Congenital Malformations: Agreement Between Diagnostic Codes in an Administrative Database and Mothers’ Reports

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Abstract

Objective: The validity of epidemiologic research on congenital malformations conducted using administrative databases relies heavily on the quality of diagnostic information. The goal of this study was to validate the diagnostic codes for major congenital malformations (MCMs) recorded in a medical service claims database against mother’s report obtained by questionnaire.

Methods: Using the Quebec Pregnancy Registry, we mailed a questionnaire to women who gave birth to a baby with an MCM and to a random sample of mothers whose infants did not have any malformation. We reviewed each infant’s database record for a diagnostic code for MCM, which we then compared with the corresponding mother’s report.

Results: Among the 3142 mother–infant pairs there was 60% agreement regarding the presence or absence of an MCM; for 456 babies, both sources reported the presence of an MCM. For 90% of these infants the two sources of information agreed with respect to the type of malformation.

Conclusion: Overall, mothers reported fewer infants with MCM than were recorded in the database; among those for whom both sources of information indicated the presence of an MCM, a very high proportion of diagnostic codes accorded with the mother’s description of the specific type of malformation.

INTRODUCTION

Health administrative databases are increasingly used to conduct medical research studies. Research on the occurrence of congenital malformations in relation to medication use during pregnancy is often conducted using such databases because they provide the large cohorts needed to study rare outcomes.1–3 However, the validity of such research depends largely on the quality of the diagnostic information held within the database. While several studies1,2,4 using the medical service claims database of the provincial health insurance program of the Province of Quebec (RAMQ) have already made important contributions to knowledge about the effects of maternal drug use during pregnancy on the health of the baby, the coding of the diagnosis of a major congenital malformation has never been validated.

The RAMQ database holds claim records for visits or procedures performed by physicians; each medical service
claim requires the specification of a diagnostic code. While the procedure codes are verified because they are linked to the level of reimbursement, the diagnostic codes are not routinely reviewed. The diagnostic codes held by this database have previously been validated against medical charts for a number of conditions. To our knowledge there has not been a study addressing the accuracy and comprehensiveness of congenital malformation codes in the RAMQ database. As the prevalence of congenital malformations is low, estimated at 3% to 5% in the general population, the validity of such physician-based codes within the RAMQ is essential for the production of robust estimates from research studies.

It has been shown in several studies that maternal reports of babies’ characteristics such as birth weight, for which a gold standard source of information exists, are valid, reliable and accurate. Considering the universal concern of mothers with the health of the newborn, a mother’s report about the presence or absence of a major congenital malformation in her baby can be expected to be similarly reliable and accurate.

Therefore, the objective of our study was to quantify the level of agreement between physician-based diagnostic codes of congenital malformations, as recorded in the provincial health administrative databases of the province of Quebec (RAMQ), and maternal reports from a self-administered questionnaire.

METHODS

The Quebec Pregnancy Registry resulted from the linkage of three administrative databases of the Province of Quebec: RAMQ, MedEcho, and ISQ. This registry has been in existence since 1998. The RAMQ database contains information on physician-based medical services (diagnoses and procedures) received by all Quebec residents as well as medication use for a subgroup of residents. Diagnoses specified by physicians are classified according to the ICD-9. The diagnostic codes are entered on claims for reimbursement forms following a visit or procedure and are therefore completed prospectively. The MedEcho database records acute care hospitalization data for all Quebec residents and is a source of data about congenital malformations; it also records gestational age for deliveries. ISQ provides demographic information about the mother, father, and baby, as well as birth weight and gestational age for live births and stillbirths.

To form the Quebec Pregnancy Registry, a linkage was made between the RAMQ and MedEcho databases using the health insurance number, which is a unique personal identifier for each person living in Quebec. Linkage between the RAMQ and ISQ databases was made using the mothers’ and infants’ names, family names, and dates of birth. Linkage between RAMQ and ISQ databases was possible in 73% of instances. Each mother–infant pair was also linked with MedEcho and ISQ databases to obtain hospital discharge information and sociodemographic characteristics of the mother. The Quebec Pregnancy Registry contains information on all pregnancies that had a first day of gestation (date of last menstrual period) between January 1, 1997, and December 31, 2003.

A questionnaire was developed by a group of experts in perinatal epidemiology and genetics to collect information not routinely available from the Quebec Pregnancy Registry and to validate congenital malformation codes. The questionnaire was first developed in French and translated into English; it was further back-translated into French. The questionnaire was pre-tested on a group of 20 pregnant women attending the obstetrics department at Sainte-Justine Hospital, and face validity was established by pregnant women, obstetricians, and pediatricians. Questionnaires were sent to mothers in either English or French according to the preferred language of correspondence indicated to the RAMQ by subjects. Mothers were asked a number of questions including whether their child had been given a diagnosis of a major congenital malformation, and if so, to specify the type of malformation. The specific questions that were the focus of this study were: “Was this child or were these children (twins/triplets) born with one or more birth defects? If yes, which birth defect(s)?” or “Est-ce que votre enfant (ou vos enfants s’il s’agit de jumeaux, triplets) a une ou plusieurs malformation(s) congénitale(s)? Si oui, laquelle/lesquelles?”

Women in the Quebec Pregnancy Registry were eligible for this validation study if they had given birth to a baby with a major congenital malformation, or if they had a previous diagnosis of diabetes or psychiatric disorder. Major congenital malformations are defined as ICD-9 categories 740.0–759.9: anomalies of the central nervous system, eye, ear, cardiovascular system, orofacial, gastrointestinal, genitourinary, musculoskeletal or chromosomal but excluding, as minor, malformations of the eyelids, lacrimal system and orbit (ICD-9 743.6); accessory auricle, macrotia or microtia, branchial cleft cyst or fistula, preauricular sinus (ICD-9 744.1–744.4); macrocheilia, microcheilia, macrostomia, microstomia (ICD-9 744.8); not otherwise specified anomalies of face and neck (ICD-9 744.9), patent ductus arteriosus (ICD-9 747.0), absence or
hypoplasia of umbilical artery (ICD-9 747.5), ankyloglossia (ICD-9 750.0); absence of, or agenesis of, or anomalous development of cervix, clitoris, vagina, or vulva, cyst of canal of Nuck, congenital vagina, embryonal vulva, congenital imperforate hymen (ICD-9 752.4); undescended testicle (ICD-9 752.5); valgus deformities of the feet (ICD-9 754.6); polydactyly (ICD-9 755.0); syndactyly (ICD-9 755.1); abnormal palmar creases (ICD-9 757.2); congenital ectodermal dysplasia, vascular hamartomas, congenital pigmented anomalies of skin, congenital accessory skin tags, congenital scar, epidermolysis bullosa, congenital keratoderma (ICD-9 757.3); specified anomalies of hair (ICD-9 757.4); specified anomalies of nails (ICD-9 757.5); specified anomalies of breast (ICD-9 757.6); other specified anomalies of the integument (ICD-9 757.8); unspecified anomaly of the integument (ICD-9 757.9); balanced autosomal translocation in normal individual (ICD-9 758.4). All women with babies with a major congenital malformation identified in the RAMQ or MedEcho were included in this study, and a random sample of women with babies with no diagnosed congenital malformations was selected from the remainder of the registry. If a woman had more than one delivery during the study period, she was asked to complete the questionnaire with respect to the first eligible delivery. Women included in this study were those who completed and returned their questionnaire.

The mailing addresses of the 8505 women sampled from the registry were obtained by the RAMQ. A first mailing of the questionnaire took place in October 2006, followed by a second mailing to the non-responders in January 2007. A toll-free telephone number was provided for those who needed help in completing the questionnaire or who wanted more information. All questionnaires used in this study were received by April 2007. The questionnaire data were linked to the Quebec Pregnancy Registry using a scrambled health insurance number. The RAMQ data used for the study were the physician-specified four-digit diagnostic codes for major congenital malformations (ICD-9 codes: 740.0–759.9) any time in the first year of life.

To measure overall agreement between the two information sources regarding the presence or absence of a major congenital malformation, we calculated the proportion of children for whom RAMQ diagnostic codes and mother’s report of congenital malformation were concordant; that is, both RAMQ and mother reported at least one major congenital malformation or both reported absence of any malformation. To measure the extent of agreement about the type of malformation, we calculated the proportion of babies in whom the malformation specified by the diagnostic code in the RAMQ was reflected by the description provided by the mother, among those for whom both mother and RAMQ reported that the baby had a major congenital malformation. We calculated 95% confidence intervals for the proportions.

This study was approved by the Sainte-Justine Hospital Ethics Committee. All women who responded to the questionnaire provided informed consent. The linkage between administrative databases and the self-administered questionnaire was approved by the Commission d’accès à l’information du Quebec.

RESULTS

Questionnaires were completed and returned by 3351 of the 8505 women to whom it had been mailed (39.4%); 9% were completed in English and 91% in French. Thirty-six percent of the women returned the completed questionnaire after the first mailing, and 4% of the remainder after the second mailing. Those who responded to this questionnaire had previously been shown to be similar to those who did not respond with respect to maternal age, use of health services before and during pregnancy (physician and prenatal visits, emergency department visits, hospitalizations), comorbidity status, prescribed medication use, mean birth weight, and sex of baby. Eighteen mothers did not answer the question relating to congenital malformations and were excluded from the analysis. We also excluded data on 76 women who had given birth to twins and two women who had given birth to triplets because only one questionnaire per pregnancy was completed. Sociodemographic information in the RAMQ database was missing for 113 women and these were excluded from analysis. Overall, 3142 mother–infant pairs met all inclusion criteria for our validation study.

The mean age of the 3142 women meeting the inclusion criteria for this study was 28±6.0 years. Twenty-nine percent were welfare recipients, 52% reported being employed, 61% had a household income of $30 000 or less, and 74% lived in an urban area.

Among the 3142 mothers included in the study, 1426 (45%) reported that their babies had no major congenital malformation and the database record for the baby held no diagnostic code for a malformation. In 14% of instances, the mother reported that her baby had been born with a major congenital malformation, and the database record held a diagnostic code of at least one major malformation. In total, the database record and information from the mother agreed with whether or not a given infant had a major congenital malformation in 60% of instances (95% CI 58% to 61%) (Table 1). In 38% of instances the mother indicated that her baby had no major congenital malformation but the RAMQ record held at least one diagnostic code for a major malformation.
The most common malformations recorded for this group were “certain musculoskeletal deformities” (ICD-9 754), 21% (n = 253); “other congenital musculoskeletal anomalies” (ICD-9 756), 15% (n = 180); “bulbus cordis anomalies and anomalies of cardiac septal closure” (ICD-9 745), 12% (n = 144); “other and unspecified congenital anomalies” (ICD-9 759), 10% (n = 120); “congenital anomalies of the eye” (ICD-9 743), 7% (n = 84); “congenital anomalies of genital organs” (ICD-9 752), 6% (n = 72); and “other congenital anomalies of nervous system” (ICD-9 742), 5% (n = 60).

In 2% of instances the mother’s report indicated that her baby had a major congenital malformation, but the RAMQ record held no code related to congenital malformation. The following are some examples of the types of malformations reported only by the mothers, with the relevant ICD-9 code for major congenital malformation in parenthesis: bicuspid aortic valve (ICD-9 746.4); club foot (754.5–754.7); paraxial ulnar hemimelia (755.2); hypospadias (752.6); Perthes disease (732.1); absence of hip (755.6); VACTERL syndrome (759.8 or as per condition being treated); Sprengel’s deformity (755.5); craniosyntosis (756.0); and partial trisomy 16 (758).

Among 456 infants for whom both RAMQ and mother reported a major congenital malformation, we assessed whether the mother’s description of the malformation was the same as that denoted by the diagnostic code held in the RAMQ database (Table 2). Five mothers did not provide a description and so were excluded from the analysis. Among the 451 mothers who provided a description of the malformation, the mother’s description was consistent with the RAMQ code in 90% (95% CI 87% to 93%) of instances; that is, the mother’s answer was consistent with at least one of the diagnostic codes held in the database for that baby or could be included in a non-specific category.

### DISCUSSION

In this study, we found that in 60% of instances the information about the presence or absence of major congenital malformations that were diagnosed in an infant’s first year of life held in the RAMQ agreed with the mother’s report from a self-administered questionnaire. Further, in 90% of cases in which both the RAMQ and mother’s report indicated that the child had been born with a malformation, the specific malformation reported by the mother was consistent with at least one of the diagnostic codes that had been assigned by a physician seeing that baby at some time during the first year of life.

In 38% of instances the RAMQ had recorded a major congenital malformation but the mother reported that the baby did not have a congenital malformation. There are a number of possible reasons for this result. Many early malformations resolve spontaneously or are corrected by treatment. Records of the presence of these early malformations remain unchanged in the database, while the mother’s information about her child is updated with time. For example, a newborn may be coded as having a heart murmur at birth, but the murmur resolves spontaneously within the baby’s first months of life. Months or years later, the mother may report that the baby had no congenital malformation, but the record of that early murmur remains
in the database. Further, some malformations may be suspected initially but ruled out after further investigation; the database record retains the original malformation code and has no mechanism for updating that information to show that the malformation was not in fact present. Mothers may be less likely to report malformations that are not visible to them. Indeed, in an earlier study, the type of malformation was found to be the most important factor in determining a mother’s ability to recall the presence and type of congenital malformation. These authors found that maternal recall was best for malformations that have life-long effects on the family and is poorest for those malformations that are amenable to early and effective treatment. Mothers may deny or may not be aware of the existence of a malformation in their infant. Some mothers may misunderstand the term congenital malformation, or may take it to mean only certain specific anomalies that their child does not exhibit.

As all mothers with babies for whom the RAMQ record held a diagnostic code of malformation were invited to complete the questionnaire, the mother–infant pairs were selected in a way to maximize the number of babies with congenital malformations (n = 1674, according to RAMQ). The mother–baby pairs in which the baby had no RAMQ diagnosis of malformation were selected by a random sample (n = 1486) of the Quebec Pregnancy Registry. This enhanced our ability to study the question of agreement between physician-based diagnostic codes in the RAMQ and maternal report of congenital malformation. This approach also allowed us to estimate the extent to which the RAMQ record missed a diagnosis of congenital malformation reported by the mother. In 4% of instances among the 1486 with no diagnostic code for congenital malformation in the RAMQ, chosen by random sample, the mother reported that the baby had a congenital malformation. Most of the malformations reported only by mothers were either minor or would not medically be considered a malformation. Given that major congenital malformation is the outcome of interest for studies of prenatal exposures, the RAMQ record appears to agree with the maternal report with respect to specific type of major congenital malformation.

Our results are similar to those of a study conducted in the Metropolitan Atlanta Birth Defects Program, in which 61% of mothers of babies with congenital malformations according to hospital medical records responded during an interview that their baby had a congenital malformation; 98% of control mothers reported no malformation. These authors found that the proportions varied widely within subgroups of type of malformation and sociodemographic characteristics of the mother. In our study, 60% of mothers of babies for whom the RAMQ held a congenital malformation diagnostic code reported by questionnaire that their baby was born with a congenital malformation; 96% of mothers of babies with no diagnosis of congenital malformation in the RAMQ reported in response to a questionnaire that their baby had no congenital malformation. We are aware of only one other study that has examined agreement between maternal questionnaire response and registry record of congenital malformation, that study was different from our study in that the registry information was ascertained only during the neonatal period, while mothers in our study completed the questionnaire some years later.

A number of studies have attempted to validate other sources of congenital malformation information using medical chart review. These have used a birth certificate as the source of information and not a medical claims database. Although medical chart review is a preferred approach to validation of diagnostic information held in administrative databases, it is not without some limitations. Indeed, in a study of the validation of diagnostic codes for asthma in the RAMQ database using the medical chart as a gold standard, medical charts were found to be inconsistently complete. Others have used medical chart review as a putative gold standard in validating diagnostic codes held in the RAMQ database for a number of conditions, but they caution that medical charts are not error-free and suggest that an important number of diagnoses may not be documented in the chart, leading to misclassification.

CONCLUSION

This is the first study to compare the diagnostic codes for congenital malformations held by the RAMQ database with a second source of information: the mother’s report. The RAMQ diagnostic codes, specified by physicians in course of making reimbursement claims, appear to provide comprehensive and highly accurate information about congenital malformations that have been diagnosed in babies. Care providers should be cognizant of the fact that the diagnostic codes entered on reimbursement claims forms are being used for research purposes. The evidence produced by this research serves to inform clinical practice; thus, diagnoses should be recorded as accurately and specifically as possible.

REFERENCES


