

The Relative Contribution of Data Sources to a Birth Defects Registry Utilizing Passive Multi-source Ascertainment Methods: Does a Smaller Birth Defects Case Ascertainment Net Lead to Overall or Disproportionate Loss?

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Abstract: Since 1998, the Florida Department of Health (FDOH) has operated the Florida Birth Defects Registry (FBDR), a statewide, population-based, passive surveillance system. Cases are identified by collecting information from extant data sources including the statewide hospital inpatient and ambulatory discharge data sets. Additional data sources include administrative, clinical, and service-related information from the FDOH's Children's Medical Services program for children with special health care needs. Like many state birth defects programs, the FBDR faces diminishing funding and resources that may restrict the registry to hospital discharge data. We conducted an evaluation to quantify the potential under-ascertainment to the FBDR resulting from loss of specific data sources, and to determine if there would be a disproportionate loss of cases by sociodemographic and perinatal characteristics. Analyses involved a series of retrospective reconstructions of the FBDR for 1998–2007 to assess the number of cases that would have been ascertained and reported based on the hypothetical loss of 1 or more of the FBDR source data sets. The reconstructed number of cases identified for each defect category was then compared to the current FBDR (constructed using all 5 source data sets) to determine the proportion of cases that would have been missed if the data sources in question were eliminated. These scenarios were constructed overall and by selected characteristics to identify potential disparities in the proportion of cases missed. The inpatient hospital discharge data set was the primary data source for identification of birth defects in the FBDR. Elimination of this single data source would cause the FBDR to miss nearly three fourths of infants diagnosed with 1 or more of the birth defects under study. Our evaluation revealed that an FBDR constructed on hospital discharge data alone would disproportionately miss more cases born to subgroups of women, including non-Hispanic blacks, Hispanics, and those born outside the US. Despite funding and resource constraints, the FBDR continues efforts to identify data sources that may contribute to completeness of case ascertainment in an effort to serve the needs of the Florida maternal and child health population.

Key words: birth defects, congenital malformations, surveillance, case ascertainment, secondary data sources, disparities

Introduction

Birth defects affect 3%–5% of live births, and collectively remain the leading cause of infant mortality in the United States.¹ The purposes and objectives of state birth defects surveillance programs are focused on provision of data to meet these public health challenges. The existence of complete, timely, and accurate prevalence data is essential to monitoring the distribution and determinants of birth defects, implementing and evaluating effective prevention and education programs, identifying children and families in need of services, and providing a basis for clinical research.^{2–4} Programs employing active case-finding methods—an intense level of case identification involving examination of clinical records from strategic locations by trained abstractors—provide the most valid

and precise data on children with birth defects.^{5,6} Although operationally different from active case-finding efforts, programs using passive case-finding methods (often relying on existing administrative and clinical data sources) can achieve comparable levels of data quality with a comprehensive approach to case ascertainment and quality assurance/control.⁴ However, many state birth defects programs, as well as the agencies that collect and share the data utilized by surveillance programs, are facing diminishing funding and resources. This has led some programs to modify operations and re-evaluate surveillance methodologies. To ensure the utility of these data for public health surveillance, the potential impact of these changes on observed prevalence rates should be assessed.

In 1998, the Florida Department of Health (FDOH)

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established the Florida Birth Defects Registry (FBDR), a statewide, population-based, passive surveillance system. Since its inception, the FBDR has monitored structural, functional, and biochemical abnormalities identified within the first year of life in live-born children of women who are Florida residents at the time of delivery. Cases are identified by collecting information from extant data sources, including the statewide hospital inpatient and ambulatory discharge data sets. Additional data sources include administrative, clinical, and service-related information from the FDOH's Children's Medical Services (CMS) program for children with special health care needs. Birth defect diagnoses are recorded using the *International Classification of Diseases, Ninth Edition, Clinical Modification (ICD-9-CM)* codes. All data sets are linked to birth certificate records and an unduplicated inventory of infants with birth defects in Florida is developed. Although the FDOH has worked with the University of South Florida (USF) to develop, pilot, and operate an enhanced surveillance system in targeted areas that incorporates defect confirmation and more complete case ascertainment efforts,⁷ all published birth defects prevalence data currently and historically submitted for Florida to the National Birth Defects Prevention Network (NBDPN) are derived from the FBDR.⁸ For several years, state funding for the FDOH, including budget for the FBDR and CMS programs, has declined, and now represents a small fraction of the support provided only a few years previously. The FDOH has attempted to leverage its remaining resources efficiently by bringing data acquisition, linking, and epidemiologic functions of the FBDR in house. Nonetheless, the FBDR faces the elimination of several CMS data sources that collectively cast a wide ascertainment net for infants with birth defects. The impact of this potential loss, which would result in the FBDR becoming a registry based exclusively on hospital discharge data, has yet to be investigated.

The relatively high cost of active surveillance, growing focus on data quality within the research community, and the increasing existence and availability of administrative and clinical computerized databases have triggered many studies investigating the completeness and accuracy of population-based data sources nationally, most notably vital records⁹⁻¹⁵ (ie, birth certificates) and hospital discharge data.^{2,5,16-20} Most studies report that the use of birth certificates as a lone case ascertainment source results in incomplete surveillance and a marked underestimation of prevalence rates.^{9-11,13,15} Although hospital discharge data provide higher yield and accuracy in identifying cases than do birth certificates^{5,10,18} when compared to national prevalence estimates generated from active surveillance programs,²¹ hospital discharge databases are estimated to miss 30%–50% of birth defects and have had a comparatively higher proportion of false-positive diagnoses.^{2,5,10,17} As the FBDR faces potential restriction to hospital discharge data (inpatient and ambulatory), we anticipate a diminished ability to identify cases of birth defects. We conducted an evaluation to quantify the potential under-ascertainment to the FBDR resulting from loss of specific data sources, and to determine if there would be a disproportionate loss of cases by sociodemographic and perinatal characteristics.

Methods

Data Sources

From 1998–2007, the FBDR was constructed through linkage of several existing data sources to identify infants with birth defects and create a comprehensive inventory of infants with birth defects in Florida. These data sources include: 1) Florida Office of Vital Statistics birth records, 2) Agency for Health Care Administration (AHCA) inpatient and outpatient hospital discharge data sets, 3) CMS Regional Perinatal Intensive Care Centers (RPICC) data, 4) CMS Early Steps program data, and 5) CMS Minimum Data Sets (MDS) (CMS service-related data sets). Each of these 5 data sources was available over the entire study period (1998–2007).

Vital Statistics birth records. The FDOH Office of Vital Statistics collects birth certificate information on all children born in Florida or to a Florida resident and creates a birth database each year. In order to be considered for inclusion in the FBDR, an infant must have a birth record as a resident event in the Florida Birth Vital Statistics database. This data source is used to establish the population of interest (all Florida-resident live births) and not for case identification. All sociodemographic and perinatal characteristics are obtained from the birth certificate.

Hospital discharge data. AHCA is the chief health policy and planning agency for the state. AHCA licenses and regulates health care facilities and health maintenance organizations in Florida and manages the Florida Medicaid program. The agency maintains a database of discharges from all Florida hospitals (inpatient, ambulatory surgery, and emergency). This database contains information on age, sex, ethnicity, ICD-9-CM diagnosis and procedure codes (up to 31 codes), source of admission, and the discharge status of all hospitalized patients. ACHA is also responsible for the Florida Medicaid program, and approximately half of all Florida resident births are covered by Medicaid. Subsets of these records, which include information on all hospitalizations occurring in the first year of life for children born in Florida, are received by the FDOH. Only inpatient and ambulatory records are used in the FBDR's construction.

Children's Medical Services data sources. Florida's program for children with special health care needs is titled Children's Medical Services. This program supports the needs of children with special needs and their families through a variety of programs and services. CMS funding supports a statewide network of RPICCs and the Florida Early Steps program for early intervention services to infants and toddlers under the age of 3. Each program maintains clinical databases containing diagnostic information.

Early Steps consists of several programs that assist families of infants and toddlers with a developmental delay or an established condition that are at high risk for developmental disabilities, and facilitates families in gaining the services and skills they need to support their children. Infants and toddlers from birth to 3 years of age are eligible for the Early Steps program if they have been diagnosed with conditions which have a high probability of resulting in disability or developmental delay: 1) genetic

Table 1. The effect of hypothetical reconstruction of the Florida Birth Defects Registry (FBDR) using various case ascertainment data source combinations, by defect category, 1998–2007.

		<i>TOP: Percent of Cases Missed (Relative to Current FBDR)</i>					
		<i>BOTTOM: Estimated Prevalence* (95% CI)</i>					
Birth Defect	Current FBDR	Without HID	Without HAD	Without ES	Without RPICC	Without MDS	With Only HID, HAD
OVERALL (any defect below)	47,949	73.6%	2.7%	0.9%	1.2%	1.9%	5.1%
	224.6	59.3	218.6	222.5	221.8	220.3	213.2
	(222.6, 226.6)	(58.3, 60.3)	(216.6, 220.6)	(220.5, 224.5)	(219.8, 223.8)	(218.4, 222.3)	(211.2, 215.2)
Anencephalus	97	91.8%	0.0%	1.0%	3.1%	1.0%	7.2%
	0.5 (0.4, 0.6)	0.0 (0.0, 0.1)	0.5 (0.4, 0.6)	0.4 (0.4, 0.5)	0.4 (0.4, 0.5)	0.4 (0.4, 0.5)	0.4 (0.3, 0.5)
Aniridia	24	33.3%	12.5%	0.0%	4.2%	8.3%	29.2%
	0.1 (0.1, 0.2)	0.1 (0.0, 0.1)	0.1 (0.1, 0.2)	0.1 (0.1, 0.2)	0.1 (0.1, 0.2)	0.1 (0.1, 0.2)	0.1 (0.0, 0.1)
Anophthalmia/ microphthalmia	198	63.1%	7.6%	3.0%	1.5%	10.1%	15.7%
	0.9 (0.8, 1.1)	0.3 (0.3, 0.4)	0.9 (0.7, 1.0)	0.9 (0.8, 1.0)	0.9 (0.8, 1.1)	0.8 (0.7, 1.0)	0.8 (0.7, 0.9)
Anotia/microtia	140	64.3%	10.0%	7.9%	2.9%	5.7%	18.6%
	0.7 (0.6, 0.8)	0.2 (0.2, 0.3)	0.6 (0.5, 0.7)	0.6 (0.5, 0.7)	0.6 (0.5, 0.8)	0.6 (0.5, 0.7)	0.5 (0.4, 0.6)
Aortic valve stenosis	347	86.2%	1.7%	0.3%	1.2%	2.9%	4.3%
	1.6 (1.5, 1.8)	0.2 (0.2, 0.3)	1.6 (1.4, 1.8)	1.6 (1.5, 1.8)	1.6 (1.4, 1.8)	1.6 (1.4, 1.8)	1.6 (1.4, 1.7)
Biliary atresia	243	64.6%	1.6%	0.4%	2.5%	4.9%	7.8%
	1.1 (1.0, 1.3)	0.4 (0.3, 0.5)	1.1 (1.0, 1.3)	1.1 (1.0, 1.3)	1.1 (1.0, 1.3)	1.1 (1.0, 1.2)	1.0 (0.9, 1.2)
Bladder exstrophy	72	56.9%	2.8%	0.0%	5.6%	2.8%	12.5%
	0.3 (0.3, 0.4)	0.1 (0.1, 0.2)	0.3 (0.3, 0.4)	0.3 (0.3, 0.4)	0.3 (0.3, 0.4)	0.3 (0.3, 0.4)	0.3 (0.2, 0.4)
Choanal atresia	329	76.3%	3.3%	0.6%	1.8%	3.0%	6.1%
	1.5 (1.4, 1.7)	0.4 (0.3, 0.5)	1.5 (1.3, 1.7)	1.5 (1.4, 1.7)	1.5 (1.4, 1.7)	1.5 (1.3, 1.7)	1.4 (1.3, 1.6)
Coarctation of aorta	1,253	72.6%	0.9%	0.7%	2.0%	2.0%	5.7%
	5.9 (5.6, 6.2)	1.6 (1.4, 1.8)	5.8 (5.5, 6.2)	5.8 (5.5, 6.2)	5.8 (5.4, 6.1)	5.8 (5.4, 6.1)	5.5 (5.2, 5.9)
Common truncus	194	57.7%	1.0%	3.1%	11.3%	9.3%	23.7%
	0.9 (0.8, 1.0)	0.4 (0.3, 0.5)	0.9 (0.8, 1.0)	0.9 (0.8, 1.0)	0.8 (0.7, 0.9)	0.8 (0.7, 1.0)	0.7 (0.6, 0.8)
Congenital cataract	312	56.4%	28.2%	0.3%	1.0%	3.5%	5.1%
	1.5 (1.3, 1.6)	0.6 (0.5, 0.8)	1.0 (0.9, 1.2)	1.5 (1.3, 1.6)	1.4 (1.3, 1.6)	1.4 (1.3, 1.6)	1.4 (1.2, 1.6)
Congenital hip dislocation	2,176	92.1%	1.8%	0.2%	0.8%	1.1%	2.3%
	10.2 (9.8, 10.6)	0.8 (0.7, 0.9)	10.0 (9.6, 10.4)	10.2 (9.7, 10.6)	10.1 (9.7, 10.5)	10.1 (9.7, 10.5)	10.0 (9.5, 10.4)
Diaphragmatic hernia	644	58.2%	0.3%	1.1%	6.7%	3.0%	14.0%
	3.0 (2.8, 3.3)	1.3 (1.1, 1.4)	3.0 (2.8, 3.2)	3.0 (2.8, 3.2)	2.8 (2.6, 3.0)	2.9 (2.7, 3.2)	2.6 (2.4, 2.8)
Down syndrome (Trisomy 21)	2,800	29.4%	0.5%	7.0%	1.1%	4.1%	16.3%
	13.1 (12.6, 13.6)	9.3 (8.9, 9.7)	13.0 (12.6, 13.5)	12.2 (11.7, 12.7)	13.0 (12.5, 13.5)	12.6 (12.1, 13.1)	11.0 (10.5, 11.4)
Ebstein's anomaly	122	67.2%	0.0%	0.8%	5.7%	2.5%	13.1%
	0.6 (0.5, 0.7)	0.2 (0.1, 0.3)	0.6 (0.5, 0.7)	0.6 (0.5, 0.7)	0.5 (0.4, 0.6)	0.6 (0.5, 0.7)	0.5 (0.4, 0.6)
Encephalocele	182	58.8%	1.1%	2.2%	3.8%	5.5%	14.3%
	0.9 (0.7, 1.0)	0.4 (0.3, 0.4)	0.8 (0.7, 1.0)	0.8 (0.7, 1.0)	0.8 (0.7, 1.0)	0.8 (0.7, 0.9)	0.7 (0.6, 0.9)
Endocardial cushion defect	864	68.6%	0.5%	0.3%	2.7%	4.4%	7.9%
	4.0 (3.8, 4.3)	1.3 (1.1, 1.4)	4.0 (3.8, 4.3)	4.0 (3.8, 4.3)	3.9 (3.7, 4.2)	3.9 (3.6, 4.1)	3.7 (3.5, 4.0)
Esophageal atresia/ tracheoesophageal fistula	495	50.9%	0.8%	1.4%	3.4%	2.4%	9.3%
	2.3 (2.1, 2.5)	1.1 (1.0, 1.3)	2.3 (2.1, 2.5)	2.3 (2.1, 2.5)	2.2 (2.0, 2.4)	2.3 (2.1, 2.5)	2.1 (1.9, 2.3)
Gastroschisis/omphalocele (abdominal wall)	1,439	47.5%	0.8%	1.3%	5.4%	2.5%	12.1%
	6.7 (6.4, 7.1)	3.5 (3.3, 3.8)	6.7 (6.3, 7.0)	6.7 (6.3, 7.0)	6.4 (6.0, 6.7)	6.6 (6.2, 6.9)	5.9 (5.6, 6.3)
Hirschsprung's disease (congenital megacolon)	523	65.0%	1.1%	0.6%	1.9%	3.4%	6.9%
	2.4 (2.2, 2.7)	0.9 (0.7, 1.0)	2.4 (2.2, 2.6)	2.4 (2.2, 2.7)	2.4 (2.2, 2.6)	2.4 (2.2, 2.6)	2.3 (2.1, 2.5)

HID=Hospital Inpatient Database; HAD=Hospital Ambulatory (Outpatient) Database; ES=Early Steps Database; RPICC=Regional Perinatal Intensive Care Centers Database; MDS=Minimum Dataset (Children's Medical Services). *Per 10,000 live births.

Table 1, cont. The effect of hypothetical reconstruction of the Florida Birth Defects Registry (FBDR) using various case ascertainment data source combinations, by defect category, 1998–2007.

Birth Defect	Current FBDR	TOP: Percent of Cases Missed (Relative to Current FBDR) BOTTOM: Estimated Prevalence* (95% CI)					
		Without HID	Without HAD	Without ES	Without RPICC	Without MDS	With Only HID, HAD
Hydrocephalus w/o spina bifida	1,590	56.9%	0.4%	4.7%	3.1%	7.0%	18.6%
	7.4 (7.1, 7.8)	3.2 (3.0, 3.5)	7.4 (7.1, 7.8)	7.1 (6.8, 7.5)	7.2 (6.9, 7.6)	6.9 (6.6, 7.3)	6.1 (5.7, 6.4)
Hypoplastic left heart syndrome	587	62.7%	0.9%	1.5%	2.9%	2.9%	8.5%
	2.7 (2.5, 3.0)	1.0 (0.9, 1.2)	2.7 (2.5, 3.0)	2.7 (2.5, 2.9)	2.7 (2.5, 2.9)	2.7 (2.5, 2.9)	2.5 (2.3, 2.7)
Hypospadias and epispadias	7,366	69.5%	11.5%	0.1%	0.7%	0.2%	1.1%
	34.5 (33.7, 35.3)	10.5 (10.1, 11.0)	30.5 (29.8, 31.3)	34.5 (33.7, 35.3)	34.3 (33.5, 35.1)	34.4 (33.6, 35.2)	34.1 (33.4, 34.9)
Microcephalus	1,404	64.9%	0.8%	6.5%	1.2%	10.8%	22.1%
	6.6 (6.2, 6.9)	2.3 (2.1, 2.5)	6.5 (6.2, 6.9)	6.1 (5.8, 6.5)	6.5 (6.2, 6.8)	5.9 (5.6, 6.2)	5.1 (4.8, 5.4)
Obstructive genitourinary defect	5,971	89.4%	1.6%	0.1%	1.5%	0.9%	2.6%
	28.0 (27.3, 28.7)	3.0 (2.7, 3.2)	27.5 (26.8, 28.2)	28.0 (27.3, 28.7)	27.5 (26.8, 28.3)	27.7 (27.0, 28.4)	27.2 (26.5, 27.9)
Orofacial clefts	2,909	44.5%	2.4%	1.2%	0.5%	3.9%	7.4%
	13.6 (13.1, 14.1)	7.6 (7.2, 7.9)	13.3 (12.8, 13.8)	13.5 (13.0, 14.0)	13.6 (13.1, 14.1)	13.1 (12.6, 13.6)	12.6 (12.1, 13.1)
Pulmonary valve atresia and stenosis	2,364	82.2%	2.4%	0.5%	1.6%	4.1%	6.8%
	11.1 (10.6, 11.5)	2.0 (1.8, 2.2)	10.8 (10.4, 11.3)	11.0 (10.6, 11.5)	10.9 (10.5, 11.3)	10.6 (10.2, 11.1)	10.3 (9.9, 10.8)
Pyloric stenosis	5,397	98.9%	0.1%	0.1%	0.1%	0.2%	0.3%
	25.3 (24.6, 26.0)	0.3 (0.2, 0.3)	25.2 (24.6, 25.9)	25.3 (24.6, 25.9)	25.3 (24.6, 25.9)	25.2 (24.6, 25.9)	25.2 (24.5, 25.9)
Rectal and large intestinal atresia/stenosis	864	64.9%	1.5%	0.2%	3.1%	1.4%	6.0%
	4.0 (3.8, 4.3)	1.4 (1.3, 1.6)	4.0 (3.7, 4.3)	4.0 (3.8, 4.3)	3.9 (3.7, 4.2)	4.0 (3.7, 4.3)	3.8 (3.5, 4.1)
Reduction deformity: lower limbs	288	85.4%	0.3%	1.0%	0.7%	6.6%	9.7%
	1.3 (1.2, 1.5)	0.2 (0.1, 0.3)	1.3 (1.2, 1.5)	1.3 (1.2, 1.5)	1.3 (1.2, 1.5)	1.3 (1.1, 1.4)	1.2 (1.1, 1.4)
Reduction deformity: upper limbs	402	80.1%	1.2%	0.5%	1.7%	3.7%	7.0%
	1.9 (1.7, 2.1)	0.4 (0.3, 0.5)	1.9 (1.7, 2.1)	1.9 (1.7, 2.1)	1.9 (1.7, 2.0)	1.8 (1.6, 2.0)	1.8 (1.6, 1.9)
Renal agenesis/hypoplasia	757	79.8%	3.0%	0.4%	1.3%	3.8%	6.2%
	3.5 (3.3, 3.8)	0.7 (0.6, 0.8)	3.4 (3.2, 3.7)	3.5 (3.3, 3.8)	3.5 (3.3, 3.8)	3.4 (3.2, 3.7)	3.3 (3.1, 3.6)
Spina bifida w/o anencephalus	699	35.3%	0.4%	3.1%	2.0%	6.4%	16.2%
	3.3 (3.0, 3.5)	2.1 (1.9, 2.3)	3.3 (3.0, 3.5)	3.2 (2.9, 3.4)	3.2 (3.0, 3.5)	3.1 (2.8, 3.3)	2.7 (2.5, 3.0)
Teratology of Fallot	1,137	61.9%	0.7%	0.6%	1.2%	5.7%	8.2%
	5.3 (5.0, 5.6)	2.0 (1.8, 2.2)	5.3 (5.0, 5.6)	5.3 (5.0, 5.6)	5.3 (5.0, 5.6)	5.0 (4.7, 5.3)	4.9 (4.6, 5.2)
Transposition of great arteries	937	66.7%	0.6%	0.5%	2.0%	3.4%	7.2%
	4.4 (4.1, 4.7)	1.5 (1.3, 1.6)	4.4 (4.1, 4.7)	4.4 (4.1, 4.7)	4.3 (4.0, 4.6)	4.2 (4.0, 4.5)	4.1 (3.8, 4.4)
Tricuspid valve atresia & stenosis	279	72.4%	1.4%	0.4%	1.4%	4.3%	6.5%
	1.3 (1.2, 1.5)	0.4 (0.3, 0.5)	1.3 (1.1, 1.5)	1.3 (1.2, 1.5)	1.3 (1.1, 1.5)	1.3 (1.1, 1.4)	1.2 (1.1, 1.4)
Trisomy 13 (Patau Syndrome)	228	59.2%	0.4%	4.4%	4.8%	7.9%	21.5%
	1.1 (0.9, 1.2)	0.4 (0.4, 0.5)	1.1 (0.9, 1.2)	1.0 (0.9, 1.2)	1.0 (0.9, 1.2)	1.0 (0.9, 1.1)	0.8 (0.7, 1.0)
Trisomy 18 (Edwards Syndrome)	340	57.4%	0.0%	2.9%	5.3%	4.7%	18.2%
	1.6 (1.4, 1.8)	0.7 (0.6, 0.8)	1.6 (1.4, 1.8)	1.5 (1.4, 1.7)	1.5 (1.4, 1.7)	1.5 (1.4, 1.7)	1.3 (1.2, 1.5)
Ventricular septal defect	10,847	86.0%	0.6%	0.2%	1.5%	1.8%	4.0%
	50.8 (49.9, 51.8)	7.1 (6.8, 7.5)	50.5 (49.6, 51.5)	50.7 (49.7, 51.7)	50.1 (49.1, 51.0)	49.9 (48.9, 50.8)	48.8 (47.8, 49.7)

HID=Hospital Inpatient Database; HAD=Hospital Ambulatory (Outpatient) Database; ES=Early Steps Database; RPICC=Regional Perinatal Intensive Care Centers Database; MDS=Minimum Dataset (Children's Medical Services). *Per 10,000 live births.

and metabolic disorders, 2) neurological abnormalities and insult, 3) severe attachment disorder, or 4) significant sensory impairments. The RPICC program is a regionalized health care delivery system designed to provide optimal obstetrical services to high risk pregnant women and care

for newborns with special health needs, such as critical illness or low birth weight. RPICC services include medical care by a team of doctors, nurses, genetic counselors, and ultrasound technicians specializing in high-risk obstetrical care, as well as health education, nutritional assessments,

and consultations during pregnancy. The third CMS data set used in the construction of the FBDR is the MDS. This data set contains demographic and service information for each active CMS patient during the agency's reporting period.

Data Analysis

To examine potential under-ascertainment to the FBDR and patterns of loss of cases by sociodemographic and perinatal characteristics resulting from the elimination of specific data sources, we retrospectively reconstructed the FBDR for 1998–2007 to assess the number of cases that would have been ascertained and reported based on the hypothetical loss of 1 or more of the FBDR source data sets. We conducted these analyses for each major defect listed in Table 1 as well as for these birth defects overall. The reconstructed number of cases identified for each defect category was then compared to the current FBDR (constructed using all 5 source data sets) to determine both the proportion of cases that would have been missed and the revised prevalence estimate if the data sources in question were eliminated. Six hypothetical scenarios were considered and compared to the current construction of the registry:

1. What if the FBDR were constructed WITHOUT the AHCA inpatient data set?
2. What if the FBDR were constructed WITHOUT the AHCA outpatient data set?
3. What if the FBDR were constructed WITHOUT the CMS Early Steps data set?
4. What if the FBDR were constructed WITHOUT the CMS RPICC data set?
5. What if the FBDR were constructed WITHOUT the CMS Minimum Data Set?
6. What if the FBDR were constructed USING ONLY the AHCA inpatient and outpatient data sets?

These scenarios were constructed overall and by selected characteristics to explore any disparities in the proportion of cases missed. Sociodemographic and perinatal characteristics were obtained from the infant's birth certificate record. Race/ethnicity is self-reported by the newborn's mother and was first grouped by ethnicity (Hispanic or non-Hispanic), with the non-Hispanic (NH) group subdivided into whites and blacks. Because of small numbers, we did not tabulate data for Asian Americans and American Indians. Maternal nativity was dichotomized as US or foreign-born (born outside the 50 US states); maternal education was classified as eighth grade or less, ninth to twelfth grade with no diploma, high school graduate or GED, some college (13–15 years), and college graduate and above (16+ years). Birth weight was grouped as very low (< 1500 grams), low (1500–2499 grams), and normal (2500+ grams); plurality was grouped into singleton, twins, and higher order multiple (3+) categories.

Finally, the potential impact of eliminating the CMS data sets as case ascertainment sources on birth prevalence (number of infants with a birth defect per 10,000 resident live births) was examined by comparing rates using the FBDR's current case ascertainment sources to recalculated

rates that included cases identified only by the inpatient and ambulatory hospital discharge data sources. Analyses were conducted using SAS software version 9.2 (SAS Institute, Inc., Cary, North Carolina).

Results

During 1998–2007, the FBDR identified 47,949 infants with 1 or more of the major birth defects listed in Table 1. As expected, the hospital inpatient data set was the major contributor of case identification by the FBDR. Overall, the inpatient data set uniquely identified approximately 74% of infants with major birth defects. For all but 5 defects studied, creating the FBDR without the inpatient data set would result in a loss of over 50% of its cases. Furthermore, if the FBDR were created using only the inpatient and ambulatory hospital discharge data sources, all but 6 of the 39 defect categories investigated would have more than 5% of its cases missed, with 14—including spina bifida; trisomies 13, 18 and 21; and abdominal wall defects (gastroschisis/omphalocele)—having between 10% and 25% missed, and 1 (aniridia) having over 25% missed.

Although the overall contribution of the non-hospital discharge data sources is relatively small, the impact of their elimination varies greatly by defect. For example, 1% of the infants in this study would be missed if the Early Steps data set were eliminated. However, approximately 8% of anotia/microtia, 7% of Down syndrome, and over 6% of microcephalus cases would be missed. Eliminating the RPICC data set would result in 5 defects with 5% to 10% of cases missed, with 1 (common truncus) missing >10%. Eliminating the CMS MDS would result in a loss of 5%–10% of cases for 9 defects and >10% for 2 other defects (microcephalus and anophthalmia/microphthalmia).

Table 2 summarizes the results of recreating the FBDR using only inpatient and outpatient hospital discharge data (eliminating the 3 CMS data sources) and indicates the effect on case ascertainment across relevant sociodemographic and perinatal characteristics. Analysis by year showed a slight decrease in the percent of cases missed over the period of the study, from approximately 7% in 1998 and 1999, to a steady 4%–5% in subsequent years. Restricting the FBDR to these data sources would result in a disproportionate loss of infants born to NH-black and Hispanic mothers. Over 7% of both NH-black and Hispanic cases would be missed compared to 3.3% of NH-white cases. This disparity was also seen at the defect level. The reconstructed FBDR would miss 5% of NH-white cases of orofacial clefts, but approximately 11% and almost 13% of NH-black and Hispanic cases, respectively. Furthermore, birth defect cases whose mothers were foreign-born were more likely to be missed compared to US-born mothers (8.8% vs 3.8%).

As the level of maternal education increased, the percent of cases missed fell from 16% in infants whose mothers had an eighth grade or less education to 2.3% in infants whose mother had a graduate degree or higher. Limiting to inpatient and outpatient hospital data for its construction, the FBDR would disproportionately miss very low, and to a lesser extent, low birth weight cases more often than normal birth weight cases (17.1%, 8.7%, and 3.5%

Table 2. The effect of hypothetical reconstruction of the Florida Birth Defects Registry (FBDR) with only hospital discharge data sources (inpatient and ambulatory), by selected sociodemographic and perinatal characteristics; 1998–2007.

Characteristic	Number of Cases Captured		Under-ascertainment (%)	Prevalence* (95% CI)	
	All FBDR Data Sources	Only HID, HAD	Only HID, HAD	All FBDR Data Sources	Only HID, HAD
Overall	47,949	45,517	5.1	22.5 (22.3, 22.7)	21.3 (21.1, 21.5)
Year of Birth					
1998	4,278	3,952	7.6	21.9 (21.3, 22.6)	20.2 (19.6, 20.9)
1999	4,360	4,057	6.9	22.2 (21.5, 22.8)	20.6 (20.0, 21.3)
2000	4,500	4,269	5.1	22.1 (21.5, 22.7)	21.0 (20.3, 21.6)
2001	4,397	4,216	4.1	21.5 (20.9, 22.1)	20.6 (20.0, 21.2)
2002	4,621	4,424	4.3	22.6 (22.0, 23.3)	21.6 (21.0, 22.3)
2003	4,985	4,763	4.5	23.6 (23.0, 24.3)	22.6 (21.9, 23.2)
2004	4,923	4,688	4.8	22.6 (22.0, 23.2)	21.5 (20.9, 22.1)
2005	5,130	4,866	5.1	22.7 (22.1, 23.4)	21.6 (21.0, 22.2)
2006	5,363	5,124	4.5	22.7 (22.1, 23.3)	21.6 (21.1, 22.2)
2007	5,392	5,158	4.3	22.6 (22.0, 23.2)	21.6 (21.0, 22.2)
Maternal Race/Ethnicity					
Non-Hispanic white	25,862	25,013	3.3	24.7 (24.4, 25.0)	23.9 (23.6, 24.2)
Non-Hispanic black	9,244	8,572	7.3	20.0 (19.6, 20.4)	18.5 (18.1, 18.9)
Hispanic	11,447	10,604	7.4	21.0 (20.6, 21.4)	19.4 (19.1, 19.8)
Maternal Nativity					
US-born	35,680	34,323	3.8	23.9 (23.6, 24.1)	23.0 (22.7, 23.2)
Foreign-born	12,188	11,115	8.8	19.1 (18.8, 19.5)	17.5 (17.1, 17.8)
Maternal education					
8th grade or less	2,122	1,780	16.1	18.6 (17.8, 19.4)	15.6 (14.9, 16.4)
9th–12th grade, no diploma	7,758	7,220	6.9	23.7 (23.2, 24.2)	22.1 (21.6, 22.6)
HS graduate or GED	16,151	15,312	5.2	23.0 (22.6, 23.4)	21.8 (21.5, 22.2)
13–15, some college or AA	11,475	11,080	3.4	22.3 (21.9, 22.8)	21.6 (21.2, 22.0)
16+, college graduate or above	10,082	9,803	2.8	21.7 (21.3, 22.2)	21.1 (20.7, 21.6)
Birth weight					
VLBW (125-1499g)	2,849	2,361	17.1	83.6 (80.6, 86.7)	69.3 (66.5, 72.1)
LBW (1500-2499g)	6,746	6,160	8.7	46.4 (45.3, 47.5)	42.4 (41.3, 43.4)
Normal (2500-6000g)	38,333	36,978	3.5	19.6 (19.4, 19.8)	18.9 (18.7, 19.1)
Plurality					
Singleton (1)	46,553	44,448	4.5	22.5 (22.3, 22.7)	21.5 (21.3, 21.7)
Twin (2)	1,290	1,001	22.4	20.8 (19.7, 21.9)	16.1 (15.1, 17.1)
Higher order multiple (3+)	103	65	36.9	31.2 (25.7, 37.9)	19.7 (15.4, 25.1)
Infant sex					
Female	17,938	16,809	6.3	17.2 (17.0, 17.5)	16.1 (15.9, 16.4)
Male	30,009	28,708	4.3	27.5 (27.2, 27.8)	26.3 (26.0, 26.6)

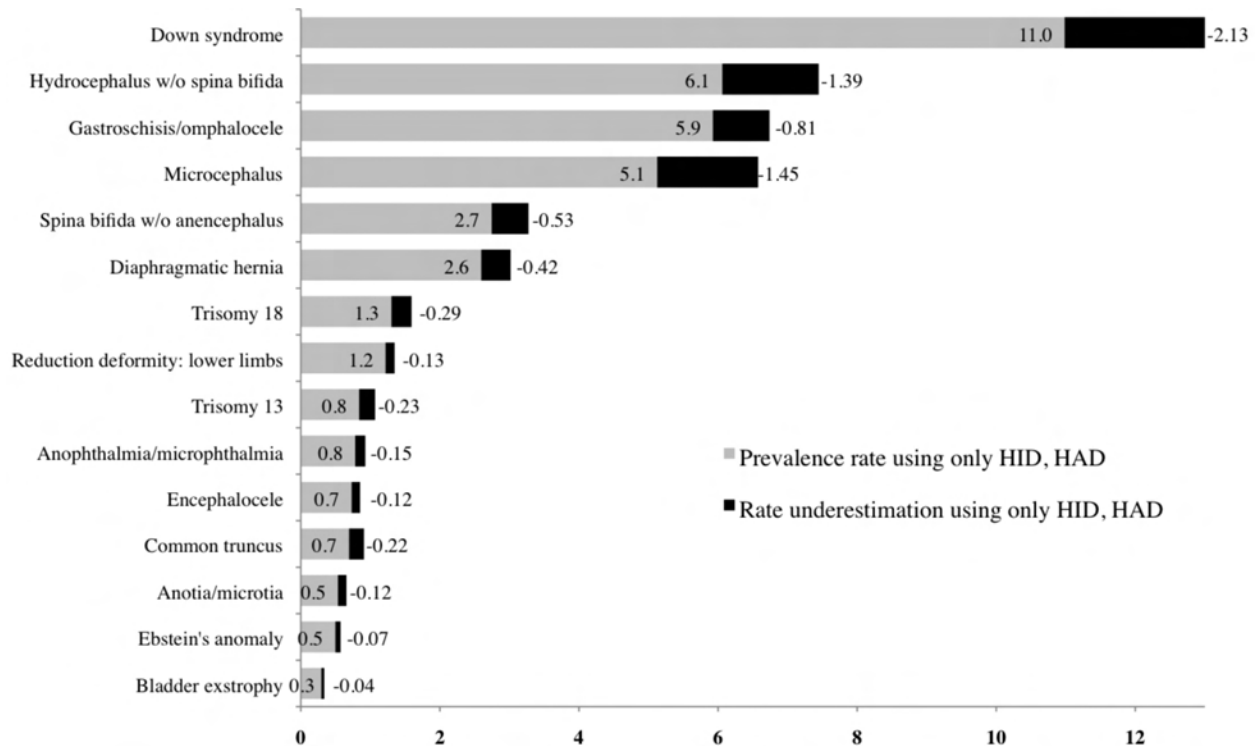
HID=Hospital Inpatient Database; HAD=Hospital Ambulatory (Outpatient) Database, *per 1,000 live births.

respectively). Lastly, limiting the FBDR would disproportionately miss cases among multiple births compared with singleton births.

From 1998–2007, the overall prevalence rate of major birth defects (listed in Table 1) in Florida, using the current FBDR's full set of data sources, was 225 cases per 10,000 live

births. A 5% decrease in rate was observed (to 213 cases per 10,000 live births) when the FBDR was limited to hospital inpatient and outpatient data sets. The underestimation of the overall birth defect rates, while small, varied by specific type of birth defect. Figure 1 provides a more graphic representation of results from Table 1 focusing on major defects

Figure 1. The effect of hypothetical reconstruction of the Florida Birth Defects Registry with only hospital discharge data sources (inpatient and ambulatory) on the Florida Birth Defects Registry's current prevalence rates, 1998–2007.



HID=Hospital Inpatient Database; HAD=Hospital Ambulatory (Outpatient) Database.

for which under-ascertainment would be >10% were the FBDR ascertainment strategy limited to hospital discharge data alone.

Discussion

The present study evaluated the value of each FBDR data source during 1998-2007 by performing a series of retrospective reconstructions of the FBDR. We investigated the number of cases that would have been missed based on the hypothetical loss of specific data sources that currently comprise the FBDR ascertainment net. The inpatient hospital discharge data set was the primary data source for identification of birth defects in the FBDR and uniquely (it was the only source to identify the defect) or jointly (it along with at least 1 other data source identified the defect) identified over 90% of infants included in this study. Elimination of this single data source would cause the FBDR to miss nearly 3 of every 4 of infants diagnosed with 1 or more of the birth defects under study. Moreover, only 5.1% of cases would have been missed by an FBDR relying solely on inpatient and outpatient hospital discharge data. Although the overall contribution of the administrative, clinical, and service-related CMS data sources was relatively small, the impact of their potential elimination varied by defect. For defects such as Down syndrome, spina bifida, and certain congenital heart defects, conditions for which the affected child often receives ongoing medical management and outpatient procedures throughout the first year of life, these CMS sources captured a larger proportion of cases that would otherwise have been missed by hospital

discharge data alone.

Many studies have investigated the accuracy and completeness of state administrative databases in ascertaining birth defects cases.^{2,5,6,9,10,13–20,22–27} Here we offer a counterfactual analysis that investigated the impact of changes to a passive birth defects surveillance system's case ascertainment net on reported prevalence rates and assess the potential for under-ascertainment that varies over time or according to maternal race/ethnicity, nativity, and other sociodemographic and perinatal characteristics. During the first 2 years of the FBDR (1998–1999), restriction to hospital discharge data alone would have resulted in somewhat greater under-ascertainment (7%–8%) than in subsequent years (4%–5%). Linkage algorithms used to create the FBDR remained the same throughout the study period. Thus, we could not determine whether this finding was due to improved ability to capture birth defects in hospital discharge data over time (due to better collection/reporting of variables used in linkage algorithms), or due to a declining ability to ascertain cases in the CMS data sources.

Our evaluation revealed that an FBDR constructed on hospital discharge data alone would disproportionately miss more cases born to subgroups of women, including NH-blacks, Hispanics, and those born outside the US. This is a critical finding in that these subgroups may be at higher risk of adverse pregnancy outcomes, including birth defects, and they are often the focus of birth defects prevention and education efforts. Births to NH-blacks, Hispanics and those born outside of the US may have fewer opportunities to link to birth certificate data as they may have fewer encounters

with various hospital care systems thus producing fewer records available to link. Hospital discharge data sets have a paucity of personal identifiers on which to link to other data sources and are most successfully linked when a maternal social security number (SSN) is available. Florida records for Hispanic, NH-black, and foreign-born mothers more often have missing and, we suspect, erroneous reporting of the SSN, making the linkage rate for these records lower than for NH-white and US-born mothers. Often, the FBDR relies on linkages to the CMS data sources to ascertain birth defect diagnoses not found in the discharge data due to the aforementioned limitations in data linkage. These CMS data sets contain additional identifiers, including names and residential addresses, that can be used to inform subsequent iterations of the linkage algorithms, thereby improving linkage rates resulting in better case ascertainment. Thus, the loss of these CMS data sources increases the likelihood that difficult-to-link records will be missed.

In addition to maternal race/ethnicity and nativity, we also observed that several categories of high-risk deliveries (notably very low and low birth weight infants, and multiple births) were more likely to be missed in an FBDR limited to using only hospital discharge data for case identification. There are several plausible explanations for these findings. Similar to records for minority populations, multiple births hospitalization records are notoriously difficult to link since the hospital discharge data sets do not contain the infant's first name. Thus, hospitalization records for same-sex twins of similar birth weight are virtually indistinguishable from one another, resulting in linkage failure with potential birth defect cases not included in the FBDR. By using the CMS data sources, which include the infant's first name, significant improvement in linkage rates occurs, particularly among same-sex multiples. Loss of these data sources would result in a greater under-ascertainment of multiples versus singletons.

Should the FDOH continue to experience diminished funding as in the past few years, elimination of the CMS data sources and reversion to a birth defects registry relying solely on hospital discharge data may become a reality. The results of this evaluation not only suggest implications for surveillance, but also for epidemiologic studies using surveillance data. We recently reported that foreign-born, NH-black, and Hispanic women were significantly less likely than NH-white and US-born women to have an offspring affected by gastroschisis.²⁸ Had that study been conducted using an FBDR based solely on hospital discharge data for identification of gastroschisis cases, the disproportionate under-ascertainment of cases born to these minority groups would have resulted in a significant exaggeration of the purported protective effect these groups experienced. In studies in which risk might be greater for minority groups, the under-ascertainment of a restricted FBDR, depending on the defect, may be strong enough to mask any true association.

Interpretation of our study results should consider the following limitations. First, the FBDR is a passive surveillance system without case confirmation. The FBDR links administrative data sets not initially created for constructing

a birth defects registry, and relies on ICD-9-CM codes for case identification. The interpretation of a clinician's diagnostic write-up, translation into an ICD-9-CM code, and entry of that code into a database is highly susceptible to human error. The potential for false positive diagnoses in this passive case ascertainment strategy is high and has been characterized and quantified in the literature.^{2,5,10,17} Without an established "gold standard," we were unable to assess the validity and reliability of the various databases used by the FBDR (eg, false positive diagnoses) in this evaluation and focused instead on completeness of ascertainment. However, the FDOH has recently partnered with the USF Birth Defects Surveillance Program to develop, pilot, and operate enhanced surveillance activities in targeted areas using a more comprehensive approach to case identification and diagnosis confirmation than the statewide FBDR.⁷ Future analyses will compare the 2 systems to assess the accuracy and positive predictive value of the FBDR. Second, the FBDR does not include spontaneous and elective terminations, nor does it include fetal deaths; thus, we were only able to evaluate the impact of losing case ascertainment data sources on ascertainment of cases among live births. Birth defects surveillance programs are used both for public health epidemiology purposes and for program planning and evaluation, and the relevance of our findings for these important but differing objectives varies. Our results are of greatest relevance for the use of birth defects surveillance to evaluate and improve service delivery programs for children and families affected by major defects.

Our analysis provides counterfactual evidence for the importance of including multiple sources of case ascertainment in passive birth defects surveillance programs. Inclusion of service delivery program sources enhances the ability of the FBDR to support assessment of program effectiveness and outcome evaluations. We do not wish to imply that the present structure of the FBDR, with the potential loss of the data sources evaluated here, will provide a satisfactory population-based surveillance program for the state of Florida. Our long-term goal remains the expansion of the FBDR into a comprehensive registry utilizing active case-finding methods supported by administrative health data sources as case-finding tools. Unfortunately the current financial climate remains inimical to the realization of this goal.

Despite funding and resource constraints, the FBDR continues efforts to identify data sources that may contribute to completeness of case ascertainment. We recently investigated the potential incorporation of infant death certificates as a new and potentially low-cost case ascertainment source²⁹ and are currently planning a validation of the birth defect-related cause-of-death codes on death certificates in Florida. Our efforts reflect a continued commitment to serving the needs of the Florida maternal and child health population, using diminished resources as effectively as possible to support reliable and valid epidemiologic data for the identification of trends in prevalence and spatial pattern of specific birth defects, and to support improvements in service delivery programs for affected children and their families.

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