The Accuracy of Hospital Discharge Diagnosis Codes for Major Birth Defects: Evaluation of a Statewide Registry With Passive Case Ascertainment

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Context: Birth defects prevention, research, education, and support activities can be improved through surveillance systems that collect high-quality data. **Objective:** To estimate the overall and defect-specific accuracy of Florida Birth Defects Registry (FBDR) data, describe reasons for false-positive diagnoses, and evaluate the impact of statewide case confirmation on frequencies and prevalence estimates. **Design:** Retrospective cohort evaluation study. **Participants:** A total of 8479 infants born to Florida resident mothers between January 1, 2007, and December 31, 2011, and diagnosed with 1 of 13 major birth defects in the first year of life. **Main Outcome Measures:** Positive predictive value: calculated overall (proportion of FBDR-identified cases confirmed by medical record review, regardless of which of the 13 defects were confirmed) and defect-specific (proportion of FBDR-identified cases confirmed by medical record review *with the same defect*) indices. **Results:** The FBDR's overall positive predictive value was 93.3% (95% confidence interval, 92.7-93.8); however, there was variation in accuracy across defects, with positive predictive values ranging from 96.0% for gastroschisis to 54.4% for reduction deformities of the lower limb. Analyses suggested that *International Classification of Diseases, Ninth Edition, Clinical Modification*, codes, upon which FBDR diagnoses are based, capture the general occurrence of a defect well but often fail to identify the specific defect with high accuracy. Most infants with false-positive diagnoses had some type of birth defect that was incorrectly documented or coded. If prevalence rates reported by the FBDR for these 13 defects were adjusted to incorporate statewide case confirmation, there would be an overall 6.2% rate reduction from 82.6 to 77.5 per 10 000 live births. **Conclusions:** A statewide birth defects surveillance system, relying on linkage of administrative databases, is capable of achieving high accuracy (>93%) for identifying infants with any one of the 13 major defects included in this study. However, the level of accuracy and the ability to minimize false-positive diagnoses vary depending on the defect.

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Despite prevention, education, support, and research efforts, birth defects remain common, costly, and critical.[1,2] In the United States, every 4.5 minutes an infant is born with a birth defect—that is 120 000 affected infants and families each year.[2] Globally, is it estimated that nearly 8 million children are born with a birth defect with genetic or partially genetic origin and at least hundreds of thousands more with postconception defects of teratogenic origin.[3] They are among the leading causes of infant mortality and have long-term health consequences that affect every organ and body system.[3] When available, surveillance programs that collect, analyze, and interpret data from their tracking systems not only play a vital role in research and education that help prevent birth defects but may also engage in various planning, support, and referral activities that lessen their negative health and economic impact.[4-6] However, the effectiveness of a surveillance program depends on the quality of its data; that is, how timely, complete, and accurate they are. Programs using active case ascertainment, a labor-intensive process involving staff finding cases through direct review of primary data sources (eg, medical records, hospital/nursery logs, autopsy reports), often have high completeness of ascertainment and accuracy. Conversely, passive ascertainment that involves physician or facility filing of case reports or identification of cases using secondary data sources (eg, hospital discharge data and vital statistics records) is often characterized by suboptimal metrics of data quality.[6,7] This is important because recent decreases in funding, the high cost of active case ascertainment, and the dramatic growth of clinical and administrative databases have led to programs relying on passive case ascertainment that uses data not originally created for the purposes of establishing a birth defects registry.[8-10] In fact, more than 60% of the states reporting population-based prevalence data to the National Birth Defects Prevention Network and the Centers for Disease Control and Prevention (CDC) reported using a primarily passive case-finding methodology.[11] Therefore, understanding the accuracy of these databases for identifying major birth defects is essential.

Florida has the second largest population-based surveillance system in the United States that relies predominantly on passive case ascertainment. Operated by the Florida Department of Health (FDOH) and a consortium of partners, the Florida Birth Defects Registry (FBDR) currently has a 14-year inventory of children with structural, functional, and biochemical abnormalities, covering a source population of more than 3 million live births. FBDR data have been used to support local investigations, epidemiologic studies,[12-15] health outcomes research,[16-19] and national collaborative projects.[20,21] However, since cases in the FBDR have historically been defined on the basis of diagnosis codes present in hospital discharge or service-related databases, which are susceptible to underascertainment and reporting errors,[8,9,22-24] there has been concern with the internal validity of results. We previously estimated the completeness of ascertainment of the FBDR for selected defects in 2003-2006 by linking the FBDR to an enhanced surveillance system in a 9-county catchment area and using capture-recapture modeling.[7] Overall completeness was nearly 87% but varied markedly across defects, from 45.6% for anencephaly to 88.6% for Down syndrome. Recently, the FDOH completed a 5-year project whereby abstractors reviewed medical records to confirm (or refute) every infant reported by the FBDR to have been diagnosed with 1 of 13 major birth defects in the first year of life. The purpose of the study is to (1) estimate the overall and defect-specific accuracy of the FBDR; (2) describe, for each defect, the most common reasons for false-positive FBDR diagnoses; and (3) evaluate the impact of implementing statewide medical record review to confirm FBDR cases on reported frequencies and prevalence estimates.

Methods and Materials

In this evaluation study, we linked data from Florida's statewide, population-based surveillance program to detailed information collected as part of a statewide case confirmation project for selected birth defects of major public health importance. The source population in which surveillance and case confirmation activities were conducted included all infants born to Florida resident mothers in the 5-year period from January 1, 2007, to December 31, 2011.

Florida birth defects registry

In Florida on July 4, 1999, congenital anomalies (henceforth referred to as *birth defects*) were added to the list of reportable conditions that may significantly impact health, under the authority of Florida Statute (F.S. 381.0031),[25] and further specified by Florida Administrative Code 64D-3.035.[26] The FBDR was created later that year by the FDOH to serve as a population-based birth defects surveillance program to protect, promote, and improve the health of people in Florida by detecting, investigating, and preventing birth defects. Details of the FBDR have been described previously.[7,27] Briefly,

the FBDR defines a "case" based on the following inclusion criteria: (1) the biological mother is a Florida resident; (2) the infant is born alive and is diagnosed in the first year of life with 1 or more structural, genetic, or other specified birth outcomes that can adversely affect the infant's health and development; and (3) the infant was delivered on or after January 1, 1998. The FBDR is a surveillance system whose passive case ascertainment methodology relies on the acquisition, cleaning, and linking of a number of administrative data sources. The underlying source population is first defined by all birth certificates in which Florida is documented as the maternal state of residence. Interstate data sharing enables collection of Florida resident births that occur outside Florida. Then, using a hierarchical, deterministic data linkage strategy,[28] supplemental data sources with clinical information are linked to the birth certificate record. Although the data sources have changed over time[27] due to funding restrictions for some data sets or the identification of new, reliable data sets, their use is the same. These supplemental data sources have included: (1) Agency for Health Care Administration inpatient (1998-2011), outpatient (1998-2011), and emergency department (2010-2011) hospital discharge records; (2) Children's Medical Services Regional Perinatal Intensive Care Center data (1998-2008), Early Steps program data (1998-2008), and Minimum Data Set (encounter-level, service-related) records (1998-2007); and (3) Florida Bureau of Vital Statistics infant death (2009-2011) records. Once linked, each record from each source contains *International Classification of Diseases, Ninth Edition, Clinical Modification* (*ICD-9-CM*), diagnosis codes that are used to identify birth defects. Infant death certificates instead contain 10th edition (*ICD-10*) diagnosis codes. The final FBDR for a given annual birth cohort consists of an unduplicated inventory of cases that have 1 or more "included" birth defect codes (primarily but not restricted to *ICD-9-CM* code range 740-759 and *ICD-10* "Q" codes). If a case has multiple birth defects, all are documented. The FBDR currently has published data covering the 1998-2011 birth cohorts available at www.fbdr.org.

Statewide case confirmation project

As funding became available, the FDOH and the University of South Florida Birth Defects Surveillance Program have collaborated on several "enhanced" surveillance projects. Although objectives have varied, a characteristic shared among all projects and indicative of the enhanced methodology has been medical record review by trained abstractors to confirm each suspected case. In 2007, with funding from the CDC (grant number U38-EH000941), Florida began operating an Environmental Public Health Tracking enhanced surveillance project in 14 of its 67 counties, covering more than 70% of resident live births in the state. The objective of the project was to enhance the quality of birth defects surveillance data to effect meaningful linkages between birth defects outcome data and environmental hazard or exposure data. The staple hallmarks of generating high-quality data were to (1) implement more active case finding by contacting hospitals, reviewing labor and delivery logs, neonatal intensive care unit admission lists, and other clinical records, and (2) confirming cases through extensive medical record review. Thirteen birth defects were chosen for the project and were selected on the basis of public health importance, consistency of reporting across states in the United States, and the potential for causal links to environmental exposures. These defects included 2 neural tube defects (anencephaly, spina bifida), 3 critical congenital heart defects (hypoplastic left heart syndrome, tetralogy of Fallot, and transposition of the great arteries), upper and lower limb reduction defects, orofacial clefts, gastroschisis, hypospadias, and Down syndrome (Table 1). In 2010, additional funds from the CDC supported building on this existing infrastructure and expanding the case confirmation aspect of the Environmental Public Health Tracking project to cover the entire state and to focus on all cases identified by the passive case-finding system (FBDR) as having 1 of the 13 included defects. Birth Defects Surveillance Program staff queried the FBDR not only to ascertain all cases and their documented defects but also to identify all medical encounters (inpatient, outpatient, emergency department) that each case had during the first year of life. This way, medical records at all facilities in which the defect may have been diagnosed would be reviewed. If review of all records for a particular case was already performed as part of the Environmental Public Health Tracking project, those data were imported into the case confirmation database.[29] For all other FBDR-identified cases, medical records at all facilities and for all encounters were requested and subsequently reviewed, either on-site or through remote access to hospital information management databases. All defects confirmed were coded using a CDC-modified version of the British Pediatric Association coding system, which provides higher specificity than *ICD-9-CM* diagnosis codes. For false-positive diagnoses—FBDR-identified cases that after medical record review were deemed to not have the documented birth defect(s)—detailed notes were recorded by the abstractor to understand what defects were actually diagnosed, if any. The case confirmation project was implemented to cover the 2007-2011 FBDR cohorts.

TABLE 1 ● Diagnosis Codes Used to Identify Individual Birth Defects and Defect Categories Included in the Florida Birth Defects Registry's Statewide Case Confirmation Project, 2007-2011

Birth Defect/Defect Group	*ICD-9-CM* Codes[ab]	Modified BPA Code(s)[ac]
Central nervous system		
Anencephaly	740.0, 740.1	740.0x, 740.1x
Spina bifida without anencephaly	741.x without 740.0, 740.1	741.x without 740.0x, 740.1x
Congenital heart defects		
Hypoplastic left heart syndrome	746.7	746.700
Tetralogy of Fallot	745.2	745.200
Transposition of the great arteries	745.1x	745.1x
Limb anomalies		
Reduction deformities, upper limb	755.2x	755.2x
Reduction deformities, lower limb	755.3x	755.3x
Orofacial clefts		
Cleft palate without cleft lip	749.0x without 749.1x, 749.2x	749.0x without 749.1x, 749.2x
Cleft lip without cleft palate	749.1x without 749.0x, 749.2x	749.1x without 749.0x, 749.2x
Cleft palate with cleft lip	749.2x or 749.0x with 749.1x	749.2x or 749.0x with 749.1x
Other defects		
Gastroschisis	756.79 with 54.71[d] or 756.73	756.710
Hypospadias	752.61	752.60x, 652.620, 652.625-652.627
Trisomy 21 (Down syndrome)	758.0	758.0x

Abbreviations: BPA, British Pediatric Association; *ICD-9-CM*, *International Classification of Diseases, Ninth Revision, Clinical Modification*.
[a]Presence of 1 or more of the listed codes serves as positive indication of the defect. Similarly, presence of 1 or more of the listed codes following "without" indicates absence of the defect regardless of other listed codes. An "x" indicates that all subcodes with the listed code prefix are part of the defect definition.
[b]*ICD-9-CM* codes were used by the statewide passive surveillance system to identify cases of birth defects. All cases identified by this system underwent medical record review as part of the statewide confirmation project to confirm/rule out each included defect.
[c]Modified BPA codes were used by trained abstractors to identify cases of birth defects following medical record review.
[d]Beginning October 1, 2009, gastroschisis is identified exclusively by a single *ICD-9-CM* diagnosis code (756.73). Prior to that cases of gastroschisis were identified using the presence of both the nonspecific 756.79 *ICD-9-CM* diagnosis code ("other congenital anomalies of abdominal wall") and the 54.71 *ICD-9-CM procedure* code indicating repair of gastroschisis.[14]

Statistical analyses

This study used the positive predictive value (PPV) as the primary measure of the accuracy of the FBDR data. The medical record review implemented by the case confirmation project served as the gold standard diagnosis (case or noncase). Two PPVs were calculated: the overall PPV and the defect-specific PPV. The overall PPV represented the proportion of all FBDR-identified cases that were confirmed by medical record review, regardless of which of the 13 defects were confirmed. In contrast, the defect-specific PPV represented the proportion of FBDR-identified cases that were confirmed by medical record review *with the same defect*. PPV, using both definitions, is presented overall, for each of the 13 individual defects, and for several groups of defects classified by body system. For these analyses, infants were unduplicated at the level of the defect; however, an infant with multiple FBDR-identified defects considered in this study will be included in more than 1 defect-specific analysis.

Whereas some infants deemed noncases following medical record review did not have any of the birth defects under study, it was quite common, particularly for orofacial clefts and critical congenital heart defects, to be misdiagnosed with another defect. In such instances, we have provided summaries of abstractor notes describing the decision to classify suspected cases and noncases, including other defects diagnosed, lack of clinical evidence to confirm defects, and suspected inaccuracies in the FBDR's record linkage. Finally, to examine the potential impact of incorporating case confirmation into the surveillance protocol for these 13 defects, we compared the FBDR's current prevalence rate (number of infants with a birth defect per 10 000 resident live births) with an adjusted rate that removes from the numerator false-positive diagnoses. We then calculated the percent rate reduction for each defect: [(adjusted rate − current FBDR rate)/current FBDR rate × 100]. All inferential tests were 2-tailed, with a 5% type I error rate, and analyses were conducted using SAS software (version 9.4; SAS Institute, Inc, Cary, North Carolina). Since this constitutes an evaluation of the accuracy of FBDR data, whose reporting and surveillance are under the authority of Florida Statue 381.0031, this study was not considered research and is exempt from institutional review board review.

Results

Of 1 026 373 infants born alive to Florida resident mothers between 2007 and 2011, the FBDR identified 8479 infants with 1 or more of the 13 defects under study. The overall and defect-specific PPVs of the FBDR are presented in Table 2. Results of this statewide case confirmation project revealed an overall PPV of 93.3% (95% confidence interval [CI], 92.7-93.8), with 7907 of the 8479 infants suspected as having at least 1 of the 13 included defects being confirmed with 1 or more of the 13. There was significant variation in accuracy across defects, with PPVs ranging from 96.0% for gastroschisis to 54.4% for reduction deformities of the lower limb. When considered together, the FBDR had high accuracy for orofacial clefts (PPV = 95.6%; 95% CI, 94.5-96.7); however, when considered separately, cleft palate without cleft lip, cleft lip without cleft palate, and cleft palate with cleft lip demonstrated markedly lower PPVs (between 81.7% and 88.2%). Similarly, when grouped, FBDR diagnoses are relatively accurate for critical congenital heart defects (PPV = 90.4%; 95% CI, 88.7-92.0), but for hypoplastic left heart syndrome, the PPV is barely above 75% (95% CI, 71.0-80.2). This suggests that *ICD-9-CM* codes, upon which FBDR diagnoses are based, capture the general occurrence of a defect quite well but often fail to identify the specific defect with high accuracy. This is exemplified further in Table 3, which captures those infants the FBDR identified as having one of the included defects but not the correct defect. With minor exceptions, most of the inaccurate diagnoses occur for orofacial clefts and critical congenital heart defects. Infants who actually had cleft palate with cleft lip were most often misdiagnosed in the FBDR as having *either* cleft palate alone (n = 13) *or* cleft lip alone (n = 22). Similarly, infants identified by the FBDR as having cleft palate with cleft lip actually had one or the other but not both (n = 82). The most common errors for critical congenital heart defects were that infants with transposition of the great arteries were incorrectly identified by the FBDR as having hypoplastic left heart syndrome (n = 22) or tetralogy of Fallot (n = 16). Also, 15 infants the FBDR identified as having transposition of the great arteries actually had tetralogy of Fallot (Table 3).

Table 4 summarizes the reasons for all false-positive diagnoses reported by the FBDR, separately for each defect. With the exception of hypospadias and

TABLE 2 Overall and Defect-Specific Positive Predictive Value of the FBDR for Selected Birth Defects, 2007-2011

Birth Defect[a]	Cases reported by the FBDR	Cases Confirmed[b] With Any Included Defect	Overall PPV[c] of the FBDR (95% CI)	Cases Confirmed[b] for the Specified Defect	Defect-Specific PPV[d] of the FBDR (95% CI)
Individual defects					
Anencephaly	60	52	86.7 (78.1-95.3)	51	85.0 (76.0-94.0)
Cleft lip without cleft palate	229	213	93.0 (89.7-96.3)	187	81.7 (76.6-86.7)
Cleft palate without cleft lip	578	547	94.6 (92.8-96.5)	510	88.2 (85.6-90.9)
Cleft palate with cleft lip	523	516	98.7 (97.7-99.6)	434	83.0 (79.8-86.2)
Gastroschisis	477	460	96.4 (94.8-98.1)	458	96.0 (94.3-97.8)
Hypoplastic left heart syndrome	332	283	85.2 (81.4-89.1)	251	75.6 (71.0-80.2)
Hypospadias	3619	3449	95.3 (94.6-96.0)	3434	94.9 (94.2-95.6)
Reduction deformities, upper limb	219	174	79.5 (74.1-84.8)	161	73.5 (67.7-79.4)
Reduction deformities, lower limb	160	95	59.4 (51.8-67.0)	87	54.4 (46.7-62.1)
Spina bifida without anencephaly	294	257	87.4 (83.6-91.2)	255	86.7 (82.9-90.6)
Tetralogy of Fallot	513	473	92.2 (89.9-94.5)	449	87.5 (84.7-90.4)
Transposition of the great arteries	489	470	96.1 (94.4-97.8)	451	92.2 (89.9-94.6)
Trisomy 21 (Down syndrome)	1363	1270	93.2 (91.8-94.5)	1255	92.1 (90.6-93.5)
Defect groups					
Any orofacial cleft	1330	1276	95.9 (94.9-97.0)	1271	95.6 (94.5-96.7)
Any included congenital heart defect	1223	1122	91.7 (90.2-93.3)	1105	90.4 (88.7-92.0)
Any limb reduction deformity	346	250	72.3 (67.5-77.0)	237	68.5 (63.6-73.4)
Any included defect	8479	7907	93.3 (92.7-93.8)	7907	93.3 (92.7-93.8)

Abbreviations: BPA, British Pediatric Association; CI, confidence interval; FBDR, Florida Birth Defects Registry (passive surveillance system); PPV, positive predictive value.
[a]Results are presented at the defect-level. Since infants may have more than 1 defect included in the case confirmation project, the sum of individuals defects in any defect group, and overall, may add to more than group/overall totals. Ordering of defects within the "individual defects" grouping is sorted alphabetically.
[b]Case confirmation protocol included medical record review by a trained abstractor, followed by documentation of each confirmed defect using modified BPA diagnosis codes.
[c]Calculated as: (number of FBDR cases confirmed with *any defect* listed in the table/number of cases of the defect reported by the FBDR) × 100.
[d]Calculated as: (number of FBDR cases confirmed with the *same defect*/number of cases of the defect reported by the FBDR) × 100.

TABLE 3 ● Suspected Cases Reported by the FBDR and Confirmed Through Medical Record Review With 1 or More Included Birth Defects[a] But Not the Correct Defect, 2007-2011[b]

Birth Defect Captured and Reported by the FBDR	Cases	Defect(s) Confirmed Following Medical Record Review[c]											
		CL	CP	CLCP	GA	HLHS	HY	RU	RL	SB	TOF	TGA	T21
Anencephaly	1	0	0	0	0	0	1	0	0	0	0	0	0
CL	26	. . .	3	22	0	0	3	0	0	0	0	0	0
CP	37	20	. . .	13	0	1	1	1	0	0	0	1	2
CLCP	82	55	27	. . .	0	0	3	1	1	0	0	0	0
GA	2	0	0	0	. . .	0	0	1	0	0	1	2	0
HLHS	32	0	2	3	0	. . .	1	1	0	1	3	22	2
HY	15	1	3	1	0	2	. . .	0	0	1	4	0	3
RU	13	0	4	0	2	0	0	. . .	3	0	1	0	3
RL	8	0	4	1	0	0	0	4	. . .	0	0	1	1
SB	2	0	0	0	0	1	0	1	0	. . .	0	1	0
TOF	24	0	1	0	0	1	4	2	0	1	. . .	16	3
TGA	19	0	1	2	0	0	0	1	0	0	15	. . .	4
T21	15	0	5	1	0	1	6	2	0	1	3	2	. . .

Abbreviations: CL, cleft lip without cleft palate; CLCP, cleft palate without cleft lip; CP, cleft palate with cleft lip; FBDR, Florida Birth Defects Registry; GA, gastroschisis; HLHS, hypoplastic left heart syndrome; HY, hypospadias; RL, reduction deformities, lower limb; RU, reduction deformities, upper limb; SB, spina bifida without anencephaly; TGA, transposition of the great arteries; TOF, tetralogy of Fallot; T21, trisomy 21 (Down syndrome).

[a]An "included defect" consists of any defect listed in the table and that was part of the statewide case confirmation project.

[b]During the case confirmation process, abstractors identified a record as a "confirmed case" if it was confirmed through medical record review as having any of the defects included in the case confirmation project. Those without any included defect were classified as "noncases". This table only includes "confirmed cases" who were found to have a different defect than the one captured by the FBDR (eg, the FBDR identified the suspected case as having HLHS, but medical record review found only evidence of TGA).

[c]Each row represents the number of suspected cases identified by the FBDR with that defect. Following medical record review, suspected cases may be confirmed as having 1 or more of the defects included in the case confirmation project; therefore, the sum of confirmed defects for a given row may add to more than the number of cases reported in the second column.

orofacial clefts, most infants with false-positive diagnoses had some type of birth defect that was incorrectly documented or coded. Six of 8 false-positive diagnoses for anencephaly had other cranial/intracranial malformations; 26 of 37 false-positive spina bifida cases had teratomas, lipomas, sacral dimples, tethered cord, and other defects that are commonly seen with spina bifida; and 12 of 17 false-positive gastroschisis cases actually had a ruptured omphalocele. The misdiagnosed critical congenital heart defects were for those infants whose medical record documented a wide range of cardiac defects but without sufficient clinical validation of the FBDR-coded defect. Similarly, those mistakenly in the FBDR for reduction deformity of the limbs often had evidence of shortened extremities, clubfoot, syndactyly, or achondroplasia but not a true reduction deformity. For false-positive Down syndrome cases (n = 93), the majority (n = 62) were infants with dysmorphic features characteristic of Down syndrome but that were ruled out by karyotyping. Abstractors were also able to identify false-positive cases that occurred because of the incorrect linkage of records during the construction of the FBDR, particularly when a hospital discharge record for a twin or triplet was incorrectly linked to a different sibling.

If prevalence rates reported by the FBDR for these 13 defects were adjusted to incorporate the results of a statewide confirmation of suspected cases, there would be an overall 6.2% rate reduction from 82.6 to 77.5 per 10 000 live births (Figure). The impact on individual defects would be varied. The prevalence rates for 3 defects (upper and lower limb reduction deformities, and hypoplastic left heart syndrome) would decrease by more than 20% and only the rates for gastroschisis and hypospadias (2 defects with easily identifiable physical characteristics present at birth) would have less than a 5% reduction. Overall, if surveillance included only these 13 defects, 572 false-positive diagnoses would be removed from the FBDR. The actual number of infants removed would be less since many had at least one of the included defects.

● Discussion

This study demonstrated that a population-based, statewide birth defects surveillance system, relying exclusively on linkage of administrative databases for case ascertainment, is capable of achieving high accuracy (>93%) for the 13 major defects of public health

TABLE 4 ● Summary of Reasons for False-Positive Diagnoses[a] Reported by the FBDR, 2007-2011

Birth Defect Captured and Reported by the FBDR	Total Noncases	Reason			Summary of Abstractor Notes Describing Decision to Classify SCs as Noncases on the Basis of Misdiagnoses
		Data Linkage Errors	No Evidence of Any Defect	Misdiagnoses	
Anencephaly	8	1	1	6	SCs had other major cranial or intracranial malformations including hydranencephaly, encephalocele, abnormal skull shape, and holoproscencephaly.
Cleft lip without cleft palate	16	1	7	8	SCs had other forms of clefts (facial and hairline) or multiple birth defects
Cleft palate without cleft lip	31	2	19	10	SCs had a "high arched palate" or multiple birth defects
Cleft palate with cleft lip	7	1	3	3	SCs had other forms of clefts (facial and hairline) or multiple birth defects
Gastroschisis	17	0	2	15	SCs had a ruptured omphalocele initially diagnosed as gastroschisis or multiple birth defects
Hypoplastic left heart syndrome	49	2	1	46	SCs had ≥1 characteristic of hypoplastic left heart syndrome (mitral valve atresia, aortic valve atresia, hypoplastic left ventricle, hypoplasia, and coarctation of the aorta), Shone syndrome or other heart defects
Hypospadias	170	3	84	83	SCs had other genitourinary anomalies including chordee, hooded foreskin, epispadias, micropenis and natural circumcision, or multiple birth defects
Reduction deformities, upper limb	45	0	3	42	SCs had shortened upper extremities, other forms of upper limb deformities (micromelia, arthrogryposis, syndactyly, brachydactyly, bowing), achondroplasia, or multiple congenital anomalies but did not meet the inclusion criteria of "missing boney components"
Reduction deformities, lower limb	65	0	4	61	SCs had shortened lower extremities, other forms of lower limb deformities (clubfoot, metatarsus adduction, hypoplastic toes, bowing), achondroplasia multiple congenital anomalies but did not meet the inclusion criteria of "missing boney components."
Spina bifida without anencephaly	37	0	4	33	SCs had other spinal defects including teratomas, lipomas, sacral sinus/dimples, and tethered cord, multiple defects one of which would typically co-occur with a spina bifida diagnosis (Arnold-Chiari malformation, hydrocephalus, Dandy-Walker syndrome), or spina bifida occulta
Tetralogy of Fallot	40	1	4	35	SCs had ≥1 component of tetralogy of Fallot (pulmonary stenosis, overriding aorta, ventricular septal defect, right ventricular hypertrophy) or other heart defects
Transposition of the great arteries	19	1	2	16	SCs had defects resembling transposition of the great arteries (eg, double inlet left ventricle, double chamber right ventricle) or other heart defects

(*continues*)

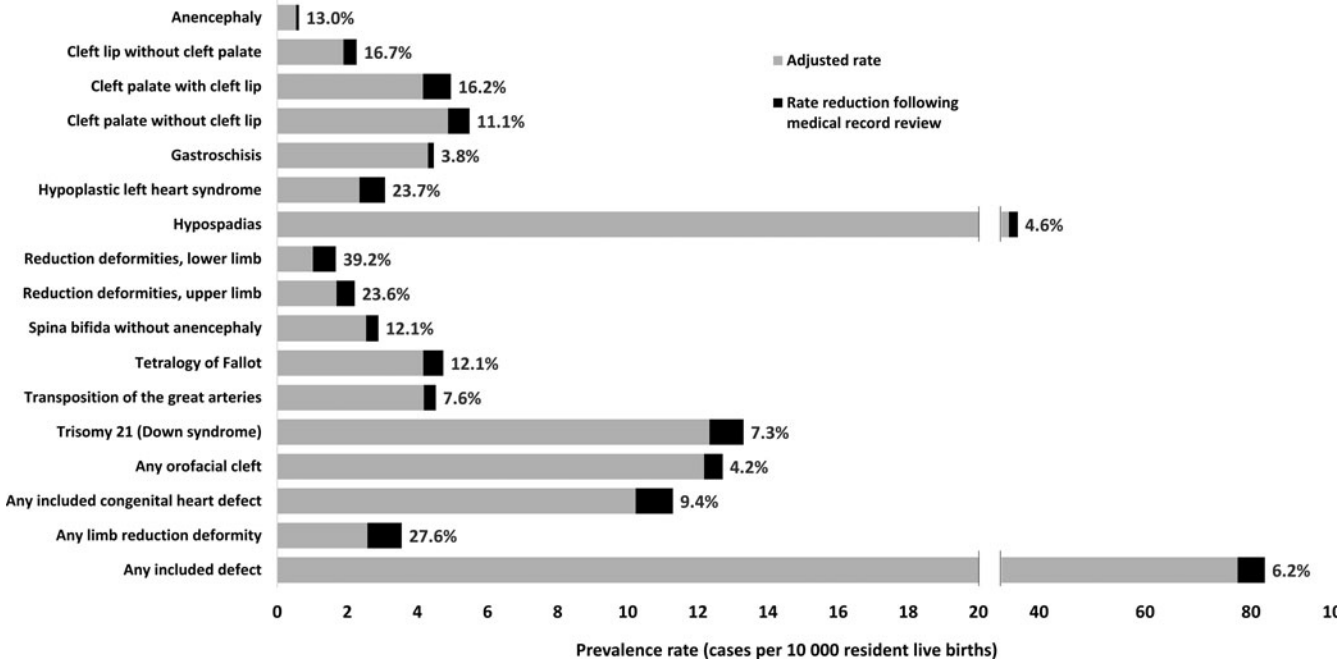
TABLE 4 ● Summary of Reasons for False-Positive Diagnoses[a] Reported by the FBDR, 2007-2011 (*Continued*)

Birth Defect Captured and Reported by the FBDR	Total Noncases	Reason			Summary of Abstractor Notes Describing Decision to Classify SCs as Noncases on the Basis of Misdiagnoses
		Data Linkage Errors	No Evidence of Any Defect	Misdiagnoses	
Trisomy 21 (Down syndrome)	93	1	23	69	SCs with dysmorphic features at birth were ruled out by karyotyping (45 had normal chromosomes, 17 had other translocations, trisomies) or had other anomalies associated with trisomy 21 (heart defects, omphalocele, jejunal atresia)

Abbreviations: FBDR, Florida Birth Defects Registry; SC, suspected case.
[a]During the case confirmation process, abstractors identified a record as a "confirmed case" if it was confirmed through medical record review as having any of the defects included in the case confirmation project. Those without any included defect were classified as "noncases." This table includes only "noncases," also referred to as false-positive diagnoses.

importance that are included in this study. However, our findings are similar to other studies investigating surveillance programs relying mostly or entirely on hospital discharge records for case ascertainment—they are valuable data sources but carry with them suboptimal completeness and accuracy.[9,23,30-33] As is often the case with quality indicators, the level of accuracy and the ability of the FBDR to minimize false-positive diagnoses varied considerably depending on the nature of the defect. Overall accuracy, which measures the FBDR's ability to accurately identify infants who have one of the birth defects included in this study, was very high (PPV >95%) for defects that have distinctive physical features present at birth (eg, gastroschisis, orofacial clefts, hypospadias). High overall accuracy corresponds to a low likelihood that babies without birth defects will be erroneously captured by a surveillance system as having a birth defect; this is particularly important for registries that conduct needs assessments and referrals to clinics and services. Defect-specific

FIGURE ● The Percent Reduction in Reported Prevalence Rates Observed Following Statewide Medical Record Review to Confirm Cases With Selected Birth Defects Identified by the FBDR, 2007-2011[a]



Abbreviation: FBDR, Florida Birth Defects Registry. [a]The entire length of each bar (gray and black portions) reflects the unadjusted FBDR prevalence rate for each defect. The numbers reported to the right of each bar represents the percent reduction in the FBDR-reported rate following removal of noncases. The horizontal axis is split to accommodate both low and high prevalence rates.

diagnostic accuracy mirrored overall accuracy for some defects, such as gastroschisis (overall: 96.4%; defect-specific: 96.0%) and spina bifida without anencephaly (overall: 87.4%; defect-specific: 86.7%), but differed substantially from overall accuracy for other defects including cleft palate without cleft lip (overall: 98.7%; defect-specific: 83.0%), cleft lip without cleft palate (overall: 93.0%; defect-specific: 81.7%), and hypoplastic left heart syndrome (overall: 85.2%; defect-specific: 75.6%). For these defects, although the system's *ICD-9-CM* codes did well to correctly identify an infant as having a birth defect, the final, unconfirmed diagnosis was often incorrect. In particular, medical record review of suspected cases found that orofacial clefts, critical congenital heart defects, and limb reduction defects included in the statewide case confirmation project were often confused for one another or for another defect in the same body system. The lack of accurate diagnostic data for specific defects adversely impacts a surveillance program's ability to provide reliable frequency and prevalence data and inhibits trustworthy comparisons across states and countries.[6]

There has been considerable variation in the overall and defect-specific accuracies reported by previous studies. Recently, in a 2014 Canadian study of 221 affected infants, Metcalfe et al[34] reported much lower overall PPVs (between 59.7% and 78.3%) for various administrative data sources and defect-specific PPVs for all sources combined from 3.6% for respiratory anomalies to 60.0% for genital defects. In another Canadian study assessing validity of *ICD-9-CM* codes in administrative databases among 553 cases, the overall PPV was 80.7% and defect-specific PPV (for defects with >20 confirmed cases) ranged from 83.3% for limb defects to 95.5% for urinary system defects.[35] A Danish study comparing 3 registries reported only the overall PPV for a wide range of birth defects among 848 affected infants, which was between 88.2% and 99.6% among the registries.[36] However, this study lacked medical record review and used presence in more than 1 registry as confirmation of the defect. Other studies were either small and focused on a single defect group (eg, neural tube defects, Hirschsprung disease)[37,38] or were completed using registry data from the 1980s or earlier.[22,31]

The majority of state-based birth defects surveillance programs in the United States use, at least primarily, passive case-finding methodologies that depend on administrative data sources. These data sources rely mostly, if not exclusively, on *ICD* codes that require an accurate clinical diagnosis, interpretation of the clinician's diagnostic write-up, translation into an appropriate *ICD-9-CM* or *ICD-10* code, and entry of that code into a database. Each step is highly susceptible to human error. Depending on funding and data availability/access issues, many of these states seek multiple data sources (eg, birth and infant death certificates, hospital discharge records, Medicaid data, insurance claims) to maximize the completeness of their case ascertainment nets and may face a data source composition that changes over time.[27] For such programs, particularly ones without case confirmation protocols in place, this study provides an understanding of the relative accuracy of diagnostic codes in administrative data for major defects of public health importance. Furthermore, it demonstrates the potential impact of medical record review on reported prevalence rates and offers a guide as to the most frequent reasons for false-positive diagnoses.

Our study is not without limitations, specifically that our assessment is restricted to 13 birth defects. Thus, not only is the defect-specific PPV for some defects not available but also the comparability of our overall PPV to other studies is limited because of differences in definitions of "any included defect." During the 2007-2011 study period, Florida had more than 1 million live births, more than 3% of which had a birth defect. To cover all birth defects, we would have had to send abstractors to more than 200 facilities across the state to review medical records for more than 35 000 infants, which was not feasible with available resources and personnel. Our study is still the largest study to date to focus on the accuracy of a surveillance program using passive case finding for identification of birth defects, with more than 8000 cases included.

The implementation of active case finding for all defects statewide, as a mechanism for improving the largely hospital discharge-based FBDR, is currently cost prohibitive in many states. The Texas Birth Defects Registry uses active surveillance methods to ascertain information about infants and fetuses with birth defects. In 2003, the total budget for the Texas Birth Defects Registry was more than $2.5 million,[39] translating into a cost of nearly $7 per live birth and $180 per infant/fetus identified with a birth defect.[40] Most states currently operate on $2 or less per live birth, which requires exclusive reliance on passive case finding or a hybrid approach that incorporates active case ascertainment only for a subset of defects, restricted to specific geographic regions, or for a limited time frame. As the use of electronic medical records increases, so too does the potential for surveillance programs to have access to maternal and fetal/infant clinical information remotely, with a substantial reduction in the costs (eg, travel, lodging) associated with more active case finding and case confirmation.

Maintaining high-quality surveillance data requires ongoing attention to improvement, primarily through quality control (retrospective assessments of deficiencies and inaccuracies) and quality assurance (proactive measures taken to prevent inaccurate and

incomplete data).[6] In addition to this study focusing on accuracy, Florida recently implemented other quality control projects focusing on the FBDR's completeness[7] and timeliness. The findings across all 3 studies have been distributed to members of the FBDR consortium, which includes representatives from the FDOH, data owners (Agency for Health Care Administration and Bureau of Vital Statistics), academic partners from the University of South Florida, and other stakeholders. A protocol is being implemented to remove from the FBDR those cases deemed not to have a birth defect after medical record review and to adjust diagnoses for misclassified cases. Also, since enhanced surveillance captures cases not identified by the FBDR, the protocol will include the addition of those cases currently missed by the FBDR. Finally, consortium members are evaluating various approaches to reporting FBDR frequency and prevalence data so that the FBDR's completeness and accuracy are reflected appropriately. Not only will this improve the quality of birth defects surveillance data in Florida but it also serves as a model for other programs on the use of results of quality control assessments to design new quality assurance procedures that will maximize the utility of their data. If programs that currently rely on passive case finding are able to allocate funds toward projects that incorporate enhanced case finding[7] or case confirmation (the current study), even if limited to selected geographic regions or time frames, measures of the completeness and accuracy of case ascertainment case can be calculated and subsequently used to generate bias-adjusted prevalence estimates of birth defects statewide.

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