2013
  - "Rapid Implementation of Statewide Mandate for Pulse Oximetry Newborn Screening to Detect Critical Congenital Heart Defects — New Jersey, 2011."
  - "Assessment of Current Practices and Feasibility of Routine Screening for Critical Congenital Heart Defects — Georgia, 2012"

- **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



Centers for Birth Defects Research and Prevention (National Birth Defects Prevention Study)

- Arkansas
- California
- Iowa
- Georgia (CDC)
- Massachusetts
- New York
- North Carolina
- Texas
- Utah

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

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

Mai CT et al. Selected birth defects data from population-based birth defects surveillance programs in the United States, 2005–2009: Featuring critical congenital heart defects targeted for pulse oximetry screening. Birth Defects Research Part A 2012:94(12);970-983.

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

State	Common Truncus	Hypoplastic Left Heart Syndrome	Pulmonary Valve Atresia and Stenosis	Pulmonary Valve Atresia	Tetralogy of Fallot	TAPVR	All TGA	d-TGA	Tricuspid Valve Atresia and Stenosis	Tricuspid Valve Atresia	Notes
Alaska[P]	12	14	80		30	11	24		9		
	2.2	***2.6***	***14.6***		***5.5***	***2.0***	***4.4***		***1.6***		
Arkansas[a]	12	68	275	14	86	22	84	74		13	
	0.6	***3.4***	***13.6***	***0.7***	***4.3***	***1.1***	***4.2***	***3.7***		***0.6***	
Arizona[a]	25	130	228	105	210		107	96			
	0.5	***2.6***	***4.6***	***2.1***	***4.3***		***2.2***	***2.0***			
Colorado[P]	16	83	298	75	144	38	100	61	53		1
	0.5	***2.4***	***8.5***	***2.1***	***4.1***	***1.1***	***2.9***	***1.7***	***1.5***		
Connecticut[P]	7	30	109		95		72		9		2
	0.4	***1.8***	***6.6***		***5.7***		***4.4***		***0.5***		
Delaware[a]	0	10	36	3	11	2	7	7		1	3
	0.0	***4.1***	***14.8***	***1.2***	***4.5***	***0.8***	***2.9***	***2.9***		***0.4***	
Florida[P]	92	351	1181	172	566	98	551	290	141		4
	0.8	***3.1***	***10.3***	***1.5***	***4.9***	***0.9***	***4.8***	***2.5***	***1.2***		
Georgia / CDC[a]	31	40	191	49	109	29	77	66	50	34	
	1.1	***1.5***	***7.0***	***1.8***	***4.0***	***1.1***	***2.8***	***2.4***	***1.8***	***1.2***	
Hawaii[a]	2	2	44		3	2	5		0		5
	1.1	***1.1***	***24.6***		***1.7***	***1.1***	***2.8***		***0.0***		
Iowa[a]	9	35	221	24	77	24	61	50	45	23	

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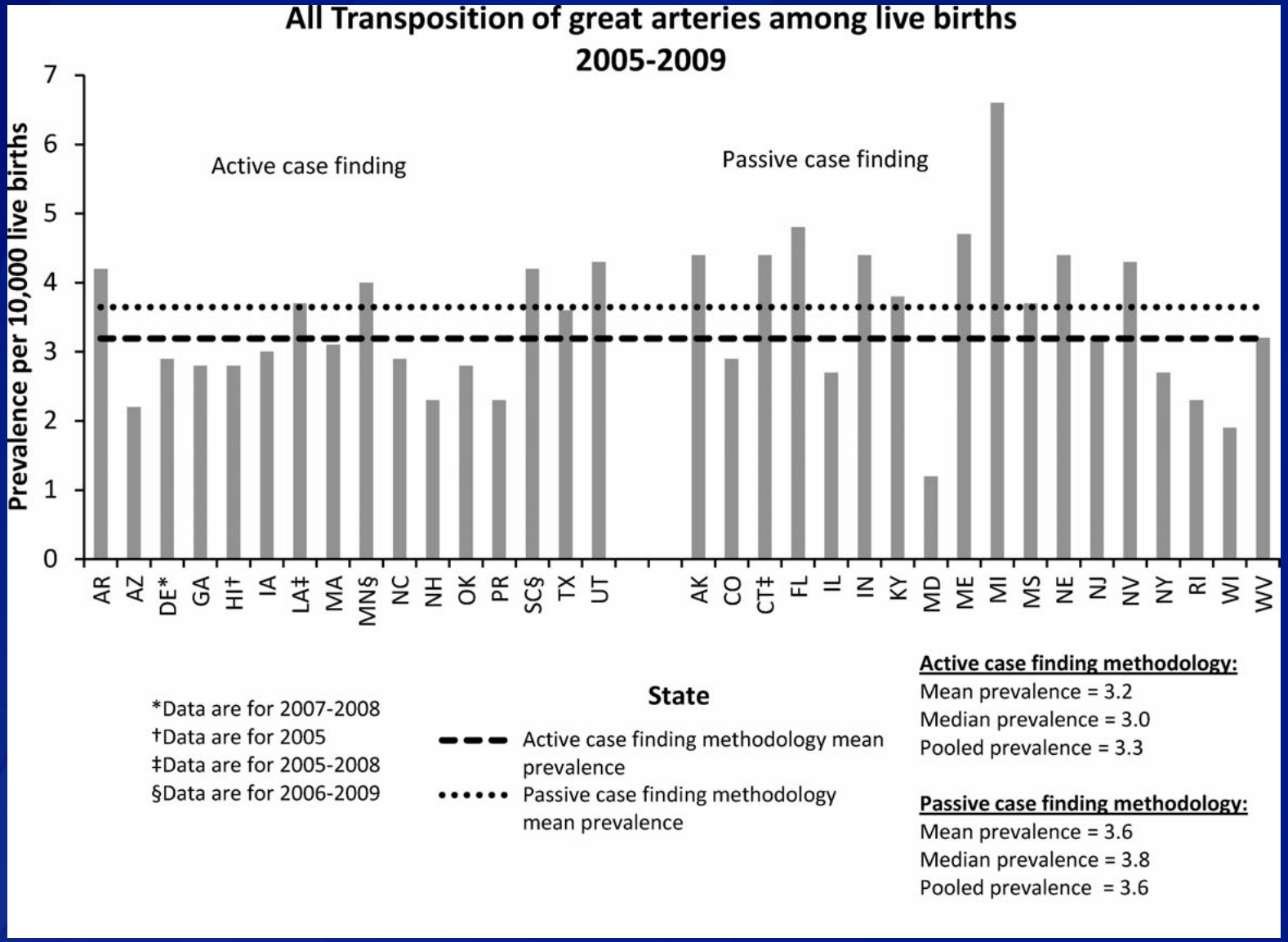

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AMCHP Meeting: State NBS and Birth Defects Program Roles

- 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

	1998-2003 Atlanta		Hoffman and Kaplan*
	n	Prevalence	Prevalence IQR
Left-to-right shunts			
Ventricular septal defect	1665	41.8	17.6-44.8
Perimembranous ventricular septal defect	423	10.6	—
Muscular ventricular septal defect	1096	27.5	—
Subarterial ventricular septal defect	20	0.5	—
Ventricular septal defect NOS†	126	3.2	—
Atrial septal defect	523	13.1	3.7-10.6
Secundum atrial septal defect	411	10.3	—
Sinus venosus atrial septal defect	15	0.4	—
Atrial septal defect NOS†	97	2.4	—
Atrioventricular septal defect	163	4.1	2.4-4.0
Complete atrioventricular septal defect	88	2.2	—
Patent ductus arteriosus	114	2.9	3.2-7.8
Cyanotic congenital heart defects			
Tetralogy of Fallot	189	4.7	2.9-5.8
Transposition of the great arteries	90	2.3	2.3-3.9
Discordant atrioventricular connections	10	0.3	—
Truncus arteriosus	24	0.6	0.6-1.4
Total anomalous pulmonary venous return	31	0.8	0.6-1.2
Tricuspid atresia	19	0.5	0.2-1.2
Ebstein's anomaly	24	0.6	0.4-1.6
Single ventricle complex	41	1.0	0.5-1.4
Heterotaxy syndrome	68	1.7	—
Left heart obstructive defects			
Coarctation of the aorta	177	4.4	2.9-4.9
Valvar aortic stenosis	45	1.1	1.6-3.9
Interrupted aortic arch type B	15	0.4	—
Hypoplastic left heart syndrome	91	2.3	1.5-2.8
Right heart obstructive defects			
Valvar pulmonic stenosis	220	5.5	3.6-8.4
Pulmonary atresia	17	0.4	0.8-1.5
Critical CHD‡	621	15.6	10.8-15.3
All CHD‡	3240	81.4	60.2-105.7

*Adapted from Hoffman JI, Kaplan S. Incidence of congenital heart disease. *J Am Coll Cardiol.* 2002;39:1890-900.
†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.

Reller MD, et al. Prevalence of congenital heart defects in metropolitan Atlanta, 1998-2005. J Pediatr 2008; 153: 807-813

MACDP CCHD Survival Study

- **Oster ME, Lee KA, Honein MA, Colarusso T, Shin M, Correa A. Temporal Trends in Survival Among Infants with Critical Congenital Heart Defects. Pediatrics 2013.**
- **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)

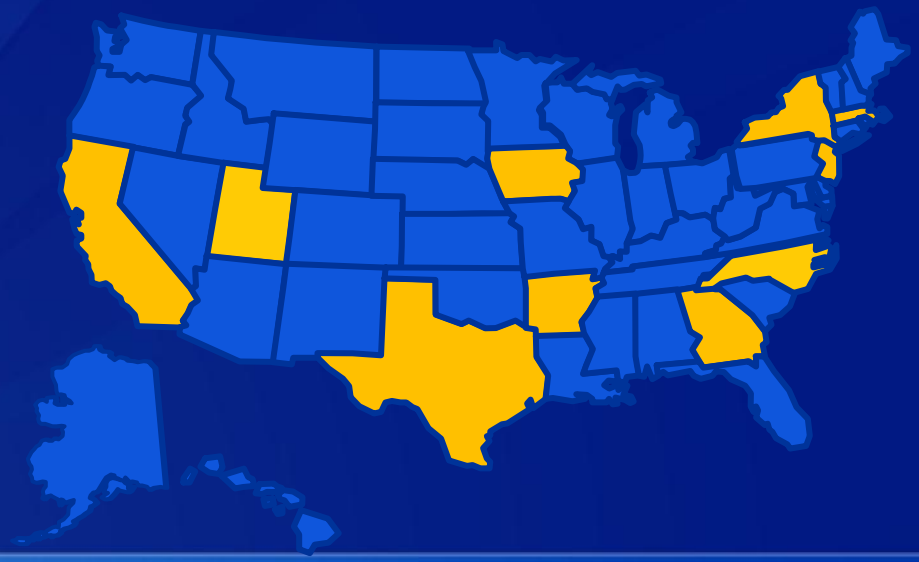
(1997–Present)

- **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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Thank you!

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