TERATOLOGY 64:S3–S7 (2001)

Effective Coding in Birth Defects Surveillance

SONJA A. RASMUSSEN* **AND CYNTHIA A. MOORE**

National Center on Birth Defects and Developmental Disabilities, Centers for Disease Control and Prevention, Atlanta, GA 30341

ABSTRACT Effective coding is critical to data collected by birth defects surveillance programs because subsequent use of the data depends on storage and retrieval of cases using codes. Hence, careful consideration needs to be given to the coding process. The primary goal of coding is to accurately, completely, and concisely represent infants with birth defects. Coding procedures need to accommodate the objectives of the surveillance program; for example, programs that focus on research may require different coding procedures from those that focus on linking infants to services. Several challenges exist in coding birth defects, including the need to distinguish infants with multiple defects and syndromes from those with isolated defects, and the need for strategies to code suspected defects for which confirmation is not available. Selection of a coding system by a birth defects surveillance program is central to the utility of the data collected. Most programs use a modification of the International Statistical Classification of Diseases and Related Health Problems-based (ICD) systems. This paper addresses ICD-based systems and the modifications used by many birth defects surveillance programs and presents examples of the problems in interpreting birth defects data because of inappropriate coding. *Teratology 64:S3–S7, 2001.*

Published 2001 Wiley-Liss, Inc.†

INTRODUCTION

Birth defects are a leading cause of neonatal and infant mortality (CDC, '96; Peters et al., '98), but little is known about the etiology of most birth defects. Since the identification of thalidomide as a teratogen, birth defects surveillance programs have been established in many states and countries to serve as early warning systems for identification of new teratogens, as well as to improve understanding of birth defects etiology (Källén, '84; Khoury et al., '94a). Effective coding is an essential component of birth defects surveillance. Although verbatim descriptions are helpful, consistent terms are often not used (for example, several different terms could be used to describe a lumbar meningomyelocele), and coding allows aggregation of similar cases. In addition, because birth defects surveillance programs process large numbers of cases, coding of defects is necessary for case storage and retrieval.

Previous studies have demonstrated problems associated with interpretation of birth defects surveillance data resulting from inappropriate coding (Cunniff et al., '94; De Wals et al., '99; Reefhuis et al., '99). This paper discusses the importance of a carefully defined case definition, goals of the coding process, selection of a coding system, and potential problems with coding in birth defects surveillance programs.

Case definition

Before a coding system can be selected, a case definition needs to be carefully delineated. Case definition for a birth defects surveillance system has two components: characteristics of the infant to be included and types of defects to be ascertained. Characteristics of the infant include demographic information (e.g., mother's residence at the time of the infant's birth), birth outcome (e.g., live-born, fetal death, elective termination), and minimum gestational age or birth weight. Whether to include elective terminations in the system is an important consideration because many defects are now identified prenatally, and exclusion of these defects can significantly alter surveillance data (Limb and Holmes, '94; Roberts et al., '95). Programs should also consider limiting ascertainment to cases in which a defect is diagnosed by a certain age.

The types of defects to be ascertained must also be carefully considered. Some programs focus on structural birth defects; others expand their case definitions to include other health problems (e.g., inborn errors of metabolism). Specific examples of exclusions should be provided, because the classification of some defects as structural or functional may not be clear to everyone involved in the surveillance system. Whether only major defects (those of medical, surgical, or cosmetic significance) will be ascertained or whether an attempt will be made to ascertain minor anomalies (those unlikely to be of medical, surgical, or cosmetic significance) also must be considered. Minor defects are important in the study of birth defects because they often suggest the presence of a syndrome of known etiology. In addition, the presence of three or more minor anomalies is predictive of a major malformation (Leppig et

*Correspondence to: Sonja A. Rasmussen, MD, MS, 4770 Buford Highway, NE, Centers for Disease Control and Prevention, MS F45, Atlanta, GA 30341. E-mail: skr9@cdc.gov

al., '87). However, recognition of minor anomalies has not been standardized (Lechat and Dolk, '93), and these anomalies often are not ascertained well in birth defects surveillance programs; therefore, ascertainment of infants with isolated minor anomalies may not be the best use of resources, given a surveillance program's goals.

Birth defects surveillance programs also need to specify how the program will address anomalies that occur commonly among premature infants, such as patent ductus arteriosus. Often programs exclude these prematurity-related anomalies in infants on the basis of gestational age (e.g., < 37 weeks) or birth weight (e.g., < 2500 g) because these are normal physiologic differences associated with gestational age, not structural birth defects.

When determining the case definition of a birth defects registry, the planned application of the data collected must be considered. Types of application of data from birth defects registries include surveillance (for calculation of rates, study of trends, identification of clusters, and evaluation of prevention activities), as a source for analytic studies of potential risk factors, for health services research and evaluation, to ensure sufficient resources are available to meet health service needs (Cordero, '92) and for referral to health-care services, such as early intervention. A program that focuses on referral of at-risk children to health-care services may define defects broadly; one focusing on analytic studies may use a narrow definition of only major structural birth defects. Some programs may focus on only a few specific defects.

Goals of coding in a birth defects surveillance program

The major goal of coding in a birth defects surveillance program is to represent as accurately as possible the defects in the infants. Källén ('88) describes the "distortion" of information that occurs in a surveillance program from the "truth" (the malformed infant) to the coded information. Information is lost or distorted as the malformed infant is viewed, described and recorded in the medical record by the physician, and abstracted and coded by the surveillance program (Källén, '88). The goal should be to minimize this distortion as much as possible, while realizing that a certain amount of distortion is inevitable.

To allow for an accurate picture of an infant with birth defects through codes, codes need to be specific. The use of specific codes allows a researcher easy retrieval of infants even with uncommon birth defects. In addition, specific coding allows classification of cases using knowledge of presumed embryologic and pathogenetic mechanisms, a process that can make case groups more homogeneous (Khoury et al., '94b). An effort should be made to minimize the number of defects that will be coded under nonspecific code categories, such as "other specified" defects of a particular organ system. Coding the defect as specifically as possible (for example, to use a code that describes not only meningomyelocele but the level of lesion and the presence or absence of hydrocephalus) is advantageous to prevent additional work at the time of analysis. However, this goal for specificity needs to be balanced against producing a coding system that becomes cumbersome; defect codes that will be used only rarely are not beneficial.

For accurate translation of medical terminology into birth defects codes, we believe case records and codes need to be reviewed by a person with knowledge of birth defects and the coding system. The protocol of many programs includes a review of cases (selected or all cases) and their assigned codes by a pediatrician with special knowledge of birth defects, as records are collected. This review can be helpful not only for optimal coding, but also to assist with other issues, such as when contradictory information is received from different ascertainment sources (e.g., autopsy vs. echocardiography).

Although translating a defect into a code is typically necessary for storage of data, retaining verbatim information about the defect in the surveillance program is advantageous. Even if a specific code is not available for a particular defect, the information is still available to a researcher willing to carefully examine case records. To allow for the researcher to analyze the accuracy of the description, documentation by the abstractor of how the defect was diagnosed is also helpful. For example, diagnosis of a ventricular septal defect (VSD) by auscultation would be viewed differently from the same diagnosis by echocardiogram. Other defects may be suspected but never confirmed (e.g., diagnosis of possible Down syndrome in an infant for whom confirmatory chromosome analysis is not available). Likewise, a defect diagnosed by prenatal ultrasound without postnatal confirmation would be viewed differently from one diagnosed by postnatal examination because some of these defects (e.g., hydronephrosis, ventriculomegaly) may resolve without treatment (Morin et al., '96; Vergani et al., '98). A standardized method of coding these suspected defects is necessary. All of these issues present challenges to the coding system, which need to be dealt with uniformly.

Another important feature of coding is that the coded data be complete. Although a child may have one defect that initially seems most important, including codes for additional defects is also crucial because these defects may shed light on etiology. In addition, complete coding is important for future case classification. Because etiologic heterogeneity can complicate epidemiologic studies designed to identify birth defects etiology (Friedman, '92; Khoury et al., '92a; Khoury et al., '92b), classification of cases into categories of isolated cases, cases with additional unrelated major defects, and cases with syndromes is important. Inclusion of cases of known etiology can dilute the magnitude of an observed association toward the null (Khoury et al., '92a), and infants with additional major defects have also been shown to have different etiologic and epidemiologic factors than isolated cases (Khoury et al., '82). The

coding process should be sufficiently complete to allow for this case classification process.

In addition to accurate and complete coding, we believe an effort should be made to avoid coding redundancy. For example, when an infant has a tetralogy of Fallot, separately designating the component defects (VSD, overriding aorta, pulmonic stenosis, and right ventricular hypertrophy) is not necessary. Redundant coding also can occur when several nonspecific codes are used to describe a specific defect. For example, a child with radial aplasia and rudimentary thumb could be described using several other terms including radial clubhand, short forearm, curved forearm, absent radius, and absent thumb. However, when available, a more concise code is preferable, such as 755.260 from the coding system used by the Centers for Disease Control and Prevention (CDC) (preaxial longitudinal reduction defects of upper limb), and provides more information about the defect. Redundant codes are inefficient and can lead to confusion. In addition, redundant coding can result in counting a case in multiple defect categories (e.g., counting an infant with tetralogy of Fallot in this defect category, as well as in the pulmonic stenosis and VSD categories, if concise coding procedures are not used). However, defects accompanying a syndrome (e.g., duodenal atresia accompanying Trisomy 21) or an association (e.g., the component defects of the VATER association) should be coded, because the phenotype of these conditions often varies widely. In addition, anomalies that are secondary to an underlying defect (e.g., clubfoot secondary to spina bifida) should also be coded, because these secondary defects are not invariably present.

Selection of a coding system

The selection of a coding system is a critical decision for a birth defects surveillance program because the program's ability to reach the goals delineated above depends on this system. Most birth defects surveillance programs use a coding system based on the International Statistical Classification of Diseases and Related Health Problems (ICD) system published by the World Health Organization and designed to allow international comparability for mortality statistics. Nearly all state programs listed in the state birth defects surveillance programs directory use a modification of the ICD system. The ninth revision of this system (ICD-9) (International Classification of Diseases, '77) has a section entitled "Congenital Anomalies" (740.0-759.9), which includes the majority of congenital defects ascertained by birth defects surveillance programs. In general, this section is arranged by organ systems, with subdivisions specifying defects of particular organs. For example, anomalies of ovaries (752.0), fallopian tubes and broad ligaments (752.1), uterus (752.2, 752.3), and cervix, vagina, and external female genitalia (752.4) are included under the section entitled "congenital anomalies of genital organs" (752). An exception to the hierarchy is the section on chromosome abnormalities, in which etiology is taken into account.

The four-digit ICD-9 coding system has insufficient detail for most birth defects surveillance programs. A clinical modification of ICD-9 (ICD9-CM) has been developed for coding diagnoses associated with hospital reimbursement in the United States (International Classification of Diseases, '98). This system provides further detail (e.g., it further divides anomalies of the cervix, vagina, and external female genitalia into the following categories: unspecified anomalies (752.40), embryonic cysts (752.41), imperforate hymen (752.42), and other anomalies (752.49)). Surveillance programs with passive case ascertainment often use ICD-9-CM codes because these codes are provided from a frequently used ascertainment source (i.e., hospital discharge diagnoses). This modification, although an improvement over ICD-9, is often still not sufficiently precise for many birth defects registry uses. For example, in ICD-9-CM, 756.79 codes for "other congenital anomalies of abdominal wall" and includes both omphalocele and gastroschisis, defects that are etiologically and epidemiologically heterogeneous (Calzolari et al., '95; Hoyme et al., '81). The British Paediatric Association (BPA) modified this system to a five-digit code (British Paediatric Association, '79), and some programs use this coding system (Lechat and Dolk, '93). Many programs in the United States use an ICD-9-based six-digit coding scheme for birth defects developed by the CDC from the BPA-modification of ICD-9 (CDC, 2000; Cordero, '92). Two examples of the additional detail provided by this coding scheme are included in Table 1. A copy of this coding system can be obtained from the authors.

Although the CDC modification allows more specific coding than other coding systems, it still cannot accommodate some defects or diagnoses. In particular, the coding of syndromes has presented a challenge. Some programs have used the classification scheme from the Online Mendelian Inheritance in Man (OMIM) (2000) for conditions with Mendelian inheritance (Lechat and Dolk, '93). Other programs have developed new systems to deal with these conditions (Zwamborn-Hanssen et al., '97).

A tenth revision of the ICD (ICD-10) has been developed (International Statistical Classification of Diseases and Related Health Problems, '92) and has been used for classifying mortality statistics in the United States since 1999. ICD-10 provides more detail than ICD-9 and is an alphanumeric code; most birth defects are included in chapter XVII (Congenital malformations, deformations and chromosomal abnormalities: Q00-Q99). The alphanumeric nature of ICD-10 allows for additional expansion of the codes, without the codes becoming unwieldy. CDC's National Center for Health Statistics is developing a clinical modification of ICD-10 (ICD-10-CM) for morbidity purposes to replace ICD-9-CM. An implementation date for ICD-10-CM in the United States has not yet been set. Once a final version of ICD-10-CM is available, a modification with further detail, suitable for birth defects surveillance programs, will need to be developed. CDC plans to

TABLE 1. Comparison of ICD-based coding systems

Example: Anomalies of abdominal wall				
Defect	ICD-9 code	ICD-9-CM code	CDC/BPA-modified code	ICD-10 code
Omphalocele	756.7	756.79	756.700	Q79.2
Gastroschisis	756.7	756.79	756.710	Q79.3
Prune belly syndrome	756.7	756.71	756.720	Q79.4
Unspecified anomalies of abdominal wall	756.7	756.70	756.790	Q79.5
Epigastric hernia	756.7	756.79	756.795	Q79.5
Example: Anomalies of diaphragm				
Defect	ICD-9 code	ICD-9-CM code	CDC/BPA-modified code	ICD-10 code
Absence of diaphragm	756.6	756.6	756.600	Q79.1
Congenital diaphragmatic hernia	756.6	756.6	756.610	Q79.0
Diaphragmatic hernia (Bochdalek)	756.6	756.6	756.615	Q79.0
Diaphragmatic hernia (Morgagni)	756.6	756.6	756.616	Q79.0
Hemidaphragm	756.6	756.6	756.617	Q79.1
Eventration of diaphragm	756.6	756.6	756.620	Q79.1
Other specified anomalies of diaphragm	756.6	756.6	756.680	Q79.1
Unspecified anomalies of diaphragm	756.6	756.6	756.690	Q79.1

develop this coding system in conjunction with the National Birth Defects Prevention Network. The development of this new coding system provides the opportunity to remedy problems present in the current version. A comparison of ICD-9, ICD-9-CM, CDC/BPA modification of ICD-9-CM, and ICD-10, using anomalies of the abdominal wall and of the diaphragm as examples, is included in Table 1.

Although identical coding systems in different birth defects surveillance programs are not necessary, some standardization is important if rates from different programs are to be presented and compared. Therefore, as programs make individual modifications to a coding system to increase its specificity, it is helpful if the codes continue to be collapsible to a more general code. For example, if a program adds codes to provide more specific information about VSDs (code 745.4 in ICD-9-CM), the codes should continue to allow for comparison to programs that use only this coding system. The CDC/BPA modification has followed this guideline, with other codes for VSDs being 745.400-498. Programs wishing to modify this coding system further could expand the CDC code 745.480 (other specified ventricular septal defect) to include other codes (e.g., 745.481-muscular VSD, 745.482-membranous VSD). One possible mechanism to increase the consistency of coding among birth defects surveillance programs is for the National Birth Defects Prevention Network to serve as a clearinghouse for proposed code changes.

An important function of birth defects surveillance programs is to follow trends in rates of birth defects; therefore, a coding system needs to be relatively static. However, in some situations, advances in medical technologies necessitate changes in coding systems. For example, a 22q11 deletion associated with DiGeorge/velocardiofacial syndrome would be coded as 758.3 (autosomal deletion syndromes) in ICD-9-CM and 758.380 (other loss of autosomal material) in the CDC modified BPA code. However, given the frequency of 22q deletions in infants with birth defects, especially congenital heart defects (Goldmuntz et al., '98; Goldmuntz et al., '93), adding a specific code for this chromosome abnormality seems appropriate.

Impact of problems with coding

Inaccurate coding has previously presented difficulties in studies of birth defects. For example, in an evaluation of a possible increase in the rate of renal agenesis (Cunniff et al., '94), medical record review of cases showed that of 59 cases, 18 (31%) were incorrectly coded (most often, the miscoded cases had isolated multicystic dysplasia, not renal agenesis). In an evaluation of the validity of diagnoses of neural tube defects in the Canadian Congenital Anomalies Surveillance System, a system that depends on hospital admission/discharge summary databases, three of 10 infants identified with neural tube defects were incorrectly coded (De Wals et al., '99). An increasing frequency of omphalocele was observed in the Netherlands, leading to further examination of these cases (Reefhuis et al., '99). Five cases had been miscoded in earlier analyses; when the coding problems were corrected, no significant increase in omphalocele was observed. These authors recommended a protocol for evaluating an increasing prevalence in a registry, with the first step being an examination of the coding or classification of cases.

Lack of specificity of the coding system frequently occurs in studies of birth defects. Källén et al. ('84) describe an example of this problem encountered in the International Clearinghouse for Birth Defects Monitoring Systems. A possible increase in the rate of femoral aplasia or hypoplasia was noted in one member program, and other member programs attempted to evaluate whether a similar increase was noted in other areas. The eighth revision of the ICD code, used by

most members at that time, was not sufficiently specific to identify this type of limb deficiency. An arduous review of cases was required before programs were able to respond to this inquiry.

Problems with coding have a major impact on the data produced by birth defects surveillance programs. Because cases are typically retrieved using codes, a case is essentially lost to the system if it is inappropriately coded. These problems can be enhanced by the rarity of some individual birth defects; thus, only a few inappropriately coded cases can greatly influence rates. Because of the importance of coding, a process to evaluate coding quality is beneficial to a surveillance program. For example, a program may institute a process in which a sample of cases is recoded to ensure that defects are being coded accurately and consistently. The concept that birth defects codes are merely surrogates for the verbatim information about the infant's defects should always be recognized. If results obtained from coded information suggest an atypical finding (e.g., an increasing trend of a defect), the information should always be confirmed by viewing the verbatim information collected about the defects. Selection of a coding system and the coding process itself need to be carefully considered, given the central role that coding plays in birth defects surveillance. Careful consideration of the issues discussed here will maximize the usefulness of data collected and coded by birth defects surveillance programs.

ACKNOWLEDGMENTS

The authors thank Dr. Paula Yoon for her helpful discussions and review of the manuscript.

LITERATURE CITED

British Paediatric Association. 1979. Classification of Diseases. London: British Paediatric Association.

Calzolari E, Bianchi F, Dolk H, Milan M. 1995. Omphalocele and gastroschisis in Europe: a survey of 3 million births 1980-1990. EUROCAT Working Group. Am J Med Genet 58:187–194.

CDC. 1996. Infant mortality—United States, 1993. MMWR Morb Mortal Wkly Rep 45:211–215.

CDC. 2000. State birth defects surveillance programs directory. Teratology 61:33–85.

Cordero JF. 1992. Registries of birth defects and genetic diseases. Pediatr Clin North Am 39:65–77.

Cunniff C, Kirby RS, Senner JW, Canino C, Brewster MA, Butler B, Hassed SJ , Murphy P. 1994. Deaths associated with renal agenesis: a population-based study of birth prevalence, case ascertainment, and etiologic heterogeneity. Teratology 50:200–204.

De Wals P, Trochet C, Pinsonneault L. 1999. Prevalence of neural tube defects in the province of Quebec, 1992. Can J Public Health 90:237–239.

Friedman JM. 1992. The use of dysmorphology in birth defects epidemiology. Teratology 45:187–193.

Goldmuntz E, Clark BJ, Mitchell LE, Jawad AF, Cuneo BF, Reed L, McDonald-McGinn D, Chien P, Feuer J, Zackai EH, Emanuel BS, Driscoll DA. 1998. Frequency of 22q11 deletions in patients with conotruncal defects. J Am Coll Cardiol 32:492–498.

Goldmuntz E, Driscoll D, Budarf ML, Zackai EH, McDonald-McGinn DM, Biegel JA, Emanuel BS. 1993. Microdeletions of chromosomal region 22q11 in patients with congenital conotruncal cardiac defects. J Med Genet 30:807–812.

Hoyme HE, Higginbottom MC, Jones KL. 1981. The vascular pathogenesis of gastroschisis: intrauterine interruption of the omphalomesenteric artery. J Pediatr 98:228–231.

International Classification of Diseases. 1977. 1975 Revision. Vol. 1. Geneva: World Health Organization.

International Classification of Diseases. 1998. International Classification of Diseases, 9th Revision, Clinical Modification. Washington, DC: National Center for Health Statistics and the Health Care Financing Agency.

International Statistical Classification of Diseases and Related Health Problems. 1992. ICD-10 10th Revision. Vol. 1. Geneva: World Health Organization.

Källén B, Hay, S., Klingberg, M. 1984. Birth defects monitoring systems: accomplishments and goals. In Issues and Reviews in Teratology, Kalter H. (ed), Vol. 2. pp. 1–22. New York: Plenum Publishing Corporation.

Källén B. 1988. Epidemiology of human reproduction. Boca Raton, FL: CRC Press, Inc.

Khoury MJ, Botto L, Mastroiacovo P, Skjaerven R, Castilla E, Erickson JD. 1994a. Monitoring for multiple congenital anomalies: an international perspective. Epidemiol Rev 16:335–350.

Khoury MJ, Erickson JD, James LM. 1982. Etiologic heterogeneity of neural tube defects: clues from epidemiology. Am J Epidemiol 115: 538–548.

Khoury MJ, James LM, Flanders WD, Erickson JD. 1992a. Interpretation of recurring weak associations obtained from epidemiologic studies of suspected human teratogens. Teratology 46:69–77.

Khoury MJ, Moore CA, Evans JA. 1994b. On the use of the term "syndrome" in clinical genetics and birth defects epidemiology. Am J Med Genet 49:26–28.

Khoury MJ, Moore CA, James LM, Cordero JF. 1992b. The interaction between dysmorphology and epidemiology: methodologic issues of lumping and splitting. Teratology 45:133–138.

Lechat MF, Dolk H. 1993. Registries of congenital anomalies: EUROCAT. Environ Health Perspect 101 Suppl 2:153–157.

Leppig KA, Werler MM, Cann CI, Cook CA, Holmes LB. 1987. Predictive value of minor anomalies. I. Association with major malformations. J Pediatr 110:531–537.

Limb CJ, Holmes LB. 1994. Anencephaly: changes in prenatal detection and birth status, 1972 through 1990. Am J Obstet Gynecol 170:1333–1338.

Morin L, Cendron M, Crombleholme TM, Garmel SH, Klauber GT, D'Alton ME. 1996. Minimal hydronephrosis in the fetus: clinical significance and implications for management. J Urol 155:2047–2049.

Online Mendelian Inheritance in Man (OMIM). 2000. McKusick-Nathans Institute for Genetic Medicine, Johns Hopkins University (Baltimore, MD) and National Center for Biotechnology Information, National Library of Medicine (Bethesda, MD) (http://www.ncbi.nlm.nih.gov/Omim/).

Peters KD, Kochanek KD, Murphy SL. 1998. Deaths: final data for 1996. Natl Vital Stat Rep 47:1–100.

Reefhuis J, de Walle HE, Cornel MC. 1999. Artefactual increasing frequency of omphaloceles in the Northern Netherlands: lessons for systematic analysis of apparent epidemics. Int J Epidemiol 28:258–262.

Roberts HE, Moore CA, Cragan JD, Fernhoff PM, Khoury MJ. 1995. Impact of prenatal diagnosis on the birth prevalence of neural tube defects, Atlanta, 1990–1991. Pediatrics 96:880–883.

Vergani P, Locatelli A, Strobelt N, Cavallone M, Ceruti P, Paterlini G, Ghidini A. 1998. Clinical outcome of mild fetal ventriculomegaly. Am J Obstet Gynecol 178:218–222.

Zwamborn-Hanssen AM, Bijlsma JB, Hennekam EF, Lindhout D, Beemer FA, Bakker E, Kleijer WJ, de France HF, de Die-Smulders CE, Duran M, van Gennip AH, van Mens JT, Pearson PL, Mantel G, Verhage RE, Geraedts JP. 1997. The Dutch Uniform Multicenter Registration system for genetic disorders and malformation syndromes. Am J Med Genet 70:444–447.