A B S T R A C T

To assess the quality and appropriateness of Canadian Congenital Anomalies Surveillance System (CCASS), a system based on routine hospital admission/separation records, we compared the congenital anomalies ascertained by CCASS for the period of January 1, 1990 to December 31, 1993 in the province of Alberta with corresponding figures obtained from Alberta Congenital Anomalies Surveillance System (ACASS), a specific-purpose surveillance program collecting information on congenital anomalies from multiple sources with mechanisms to evaluate diagnosis. Rates of congenital anomalies estimated by CCASS tended to be higher. Agreement between CCASS and ACASS depended on diagnosis: for the International Clearinghouse for Birth Defects Monitoring System standard categories of congenital anomalies (except for anomalies of abdominal wall), agreement usually exceeded 50%; for less clear-cut diagnoses, it was well below 50%. We conclude that routine medical records can be used for surveillance purposes for major congenital anomalies with clear-cut diagnosis.

A B R É G É

Pour évaluer la qualité et l’utilité du Système canadien de surveillance des anomalies congenitales (SCSAC) qui se sert des dossiers d’admission et de sortie des hôpitaux, nous avons fait une comparaison entre les anomalies congenitales constatées par le SCSAC entre le 1er janvier 1990 et le 31 décembre 1993 dans la province de l’Alberta et les statistiques correspondantes fournies par le système albertain de surveillance des anomalies congenitales, un programme de surveillance ciblé qui recueille des données sur les anomalies congenitales provenant de sources multiples avec des mécanismes pour évaluer le diagnostique. Les estimations des taux d’anomalies congenitales selon le SCSAC ont eu tendance à être plus élevées. La concordance entre les données du SCSAC et celles du système albertain dépendaient du diagnostic; pour les catégories standard d’anomalies congénitales (sauf celles de la paroi abdominale) de l’Organisation internationale des systèmes pour la monitorisation des défauts congenitaux, la concordance était en général supérieure à 50%: pour les diagnostics moins évidents, elle était bien inférieure à 50%. Nous en concluons que les dossiers médicaux peuvent servir à la surveillance des grandes anomalies congénitales dans les cas de diagnostique clair et net.

Congenital Anomalies Ascertained by Two Record Systems Run in Parallel in the Canadian Province of Alberta

Shi Wu Wen,1 Jocelyn Rouleau,1 Robert Brian Lowry,2 Brenda Kinakin,2 Stacey Anderson-Redick,2 Barb Sibbald,2 Tanya Turner2

Since the 1960s, congenital anomaly surveillance systems have been set up in many industrialized countries in response to the tragedy that followed the introduction of thalidomide.1,2 There is no ideal model in congenital anomaly surveillance. Because of enormous costs for sustained surveillance in large populations, many congenital anomaly surveillance systems, including the Canadian Congenital Anomaly Surveillance System (CCASS), have been based on routine medical records.3-6 However, because routine data are usually collected for other purposes (e.g., administration), it would be natural for people to be sceptical about their quality and appropriateness for disease surveillance purposes. The existence of two data sets in the Canadian province of Alberta provided us a unique opportunity to assess the quality of CCASS.

The Alberta Congenital Anomalies Surveillance System (ACASS) was established in 1980.3 Prior to that year, congenital anomalies were registered as part of a General Handicapped Children’s Registry which was established in 1966 and disbanded in 1980. This system uses multiple sources of data, including the Alberta Congenital Anomaly Reporting Form (that is forwarded from hospitals and special outpatient clinics to Alberta Vital Statistics); birth, stillbirth and death certificates; and reports from cytogenetics laboratories, genetic clinics, public health units, and the provincial newborn screening program.4 ACASS is able to use personal identifiers, thus duplicate cases are virtually eliminated and additional information can be included. Diagnoses with a query are shown to a medical geneticist, who writes to the responsible physician for more information and makes necessary correction of the diagnosis accordingly.5 Like CCASS, ACASS follows infants up to one year old. Hospitals in Alberta send admission/separation records to CIHI, and the Laboratory Centre for Disease Control receives a copy. The main objective of this study is to assess the quality and appropriateness of CCASS for policy and scientific issues related to congenital anomalies, based on its comparison with ACASS.

METHODS

A total of 21 categories of congenital anomalies were used in the comparison including the International Clearinghouse for Birth Defects Monitoring System’s (ICBDSM) 14 standard categories of congenital anomalies6 and 7 other categories (Table I). We have set in prior to assess certain diagnoses with large discrepancies between the two systems. The 7 “other” categories had large discrepancies and had a sufficient number of cases for reasonable assessment. Total births in the province of Alberta (including both live births and stillbirths) registered by Statistics Canada were used for the rate calculation. A revised melding procedure for CCASS data using date of birth, sex, and four-digit ICD-9 codes to identify re-admission of the same individual was used in this comparison. Since the information on date of birth and sex in CCASS is quite reliable,
and since the number of cases for a specific category of congenital anomalies listed in Table I in the province of Alberta seldom exceeds 100, this melding procedure could reduce the possibility of residual duplication caused by errors in variables used for melding, while limiting the chance of lumping two or more individuals with the same diagnosis together. To increase statistical stability, data were combined for the 4-year period from January 1, 1990 to December 31, 1993.

Rates for the 21 categories of congenital anomalies estimated from CCASS and from ACASS were compared first. Cases ascertained by the two systems can differ in several ways: a case can be misdiagnosed by CCASS (or missed by ACASS) or vice versa, or misdiagnosed by both systems (or missed by both systems). To further assess the actual source of misdiagnosis (or missing), an individual matching between ACASS and CCASS, based on date of birth, sex, and ICD-9 code, was performed. In a typical assessment of diagnostic accuracy, a “gold standard” diagnosis is used to classify disease status of the participants, with four possible results for the “new” diagnosis: true positive, false positive, true negative, and false negative. However, for the current study, because the number of “true negatives” is too large, and there are possibilities of under-reporting in any surveillance system, the assessment of “true negative” status becomes an insurmountable obstacle. Although it is possible to estimate the “true negative” cases by using the capture-recapture method, the estimation is likely to be invalid, as the fundamental assumption required for this estimation, the independence of the two systems, is unlikely to be held. As a result, agreement for ascertained cases between ACASS and CCASS has been used instead as a measure of the quality of CCASS. Only three results for the individual matching between ACASS and CCASS are possible: a case is recorded by both systems, a case is recorded by ACASS but missed by CCASS, and a case is recorded by CCASS but missed by ACASS. The agreement is thus calculated as the percentage of cases recorded by both systems of the total cases recorded by either of the systems.

We performed supplement analysis using original melding procedure by CCASS, and compared the results with the revised melding procedure.

RESULTS

During the four years being examined and compared, a total of 169,000 births (including 889 stillbirths and 168,111 live births) in the Canadian province of Alberta were registered by Statistics Canada. Except for hypoplastic left heart syndrome, common ventricle and endocardial cushion defects, birth prevalences of congenital anomalies estimated by CCASS were higher than rates estimated from ACASS (Table II).

Table III shows the agreements between CCASS and ACASS. It can be seen from
this table that the agreements tended to be higher for those anomalies which can be diagnosed earlier without much difficulty (e.g., anencephaly and spina bifida) but tended to be lower for more difficult diagnoses (e.g., anomalies of abdominal wall). ACASS recorded more cases of common ventricle and endocardial cushion defects than CCASS; but CCASS recorded more cases for other categories of defects (Table III).

Results obtained by using the original CCASS melding procedure were almost identical to the results obtained by using the revised melding procedure (data available upon request).

**DISCUSSION**

Rates of congenital anomalies estimated by the CCASS tended to be higher than those estimated by the ACASS. This observation is consistent with findings by a previous report which showed a comparable rate for ACASS while a slightly increased rate for CCASS, as compared to other jurisdictions. As we will discuss below, because of potential problems in the diagnosis and coding and the lack of validation for anomalies, rates for anomalies estimated from routine medical records may be artificially inflated, especially for those anomalies with no clear-cut diagnosis, and caution should be applied in their interpretations.

The agreement between CCASS and ACASS depends on diagnosis: for the ICBDMS standard categories of congenital anomalies (except for anomalies of abdominal wall), it usually exceeds 50%; for less clear-cut diagnoses, it is well below 50% (Table III). We realize that for the current study, there is no “gold standard” because both systems can miss or misdiagnose some cases, although the diagnosis in ACASS may be more reliable. However, if we can assume that a high degree of agreement between data collected from different systems suggests a high degree of validity, results obtained from this comparison can help us to select the categories of congenital anomaly for surveillance by routine medical records.

The degree of agreement in our study, while not very satisfactory, compared favourably with routine data sets assessed by other congenital anomalies surveillance systems. There are two possible explanations for the superiority of CCASS over other routine data sets. First, although CCASS is not specifically designed for disease surveillance purposes, it contains a large volume of clinical information (up to 16 diagnoses), a feature which could be helpful for the surveillance system to pick up the needed information but which is unlikely to be shared by other routine data such as birth and death certificates. Second, previous studies comparing routine data with specific-purpose registration data were conducted some years ago. Prenatal diagnosis has progressed rapidly during the last decade, and therefore more recent routine medical records may be able to yield more reliable information. The improvement in diagnosis over time may be an encouraging sign for more comfortable use of routine data for surveillance in the future.

Although generally superior to other routine data sets, the differences between CCASS and ACASS remain substantial and pose some concerns over its uses. Understanding the reasons for the differences would be helpful to resolve some of the concerns and make more rational use of the data in the future. ACASS could be expected to underestimate congenital anomaly rates. Because ACASS is a voluntary reporting system, if the record of the case fails to be sent out for various reasons, “true” case may be missed. Failure to send reports did happen in other similar register systems, and it could happen in ACASS as well. If ACASS missed some cases, the “true” agreement between CCASS and ACASS for congenital anomalies with an obvious diagnosis may be even higher. CCASS rates could underestimate the true rates as well. For example, cases diagnosed and seen solely in outpatient settings could be recorded in ACASS but not appear in CCASS. Artificial inflation by misdiagnosis, miscoding, and residual duplication arising from change in diagnosis over time is the main concern for CCASS. Because there is no mechanism to validate the diagnosis in CCASS, if the original diagnosis by the responsible physician is wrong, it is not possible for CCASS to make a correction. For example, one author of the current study from the ACASS team (Lowry) recently received a Death Registration Notice that coded for tracheoesophageal fistula. An autopsy was done and the
author examined it. The autopsy revealed no tracheoesophageal fistula but a blockage due to a large blood clot that had occurred in the delivery process. A wrong diagnostic code could happen even if the original diagnosis by the responsible physician is correct, because the ICD system used by ACASS may not be discriminating enough for complicated cases, which could pose difficulties for the coding clerk. The ACASS team often received data on congenital anomalies both in ICD-9 codes and word descriptions, and found many codes by hospital clerks are wrong (Lowry). ACASS also has the capacity to reject diagnoses by combining other information. We have performed a chart review on some of the discrepant records, and found that the above-mentioned reasons explain a large part of the discrepancies (data available upon request).

Despite the above discussed problems, there are still merits to maintaining a routine medical record-based system to monitor the occurrence of major congenital anomalies at the national or regional level. Such a system is less labour intensive and has large population coverage. Errors in diagnosis and coding occur more frequently in this system than in a labour-intensive specific-purpose surveillance system, however if these errors occur randomly (which we assume is the case unless there is legitimate reason to doubt it), rates obtained from such a system may assist in general monitoring of temporal trends and geographic variations, strategic planning, or program evaluation for congenital anomalies with known etiology. Because of potential problems in the diagnosis and coding and the lack of validation for anomalies with no clear-cut diagnosis, rates for these anomalies estimated from routine medical records may be artificially inflated and caution should be applied in their interpretations. Because other routine medical records based on congenital anomalies may share features with ACASS, findings from this study may be applicable to other systems as well.

REFERENCES