Using ICD-9 Codes to Establish Prevalence of Malformations in Newborn Infants

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BACKGROUND: The International Classification of Disease (ICD-9) codes are used to identify, after discharge, diagnoses from a review of the medical record and provide the basis for reimbursement. These codes have been used to establish the prevalence of malformations and to assess potential teratogens. We have analyzed the accuracy of codes 740 to 759.9 to identify newborn infants with malformations. **METHODS:** The diagnoses and ICD-9 codes in the medical records of 1000 consecutive live-born and stillborn infants were compared to the information provided in the medical records of those infants. **RESULTS:** One hundred twenty-seven of the 1000 infants had ICD-9 codes between 740 and 759.9. 67 (52.8%) of the codes identified minor features, such as birth marks and minor anomalies. Twenty-three (18.1%) of the codes designated a malformation and were correct. Two types of errors were identified in another 33 infants (26%) whose codes designated a malformation: either the pediatricians' notes described a less severe finding or the fact that there was no such abnormality. In addition, four malformed infants were missed in pregnancies that were either terminated electively or stillborn, as they did not have medical records. **CONCLUSION:** The ICD-9 codes 740 to 759.9 identified accurately some infants **(**18%**)** with malformations, but identified incorrectly many others. The accuracy of the coding for identifying malformations would be improved if (1) the findings of the examining pediatricians were considered; (2) normal features of prematurity, such as patent foramen ovale and patent ductus arteriosus, were not considered malformations; (3) minor physical features were not assigned ICD-9 codes within the 740 to 759.9 sequence. *Birth Defects Research (Part A) 94:208–214, 2012.* © 2012 Wiley Periodicals, Inc.

Key words: ICD-9 codes; malformations; exclusion criteria

INTRODUCTION

The International Statistical Classification of Diseases and Related Health Problems began in 1893 as an International List of Causes of Death. The World Health Organization began in 1948 to coordinate the revisions of this listing for the sixth revision. The scope was extended to include nonfatal diseases. The ninth revision was published in 1975. Work on the tenth revision began in 1983. The title has been amended over the years, and the abbreviation ICD has been retained (International Statistical Classification of Diseases, 1992). ICD-9-CM was used as a modification to designate the clinical modification of the World Health Organization's ICD-9 (Castillo et al., 1999). By October 1, 2013, physicians and hospitals must start using the ICD-10 codes instead of the ICD-9 codes.

The ICD-9 code numbers 740.0 through 759.9 include all malformations from *anencephaly* through *congenital anomaly, unspecified*. This digitized system is used by certified coding specialists, who have been accredited by the American Health Information Management Association, to identify the malformations recorded in the medical record. These diagnoses are based on the findings recorded by the examining physicians, nurses or consultants, the reports of procedures, the results of imaging studies, and the findings in diagnostic testing.

The hospital uses the ICD-9 codes on discharge as a measure of the complexity of an individual's diagnoses

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and for a list of procedures performed. These codes provide the basis for the invoice submitted to the insurance company for reimbursement for the hospital admission. The ICD-9 codes have also been used to establish computerized databases to determine the occurrence of malformations in newborn infants who have been exposed to medications during pregnancy (Andrade et al., 2005; Frohnert et al., 2005; Cooper et al., 2008; Davis et al., 2011) and as a basis for establishing the prevalence of malformations (Feldkamp et al., 2005).

Inaccuracies in studies using ICD-9 codes have been noted in the rate of detection of specific common malformations. For example, there has been higher accuracy in the detection of cleft lip compared with the rate of detection of heart defects and abnormalities of the spine, as well as problems with many false-positive findings (Faciszewski et al., 1997; Feldkamp et al., 2005; Frohnert et al., 2005).

We have evaluated the accuracy of ICD-9 codes for identifying malformations in newborn infants. This has been done by comparing the ICD-9 codes assigned with the findings recorded for the same infants by the examining pediatricians in the medical records of a consecutive sample of 1000 live-born and stillborn infants.

MATERIALS AND METHODS

This project was carried out within the context of the Active Malformations Surveillance Program (Nelson and Holmes, 1989; Peller et al., 2006), which was established at the Boston Lying-In Hospital on February 16, 1972. It has been carried out since then, except for a hiatus from February 15, 1975, until December 31, 1978, which was caused by a lack of office space. In 1981, this surveillance program moved to the new Brigham and Women's Hospital, which was formed from the merger of the Boston Lying-In Hospital with three other hospitals. This survey has been conducted 6 days per week and on holidays by research assistants who have read the findings recorded by the examining physicians in the medical records of each live-born and stillborn infant.

In this analysis, a malformation was defined as a structural abnormality with surgical, medical, or cosmetic importance. The word *malformation* is considered the same as a major malformation or a significant malformation.

The senior research assistant and coordinator of the surveillance program (M-N.W.) identified each infant born at this hospital from a review of the Labor and Delivery Birth Log, which lists each delivery each day. A consecutive sample of 1000 live-born and stillborn infants was identified between January 1, 2008, and February 18, 2008. The features recorded in the medical records of live-born infants were those identified between birth and discharge. Information about the findings in prenatal screening by ultrasound was included in the pregnancy histories recorded by the pediatricians. During that study period, 30 to 40 pediatricians could have been examining one or more infants each day. This is an evaluation of the ICD-9 codes assigned during this hospitalization in the period after birth and does not consider any abnormality detected at an older age.

Stillborn infants and elective terminations were identified in the delivery log by the fact that the Apgar scores are listed as 0 at 1 min and 0 at 5 min. If the malformed infant was not born alive, a medical record number was not assigned. The mother's record was read to ascertain whether her infant had anomalies. From this review, it could be established that her infant was malformed and was either stillborn or that an elective termination had been performed. The malformations were confirmed by the findings in the post mortem examinations and the prenatal screening by ultrasound.

The codes 740 to 759.9 include chromosome abnormalities and genetic syndromes. The presence of an associated chromosome abnormality was determined from a review of the reports from chromosome analysis. The presence of a specific genetic diagnosis was established from a review of the reports of the pediatricians, and consultations by geneticists, endocrinologists, surgeons, and other specialists. The results of mutation analyses and other diagnostic tests were used to confirm some diagnoses, whenever available.

The types of minor physical features to be identified and excluded in this analysis were established in the same sample of 1000 infants (Holmes and Westgate, 2011). The delineation of all types of minor physical features was based on our previous experience in this project with identifying the presence of minor anomalies and normal variations (Holmes et al., 1987; Leppig et al., 1987), birth marks (Alper and Holmes, 1983), and genetic abnormalities (Nelson and Holmes, 1989; Rasmussen et al., 1996; Lin et al., 1998) in systematic study examinations. The physical features excluded were (1) minor anomalies and normal variations, such as syndactyly of the second and third toes and clinodactyly of the fifth finger; (2) birth marks, such as hemangiomas and mongolian spots; (3) positional deformities, such as torticollis; (4) prematurity-related physical features, patent ductus arteriosus in an infant <36 weeks gestational age; (5) physiologic findings, such as a small, asymptomatic muscular ventricular septal defect; (6) abnormalities identified by ultrasound during pregnancy, but not confirmed by the examining physician in the newborn infant, such as hydronephrosis; (7) functional abnormalities identified by newborn screening, such as hearing loss; (8) abnormalities of serum amino acids or hemoglobins identified by newborn screening, such as elevated phenylalanine in an infant with phenylketonuria.

Each physical feature on each newborn infant with an ICD-9 code between 740 and 759.9 was identified from a manual review of the discharge abstract for that infant's printed hospital record, which was available on line. The progress notes recorded by the examining pediatrician, the reports by each consultant, and all results of diagnostic tests were reviewed to establish the diagnoses for each infant. A comparison was made between the diagnoses designated by the ICD-9 codes assigned by the coders and the information recorded by the examining pediatricians and all consultants in that infant's medical record.

RESULTS

The infants whose medical records were reviewed were hospitalized for varying periods of time: 50% for 2 days or less, 32% for 4 days, and 18% for more than 4 days. Seventy-one percent of the infants were examined by either their private pediatrician or a staff hospitalist; 21% were examined initially by a resident in pediatrics, followed by a staff hospitalist. The remainder (8%) were transferred immediately to Children's Hospital–Boston

Table 1
Malformations Identified Correctly

Malformation	No. affected	ICD-9 code
Trisomy 21	3	758.0
Trisomy 13	2	758.1
Cleft Palate	2	749.0
Congenital diaphragmatic hernia	1	756.6
Epispadias	1	752.62
Double outlet right ventricle	1	747.1
Double outlet right ventricle with transposition of the great arteries and coarctation of the aorta	1	745.1, 745.11, 747.1
Transposition of the great arteries	2	745.1
Hypoplastic left heart syndrome	2	746.7
Tricuspid atresia	1	746.1
Ventricular septal defect, membranous type	1	745.4
Ventricular septal defect, conoventricular type	1	745.4
Microtia	1	744.2
Multiple congenital anomalies, unknown etiology	2	759.7
Polydactyly postaxial, type b, hands and feet	1	755.01, 755.02
Venovenous malformations of the liver	1	747.4
TOTAL	23/1000 (2.3%)	

for treatment. Those medical records were reviewed to establish the diagnosis of the affected infants.

One hundred twenty-seven of the 1000 infants had ICD-9 codes between 740 and 759.9. The review of the pediatricians' notes and the other reports in the medical record showed that there were more errors in the ICD-9 codes used than correct diagnoses:

1. Twenty-three (18.1%) of the 127 infants with codes in this range had significant malformations that were identified accurately (Table 1).
2. One (0.8%) of the 127 infants was identified as malformed, but the correct diagnosis was not provided. The ICD-9 code used (753.14) was for polycystic kidney–autosomal recessive. The infant had glutaric aciduria, type II in association with hypotonia, polycystic kidneys and genital defects, a rare disorder (Mendelian Inheritance in Man no. 231680).
3. Fifteen (11.8%) of the infants had ICD-9 codes that suggested a significant malformation, but the records showed that these findings were either not significant findings (i.e. the infant did not have a malformation) or the ICD-9 codes designated diagnosis was not correct (Table 2).
4. Eighteen (14.2%) of the infants had physical findings that should not have been listed as a malformation, as the findings were either minor in nature or excluded because they were normal characteristics of a premature infant, such as undescended testes or the presence of a patent ductus arteriosus (Table 3).
5. Three infants (2.4%) had physical findings that could not be classified, because of inadequate information in the medical records: one infant with hypospadias, and two infants with a ventricular septal defect. Hypospadias can be mild, such as glandular hypospadias, which would be excluded by the criteria used in this analysis (Holmes and Westgate, 2011). If the location of the abnormal urethral opening is not recorded by the examining pediatrician, this finding cannot be classified as either an exclusion or a malformation. Likewise, there are several types of ventricular septal defects: muscular (excluded), membranous and conoventricular (both included). If the type has not been determined, this finding cannot be classified.
6. Seventy-one of the codes in 67 infants (52.8%) were used for minor physical features that were excluded in this analysis. The specific exclusions were: minor anomalies (16), birth marks (24; Table 4), ''ultrasound only'' findings (15), prematurity-related (12; Table 5), physiologic findings (3), and positional deformations (1). The specific exclusions noted seemed to reflect the special interests of the examining pediatricians. For example, eight infants were designated as having tongue-tie (ICD-9 code 750.0). This term designates a shortening of the frenulum beneath the tip of the tongue, which is defined subjectively (Cho et al., 2010). The nursing staff reported that this diagnosis was suggested often when the infant had difficulty with breast feeding.

The most common birth mark noted was mongolian spot, a blue-black macule with ill-defined borders located most often on the buttocks or lower back. This finding has no medical significance. There are dramatic ethnic differences in the prevalence of this birth mark: 89% of African American infants and 5% of white infants (Table 4; Alper and Holmes, 1983).

Two additional malformations were identified with ICD-9 codes outside of 740 to 759.9:

1. ICD-9 code 665.81: designated *suspected fetal abnormality* in the mother's medical record; this was an infant

Table 2
Malformations Coded but Not Correct, Based on Pediatrician Findings

Malformations (n)	ICD-9 code	Pediatrician findings
Chondrodystrophy (1)	756.4	Protuberant scapula only
Chromosome anomaly (1)	745.5	No chromosome abnormality identified
Congenital skull/face/jaw deformity (1)	759.0	Asymmetry of head in twin due to crowding in utero
Coarctation of aorta, suspected during pregnancy (2)	747.	Coarctation not confirmed in postnatal imaging
Patent ductus arteriosus (4)	747.	Premature infant, murmur resolved
Other heart defects (3)	747.	Murmur resolved (1), no heart defect (2)
Hydronephrosis, diagnosed during pregnancy (1)	753.2	Resolved
Hypospadias (1)	752.61	No hypospadias
Undescended testicle (1)	752.51	Testis palpable
Total: 15/1000 (1.5%)		

Table 3
Listed as Malformation but Code not Adequate

Diagnosis (n)	ICD-9 Code	Pediatrician's description
Accessory Auricle (5)	744.1	Preauricular tags or sinus
Brain anomaly (3)	742.4	Choroid plexus cysts
Brain anomaly (1)	742.2	29-week gestational age; germinal matrix hemorrhage; resolved
Congenital anomaly of the breast (1)	757.6	Accessory nipple
Heart defects (8)	747.	
Atrial septal defect (3)	745.5	Premature infant with patent foramen ovale
Ventricular septal defect (1)	745.4	Tiny apical ventricular septal defect
Patent ductus arteriosus (4)	747.0	Premature infant
Pulmonary artery anomaly (6)	747.3	Peripheral pulmonary stenosis
Laryngeal anomaly (2)	748.3	Laryngomalacia
Upper limb anomaly (1)	753.4	Clinodactyly of fifth finger
Total: 18/1000 (1.8%)		

with sacrococcygeal teratoma, which had been detected by prenatal screening; the pregnancy was terminated.

2. ICD-9 code 255.2: designated *adrenogenital disorders*; this was a female infant with virilization owing to congenital adrenal hyperplasia; this infant was treated successfully after birth.

Four infants with malformations were not assigned ICD-9 codes, as none of these infants had a medical record. Three of these infants were identified in prenatal screening and those pregnancies were terminated:

1. Sacrococcygeal teratoma (noted previously; ICD-9: 665.81)
2. Renal agenesis, bilateral (ICD-9: 753.0).
3. Spondyloepiphyseal dysplasia congenita (with mutation in *COL2A1* gene; ICD-9: 756.5).

The fourth infant with a malformation that was not identified was a stillborn infant with trisomy 21 (Down syndrome; ICD-9: 758.0).

In the review of the medical record of these 1000 liveborn and stillborn infants, no malformations were identified that did not have an ICD-9 code.

DISCUSSION

This analysis of the coding among 1000 consecutive births showed that: (1) the codes were correct for 23 infants with malformations (ICD-9 codes 740 to 759.9), which were 18.1% of the codes listed for 127 of the infants surveyed; (2) an additional 26% of the physical features were coded as malformations, but were not correct; and (3) the code numbers 740 to 759.9 were used most often to identify 67 infants (52.8%) who had only minor physical features, which would be excluded as not being a malformation.

If ICD-9 codes recorded for these 1000 infants had been used as a proxy for the identification of all infants with significant or major malformations, that approach would have been highly inaccurate. However, many of the errors that designated incorrectly an infant as mal-

Table 4
Common Birth Marks in Newborn Infants

Birth mark name	Description
Mongolian spot	Blue-black macules; often large and ill defined; most common on buttocks and back; present in 4.8% of white infants, 88.7% of black infants, 65% of Hispanic infants
Nevus simplex (salmon patch; stork bite)	Bright red, well-defined macules; most common on glabella, upper eyelid and nape of neck; present in 70.3% of white infants, 59.2% of black infants, 67.9% of Hispanic infants
Café-au-lait spot	A light to dark brown evenly pigmented macule with sharply defined margins; the skin markings are continuous with those of the surrounding skin; one café-au-lait spot present in 0.3% of white infants, 12% of black infants, 3.2% of Hispanic infants.
Infantile hemangiomas	In the first 4 days of life, these lesions are not elevated or easy to identify, appearance varies from blue-red macules with numerous fine telangiectases to faint bluish-white macules with ill-defined borders; within the first weeks of life an elevated red hemangioma may develop; present in 1.7% of white infants, 0.6% of black infants, and 1.4% of Hispanic infants
Nevocellular nevus	Black and elevated, with increased skin markings and often with speckling in periphery; typically less than 1 × 0.5 cm in size; present in 1% of white infants, 1.8% of black infants, 0.8% of Hispanic infants
Depigmented nevus	Well-defined areas with decreased pigment in comparison to adjacent areas; skin markings normal; present in 0.4% of white infants, 2.4% of black infants, 0.4% of Hispanic infants

Based on the findings in the systematic examination of 4641 liveborn and stillborn infants (Alper and Holmes, 1983; Alper et al, 1979). The ethnic and racial distributions were: 2,682 white infants, 492 black infants, 250 Hispanic infants, 1058 infants of mixed ancestry, and 159 infants from other ethnic groups.

Table 5
Prematurity-Associated Findings in Newborn Infants

Finding	Description
Patent ductus arteriousus (PDA)	A normal physiologic connection between the aorta and pulmonary artery remains open in most low-birth-weight premature infants; in term infants, closes within hours of birth. A persistent PDA in a term infant is considered a malformation that requires treatment (Koch et al, 2006).
Patent foramen ovale (PFO)	A normal physiologic connection between the right and left atria of the heart; most preterm, low-birth-weight infants have some atrial shunting through the patent foramen ovale, especially those supported by mechanical ventilation (Evans et al, 1994); closes soon after birth in most, but not all, term infants.
Undescended testicles (UDT)	Testes descend normally after 23 weeks of gestation; 98% have descended after 32 weeks of gestation (Rotondi et al, 2001). Failure to descend into scrotum of a term infant is considered a malformation.
Inguinal hernia	Each testicle descends from the abdomen into the scrotum. The persistence of the processus vaginalis becomes the route of the inguinal hernia; 13% of infants born at less than 32 weeks gestational age have an inguinal hernia; 30% of infants weighing less than 1000 gm at birth have an inguinal hernia (Peevy et al, 1986).

formed would have been corrected if the findings and interpretations provided by the pediatricians' observations recorded in the medical record had been considered by the coder and the discharge diagnoses had been changed accordingly.

The other specific limitations of the ICD-9 coding were:

1. Twelve infants had ICD-9 codes that designated features related to prematurity, such as the presence of a patent ductus arteriosus (PDA) or a patent foramen ovale (PFO) and a soft immature trachea, which should be considered a normal finding in an infant of that gestational age (Table 5). The medical progress notes of very premature infants will be expected to include one or more of the common features of prematurity (Table 5). These characteristics of prematurity are expected to be treated, in the case of the PDA, or to resolve as the infant develops and reaches the equivalent of a term gestation. However, if a PDA or undescended testis persists, those findings in a newborn infant at 37 or more weeks of gestation would be considered a malformation. Inguinal hernias are also more common in premature infants than in term infants (Peevy et al., 1986). Surgical repair is needed.
2. The coders had a limited number of options for the minor physical findings recorded by the pediatricians. For example:

   a. The typically minor physical feature, preauricular tags, is designated with the more general term *accessory auricle*, which sounds more significant medically. Fortunately, there is no phenotype of an ''extra'' ear in the spectrum of congenital anomalies. Preauricular tags are usually a minor anomaly, which would be excluded as not a malformation.
   b. The sonographic ''soft marker'' or anatomic variant, such as choroid plexus cyst, cannot be listed by a code for minor anatomic variants. The more general term *brain anomaly* sounds more serious or significant than an anatomic variant. These soft markers would be excluded as not being a malformation (Table 6).
   c. The immature, more compliant trachea, referred to by clinicians as *laryngomalacia*, was coded instead as a definite abnormality: *laryngeal anomaly*. Laryngomalacia would be considered a complication of prematurity and not a malformation.

3. There are a limited number of specific diagnoses listed among the ICD-9 codes 740 to 759.9. As a result, an incorrect diagnosis will be listed because the more precise diagnosis is not an option for the coder. For example, the diagnosis of a specific type of skeletal dysplasia, such as spondyloepiphyseal dysplasia congenita, cannot be listed with a specific ICD-9 code. Instead, all skeletal dysplasias are listed under the general code for osteochondrodystrophy (no. 756.5), with only six of the many different skeletal dysplasias assigned separate ICD-9 codes.
4. No medical record is created for the malformed fetuses in terminated pregnancies; therefore, these malformed infants were not identified by ICD-9 codes. A previous study (Peller et al., 2006) of the malformed infants born at this hospital between 1974 and 1999 showed that 20% were in pregnancies terminated by a parent's choice. These abnormalities were predominantly lethal or severe handicapping conditions. It is possible that during the time period of this analysis in

Table 6
Common Nonstructural Soft Markers Identified by Ultrasound at 15 to 20 Weeks' Gestation in Control Fetuses

Marker	Nyberg et al, 2001 (n = 8675) (%)	Bromley et al, 2002 (n = 656) (%)	Nicolaides, 2003 (n = 9384) (%)
Increased nuchal fold	0.5	0.5	0.6
Short femur	3.9	5.3	5.2
Short humerus	0.4	2.1	1.5
Pyelectasis/ hydronephrosis	2.2	2.4	2.6
Intracardiac echogenic focus	0.5	4.3	4.4
Echogenic bowel	0.5	0.9	0.6
One or more, total	11.3	12.4	14.9

2008 some elective terminations for fetal anomalies identified in prenatal testing occurred at other medical facilities.

5. The inability to designate specific genetic conditions by ICD-9 codes limits any analysis for a postulated teratogenic effect. Consider, for example, the Mendelian or monogenic disorders, which have not been shown to be caused by any teratogenic exposure. These should be excluded as potential teratogenic effects. How common are these? In the analysis of the apparent etiologies of malformations among 69,277 newborn infants at this hospital, 48 (3.1%) of 1549 were attributed to autosomal or X-linked gene mutations. An additional 157 (10.1%) were associated with chromosome abnormalities (Nelson and Holmes, 1989).

Will the use of the ICD-10 codes solve the problems identified in this analysis? There will be many more codes available: 14,025 in ICD-9 compared with 68,069 in ICD-10. There will be separate codes for omphalocele (Q79.2), gastroschisis (Q79.3), and prune belly syndrome (Q79.4), whereas a single code (756.7) was used for these three abnormalities in the ICD-9 codes. In addition, these are separate codes for unilateral (Q60.0) and bilateral (Q60.1) renal agenesis. However, several limitations noted in this analysis have not been resolved:

1. There is no designation of normal, prematurity-related findings (Table 6), such as PDA or PFO.
2. The code for atrial septal defect (Q21.1) is the same as the code for PFO.
3. The most common type of polydactyly, with the extra digit on the postaxial (ulnar or fibular) side of the hand or foot, cannot be designated. This type of polydactyly is hereditary and occurs in 1% of African American infants and 0.1% of white infants (Temtamy and McKusick, 1978).
4. There is no designation for the common anatomic variants, such as short femur and pyelectasis, which have been detected in 11 to 15% of pregnancies (Table 6).
5. Only a limited number of specific malformation syndromes and genetic disorders can be designated specifically.

This analysis showed that ICD-9 codes alone cannot be used to determine the prevalence of malformations in newborn infants. To be reliable, diagnoses must be validated. If the medical record is read and used to clarify the information being designated by the ICD-9 code, the quality of the information being tabulated will be improved significantly. However, even this labor-intensive adjustment would not make the use of ICD-9 codes in epidemiologic studies as accurate as the findings developed in active malformation surveillance programs (Nelson and Holmes, 1989; Rasmussen et al., 2003; Feldkamp et al., 2005). These programs, which rely on a review of the findings in the medical record by trained research assistants and birth defect specialists rather than relying on ICD-9 codes, make it possible to establish apparent etiologies for many malformations and make correlations with exposures in pregnancy much more reliable.

Given the inaccuracy of the ICD-9 codes for identifying malformations in newborn infants, the findings in any study of potential teratogenicity that rely solely on ICD-9 codes for diagnoses should be considered preliminary. Confirmation of the alleged association should be established by a more accurate system, such as determining phenotypes through an active malformations surveillance program or a cohort study in which the exposed and unexposed infants are examined systematically (Holmes LB et al., 2001).

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