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

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INTRODUCTION

Since 1997, the National Birth Defects Prevention Network (NBDPN), in collaboration with the Centers for Disease Control and Prevention (CDC), has published data on major birth defects affecting the central nervous, eye, ear, cardiovascular, orofacial, gastrointestinal, genitourinary, and musculoskeletal systems, as well as trisomies, amniotic bands, and fetal alcohol syndrome, from population-based birth defects surveillance programs in the United States. Annually, the NBDPN Data Committee issues a data request to population-based birth defects programs for data on 47 major birth defects; the specific defects with accompanying diagnostic codes are detailed in Appendix 1 on page 10. This year's report containing data from 41 population-based birth defects surveillance programs for births occurring from January 1, 2005, through December 31, 2009, is available as a supplement on pages. The data are presented by racial/ethnic groups for all defects and additionally by maternal age for trisomies 13, 18, and 21.

To calculate prevalence, programs were also asked to provide the number of total live births and male live births for each calendar year submitted. The standard method for calculating birth defects prevalence is to divide the number of cases (birth defect for any pregnancy outcome) by total live births for the catchment area and then multiply by 10,000 to obtain the prevalence per 10,000 live births; Mason et al. (2005) provide further detail and rationale for this approach. This methodology is used for all defects except hypospadias, which is calculated using a denominator of total male live births. An attempt was made to standardize both the submitted data and presentation of state surveillance data, however, differences in the way programs collect and report birth defects data are listed in the footnotes of the accompanying tables and may be referenced in the program directories on pages S121-S169 (online). Some programs were able to only provide data for selected years, were unable to report counts and prevalence by race/ethnicity, or were unable to provide data for each specific defect requested due to differences in the coding systems (i.e., International Classification of Diseases, 9th Revision, Clinical Modification [ICD-9-CM] vs CDC/British Pediatric Association [BPA]) Classification of Diseases used to classify birth defects.

Critical Congenital Heart Defects Targeted for Pulse Oximetry Screening

This year's data report includes several enhancements to address the interest in pulse oximetry screening of newborns for critical congenital heart defects (CCHDs). Con-

Additional Supporting Information may be found in the online version of this article.

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The findings and conclusions in this report are those of the authors and do not necessarily represent the official position of the Centers for Disease Control and Prevention.

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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	
	0.4	***1.7***	***11.0***	***1.2***	***3.8***	***1.2***	***3.0***	***2.5***	***2.2***	***1.1***	
Illinois[P]	35	166	244	40	271	56	243	124	135	120	
	0.4	***1.9***	***2.7***	***0.5***	***3.1***	***0.6***	***2.7***	***1.4***	***1.5***	***1.4***	
Indiana[P]	15	76	315		129	29	190		34		6
	0.3	***1.7***	***7.2***		***3.0***	***0.7***	***4.4***		***0.8***		
Kentucky[P]	12	77	209		92	7	108		26		7
	0.4	***2.7***	***7.4***		***3.3***	***1.3***	***3.8***		***0.9***		
Louisiana[a]	18	27	135	21	74		63	58	17	15	2
	1.1	***1.6***	***8.0***	***1.2***	***4.4***		***3.7***	***3.4***	***1.0***	***0.9***	
Massachusetts[a]	16	55	253	23	175	34	120	105		22	8
	0.4	***1.4***	***6.6***	***0.6***	***4.5***	***0.9***	***3.1***	***2.7***		***0.6***	
Maryland[P]	2	27	8	3	35		45	15			
	0.1	***0.7***	***0.3***	***0.2***	***0.9***		***1.2***	***0.5***			
Maine[P]	6	19		3	22		32	22	2		9
	0.9	***2.8***		***0.4***	***3.2***		***4.7***	***3.2***	***0.3***		
Michigan[P]	88	296	732	161	386	94	407	256	98		
	1.4	***4.8***	***11.8***	***2.6***	***6.2***	***1.5***	***6.6***	***4.1***	***1.6***		
Minnesota[a]	5	23	55	10	43		39	38		7	10
	0.5	***2.4***	***5.7***	***1.0***	***4.4***		***4.0***	***3.9***		***0.7***	
Mississippi[P]	9	54	170		111		83		43		
	0.4	***2.4***	***7.6***		***5.0***		***3.7***		***1.9***		
North Carolina[a]	51	160	490	93	254	81	185	154	125	106	
	0.8	***2.5***	***7.7***	***1.5***	***4.0***	***1.3***	***2.9***	***2.4***	***2.0***	***1.7***	
Nebraska[P]	7	49	81	19	44	13	61	57		13	
	0.5	***3.5***	***5.8***	***1.4***	***3.1***	***0.9***	***4.4***	***4.1***		***0.9***	
New Hampshire[a]	5	8	53		31		16		2		
	0.7	***1.1***	***7.6***		***4.4***		***2.3***		***0.3***		
New Jersey[P]	29	93	523	53	201	47	180	104	56		
	0.5	***1.7***	***9.3***	***0.9***	***3.6***	***0.8***	***3.2***	***1.8***	***1.0***		
Nevada[P]	18	44	183	33	113	17	85	38	25		
	0.9	***2.3***	***9.4***	***1.7***	***5.8***	***0.9***	***4.3***	***1.9***	***1.3***		
New York[P]	78	329	1091	92	579	118	328	316	168	80	8
	0.6	***2.7***	***8.9***	***0.8***	***4.7***	***1.0***	***2.7***	***2.6***	***1.4***	***0.7***	
Oklahoma[a]	28	52	172	28	119	46	74	67	28	24	
	1.0	***1.9***	***6.4***	***1.0***	***4.4***	***1.7***	***2.8***	***2.5***	***1.0***	***0.9***	
Puerto Rico[a]	17	51	248	27	86	14	55	54		33	11
	0.7	***2.2***	***10.5***	***1.1***	***3.6***	***0.6***	***2.3***	***2.3***		***1.4***	
Rhode Island[P]	0	6	49	8	26	5	14	11		4	
	0.0	***1.0***	***8.1***	***1.3***	***4.3***	***0.8***	***2.3***	***1.8***		***0.7***	
South Carolina[a]	15	59	132	23	97		105	55	21		10
	0.6	***2.4***	***5.3***	***0.9***	***3.9***		***4.2***	***2.2***	***0.8***		

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

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
Texas[a]	137	417	1976	285	787	354	712	702	365	155	12
	0.7	***2.1***	***9.9***	***1.4***	***3.9***	***1.8***	***3.6***	***3.5***	***1.8***	***0.8***	
Utah[a]	16	89	377	35	88	32	116	58		30	
	0.6	***3.3***	***14.0***	***1.3***	***3.3***	***1.2***	***4.3***	***2.2***		***1.1***	
Wisconsin[P]	21	62	110	12	81	5	65	42	22	2	
	0.6	***1.8***	***3.2***	***0.4***	***2.4***	***0.1***	***1.9***	***1.2***	***0.6***	***0.1***	
West Virginia[P]	37	17	30	10	36	6	32	15	13		
	3.7	***1.7***	***3.0***	***1.0***	***3.6***	***0.6***	***3.2***	***1.5***	***1.3***		

[a]Active case finding.
[P]Passive case finding (with or w/o case confirmation).
Data in bold are the calculated prevalence data. Non-bold data are the case data.

ICD-9-CM, International Classification of Diseases, 9th Revision, Clinical Modification; CDC/BPA, Centers for Disease Control and Prevention/British Pediatric Association; TAPVR, total anomalous pulmonary venous return; d-TGA, dextro-transposition of the great arteries; TGA, transposition of the great arteries.

Notes:

1. Tricuspid valve atresia and stenosis: tricuspid stenosis and hypoplasia included.
2. Data are for 2005–2008.
3. Data are for 2007–2008. All heart defects require an echocardiogram report. Pulmonary valve atresia and stenosis and pulmonary valve atresia: peripheral, branch, trivial, or limited are not included. Tetralogy of Fallot: a ventricular septal defect with an overriding aorta is counted as tetralogy of Fallot. Tricuspid valve atresia: trivial or limited are not included.
4. Tricuspid valve atresia and stenosis: includes probable cases.
5. Data are for 2005.
6. Transposition of great arteries: data includes entire coding range of 745.10–745.19 (2005–2009). Tricuspid valve atresia and stenosis: data does not distinguish British Pediatric Association (BPA) codes 746.105 or 746.106 (2005–2009).
7. The 2007–2009 data are preliminary.
8. The 2009 data are provisional.
9. Tetralogy of Fallot: includes pulmonary atresia with ventricular septal defect.
10. Data are for 2006–2009.
11. Transposition of great arteries: excludes 745.11 (double outlet right ventricle).
12. Pulmonary valve atresia: excludes tetralogy of Fallot. Transposition of great arteries: Texas does not use the updated CDC/BPA code that has the exclusion criteria '745.180'; those defects of 'double outlet right ventricle' which Texas has coded into 745.180 will not be counted in this defect. The dextro-transposition of great arteries: data are provisional.

genital heart defects (CHDs) occur in an estimated 1 in 110 births in the United States (Reller et al., 2008) and approximately 25% of CHDs are considered CCHDs, defined as requiring surgery or catheter intervention within the first year of life (Mahle et al., 2009). Children with CCHDs are at risk for death or disability if the defect is not detected shortly after birth (Mahle et al., 2009). Thus, in 2011, the Secretary of Health and Human Services recommended that CCHDs be added to the U.S. Recommended Uniform Screening Panel for newborns (Secretary's Advisory Committee on Heritable Disorders in Newborns and Children, 2011). When implemented, newborns would undergo pulse oximetry screening for CCHDs after 24 hours of life, which detects low blood oxygen levels (hypoxemia).

The Secretary's Advisory Committee on Heritable Disorders in Newborns and Children (2011) named seven CCHDs as primary targets for screening: common truncus, d-transposition of the great arteries, tetralogy of Fallot, pulmonary valve atresia, tricuspid valve atresia, hypoplastic left heart syndrome, and total anomalous pulmonary venous return (Mahle et al., 2009; Kemper et al., 2011). Other CCHDs may also be detected using pulse oximetry screening, but because they may not consistently have hypoxemia at or soon after birth, their identification would be variable and incomplete. The following CCHDs are considered secondary targets of pulse oximetry screening: coarctation of the aorta, double outlet right ventricle, Ebstein anomaly, interrupted aortic arch, single ventricle, severe aortic stenosis and severe pulmonary stenosis (Mahle et al., 2009).

The implementation of pulse oximetry screening for CCHDs is currently underway in a few states, with more considering legislation and implementation (Olney and Botto, 2012). Surveillance case definitions, including both ICD-9-CM and CDC/BPA diagnostic codes, for each of the seven primary CCHD targets of pulse oximetry screening are presented in Appendix 2. In using these case definitions, it is important to consider as a potential limitation the coding system's ability to capture cases of CCHDs. Three of the seven conditions ('pulmonary valve atresia and stenosis', 'tricuspid valve atresia and stenosis', and 'transposition of the great arteries') include both broad codes to capture all possible cases that are generally collected for surveillance purpose and more refined codes that are targeted for newborn screening of CCHDs using pulse oximetry. For example, the surveillance category of 'tricuspid valve atresia and stenosis' encompasses both tricuspid valve atresia (one of the seven targeted CCHDs) as well as milder cases of tricuspid stenosis.

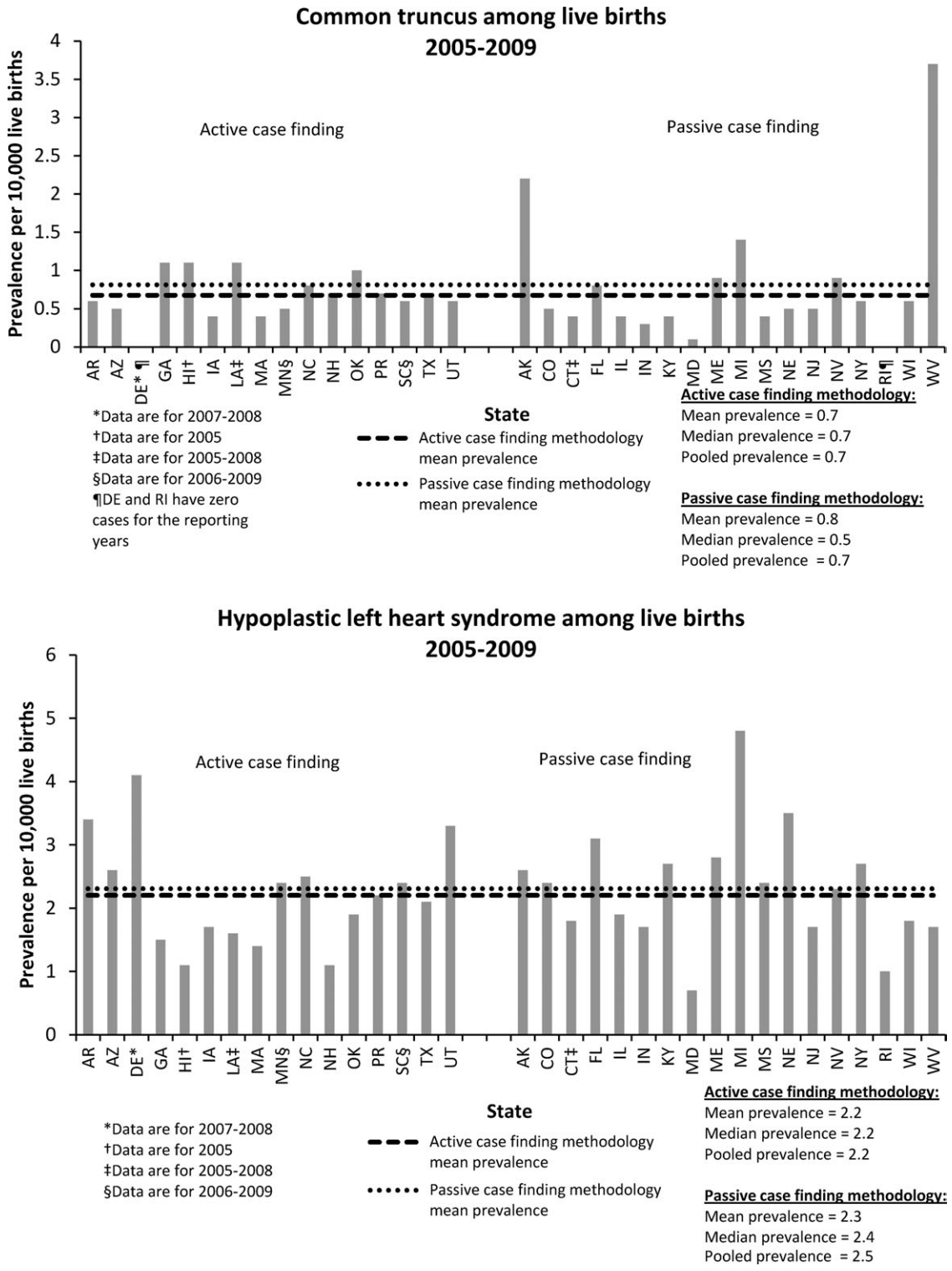


Figure 1. Critical congenital heart defects targeted for newborn screening: prevalence among live births by U.S. population-based surveillance programs.

Programs that are able to use more refined codes provided data separately for pulmonary valve atresia, tricuspid valve atresia, and d-transposition of the great arteries.

CCHD Data Presentation

Table 1 presents the counts and live birth prevalence for the seven primary targeted CCHDs from 34 popula-

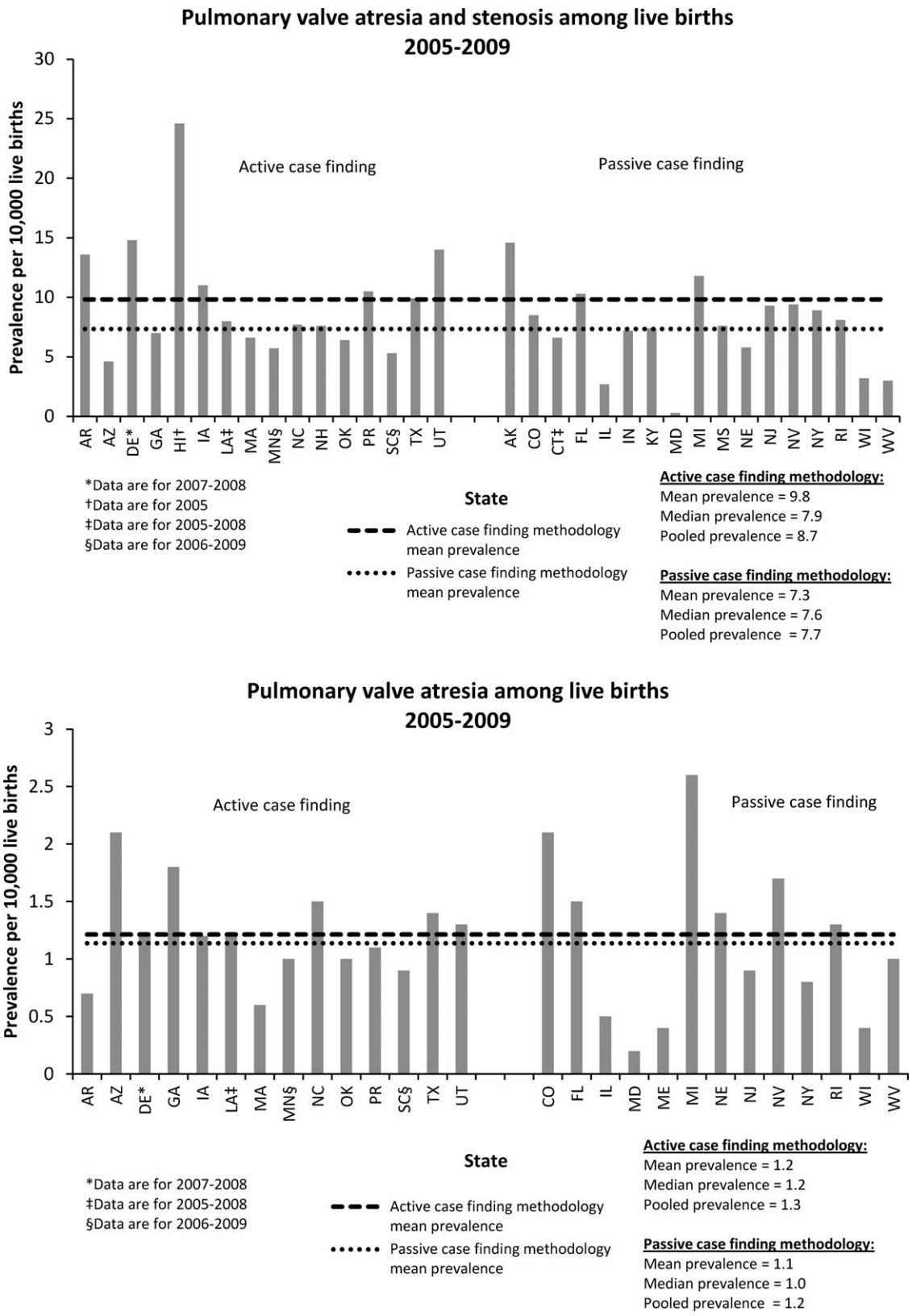


Figure 1. (Continued)

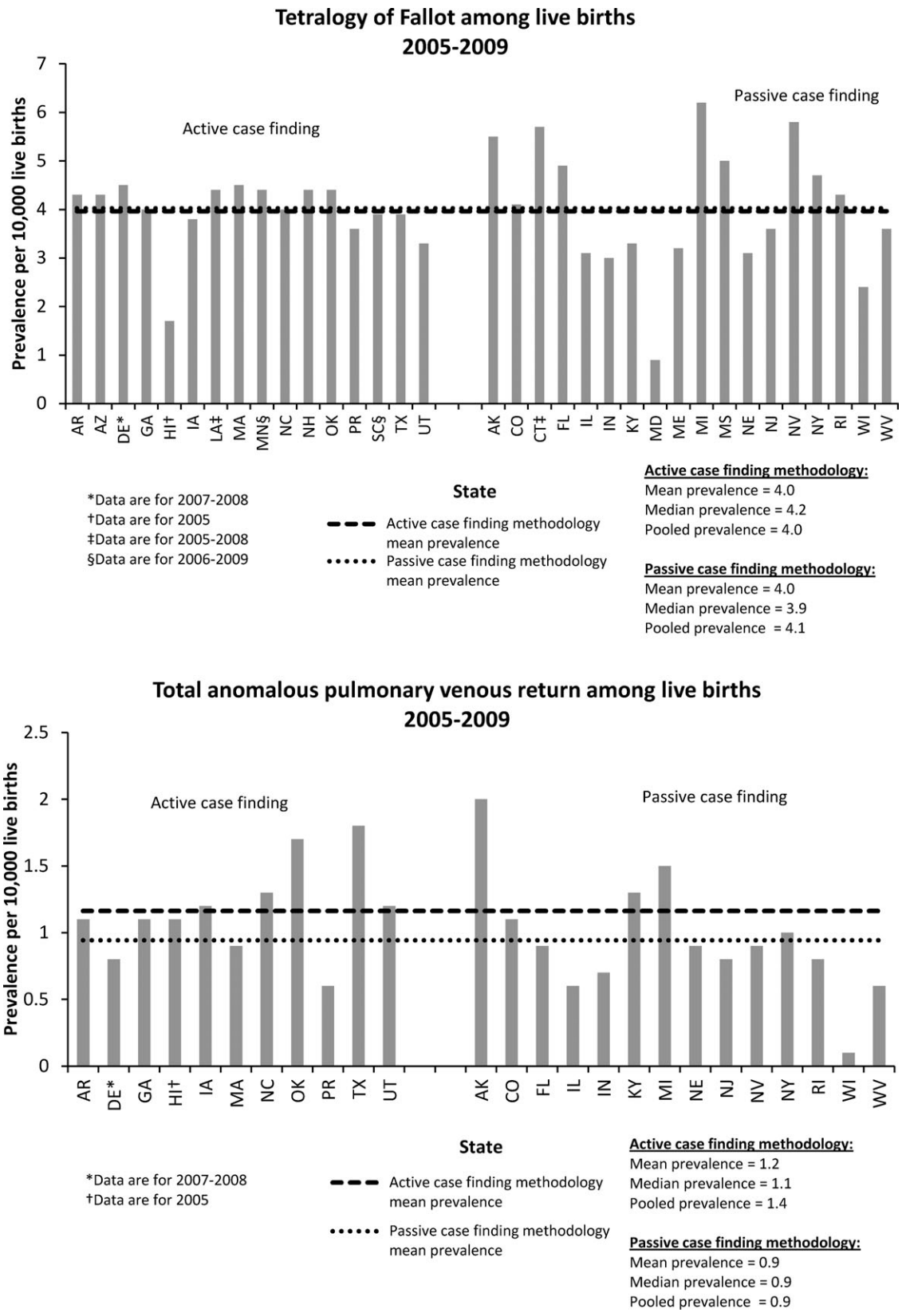
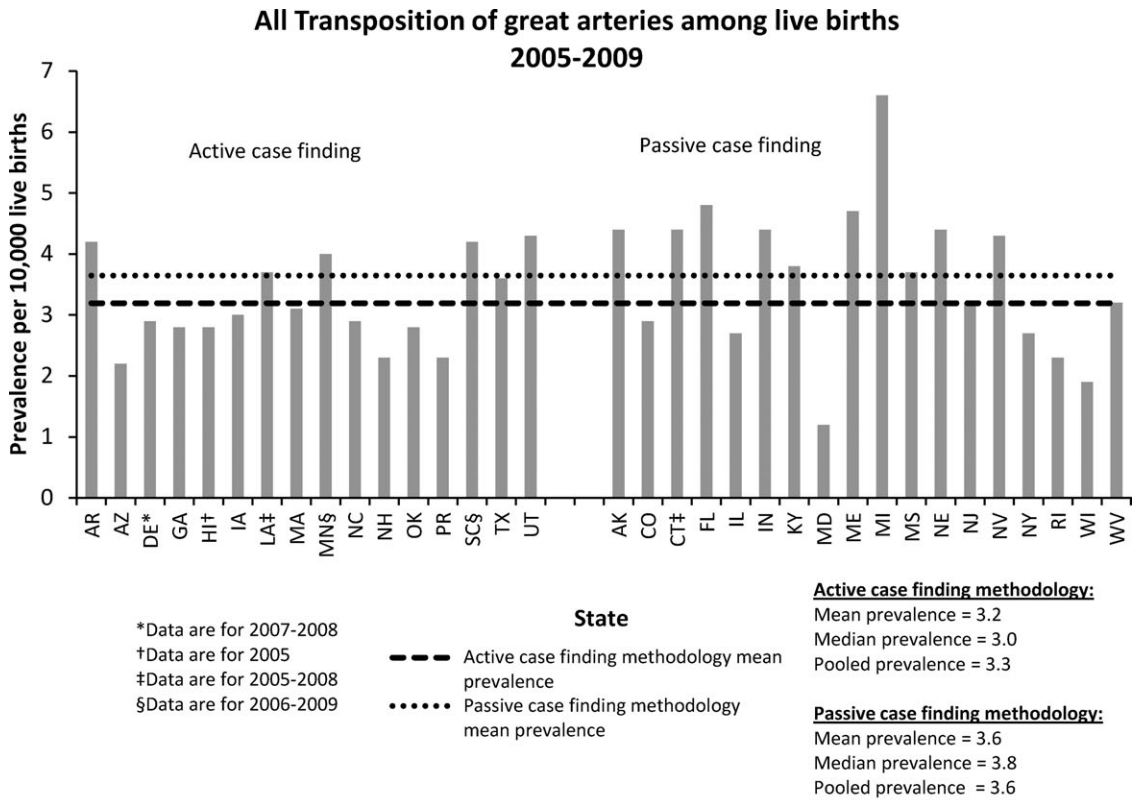


Figure 1. (Continued)



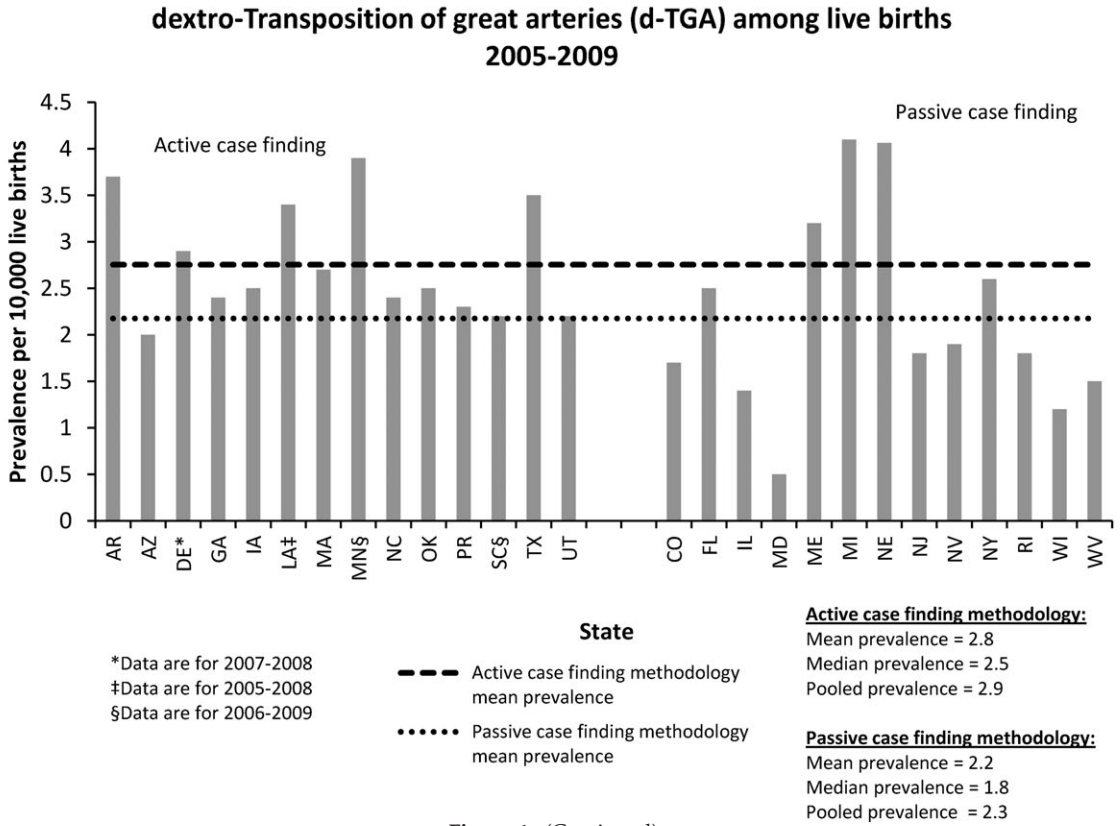
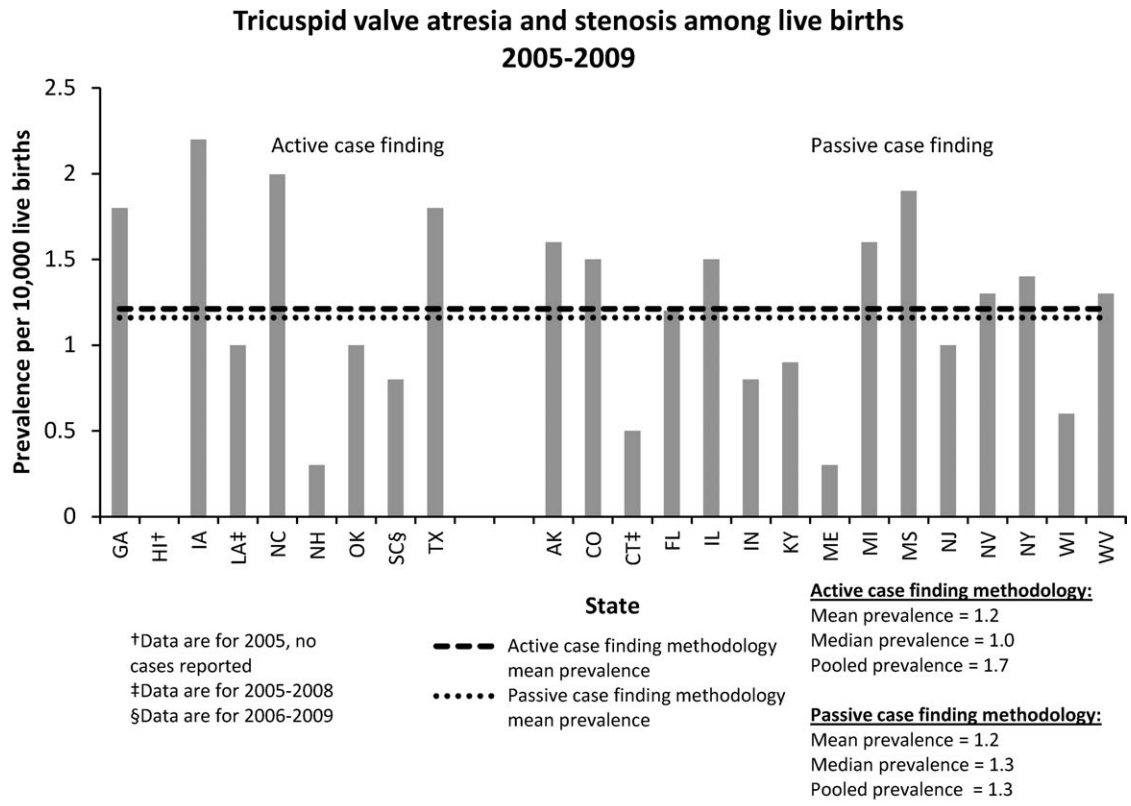


Figure 1. (Continued)



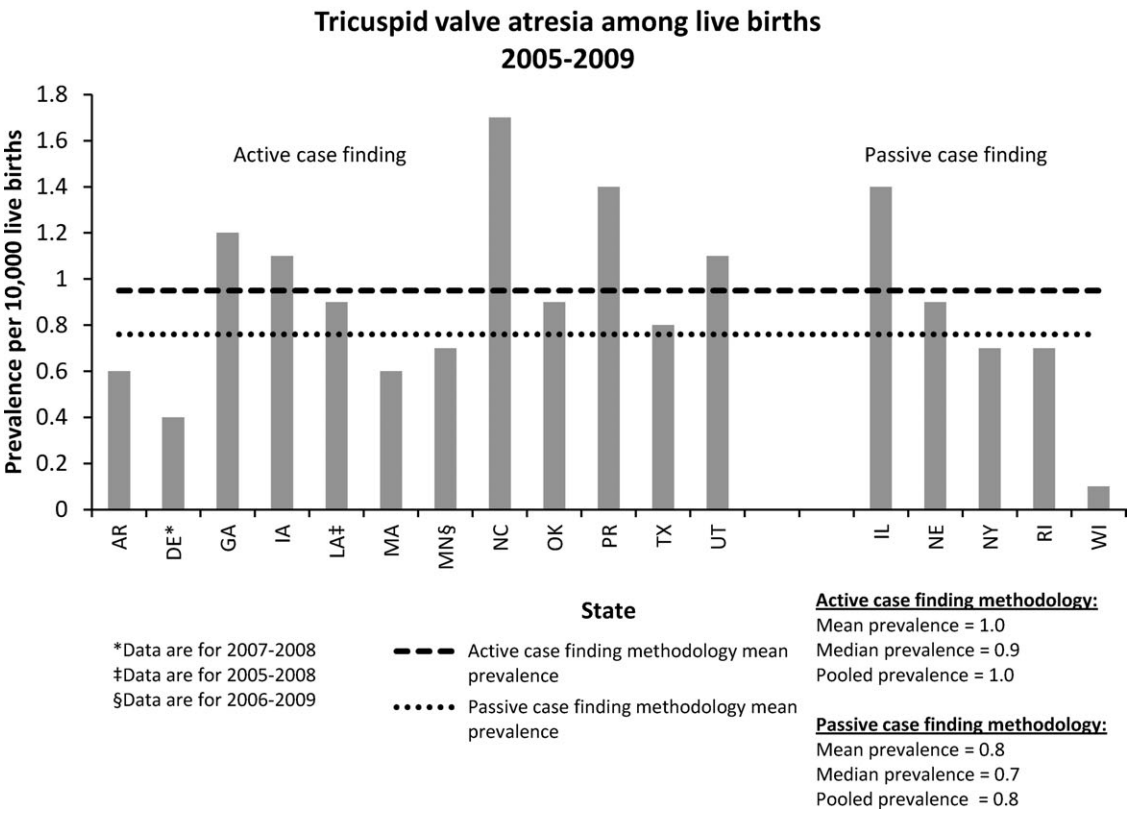


Figure 1. (Continued)

tion-based birth defects surveillance programs in the United States. Because pulse oximetry screening occurs in newborns, data presented in Table 1 include live births only; accompanying state-specific data tables available at S1-S120 include cases resulting from any pregnancy outcome (i.e., live births, stillbirths, and/or pregnancy terminations). For the three CHD diagnoses mentioned above, data are presented for both the broad surveillance category and the stricter definition for pulse oximetry screening.

Figure 1 presents the data from Table 1 graphically for each CCHD, grouped by the program's primary case finding approach (active or passive). Three 'central tendency statistics' are presented: (1) mean prevalence defined as the arithmetic average of the individual program prevalences; (2) median prevalence representing the middle value of the individual program prevalences; and (3) pooled prevalence calculated by dividing the total number of cases across programs by total number of combined live births.

DISCUSSION

Variability in the observed prevalence of CCHDs across states could be due to true differences in prevalence; however, other reasons may account for the variability. Some programs could only provide data for select years. Others were unable to provide data on specific defects, because the conditions are not part of their program case inclusion or due to limitations in the coding systems. Another consideration for variability in the reported prevalence includes pregnancy termination practice for prenatally diagnosed cases. Pregnancy terminations likely vary among states based on cultural differences, race/ethnicity, and other factors (Peller et al., 2004). Higher birth prevalence in a given state may reflect fewer pregnancy terminations of prenatally diagnosed cases.

Table 1 indicates whether the program primarily used an active or passive case finding approach to collect the data. Case finding methodology can influence the magnitude of the prevalence estimates (Parker et al., 2010). Programs with active case finding generally have the ability to use a more refined case classification than programs with passive case finding that rely on ICD-9-CM codes alone; Strickland et al. (2008) demonstrated that compared to clinical nomenclature, ICD-9-CM coding can have relatively low sensitivity and high false-positive fraction for some CHDs. The lack of refinement in the coding leads to the inability to distinguish specific CCHDs (e.g., one code for tricuspid valve atresia and stenosis instead of separate codes for each subtype). Programs can improve their data quality through case confirmation and expert case review. In addition, inclusion of pediatric cardiology centers as a data source can improve and refine case ascertainment (Bedard et al., 2012). Given that individual CCHDs are rare, missing or over-reporting even a few cases may strongly affect the reported prevalence. Figure 1 shows that although prevalence among the programs can vary greatly, the mean prevalences are similar for some CCHDs (e.g., pulmonary valve atresia).

CONCLUSION

As CCHD screening is being implemented across the United States, population-based birth defects surveillance programs can provide useful data to assist with the evaluation of CCHD screening by providing the ongoing and timely evidence to assess the sensitivity, specificity, and positive predictive value (Olney and Botto, 2012). The information gathered by birth defects surveillance programs could also provide an initial basis to assess outcomes, beginning from infant mortality and expanding, depending on a program's ability and resources, to include ultimately an evaluation of the cost effectiveness of CCHD screening.

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Appendix Table 1
ICD-9-CM and CDC/BPA Procedure Codes for 47 Birth Defects Reported in the NBDPN Annual Report

Birth Defects	ICD-9-CM Codes	CDC/BPA Codes
Central nervous system		
Anencephalus	740.0–740.1	740.00–740.10
Spina bifida w/o anencephalus	741.0–741.9 w/o 740.0–740.10	741.00–741.99 w/o 740.0–740.10
Hydrocephalus w/o spina bifida	742.3 w/o 741.0, 741.9	742.30–742.39 w/o 741.00–741.99
Encephalocele	742.0	742.00–742.09
Microcephalus	742.1	742.10
Eye		
Anophthalmia/microphthalmia	743.0, 743.1	743.00–743.10
Congenital cataract	743.30–743.34	743.32
Aniridia	743.45	743.42
Ear		
Anotia/microtia	744.01, 744.23	744.01, 744.21
Cardiovascular		
Common truncus	745.0	745.00
Transposition of great arteries	745.10, 745.11, 745.12, 745.19 (Note: for CCHD screening, 745.10 only)	745.10–745.19 (excluding 745.13, 745.15, 745.18) (Note: for CCHD screening, only 745.10, 745.11, 745.14, 745.19)
Tetralogy of Fallot	745.2	745.20–745.21, 747.31
Ventricular septal defect	745.4	745.40–745.49 (excluding 745.487, 745.498)
Atrial septal defect	745.5	745.51–745.59
Atrioventricular septal defect (endocardial cushion defect)	745.60, 745.61, 745.69	745.60–745.69, 745.487
Pulmonary valve atresia and stenosis	746.01, 746.02 (Note: for CCHD screening, 746.01 only)	746.00–746.01 (Note: for CCHD screening, 746.00 only)
Tricuspid valve atresia and stenosis	746.1	746.10 (excluding 746.105) (Note: for CCHD screening, 746.10 excluding 746.105 and 746.106)
Ebstein anomaly	746.2	746.20
Aortic valve stenosis	746.3	746.30
Hypoplastic left heart syndrome	746.7	746.70
Patent ductus arteriosus	747.0	747.00
Coarctation of aorta	747.10	747.10–747.19
TAPVR	747.41	747.42
Orofacial		
Cleft palate w/o cleft lip	749.0	749.00–749.09
Cleft lip with and w/o cleft palate	749.1, 749.2	749.10–749.29
Choanal atresia	748.0	748.0
Gastrointestinal		
Esophageal atresia/tracheoesophageal fistula	750.3	750.30–750.35
Rectal and large intestinal atresia/stenosis	751.2	751.20–751.24
Pyloric stenosis	750.5	750.51
Hirschsprung disease (congenital megacolon)	751.3	751.30–751.34
Biliary atresia	751.61	751.65
Genitourinary		
Renal agenesis/hypoplasia	753.0	753.00–753.01
Bladder exstrophy	753.5	753.50
Obstructive genitourinary defect	753.2, 753.6	753.20–753.29 and 753.60–753.69
Hypospadias	752.61	752.60–752.62 (excluding 752.61 and 752.621)
Epispadias	752.62	752.61
Musculoskeletal		
Reduction deformity, upper limbs	755.20–755.29	755.20–755.29
Reduction deformity, lower limbs	755.30–755.39	755.30–755.39

Appendix Table 1
ICD-9-CM and CDC/BPA Procedure Codes for 47 Birth Defects Reported in the NBDPN Annual Report (Continued)

Birth Defects	ICD-9-CM Codes	CDC/BPA Codes
Gastroschisis	756.79	756.71
Omphalocele	756.79	756.70
Congenital hip dislocation	754.30, 754.31, 754.35	754.30
Diaphragmatic hernia	756.6	756.61
Chromosomal		
Trisomy 13	758.1	758.10–758.19
Down syndrome	758.0	758.00–758.09
Trisomy 18	758.2	758.20–758.29
Other		
Fetus or newborn affected by maternal alcohol use	760.71	760.71
Amniotic bands	No code	658.80

ICD-9-CM, International Classification of Diseases, 9th Revision, Clinical Modification; CDC/BPA, Centers for Disease Control and Prevention/British Pediatric Association; NBDPN, National Birth Defects Protection Network; w/o, without; CCHD, critical congenital heart defect; TAPVR, total anomalous pulmonary venous return.

Appendix Table 2
Case Definition for the Seven Primary Targeted Critical Congenital Heart Defects

Common Truncus (Truncus Arteriosus or TA)	
Description	Failure of separation of the aorta and the pulmonary artery, resulting in a single common arterial trunk carrying blood from the heart to both the body and lungs.
Inclusions	Common truncus Truncus arteriosus (TA) Persistent truncus arteriosus
Exclusions	Aorto-pulmonary window
ICD-9-CM codes	745.0
CDC/BPA codes	745.00 (remove 745.01, aortic septal defect including aorto-pulmonary window)
Diagnostic methods	Truncus arteriosus is conclusively diagnosed only through direct visualization of the heart by cardiac imaging (typically echocardiography but also MRI), catheterization, surgery, or autopsy. A clinical diagnosis is considered insufficient to make the diagnosis.
Prenatal diagnoses not confirmed postnatally	These conditions may be included as cases when only diagnosed prenatally with some degree of certainty by a pediatric cardiologist through fetal echocardiography. Live-born children who survive should always have confirmation of the defect postnatally.

Additional information:
A ventricular septal defect is often present in association with truncus defects and should be coded separately.
Truncus arteriosus is one of several abnormalities of the outflow tract of the heart known as conotruncal defects. Some infants (1 in 5 to 1 in 3) with these defects have an interstitial deletion on the short arm of chromosome 22 (22q11.2 deletion). This deletion is reliably diagnosed by fluorescent in situ hybridization (FISH) or microarray technology and may be missed by routine karyotype analysis.

Transposition of the Great Arteries (TGAs)	
Description	Transposition of the aorta and the pulmonary artery such that the aorta arises from the right ventricle (instead of the left) and the pulmonary artery arises from the left ventricle (instead of the right).
Inclusions	Complete transposition (dextro-TGA [d-TGA] w/o a ventricular septal defect [VSD]). Corrected transposition (levo-TGA [L-TGA] (exclude for CCHD screening). Incomplete transposition (dextro-TGA [d-TGA] with a VSD). Transposition of the great arteries (TGAs), not otherwise specified transposition of the great vessels (TGVs).
Exclusions	N/A.
ICD-9-CM codes	745.10, 745.11, 745.12, 745.19
For CCHD screening	745.10 (d-TGA only)
CDC/BPA codes	745.10–745.19, excluding 745.13 (Double outlet right ventricle [DORV] with normally related great vessels), 745.15 (DORV, relationship of great vessels not specified), 745.18 (other specified TGA).
For CCHD screening	745.10 (TGA, complete, no VSD), 745.11 (TGA incomplete, with VSD), 745.14 (DORV with transposed great vessels), 745.19 (unspecified TGA).
Diagnostic methods	d-TGA is conclusively diagnosed through direct visualization of the heart by cardiac echo (echocardiography), catheterization, surgery, or autopsy.

Appendix Table 2
Case Definition for the Seven Primary Targeted Critical Congenital Heart Defects (Continued)

Prenatal diagnoses not confirmed postnatally	These conditions may be included as cases when only diagnosed prenatally with some degree of certainty by a pediatric cardiologist through fetal echocardiography. Live-born children who survive should always have confirmation of the defect postnatally.

Additional information:

d-TGA is a defect in which the right ventricle connects to the aorta and the left ventricle connects to the pulmonary artery (ventriculo-arterial discordance). An associated communication between the pulmonary and systemic circulations may be present, such as a ventricular septal defect (incomplete TGA). If a VSD is not present (intact ventricular septum) it is complete TGA. If the coding system does not include unique codes to differentiate TGA with and w/o a VSD (complete vs. incomplete), the VSD should be coded separately when present. If a communication (e.g., ASD) is created during catheterization or surgery, the ASD is not coded as a defect.

DORV is a distinct defect coded within this category. DORV can occur with normally or malposed great vessels. Strictly speaking, the great arteries in DORV cannot be transposed, and are malposed, although ICD-9 coding is limited. If a coding system (e.g., CDC/BPA), can distinguish these phenotypes, only the DORV with transposed vessels is included in the larger transposition category or the CCHD subcategory. If codes cannot distinguish phenotypes, all DORVs are included in the transposition category, but not CCHD subcategory.

L-TGA (corrected transposition) is a defect in which the right atrium connects to anatomic left ventricle (atrio-ventricular discordance) and this ventricle connects to the pulmonary artery (ventriculo-arterial discordance). Because oxygen-poor blood goes to the lungs and oxygen-rich blood goes to the body, circulation is normal (corrected transposition) as long as there are no other defects. L-TGA is included in the broader transposition category for surveillance, but excluded from CCHD subcategory because it is not considered critical.

Transposition of the great arteries is one of several abnormalities of the outflow tract of the heart known as conotruncal defects. Very few infants with these defects have an interstitial deletion on the short arm of chromosome 22 (22q11.2 deletion). This deletion is reliably diagnosed by fluorescent in situ hybridization (FISH) or microarray technology and may be missed by routine karyotype analysis.

Tetralogy of Fallot (TOF)

Description	The simultaneous presence of a VSD, pulmonic and subpulmonic stenosis, a malpositioned aorta that overrides the ventricular septum, and right ventricular hypertrophy.
Inclusions	Pentalogy of Fallot – Tetralogy of Fallot with an associated inter-atrial communication (patent foramen ovale [PFO] or atrial septal defect [ASD]). Tetralogy of Fallot Tet TOF Pulmonary atresia with VSD (see 'Additional information')
Exclusions	Simultaneous occurrence of a VSD and pulmonary stenosis that has TOF physiology but has not been diagnosed as Tetralogy of Fallot. Some coding systems may also include Trilogy of Fallot, or Fallot's Triad – the simultaneous presence of an ASD, pulmonic stenosis, and right ventricular hypertrophy. This is ***not*** to be included as TOF.
ICD-9-CM codes	745.2
CDC/BPA codes	745.20–745.21, 747.31 (Note: code 746.84 has been removed).
Diagnostic methods	While Tetralogy of Fallot may be suspected by clinical presentation, it may be conclusively diagnosed only through direct visualization of the heart by cardiac echo (echocardiography), catheterization, surgery, or autopsy.
Prenatal diagnoses not confirmed postnatally	These conditions may be included as cases when only diagnosed prenatally with some degree of certainty by a pediatric cardiologist through fetal echocardiography. Live-born children who survive should always have confirmation of the defect postnatally.

Additional information:

Tetralogy of Fallot is one of several abnormalities of the outflow tract of the heart known as conotruncal defects. Some infants (approximately 1 in 7) with this defect have an interstitial deletion on the short arm of chromosome 22 (22q11.2 deletion). This deletion is reliably diagnosed by fluorescent in situ hybridization (FISH) or microarray technology and may be missed by routine karyotype analysis.

Tetralogy of Fallot is on a spectrum with other defects having important physiologic and coding differences among systems as seen here in the Table.

CCHD	ICD-9	CDC/BPA
Pulmonary valve stenosis	746.02	746.01
Pulmonary atresia with intact ventricular septum	746.01	746.00
Pulmonary atresia with VSD (like Tetralogy of Fallot)	–	747.31
Tetralogy of Fallot	745.2	745.20–21

Pulmonary atresia with a VSD is similar to severe forms of Tetralogy of Fallot and is included for surveillance. There is no specific code depicting valvular pulmonary atresia with VSD; hence in CDC/BPA the code 747.31 ('pulmonary artery atresia with septal defect') is used. For pulmonary valvular atresia w/o a VSD (intact ventricular septum), the code 746.00 ('atresia, hypoplasia of pulmonary valve') is used – see separate section on Pulmonary valve atresia/stenosis.

Appendix Table 2
Case Definition for the Seven Primary Targeted Critical Congenital Heart Defects (Continued)

Pulmonary valve atresia and stenosis

Description	Pulmonary valve atresia – Lack of patency, or failure of formation altogether, of the pulmonary valve, resulting in obstruction of blood flow from the right ventricle to the pulmonary artery. Pulmonary valve stenosis – Obstruction or narrowing of the pulmonary valve, which may impair blood flow in varying degrees of severity from the right ventricle to the pulmonary artery.
Inclusions	Pulmonary valve atresia with intact ventricular septum. Pulmonary valve stenosis (PS) (most cases of PS). Pulmonic stenosis (PS – valve not specified).
Exclusions	Atresia or stenosis of the main or branch (right or left) pulmonary arteries, not involving the pulmonary valve. Pulmonary stenosis that occurs as part of Tetralogy or Pentalogy of Fallot. Supra-valvular or sub-valvular pulmonic stenosis.
ICD-9-CM codes	746.01 (Pulmonary atresia), 746.02 (pulmonary valve stenosis).
For CCHD screening	746.01 only (pulmonary atresia, intact ventricular septum).
CDC/BPA codes	746.00 (pulmonary valve atresia), 746.01 (pulmonary valve stenosis).
For CCHD screening	746.00 only (pulmonary atresia, intact ventricular septum).
Diagnostic methods	Although pulmonary valve atresia or stenosis may be suspected by clinical presentation, it may be conclusively diagnosed only through direct visualization of the heart by cardiac echo (echocardiography), catheterization, surgery, or autopsy.
Prenatal diagnoses not confirmed postnatally	Although these conditions may be identified by prenatal ultrasound, they should not be included in surveillance data w/o postnatal confirmation. In addition, the absence of pulmonary valve atresia or stenosis on prenatal ultrasound does not necessarily mean that it will not be diagnosed after delivery.

Additional information:

These defects have important physiologic and coding differences among systems as seen here in the Table, which is also discussed in the Tetralogy of Fallot section.

CCHD	ICD-9	CDC/BPA
Pulmonary valve stenosis	746.02	746.01
Pulmonary atresia with intact ventricular septum	746.01	746.00
Pulmonary atresia with VSD (like Tetralogy of Fallot)	–	747.31
Tetralogy of Fallot	745.2	745.20–21

Pulmonary valve atresia or stenosis may occur with or w/o a coexisting ventricular septal defect. For pulmonary valve atresia w/o a VSD (intact ventricular septum), the CDC/BPA code 746.00 (''atresia, hypoplasia of pulmonary valve'') is used, corresponding to the ICD-9-CM code 746.01. However, in CDC/BPA, 746.01 refers to pulmonary valve stenosis.

Pulmonary atresia with a VSD is similar to severe forms of Tetralogy of Fallot, and is included with Tetralogy of Fallot for surveillance. There is no specific code depicting valvular pulmonary atresia with VSD; hence in CDC/BPA the code 747.31 ('pulmonary artery atresia with septal defect') is used. If a case has codes for both Pulmonary atresia and Tetralogy of Fallot, it should be counted as Tetralogy of Fallot.

Tricuspid valve atresia and stenosis

Description	Tricuspid valve atresia – Lack of patency, or failure of formation altogether, of the tricuspid valve, resulting in obstruction of blood flow from the right atrium to the right ventricle. Tricuspid valve stenosis – Obstruction or narrowing of the tricuspid valve, which may impair blood flow from the right atrium to the right ventricle.
Inclusions	Tricuspid atresia. Tricuspid stenosis.
Exclusions	Tricuspid regurgitation w/o specific mention of tricuspid atresia or stenosis.
ICD-9-CM codes	746.1
CDC/BPA codes	746.100 (tricuspid atresia), 746.106 (tricuspid stenosis) (excluding 746.105 – tricuspid insufficiency).
For CCHD screening	746.100 only. Note: Only the tricuspid atresia is a CCHD. Many cases of tricuspid stenosis are not critical.
Diagnostic methods	Although tricuspid valve atresia or stenosis may be suspected by clinical presentation, it may be conclusively diagnosed only through direct visualization of the heart by cardiac echo (echocardiography), catheterization, surgery, or autopsy.
Prenatal diagnoses not confirmed postnatally	Although these conditions may be identified by prenatal ultrasound, they should not be included in surveillance data w/o postnatal confirmation. In addition, the absence of tricuspid valve atresia or stenosis on prenatal ultrasound does not necessarily mean that it will not be diagnosed after delivery.

Appendix Table 2
Case Definition for the Seven Primary Targeted Critical Congenital Heart Defects (Continued)

Additional information:
In the ICD-9-CM coding system, it is impossible to distinguish mild cases of tricuspid stenosis, from the critical CHD of tricuspid atresia. Reports using only the code 745.1 (ICD-9-CM) may include both defects, and therefore may not be an accurate estimate of the number of cases of either defect individually.

Hypoplastic left heart syndrome (HLHS)	
Description	A condition in which the structures on the left side of the heart and the aorta are extremely small. Classically, this condition includes hypoplasia of the left ventricle, atresia, or severe hypoplasia of both the mitral and aortic valves, hypoplasia of the aortic arch, and coarctation of the aorta.
Inclusions	Any diagnosis of hypoplastic left heart syndrome, regardless of whether all conditions in the classic definition are present.
Exclusions	Hypoplasia or diminished size of the left ventricle alone w/o involvement of other structures on the left side of the heart or the aorta. Hypoplastic left heart or small left ventricle that occurs as part of another complex heart defect, such as an endocardial cushion defect (''unbalanced'' AV canal).
ICD-9-CM codes	746.7
CDC/BPA codes	746.70
Diagnostic methods	Although hypoplastic left heart may be suspected by clinical presentation, examination, and echocardiogram (EKG) changes, it may be conclusively diagnosed only through direct visualization of the heart by cardiac echo (echocardiography), catheterization, surgery, or autopsy.
Prenatal diagnoses not confirmed postnatally	These conditions may be included as cases when only diagnosed prenatally with some degree of certainty by a pediatric cardiologist through fetal echocardiography. Live-born children who survive should always have confirmation of the defect postnatally.

Total anomalous pulmonary venous return (TAPVR)	
Description	A condition in which all four pulmonary veins connect anomalously into the systemic venous circulation to the right atrium or the body (systemic veins) instead of the left atrium; often occurs with other cardiac defects.
Inclusions	TAPVR (total anomalous pulmonary venous return). TAPVC (total anomalous pulmonary venous connection). TAPVD (total anomalous pulmonary venous drainage).
Exclusions	If not all 4 veins are visibly connecting/draining anomalously (e.g., Partial Anomalous Venous Return, ICD-9-CM code 747.42).
ICD-9-CM codes	747.41
CDC/BPA codes	747.42
Diagnostic methods	Although TAPVR may be suspected by clinical presentation, it may be conclusively diagnosed only through direct visualization of the heart by cardiac echo (echocardiography), catheterization, surgery, or autopsy. The difficulty in viewing all four veins may mean that several echocardiograms may be needed to confirm the diagnosis.
Prenatal diagnoses not confirmed postnatally	TAPVR is difficult to identify prenatally. If identified by prenatal ultrasound, it should not be included in surveillance data w/o postnatal confirmation. In addition, the absence of TAPVR on prenatal ultrasound does not necessarily mean that it will not be diagnosed after delivery.

Additional information:
Total anomalous pulmonary venous return and partial anomalous pulmonary venous return have not been shown to be developmentally related, although they share a similar description. Also, there are subtle differences in the meaning of anomalous venous connection, return, and drainage, but the terms are often used interchangeably.

ICD-9-CM, International Classification of Diseases, 9th Revision, Clinical Modification; CDC/BPA Codes, Centers for Disease Control and Prevention/British Pediatric Association Classification of Diseases; w/o, without; CCHD, critical congenital heart defect; N/A, not applicable; ASD, atrial septal defect.