Problems in Using Birth Certificate Files in the Capture-Recapture Model to Estimate the Completeness of Case Ascertainment in a Population-Based Birth Defects Registry in New York State

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BACKGROUND: The limitations and underlying assumptions of the capture-recapture methods have hindered their application in epidemiological settings, especially in evaluating the completeness of birth defects registries. This study explored the possibility of using birth certificates as the secondary data source in a simple two-source capture-recapture model to estimate the completeness of case ascertainment of the Congenital Malformations Registry (CMR) for selected major birth defects. METHODS: The CMR and the birth certificates were used as the primary and secondary sources, respectively. Children who were born in 1996–2001 and had selected major birth defects were identified from the two sources. The accuracy of the diagnoses was examined by comparing the individual birth defect categories of the children from the two sources. RESULTS: Discrepancies in birth defect categories in the two data sources and false positives in the birth certificates were the major problems encountered in estimating the completeness of the CMR using the simple two-source capture-recapture method. The estimated completeness for selected major birth defects was only about 71%. Stratified analyses resulted in relatively high estimated completeness for oral clefts (90%) and Down syndrome (88%). CONCLUSIONS: Although the birth certificate data was not a good source for estimating the completeness of case ascertainment of the CMR using capture-recapture methods, the analyses provided reasonable estimates for some conditions that were relatively easy to identify and diagnose at birth, such as oral clefts and Down syndrome. Birth Defects Research (Part A) 76:772–777, 2006. © 2006 Wiley-Liss, Inc.

Key words: birth defects; congenital malformations registry; capture-recapture; completeness; hospital discharge data; birth certificates

INTRODUCTION

Evaluating the completeness of registration of birth defect cases has been an especially important concern and a priority activity for birth defects registries. The completeness of a registry, that is, the ability to identify and register all new cases diagnosed within a population, is essential to produce accurate statistics and conduct valid studies on birth defects in a population. In the past decades, a number of studies were conducted to assess the completeness of birth defects registries’ data (Boyd et al., 2005; Czeizel, 1997; Honein and Paulozzi, 1999; Larsen et al., 2003; Wang et al., 2001; Wen et al., 2000; Berghold et al., 2001; Cronk et al., 2003; Knox et al., 1984). Interestingly, only a few of these studies used the capture-recapture methods (Honein and Paulozzi, 1999; Berghold et al., 2001).

Capture-recapture methods, originally developed to estimate the size of a closed animal population (Cormack, 1968), have been used increasingly in epidemiological studies to assess the completeness of cancer registries (McClish and Penberthy, 2004; Silcocks and Robinson, 2004;
The birth certificate files were used as the major source of information for identifying major birth defects that are relatively easy to identify and correctly diagnose at birth. The purpose of this exclusion was to remove the source dependency so that cases identified by one source should not have a higher (or lower) chance of being included in another source, leading to source dependence and violating the first assumption. Severe cases are more likely than mild cases to be captured within any source, violating the second assumption. Moreover, it is often difficult to find reliable data sources for comparison in the capture-recapture models, mostly due to differences in case definition or coding, such as including different birth defects in ICD-9 code groups.

The Congenital Malformations Registry (CMR) of the New York State Department of Health (NYSDOH) was established and began operation in late 1982. It is one of the largest statewide, population-based birth defects registries in the nation, and relies on reports from physicians and hospitals regarding new cases of structural birth defects. In the past decade, efforts have been made by the CMR staff to improve the completeness of the registry (Croce et al., 1996; Druschel et al., 2001; Forand et al., 2002), including a monitoring system used since 1995 to audit all reporting hospitals using hospital discharge data from the Statewide Planning and Research Cooperative System (SPARCS). A previous study has shown that using hospital discharge data to improve the CMR’s case ascertainment was a valuable and effective method of enhancing birth defect surveillance in New York State; new reports resulting from hospital discharge audits comprised about 21% of all CMR reports (Wang et al., 2005).

Because it is impossible to ascertain all cases for a population-based birth defects registry like the CMR, it is important and necessary to explore, develop, and validate methods for estimating the completeness of case ascertainment of the registry and, therefore, to provide an accurate and unbiased estimate of the number of birth defects in the population. The objective of this study was to explore the possibility of using birth certificates as the secondary data source in the two-source capture-recapture model to estimate the completeness of case ascertainment for selected major birth defects that are relatively easy to identify and diagnose at birth. The birth certificate files were used as the comparison data source in this study because no other data sources that collect birth defect information independently were available.

## MATERIALS AND METHODS

### Data Sources

**CMR database.** Hospitals and physicians are required to report to the CMR all children 2 years of age or younger who were born or reside in New York State and were diagnosed with major birth defects. Annually, the CMR receives birth defect reports for more than 10,000 children of New York State residents, which comprise about 4% of live births. CMR case ascertainment consists of: (1) mandatory reporting from hospitals and physicians; and (2) supplementary hospital audits by the CMR staff using SPARCS hospital discharge files (Wang et al., 2005).

**Birth certificate files.** The birth certificate files are maintained in the Vital Records Bureau of the NYSDOH, which annually records more than 255,000 live births in the State of New York. If a baby is diagnosed with birth defects at the time of birth, the birth certificate should indicate these malformations. One or more birth defects from a list of 27 conditions could be recorded on a newborn infant’s birth certificate (New York State Department of Health, 2001).

### Birth Defects Selected for the Study

Not all major birth defect categories were recorded in the birth certificate files. Moreover, some of the birth defect codes indicated on the birth certificates were not specific enough for classification. Thus, a list of selected major birth defects, which were in both sources and were relatively easy to identify and correctly diagnose at birth, was constructed for identifying cases in this study. This list, which accounted for about 13% of all cases in the CMR, included major congenital malformations in the central nervous, digestive, and musculoskeletal systems, oral clefts, and chromosomal anomalies. The selected major birth defects were then grouped into 11 categories. Children (not the defects) with one or more of these defects were counted because not all major malformations of a newborn were recorded in the birth certificate files.

Birth defects such as congenital anomalies of the cardiovascular system, which comprised about 30% of all cases in the CMR, were excluded from the study because some categories of these defects in the birth certificate files were not specific and some were less likely to be identified and diagnosed accurately at birth. The purpose of this exclusion was to remove the source dependency so that cases identified by one source should not have a higher (or lower) chance of being included in another source.

### Data Matching

Matching cases in the CMR to the birth certificate files has been a routine procedure to obtain various birth variables including parents’ demographic information, potential risk factors, and birth certificate number. The identifying variables such as the hospital’s Permanent Facility Identifier (PFI), both infant’s and mother’s name, date of birth, medical record number, and mother’s social security number and residential information are used as matching variables. Extensive matching with multiple matching variables results in more than 95% of all CMR cases and 99.5% of CMR cases of New York State residents matched to the birth records.

_Croce et al., 2001; Ballivet et al., 2000; Kim et al., 1999; Dockerty et al., 1997; Brenner et al., 1994, 1995; Brenner, 1994; Schouten et al., 1994; Robles et al., 1988) and birth defect registries (Honein and Paulozzi, 1999; Berghold et al., 2001), and to estimate the prevalence of some specific birth defects (Campbell et al., 2002; Orton et al., 2001; Rahi and Dezateux, 2000; Egeland et al., 1995; Bobo et al., 1994). This methodology attempts to estimate or adjust for the extent of incomplete ascertainment using information from overlapping lists of cases from distinct, independent sources. However, some limitations and underlying assumptions of the capture-recapture methods have hindered their application in most epidemiological settings (Hook and Regal, 1995, 1999; Papoz et al., 1996; Cormack, 1999; Tilling, 2001; Brenner, 1995), especially in evaluating the completeness of birth defects registries.

The two fundamental assumptions of the simple two-source capture-recapture method are the independence of the sources and the equal probability of individual cases being captured within any source (Hook and Regal, 1995). These assumptions may not hold in most epidemiological settings. For instance, some cases identified by one source have a higher (or lower) chance of being included in another source, leading to source dependence and violating the first assumption. Severe cases are more likely than mild cases to be captured within any source, violating the second assumption. Moreover, it is often difficult to find reliable data sources for comparison in the capture-recapture models, mostly due to differences in case definition or coding, such as including different birth defects in ICD-9 code groups. The Congenital Malformations Registry (CMR) of the New York State Department of Health (NYSDOH) was established and began operation in late 1982. It is one of the largest statewide, population-based birth defects registries in the nation, and relies on reports from physicians and hospitals regarding new cases of structural birth defects. In the past decade, efforts have been made by the CMR staff to improve the completeness of the registry (Croce et al., 1996; Druschel et al., 2001; Forand et al., 2002), including a monitoring system used since 1995 to audit all reporting hospitals using hospital discharge data from the Statewide Planning and Research Cooperative System (SPARCS). A previous study has shown that using hospital discharge data to improve the CMR’s case ascertainment was a valuable and effective method of enhancing birth defect surveillance in New York State; new reports resulting from hospital discharge audits comprised about 21% of all CMR reports (Wang et al., 2005).

Because it is impossible to ascertain all cases for a population-based birth defects registry like the CMR, it is important and necessary to explore, develop, and validate methods for estimating the completeness of case ascertainment of the registry and, therefore, to provide an accurate and unbiased estimate of the number of birth defects in the population. The objective of this study was to explore the possibility of using birth certificates as the secondary data source in the two-source capture-recapture model to estimate the completeness of case ascertainment for selected major birth defects that are relatively easy to identify and diagnose at birth. The birth certificate files were used as the comparison data source in this study because no other data sources that collect birth defect information independently were available.
For this study, a dataset containing information about children who were born in 1996–2001 to New York State residents and had selected major birth defects noted on their birth certificates was abstracted from the birth certificate files. This dataset, used as the secondary source in the capture-recapture analysis, was linked to the primary source, the CMR records of children who had the same selected major birth defects and were born to New York State residents, by birth year and birth certificate number. The matched cases from the linkage were identified as the cases captured by both sources. The accuracy of the diag-

Table 1

<table>
<thead>
<tr>
<th>Cases ascertained by secondary source: birth certificate</th>
<th>Yes</th>
<th>No</th>
<th>Total</th>
</tr>
</thead>
<tbody>
<tr>
<td>Cases ascertained by primary source</td>
<td>Yes</td>
<td>A</td>
<td>B</td>
</tr>
<tr>
<td></td>
<td>No</td>
<td>C</td>
<td>D</td>
</tr>
<tr>
<td>Total</td>
<td>n</td>
<td>$n = A + B + C + D$</td>
<td></td>
</tr>
</tbody>
</table>

$A$: Cases captured by both sources.
$B$: Cases captured only by the primary source.
$C$: Cases captured only by the secondary source.
$D$: Cases missed by both sources, estimate based on the assumption that the probability of ascertainment from both sources is equal, that is, $A \times D = B \times C$. Thus, $D = (B \times C)/A = (n_1 - A)/(n_2 - A)$. $n$: The total number of cases captured by the secondary source. $n_2$: The total number of cases captured by the secondary source. $n$: The estimated total number of cases in a population, $n = A + B + C + D = (n_1 \times n_2)/A$.

For this study, a dataset containing information about children who were born in 1996–2001 to New York State residents and had selected major birth defects noted on their birth certificates was abstracted from the birth certificate files. This dataset, used as the secondary source in the capture-recapture analysis, was linked to the primary source, the CMR records of children who had the same selected major birth defects and were born to New York State residents, by birth year and birth certificate number. The matched cases from the linkage were identified as the cases captured by both sources. The accuracy of the diag-

Table 2

<table>
<thead>
<tr>
<th>Cases ascertained by secondary source: birth certificate</th>
<th>Yes</th>
<th>No</th>
</tr>
</thead>
<tbody>
<tr>
<td>Cases ascertained by primary source: CMR</td>
<td>Yes</td>
<td>2,824</td>
</tr>
<tr>
<td>No</td>
<td>1,178</td>
<td>2,025</td>
</tr>
<tr>
<td>Total</td>
<td>4,002</td>
<td>10,882</td>
</tr>
</tbody>
</table>

Results are from birth years 1996–2001.
*Estimated completeness of the data source.

Table 3

<table>
<thead>
<tr>
<th>Agreement of the birth defects</th>
<th>Completely agree*</th>
<th>Partially agree†</th>
<th>Completely disagree‡</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>n</td>
<td>%</td>
<td>n</td>
</tr>
<tr>
<td>Birth year</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>1996</td>
<td>487</td>
<td>84.4</td>
<td>52</td>
</tr>
<tr>
<td>1997</td>
<td>489</td>
<td>84.3</td>
<td>53</td>
</tr>
<tr>
<td>1998</td>
<td>488</td>
<td>85.5</td>
<td>41</td>
</tr>
<tr>
<td>1999</td>
<td>475</td>
<td>84.2</td>
<td>44</td>
</tr>
<tr>
<td>2000</td>
<td>434</td>
<td>82.7</td>
<td>43</td>
</tr>
<tr>
<td>2001</td>
<td>451</td>
<td>90.5</td>
<td>23</td>
</tr>
<tr>
<td>Hospital location</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Upstate New York§</td>
<td>2,054</td>
<td>86.0</td>
<td>180</td>
</tr>
<tr>
<td>New York City</td>
<td>770</td>
<td>83.1</td>
<td>76</td>
</tr>
<tr>
<td>Childern with birth defects in CMR</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Multiple</td>
<td>204</td>
<td>27.5</td>
<td>136</td>
</tr>
<tr>
<td>Single</td>
<td>2,620</td>
<td>89.7</td>
<td>120†</td>
</tr>
<tr>
<td>Malformation categories in CMR</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Neural tube defects</td>
<td>227</td>
<td>58.1</td>
<td>69</td>
</tr>
<tr>
<td>Encephalus</td>
<td>37</td>
<td>48.6</td>
<td>9</td>
</tr>
<tr>
<td>Hydrocephalus</td>
<td>242</td>
<td>79.3</td>
<td>37</td>
</tr>
<tr>
<td>Oral clefts</td>
<td>968</td>
<td>93.8</td>
<td>49</td>
</tr>
<tr>
<td>Rectal atresia/stenosis</td>
<td>131</td>
<td>74.0</td>
<td>27</td>
</tr>
<tr>
<td>Tracheoesophageal fistula/esophageal atresia</td>
<td>84</td>
<td>84.5</td>
<td>10</td>
</tr>
<tr>
<td>Limb reduction</td>
<td>52</td>
<td>90.4</td>
<td>1</td>
</tr>
<tr>
<td>Diaphragmatic hernia</td>
<td>104</td>
<td>90.4</td>
<td>5</td>
</tr>
<tr>
<td>Omphalocele/gastrochisis</td>
<td>213</td>
<td>98.1</td>
<td>4</td>
</tr>
<tr>
<td>Down syndrome</td>
<td>596</td>
<td>84.6</td>
<td>34</td>
</tr>
<tr>
<td>Other chromosomal anomalies</td>
<td>170</td>
<td>79.4</td>
<td>11</td>
</tr>
<tr>
<td>Total</td>
<td>2,824</td>
<td>85.2</td>
<td>256</td>
</tr>
</tbody>
</table>

Sources were CMR and birth certificates, birth years: 1996–2001.
*All defects agree.
†One or more but not all defects agree.
‡None of the defects agree.
§New York State excluding New York City.
These 120 children had a single birth defect in the CMR but were identified as having multiple birth defects in the birth certificate files.
Two-Source Capture-Recapture Analysis

A simple two-source capture-recapture model was used for estimating the total number of cases (Brenner, 1994). The number of cases captured by both sources was determined by linking the two data sources. The number of cases missed by both sources was estimated based on the assumption that the probability of ascertainment from both sources was equal. The estimated completeness of the case ascertainment for the primary source can be calculated by dividing the number of cases captured by the total number of cases estimated from the model. The 95% CI of the estimated completeness was calculated using the normal distribution-based CI of the estimated total number of cases that occurred during the study period (Pollock et al., 1990).

RESULTS

Crude Analyses Using the Two-Source Capture-Recapture Model

Table 1 illustrates the simple two-source capture-recapture model for estimating the total number of selected major birth defects. The analysis was performed using the CMR and the birth certificate data as the primary and the secondary sources, respectively. The results are shown in Table 2. Among live births for the years of 1996 through 2001, 2,824 cases with selected major birth defects were captured by both sources. There were 1,178 cases that were captured by the birth certificate files but not by the CMR. According to the model, 2,025 cases were missed by both sources (the CMR and the birth certificate data as the primary and the secondary sources, respectively). The results are shown in Table 2. Among live births for the years of 1996 through 2001, 2,824 cases with selected major birth defects were captured by both sources. There were 1,178 cases that were captured by the birth certificate files but not by the CMR. According to the model, 2,025 cases were missed by both sources. The estimated total number of cases was 10,882. The estimated completeness of case ascertainment of the CMR for selected major birth defects was 70.6%.

Discrepancies of the Birth Defects in the Two Sources

To compare the birth defects of the children captured by both sources (the CMR and the birth certificates) and to examine factors that affect the discrepancies in birth defect categories defined in the two sources, analyses were performed by birth year, geographic location of the reporting hospitals, the number of birth defects (single or multiple), and birth defect category. The results are shown in Table 3. Out of 2,824 children captured by both sources, 85.2% had birth defect categories that completely agreed in both sources, 9.1% partially agreed (one or more but not all malformations agreed), and 5.7% completely disagreed. Children with a single birth defect had much higher per-
likely to be diagnosed correctly on both sources. The over-
gastroschisis, and omphalocele combined were more
categories, defects that were visible, such as oral clefts,
gastrostomosis, and omphalocele combined were more
likely to be diagnosed correctly on both sources. The over-
all agreement (completely and partially agreed) of the
birth defect categories in the two sources did not change
significantly over the study years, 1996–2001.

**Stratified Analyses Using the Capture-Recapture Method**

Stratified analyses were performed by the number of
birth defects (single or multiple) and by birth defect catego-
ries for the children with a single birth defect, using the
simple two-source capture-recapture model. The birth
defect categories defined by the CMR were used for the
children who were captured by both sources but had birth
defect categories that disagreed in the two sources. The
results are presented in Table 4. The estimated complete-
ness of case ascertainment for children with multiple de-
fects was significantly higher (88.7%, 95% CI 85.9–91.6%) than
that for children with a single defect (69.5%, 95% CI
68.3–70.7%). Moreover, results from stratified analysis
among children with a single birth defect by birth defect
category showed that the estimates of completeness
strongly depend on the specific birth defect. The highest
estimates were 90.1 and 88.1% for oral clefts and Down
syndrome, respectively, and the lowest estimate was 23.6% for
encephalus. The estimate for omphalocele and gastro-
chosisis was surprisingly low (52%), even though these
defects are relatively easy to identify and correctly diag-
nose at birth.

**DISCUSSION**

Using birth certificate files as the comparison source in a
simple two-source capture-recapture model, the esti-
mated completeness of case ascertainment of the CMR for
selected birth defects was about 71%. This relatively low
estimate was most likely attributable to the false-positive
reports in the birth certificate files (Olsen et al., 1996; Pol-
lock et al., 1990). A previous study conducted by our
CMR staff to determine whether birth certificates could be
used to ascertain unreported cases to the CMR found that
about 45% of the children with one or more birth defects
noted on their birth certificates were normal, that is, there
was no mention of a malformed in their medical
records (Olsen et al., 1996).

In order to evaluate the effect of false positives on the
results of the capture-recapture analysis, we matched the
cases that were found in the birth certificate files but not
in the CMR to SPARCS hospital discharge data to verify
the cases. When we assumed that the unmatched cases
were false positives and removed them from the analysis,
the estimated completeness was approximately 82% (data
not shown). Although the use of SPARCS data to filter
the false positives sacrifices the independence of data sources
because the SPARCS data were used as a supplementary
data source to the CMR, the finding demonstrates that the
quality of the comparison data source is critical for evalu-
ating the completeness of case ascertainment using the
capture-recapture analysis. In order to identify false posi-
tive cases from the birth certificate files and ascertain
unreported cases, we would need to request individual
medical records from reporting hospitals. This would add
an extra burden to the reporting hospitals and the CMR
staff. The CMR staff has been actively seeking more prac-
tical and less expensive measures to identify unreported
cases from other sources.

The current study found that although false positives in
the birth certificate files lead to an overestimate of the total
number of cases and thus, the completeness of the case
ascertainment for the birth defects of interest was underes-
timated, they did not significantly affect the estimates for
some birth defects that were relatively easy to identify and
diagnose at birth, such as oral clefts and Down syndrome.
The estimated completeness for oral clefts and Down syn-
drome was 90 and 88%, respectively. Our estimate for
Down syndrome was consistent with that reported by
Berghold et al. (2001) for the Styrian Malformation registry.
They estimated that Down syndrome cases reported to the
registry between 1985–1992 were 88% complete, using the
two-source capture-recapture method allowing for time-
varying detection probabilities (Berghold et al., 2001).

The discrepancy in the case definition of data sources has
been shown to be one of the major problems encountered in
estimating the completeness of case ascertainment using the
capture-recapture methods (Hook and Regal, 1999). The
results from our study show that among children captured
by both sources, about 6% had birth defects that totally
failed to agree. In this study, hospitals reported birth defect
cases to the CMR using a standard reporting card to pro-
vide ICD-9-CM codes and narratives, as well as the infor-
mation about the child and parents. On the other hand,
there were only 27 one-digit fields (with the value of 1 or 0)
used for recording major anomalies (New York State
Department of Health, 2001) in the birth certificate files and
there were no narratives available, resulting in discrep-
cancies in birth defect categories in the two data sources.
Because we do not confirm all of the diagnosed cases
reported to the CMR due to limited resources, the CMR
staff has initiated an on-site auditing program to focus on
improving the accuracy of diagnoses by visiting hospitals
that have an unreasonably low number of case reports and/
or insufficient diagnostic information for the reported cases.

There was no strong evidence of dependence between the
two sources in the current study. Our study selected
only children with major birth defects that were relatively
easy to identify and diagnose correctly at birth, because the
CMR receives case reports of children up to 2 years of age
whereas the birth certificate files collect birth defects infor-
mation at birth. This exclusion greatly reduced (if not elim-
inated) the chance of positive source dependency, that is,
cases identified by one source (the CMR) having a higher
chance of being included in another source (the birth certifi-
cates). It should also be noted that our estimate of com-
pleteness of case ascertainment for selected birth defects
might not be generalized to all the birth defects in the
CMR, because only a portion of birth defects in the CMR,
which comprised 13% of all CMR cases, was included in the
study.

In conclusion, discrepancies in birth defect categories
defined in the two sources (the CMR and the birth certifi-
cates) and false positives in the birth certificate files were
the major problems encountered in estimating the com-
pleteness of case ascertainment using the simple two-source
capture-recapture method. False positives in the birth certifi-
cate files lead to the overestimation of the total number of
cases and thus, the underestimation of CMR’s completeness of case ascertainment for selected birth defects. Although the birth certificate data were not a good source for estimating the completeness of case ascertainment of the CMR using the capture-recapture methods, our results from the capture-recapture analyses provided reasonable estimates for some birth defects that are relatively easy to identify and correctly diagnose at birth, such as oral clefts and Down syndrome. The exploratory analysis and the findings of the current study should be helpful to the registries and researchers in the birth defects research community; our study has shown the importance of the quality of the data sources and has suggested that the two-source capture-recapture model should be used with caution in estimating the completeness of case ascertainment of birth defects if the quality of the comparison data source, such as the birth certificates, is in question.

REFERENCES


