Original Article

Validating Obstructive Genitourinary Defects in an Environment of Potential Overdiagnosing

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Abstract: Introduction: The mission of the Department of Defense Birth and Infant Health Registry (Registry) is to conduct ongoing surveillance of birth defects among military families, following the National Birth Defects Prevention Network (NBDPN) case definitions. From 2009 to 2011, a 30% increase in diagnoses of obstructive genitourinary defects (OGDs) was observed in the Registry. To explore the source of this finding, we identified the location with the highest increase and conducted a validation study for OGD cases. Methods: The study population consisted of a random sample of 30 infants defined as OGD cases in the Registry (2010–2011), born at 1 specific military hospital. OGD cases were defined by the presence of the *International Classification of Diseases, Ninth Revision, Clinical Modification* (ICD-9-CM) diagnostic codes 753.2 and/or 753.6 in administrative claims data. A physician reviewed electronic medical records for each infant to confirm true OGD cases and a positive predictive value and 95% CI was calculated. Results: Physician review confirmed only 10% (95% CI, 2%–27%) of the random sample of OGD cases (n = 30) from the identified hospital as true cases. Approximately 97% of infants in this sample were assigned the less severe ICD-9-CM code, 753.2. Conclusions: These results support the 2014 modifications to the NBDPN OGD case definition, which excluded from reporting ICD-9-CM diagnostic code 753.2, which is the less severe OGD diagnosis that may spontaneously resolve without the need for intervention and is more likely to increase the number of false positive cases.

Key words: *administrative claims data, birth defects surveillance, ICD-9-CM diagnostic codes, medical records, obstructive genitourinary defects, validation study*

Introduction

The National Birth Defects Prevention Network (NBDPN) has produced guidelines for population-based birth defects surveillance programs.[1] The Department of Defense Birth and Infant Health Registry (Registry) is a population-based registry[2,3] specific to military families, and reports birth defect rates to the NBDPN annually.[4] Using diagnostic codes from the *International Classification of Diseases, Ninth Revision, Clinical Modification* (ICD-9-CM) as defined in the NDBPN guidelines, the Registry identifies major birth defects captured in administrative medical claims data through the first year of life. Birth defect diagnoses identified by ICD-9-CM codes alone have been shown to have adequate sensitivity and specificity, but lower-than-expected positive predictive values (PPVs).[5] These results vary by birth defect category, depending on the level of diagnostic testing required, and data source comparison (eg, birth certificates and medical records review).[5,6]

Between 2009 and 2011, an approximate 30% increase was observed in obstructive genitourinary defects (OGDs) (1 category of major birth defects reported to the NBDPN) among infants in the Registry.[4] Specifically, the rate of OGD increased from 4.3 in 1,000 live births in 2009 to 5.3 in 1,000 live births in 2010, and continued to reach 5.7 in 1,000 live births in 2011 and 2012. *OGDs* are defined as obstruction via physical blockage or narrowing of the urinary tract,[1] and these rates were observed during a time when OGD cases were defined by the reporting of 1 or both of the following ICD-9-CM diagnostic codes: 753.2 and 753.6.[7] These diagnostic codes include obstructive defects of the renal pelvis and/or ureter (753.2), which vary greatly in severity and location along the urinary tract, and posterior urethral valve (753.6), a condition that almost exclusively affects male infants.[8] The NBDPN reported OGD prevalence rates ranging from 0.06 to 8.9 per 1,000 live births for data collected between 2006 and 2010, which vary because of different methodologies and capabilities of individual states.[4,9] One study that systematically averaged rates reported to the NBDPN found nearly 2 infants per 1,000 live births are affected.[10] The following study was conducted to

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Informed consent was waived in accordance with criteria set forth by 32 CFR 219. This research has been conducted in compliance with all applicable federal regulations governing the protection of human subjects in research (Protocol NHRC.1999.0003).

identify the source of high OGD rates in the Registry and validate a sample of the reported cases through available electronic medical records.

Methods

One military hospital, with readily available medical records, had consistently higher rates of OGD diagnoses, with a 50% increase from 2009 (18.1/1,000 live births) to 2010 (27.1/1,000 live births), and a 2-fold increase from 2009 to 2011 (36.3/1,000 live births). Therefore, a random sample of 30 infants was selected from the 194 Registry-defined OGD cases born at the hospital of interest during 2010–2011, by using the RANUNI function in the SAS, version 9.4, procedure SURVEYSELECT (SAS Institute Inc). The sample size of 30 was selected based on available resources and personnel for data abstraction. This study used data from the Registry and the Armed Forces Health Longitudinal Technology Application (AHLTA), the Department of Defense's electronic medical record system. Briefly, the Registry collects ICD-9-CM diagnostic codes from administrative medical claims data maintained by the Military Health System Data Repository, and personnel and demographic data from the Defense Manpower Data Center.[3]

Specifically, the Military Health System Data Repository houses data from all TRICARE-sponsored medical encounters at military and civilian hospitals, both inpatient and outpatient, nationally as well as internationally. ICD-9-CM diagnostic and procedure codes are collected for infants through the first year of life. Same-sex multiple gestations (eg, twins) are excluded from the Registry due to difficulty in distinguishing between their neonatal medical records. Estimated gestational age is derived from ICD-9-CM codes, and last menstrual period is calculated by subtracting estimated gestational age from delivery date. Further details on the development and methodology of the Registry can be found in Ryan et al[2] and Bukowinski et al.[3] All data were collected with approval from the Institutional Review Board of the Naval Health Research Center (protocol NHRC.1999.0003), and informed consent was waived in accordance with criteria set forth by 32 CFR 219.

Obstructive Genitourinary Defects Definitions

Infants in the Registry were defined as OGD cases if they were assigned an ICD-9-CM code 753.2 and/or 753.6 at 1 inpatient visit or 2 outpatient visits on different days in administrative medical claims data through the first year of life. Many studies implement a 2–diagnostic code rule for outpatient visits,[10, 11] to reduce the capture of false positive cases, as was done here. Using AHLTA, electronic medical record data abstracted to validate OGD diagnoses included: specific anomaly, prenatal diagnosis, postnatal renal ultrasound diagnosis, contrast study results, surgical intervention, resolution date, and information regarding other congenital defects. The reference standard was physician review of abstracted information to confirm OGD diagnosis. If needed to validate the initial OGD diagnosis, further clarification was obtained by reviewing additional medical record notes made by the diagnosing physician. Current Registry definitions were supplemented with additional ICD-9-CM procedure codes to assess if surgical intervention occurred.

Statistical Analyses

Frequencies and medians, with respective percentages and ranges, were calculated to describe select categorical and continuous characteristics of OGD cases in the validation sample, at the hospital of interest overall, and in the Registry. For the validation sample, PPV, the number of physician-confirmed OGD cases out of the random sample of 30 Registry-defined OGD cases, and an exact 95% CI was calculated. Estimated gestational age determined from ICD-9-CM codes has previously been validated and found to be an accurate representation of estimated gestational age reported in medical records.[11] To ensure discrepancies observed between OGD definitions were not caused by systematic errors throughout medical records in this sample, estimated gestational age was compared between the 2 data sources using the Spearman rank correlation coefficient. All data management and statistical analyses (using PROC FREQ and PROC CORR) were performed using SAS, version 9.4.

Results

Select characteristics for OGD cases, born in 2010 and 2011, in the validation sample (n = 30), hospital of interest (n = 194), and the Registry (N = 1,352) are provided in Table 1. The median age of mothers at delivery and median infant estimated gestational age at birth were the same for all populations. Most infants were male, as expected with OGD cases (validation sample, 63.3%; hospital of interest, 68.6%; Registry, 69.6%). The majority of the validation sample included cases with spontaneous resolution (n = 18) and 2 cases only had 1 month of follow-up after birth, though no interventions were noted in medical records. One infant was also diagnosed with other severe birth defects, in addition to an OGD, that resulted in death soon after birth. Of the infants in the validation sample, 42.9% had hydronephrosis, 21.4% had pyelectasis, and 14.3% had vesicoureteral reflux.

One infant in the validation sample, 1 at the hospital of interest, and 40 in the Registry received both OGD diagnoses (ICD-9-CM codes 753.2 and 753.6). Most infants were diagnosed with the less severe 753.2 diagnostic code only (validation sample, 96.7%; hospital of interest, 99.0%; Registry, 96.2%). Of the 30 infants in the validation sample, physician review of medical records confirmed that 3 were true OGD cases and 27 were not true cases. The PPV for the Registry-defined OGD cases was 10% (95% CI, 2%–27%) for the validation sample. Only 2 infants were classified as OGD cases when the Registry definition for OGD cases was modified to additionally require an ICD-9-CM procedural code for genitourinary repair (eg, 56.89, other repair of ureter), and were 2 of the 3 infants who were confirmed as true cases through medical record review. The Spearman rank correlation coefficient was 0.94 ($P < .0001$) for estimated gestational age defined using the Registry compared with medical record review.

Table 1. Select Infant and Maternal Characteristics of OGD Cases Among the Validation Sample, the Hospital of Interest, and the Department of Defense Birth and Infant Health Registry, 2010–2011

Select Characteristics	*Validation Sample (n = 30)*		*Hospital of Interest (n = 194)*		*Registry (N = 1,352)*	
	n	*%*	*n*	*%*	*n*	*%*
AHLTA review						
Type of diagnosis in AHLTA[a]						
Hydronephrosis	12	42.9	–	–	–	–
Pyelectasis	6	21.4	–	–	–	–
Vesicoureteral reflux	4	14.3	–	–	–	–
Other/missing[a]	8	26.7	–	–	–	–
Physician-confirmed OGD diagnosis						
Yes	3	10.0	–	–	–	–
No	27	90.0	–	–	–	–
Estimated gestational age (median, range)	(39.6)	(33.4, 42.0)	–	–	–	–
Registry data						
ICD-9-CM diagnostic code						
753.2	29	96.7	192	99.0	1,300	96.2
753.6	0	0.0	1	0.5	12	0.9
753.2 and 753.6	1	3.3	1	0.5	40	3.0
Estimated gestational age (median, range)	(40)	(34, 42)	(40)	(30, 42)	(40)	(24, 42)
Infant sex						
Male	19	63.3	133	68.6	941	69.6
Female	11	36.7	61	31.4	411	30.4
Multiparity						
Yes	3	10.0	6	3.1	26	1.9
No	27	90.0	188	96.9	1,326	98.1
Maternal age, years (median, range)[b]	(26)	(20, 46)	(26)	(19, 46)	(26)	(16, 46)
Marital status						
Married	26	86.7	181	93.3	1,226	90.7
Unmarried	4	13.3	13	6.8	126	9.4
Race/ethnicity						
Non-Hispanic white	18	60.0	93	47.9	925	68.4
Non-Hispanic black	4	13.3	16	8.3	131	9.7
Hispanic	8	26.7	52	26.8	189	14.0
Other/unknown	0	0.0	33	17.0	107	7.9

AHLTA, Armed Forces Health Longitudinal Technology Application; ICD-9-CM, *International Classification of Diseases, Ninth Revision, Clinical Modification*; OGD, obstructive genitourinary defect. A dash indicates the field is not applicable.

[a]Includes bladder disorders, ureterocele, left duplicated collecting system with hydronephrosis of left upper pole, vesicoureteral reflux, and congenital ureterocele.

[b]Missing 1 in validation sample, 3 in hospital of interest, and 39 in the Registry sample.

Conclusion

This study was conducted to identify a potential cause of the higher rate of OGD identified in the Registry in 2010 and 2011 compared with previous years and to validate diagnoses in a sample of these cases. High OGD prevalence rates in the Registry were noted among live births in both 2010 and 2011,[4] and 1 specific hospital was found to have increasing rates over this time period and consistently higher rates in comparison to other military hospitals. Estimated gestational age in the Registry and AHLTA records served as a control for comparison of the 2 sources of information, and provided reassurance that the selected sample had reliable and complete AHLTA records.

In our validation sample, we observed a very low PPV (10%; 95% CI, 2%–27%), which may be due to overdiagnosis of cases identified during prenatal ultrasound that were unconfirmed after birth,[8] either because of erroneous prenatal diagnosis or spontaneous resolution. Thus, confirmation after birth is important and recommended in the NBDPN guidelines,[1] but difficult to do when only identifying cases through ICD-9-CM diagnostic codes. Specifically, the 753.2 ICD-9-CM diagnostic code comprises a variety of OGD manifestations, a majority of which spontaneously resolve, and the potential for overestimation of OGD cases in birth defect registries has been remedied by removing the reporting of this diagnosis as outlined in the 2014 NBDPN guidelines.[7] Prior to 2014, the OGD category included 2 ICD-9-CM diagnostic codes (753.2 and 753.6); however, in 2014, the NBDPN modified this category, retaining only the more severe diagnostic code, 753.6, for posterior urethral valve cases.[7]

To our knowledge, 1 validation study has been conducted for the diagnosis of OGD. Cooper et al[6] found OGDs identified by ICD-9-CM codes in Tennessee Medicaid data from 1985 to 2000 had a PPV of 68.9% compared with medical record review. Though this PPV is still not optimal, significant overestimation was not an issue, as we found in our validation sample. Additionally, it has been found that including procedure codes in case identification increased the PPV, but led to reduced sensitivity,[6] which is similar to our findings. Using more stringent case definitions (ie, diagnostic and procedure codes, inpatient records only) is useful for creating a more homogenous outcome in risk assessment analyses, but it may lead to underreporting in surveillance efforts.

Given our small sample size, we were unable to assess PPVs stratified by diagnostic codes. Therefore, we could not confirm that the removal of the 753.6 diagnostic code would increase PPV. Also, less follow-up time for some infants may have led to a lower overall PPV. Still, we were able to assess the impact of using ICD-9-CM diagnostic and procedure codes together to identify OGD cases. Additionally, our sample may not be generalizable to the United States overall because it focused on 1 military hospital and we did not have the ability to review medical records at civilian hospitals. Further, the Registry population is focused on military families who are provided readily available access to health care, which is not available for all US citizens. Lastly, our study does not have the ability to determine if our findings are based on a systematic error in the OGD definition, or error specific to diagnosis and reporting at the hospital of interest.

While our study does pose certain limitations, there are strengths to it as well. Our findings support the NBDPN guidelines modification made in 2014 to exclude reporting ICD-9-CM diagnostic code 753.2 for OGD. Access to both administrative medical claims data and electronic medical records gave us the ability to detect and identify overdiagnosing of OGD. Future surveillance efforts in the Registry will closely monitor the prevalence of the 753.6 ICD-9-CM diagnostic code. Additionally, the reporting of this case definition will continue to be limited to the comparable ICD-10-CM code (Q64.2) for live births occurring on or after October 01, 2015. Though we believe validity of this measure will increase, future validation efforts for the updated NBDPN guidelines will be conducted to confirm.

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14. ABSTRACT

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