Congenital Anomaly Surveillance in Canada
Surveillance des anomalies congénitales au Canada
National Workshop May 11-12 2000 Atelier national les 11 et 12 mai 2000

Report of the National Workshop

Chateau Cartier Hotel
Aylmer, Quebec

Canadian Perinatal Surveillance System

Health Canada Santé Canada

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Executive Summary

On May 11-12, 2000, Health Canada hosted a national workshop on congenital anomaly surveillance in Canada. The workshop brought together international, national and provincial/territorial representatives from the fields of public health, genetics, paediatrics, obstetrics and radiology.

The workshop provided an opportunity to review current surveillance activities at the national and provincial/territorial levels. Three presentations from an international perspective provided valuable lessons for future surveillance initiatives. Small group brainstorming sessions on responding to clusters of congenital anomalies, prenatal diagnosis and neural tube defect surveillance addressed important issues in these key components of congenital anomaly surveillance. Finally, there was an opportunity to identify the main objectives of an ideal set of congenital anomaly surveillance activities in Canada.

In concluding the workshop, Health Canada proposed three areas of work through which the federal government may contribute to meeting the identified objectives. These included: working to enhance the current national surveillance system—the Canadian Congenital Anomaly Surveillance System (CCASS), supporting a network of provincial/territorial representatives and other interested stakeholders in the field of congenital anomaly surveillance, and developing a formal process by which technical support can be provided to provincial/territorial and local jurisdictions in the response to clusters of congenital anomalies.

Report of the National Workshop
Chateau Cartier Hotel, Aylmer, Quebec

1. Introduction

Background

On May 11 and 12, 2000, a national workshop on congenital anomaly surveillance in Canada was hosted by the Bureau of Reproductive and Child Health, Health Canada. Approximately thirty-five participants met in Aylmer, Quebec to share information about their experiences with surveillance of congenital anomalies and to determine how best to enhance surveillance activities across Canada. Participants were invited from every province and territory and included representatives from provincial, territorial and regional public health authorities, universities, professional organizations, institutes, hospitals, Health Canada, and surveillance systems in other countries. (See Appendix A for the list of participants.)

Objectives

The objectives of the workshop were:

- to develop strategies to enhance congenital anomaly surveillance activities in Canada
- to provide a forum to discuss current provincial and territorial initiatives in congenital anomaly surveillance
- to develop strategies to address particular issues in congenital anomaly surveillance, namely cluster management, prenatal diagnoses and neural tube defect surveillance.

Agenda

The workshop process included presentations with question and answer sessions, large group brainstorming and discussion, and facilitated small group discussions. (See Appendix B for the workshop agenda.)

Welcoming Remarks

Paul Gully, Deputy-Director General of the Laboratory Centre for Disease Control (LCDC), Health Canada

Paul Gully welcomed the participants to the workshop. He noted that the ongoing re-alignment at Health Canada, which will combine the Health Protection Branch (HPB) with the Health Promotion and Programs Branch (HPPB), will result in a shift in the business of LCDC. The focus will be on surveillance, targeted research, and dissemination of information to enhance public health action for disease prevention and control, and for health promotion.

Surveillance of disease, especially congenital anomalies, presents many challenges including the quality and consistency of data sources, diagnosis of anomalies, and determining the anomalies on which to focus. The current issues surrounding neural tube defects (NTDs) and folic acid supplementation and food fortification make it critical to have a system of surveillance for NTDs.

The recent report of the Auditor General's Office on surveillance has been critically helpful. LCDC needs to evaluate its surveillance systems, set up new ones, and continue to evaluate them to ensure that data are valuable, reliable, and timely and provide the information which is needed for action. This meeting is an important part of the process of evaluation of the Canadian Congenital Anomalies Surveillance System (CCASS).

Paul thanked I.D. Rusen and Catherine McCourt for organizing and hosting the workshop. He also thanked the participants for contributing to the process.

Catherine McCourt, Director of the Bureau of Reproductive and Child Health

Catherine McCourt welcomed the participants and thanked them for joining in the workshop. She explained that federal government surveillance of congenital anomalies happens within the context of the Canadian Perinatal Surveillance System (CPSS). The Bureau of Reproductive and Child Health manages the CPSS program to monitor trends and patterns in the health of pregnant women and infants in Canada. (Appendix C provides an organizational chart of the Bureau of Reproductive and Child Health.)

The areas of maternal health, fetal and infant health, abortion and congenital anomalies, and maternity experiences are each supported, guided and promoted by a Study Group. Resources for perinatal surveillance, including the surveillance of congenital anomalies, were recently strengthened in the 1999 federal budget.

Catherine noted that this workshop presents an opportunity for the participants to guide the appropriate expenditure for congenital anomaly surveillance at the national level.

1. Congenital Anomaly Surveillance in Canada

Introduction

I.D. Rusen, from the Bureau of Reproductive and Child Health, presented the history and current activities in congenital anomaly surveillance at Health Canada. Then participants from the provinces and territories described their systems for surveillance of congenital anomalies based on the following three questions:

- What are the current activities in terms of surveillance and reporting of congenital anomalies in your region?
- Are there any new initiatives being developed or considered?
- What is the capacity of the province/territory to collect information on fetal anomalies that are diagnosed prenatally?

Nova Scotia, Alberta and British Columbia, which have the most comprehensive systems among the provinces and territories, provided separate presentations on their surveillance activities.

Congenital anomalies surveillance varies across the country in terms of the rationale for its creation, the anomalies which are registered and monitored, data sources, timeliness and nature of reporting, their clients and the services offered to them, partners, and sources and types of support. The following summarizes the information presented about surveillance by each jurisdiction. (Representatives from Yukon and Nunavut were unable to attend the workshop.)

N.B. Throughout this report, the terms used reflect usage by the speaker. As noted by Moore and Persaud,[1] "Congenital anomalies, birth defects and congenital malformations are terms currently used to describe developmental disorders present at birth."

National Congenital Anomalies Surveillance Activities at Health Canada

I.D. Rusen reported on the three primary congenital anomaly surveillance activities at Health Canada:

- the Canadian Congenital Anomaly Surveillance System (CCASS)
- research related to congenital anomalies, and
- technical support related to congenital anomalies.

The Canadian Congenital Anomaly Surveillance System (CCASS)

CCASS is the only national population-based information source on anomalies in Canada. CCASS was founded in 1966 and was a founding member of the International Clearinghouse for Birth Defects (ICBD). The system is based on hospital records and ICD codes, with 57 specific categories and 15 summary categories for routine analyses. CCASS captures stillbirths and live births up to one year of age. Details of the categories and data on time trend analysis 1974-96 were provided for participants in the background materials.

CCASS is capable of calculating birth prevalence of specific anomalies per 10,000 total births at the national, provincial, and for some provinces, census and census subdivision levels. It produces trends over time although the years available vary by site. The system can also produce statistical comparisons of prevalence between jurisdictions.

I.D. noted the limitations of CCASS as follows:

- The data are for birth prevalence only and do not include anomalies which are prenatally diagnosed and terminated.
- No maternal information is reported.
- Timeliness of reporting is not good (3-year delay).
- The system relies on ICD codes and hospitalization information. Codes are not checked, diagnoses are not confirmed, and data on out-patients are not included.
- Statistical limitations exist.

Planned enhancements for CCASS include:

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[1] Moore, Keith L. and Persaud, T.V.N. *Before We Are Born: Essentials of Embryology and Birth Defects*, 5th Edition. Philadelphia: W.B. Saunders Company, 1998.

- Increased dissemination of information is planned especially to public health units, academic institutions and provincial health authorities. A perinatal health report covering a broad range of topics on maternal and child health will be released in July 2000 and a report on congenital anomalies in Canada is planned for April 2001.
- CCASS will explore ways to include data on induced abortions through data from the Canadian Institute for Health Information (CIHI) and/or projections from provinces and territories with data on fetal anomalies.
- Any recommendations from this workshop for making CCASS more useful will be considered.

Participants asked about the contributions of CCASS over the years. The work has been primarily looking for patterns of occurrences and not hypothesis testing. The system has been able to correct inaccurate media information about clusters of cases. As with any other congenital anomaly surveillance system, no major discoveries have been made by CCASS.

Research Efforts Related to Congenital Anomalies

Health Canada undertakes research related to congenital anomalies using various databases including:

- comparisons of data from CCASS and the Alberta Congenital Anomaly Surveillance System (ACASS)
- analyses of vital statistics data to examine patterns of infant mortality caused by major congenital anomalies
- participation in ICBD studies, e.g., the international study of policies related to folic acid and NTDs
- carrying out special research projects, e.g., impact of food fortification with folic acid.

Technical Support Related to Congenital Anomalies

The types of technical support available from Health Canada related to congenital anomalies include:

- responses to requests from provinces/territories and local levels in terms of epidemiological expertise and data provision (e.g., response to nickel concerns in a local community; updated literature; analysis of data; participation on panel on environment and human health)
- responses to public and media requests for information.

Newfoundland

Sharon Alexander reported on the Newfoundland Provincial Perinatal Program. Newfoundland registers approximately 5008 live births a year. The Perinatal Program has undertaken a 16-week pilot at the tertiary centre which is responsible for approximately half of the provincial births. The pilot will be expanded to the rest of the province at a later date. The pilot revolves around a perinatal database of 250 variables, modeled on the minimum database of the CPSS. The data comes from hospitals' standardized prenatal records, labour and delivery records, and the live birth form. Currently the software system for the data collection is undergoing changes. The Newfoundland Genetics Program collects information on NTDs but it is not collected

electronically, only on paper. The potential for monitoring prenatal diagnoses exists because all amniocentesis procedures for the province are conducted in St. John's.

The province recently established the Newfoundland Centre for Health Information. The Centre plans to establish a unique identifier for everyone in the province. Presently an individual's health number changes if they move away and back into the province.

Prince Edward Island

Janet Bryanton of the PEI Department of Health reported on surveillance in the province. PEI registers approximately 1500 live births a year. They have a provincial perinatal database dating back to 1990. Data on congenital anomalies are captured along with other perinatal data from codes on hospital discharge records using the ICD-9 CM coding system. One coder goes to every hospital in the province to collect the data. An annual report is produced but numbers are too small to do any regional breakdowns related to congenital anomalies. No terminations are done in PEI. Most women wanting terminations go to Nova Scotia for the procedure and data on those terminations are included in the Nova Scotia database.

At one time, work on a congenital anomalies database was operational and funded through Health Canada but was discontinued when the funding ended as it could not be sustained through the province.

Nova Scotia

Rebecca Attenborough, Coordinator of the Reproductive Care Program of Nova Scotia at the IWK Grace Health Centre, reported on surveillance of congenital anomalies in Nova Scotia. The province registers approximately 10,000 births (live births and stillbirths) a year. Two separate databases exist in the province, one with the Reproductive Care Program (RCP) and the other within the Division of Maternal-Fetal Medicine in the Department of Obstetrics and Gynaecology at the IWK Grace Health Centre. Both surveillance systems were originally created for clinical support rather than surveillance. The two systems work together to provide clinicians and others with the information they want. A group of clinicians is now reviewing what are considered major and minor anomalies. A handout with data management principles was provided to workshop participants on the resource table.

Surveillance by the Reproductive Care Program

Surveillance by the RCP began as a hospital-based effort at the Grace Maternity Hospital in 1980 and expanded to the whole province in 1988. The system captures data from all hospitals in Nova Scotia and two New Brunswick hospitals near the Nova Scotia- New Brunswick border where Nova Scotia women may receive health care. Capture of data depends on care providers across the province, personnel in health records departments, and staff at the RCP. In return, they receive reports on their own analysed data. The principle of the system is to protect the confidentiality of the woman, the infant, the care provider and the health facility.

The system uses the Atlee Coding System which is unique to Nova Scotia and was developed by clinicians with a focus on clinical care. Timing of data submissions is variable depending on the source. The plan is to link to ICD-10-CA when it becomes available for the Nova Scotia provincial database in order to build on the legislated database.

The database contains data on all pregnancies of 20 weeks or more. It includes demographic information, major and minor anomalies (10 major anomaly categories, and more than 200 major anomalies), still births, terminations, and infant death information up to one year of age.

Fetal Anomaly Surveillance

The Fetal Anomaly Database was established in 1992 through the Division of Maternal-Fetal Medicine in the Department of Obstetrics and Gynaecology at the IWK Grace Health Centre. The database was established out of a growing need to capture all prenatally diagnosed fetal anomalies whether they are ongoing pregnancies or terminated pregnancies. At a weekly meeting, all new prenatally diagnosed anomalies are reviewed, as well as some anomalies diagnosed in the neonatal period. The database also includes pregnancies referred to the Centre from Prince Edward Island or New Brunswick. The system captures all pregnancies regardless of the outcome or gestational age. Data include demographic information, risk factors (e.g., age, ethnicity, consanguinity, alcohol or substance abuse), reason for referral (e.g., ultrasound, maternal age, exposure, amniocentesis, abnormal blood serum), antenatal diagnosis, infant birth information, and postnatal diagnosis of pathology.

Related Activities

The two Nova Scotia databases are used for regular surveillance and for major epidemiological studies. Data linkages are conducted with the two databases and others. For example, the RCP has an annual report containing some information on congenital anomalies by region. Also, the Maternal Serum Testing (MST) program follows all women who receive testing in the province.

New Brunswick

Chris Heisner of the New Brunswick Department of Health and Wellness reported on the status of surveillance in the province. The provincial government recently underwent significant cuts in funding and has no congenital anomaly surveillance system. In the past, New Brunswick participated in CCASS by submitting hard copies of physician's notice of birth, etc. but stopped when electronic submissions were required.

Therapeutic abortions in New Brunswick are split about 50-50 between hospitals and private clinics. Hospitals could provide information on whether the termination was for an anomaly, whereas private clinics do not provide access to any data.

Quebec

Philippe De Wals, from the University of Sherbrooke, spoke about surveillance of congenital anomalies in Quebec. The province registers approximately 80,000 live births annually. Surveillance of birth defects is not a high priority for public health authorities, and the province has no plan for surveillance of congenital anomalies. Med-Echo, which collects hospitalization data, was evaluated for its potential for surveillance. Improvements were recommended but have not been implemented yet. The new Institute for Public Health, with which he is associated, has plans for a perinatal surveillance unit.

Terminations for fetal malformations are usually performed as day surgery in a hospital and are captured by Med-Echo. Consequently, it is possible to identify cases but the ICD code does not permit much detail, e.g., all central nervous system anomalies are grouped under one category

requiring the abstractor to go back to the record for the diagnosis. Also, microbiology laboratory data allow capture of information on congenital infections, including rubella, parvovirus and varicella. In Quebec, the law allows researchers to access medical records without authorization of individuals.

In his work at the University, Philippe uses the Med-Echo system to look at NTDs. He has plans to look at the impact of food fortification on NTDs in Quebec by studying 1990-99 trends in rates of NTDs. He suspects that trends will be seasonal and related to the increasing use of multivitamins and improvement in nutrition. He is collaborating with the food industry to look at the timing and dosage of folic acid fortification. If fortification is having an impact, then the transition period when both fortified and non-fortified products were available to the public should be reflected in a step decrease in the rate of NTDs. Philippe indicated that he would be pleased to collaborate with others especially in the area of NTDs. Elizabeth Rael expressed interest in collaborating on the NTD studies to get a larger sample.

Ontario

Elizabeth Rael, Senior Epidemiologist with the Population Health Service, Public Health Branch of the Ontario Ministry of Health and Long-Term Care, reported on the status of surveillance of congenital anomalies in her province. Within Public Health in Ontario, the greatest awareness about the importance of surveillance has been in the area of infectious conditions.

[Addendum since the meeting: The Public Health Information Strategy Advisory Committee (PHISAC) was recently established "to ensure that a vision and strategy be developed… for the development and deployment of information, information technology and information systems." (PHISAC Newsletter May 2000,1;(1):1). The Population Health Service affirms the importance of data in support of all our Mandatory Health Programs and Services.]

Ontario and its bureaucracy are large and complex, so a key challenge is to learn more about existing initiatives and the data or surveillance systems pertinent to Public Health. Then the challenges are to ensure the information is available and accessible for Public Health, and that systems are developed in an integrated way. Within the Public Health Branch, access to data regarding abortions and the Maternal Serum Screening Database has not yet been fully resolved.

The Ontario program Healthy Babies, Healthy Children (HBHC) instituted a universal postpartum contact for all new mothers in October 1999. Public health units are mandated to contact all new mothers with a telephone call within 48 hours of discharge from hospital and the offer of a home visit. The local Health unit data is reported to the province on a quarterly basis through the Integrated Services for Children Information System (ISCIS).

The Public Health Branch is expecting two placements from the federal field epidemiologist program, one of which will be within the Population Health Service. It is hoped that the individual will be able to explore the issues and/or develop plans for surveillance.

Elizabeth is also seeking to develop relationships with staff in other Branches to gain a better understanding of what initiatives exist. One example is the Women's Health Council, which has provided support for developmental work by Anne Pastuszak. The Public Health Branch will be interested to receive more detailed developments of her proposed work.

Anne Pastuszak reported that the proposal would integrate the many Ontario players, genetics labs, the 106 birth hospitals, pediatricians, ultrasound labs, etc., to create a system operating in

realtime to follow each case of a fetal anomaly through the health care system. Data analysis would be centralized but the results would be fed back to all data sources. The focus of the system would be service to families. When a prenatal diagnosis is made, parents are looking for services, which may exist but are not linked.

Manitoba

Lawrence Elliott, from the Epidemiology Unit of Manitoba Health, reported that Manitoba's congenital anomalies registry has been dormant since the mid 1990s. Data from the registry were used to identify genetic syndromes and to ensure genetic counseling follow-up; however, collection of data has been reduced somewhat by provincial government cuts.

The Epidemiology Unit would like to resume congenital anomaly surveillance by adding congenital anomalies to the perinatal surveillance system which exists in Manitoba.

For the last two years, the province has linked records for infant births and deaths up to the first year of life to mothers' records. The data include in-hospital terminations and stillbirths. The 1999 perinatal surveillance report did not include congenital anomalies but the plan is to add them for both pre- and post-natally diagnosed anomalies and link to the clinical geneticists' database. The challenge is inadequate resources. The perinatal research team submitted a proposal to the March of Dimes in the U.S. to get staff to conduct research.

Saskatchewan

James Irvine, a Medical Health Officer for several of the northern health districts in Saskatchewan, reported on the situation in Saskatchewan. Passive surveillance of congenital anomalies was initiated in the 1960s but this system was not supported in an organized way over the last two decades. A PKU registry is maintained but there is no other formal congenital anomalies surveillance system. The Population Health Branch of Saskatchewan Health is presently studying trends in infant mortality in the province, by health district area and by First Nations status. This study will also include causes of infant mortality including the proportion of deaths from congenital anomalies. Results of the study will be available for a provincial conference focusing on infant mortality planned for the fall in Saskatoon.

The feasbility of studying congenital anomalies utilizing the Saskatchewan hospitalization and physician administrative data is presently being assessed. Potential exists to assess trends over time as well as to describe the rates across health regions and for First Nations. This assessment of the extent of the issue, as well as the information arising from the infant mortality study and the fall conference including birth defects, will stimulate more interest in a longer term surveillance process for congenital anomalies in Saskatchewan.

In response to participant questions: 1) The new *Public Health Act* in Saskatchewan includes the capacity to add non-communicable conditions to the list of reportable diseases; and 2) in Saskatchewan, capacity exists to capture some information on induced abortions through hospitalization data utilizing ICD-9 and procedural codes (day-surgery procedures have been captured more recently). However, it is uncertain whether there is complete ascertainment of the diagnostic code for inducement done because of a congenital anomaly.

Alberta

Brian Lowry, from the Department of Medical Genetics at the Alberta Children's Hospital, reported on ACASS. ACASS has been in place since 1980 and was based on what then remained of the province's Handicapped Children's Registry. Ascertainment of birth defects includes infants up to one year of age and stillbirths with a gestational age of 20 weeks or more. ACASS uses a passive system of ascertainment but does try to verify diagnoses. The primary source of data is Vital Statistics, hospital reporting and special communications with genetics clinics, specialty paediatric clinics and laboratories. Infants are given a unique birth number to avoid duplicate reporting from different sources.

Vital Statistics has no requirements for registering anomalies for fetuses less than 20 weeks. Data on fetuses is mainly from hospital terminations plus natural terminations if they come to autopsy. Only three hospitals do terminations making it simple for a clerk to go to the hospitals to get termination data and to the pathology log for the results of autopsies. The system does not accept diagnosis by ultrasound only. It requires a physical diagnosis.

The database originally was a Paradox system but recently changed to an Access database which uses ICD-9, ICD-10, and McKusick MIM codes. ACASS has an Advisory Committee consisting of a geneticist, paediatric cardiologist, neonatal paediatrician, and paediatric pathologist. Recent policy changes have resulted in the coding of all anomalies including conditions that might need repair.

Brian indicated that he will send the ACASS Annual Report to anyone wishing a copy. The report includes trend graphs. In Alberta, the only anomaly with a decreasing rate is anencephaly. Spina bifida and cleft lip are remaining level, and cleft palate is up somewhat. Down syndrome is up slightly but not significantly, possibly the result of the increase in births to later age mothers and the lower rate of terminations in Alberta because of the culture. These reports compare to other members of ICBD (Alberta is a separate member).

ACASS follows up on apparent clusters, e.g. 8 cases of hydrocephalus in 8 months which on examination were not the same condition and were within baseline rates; an apparent cluster of congenital heart disease was the result of the increased use of echoes on newborns—these very early diagnoses often disappear at 4 weeks after birth.

Brian reported on the reasons for CCASS and ACASS differences: ACASS conducts a rigorous review of items to be coded, does not accept query or possible diagnoses, has the ability to follow up all cases for clarification and does not code all components of all syndromes.

Plans to improve ACASS include:

< personal health number (PHN) to be assigned by hospitals at the time of birth

< improved linkage possibilities due to a reinstatement of government support in 1996; may permit linkages to other databases on maternal history, obstetric history.

British Columbia

Soo-Hong Uh, Manager of Information and Resource Management with the British Columbia Vital Statistics Agency, provided an update on the BC Health Status Registry. The HSR evolved from the crippled children's registry which was established 1952. It took its current form in

1992. HSR has the authority to register all individuals with congenital anomalies. The objective and mandate of the HSR are included in his handout which was provided on the resource table.

Soo-Hong provided information on the technical aspects of the HSR system. He noted the sources of data for the HSR as follows:

< notice of birth (NOB) from physicians and midwives

< hospitals

< medical genetics clinics

< health regions (20) for after-birth diagnoses

< others.

The different types of reports from these various sources are revised into a common data structure. The data are reviewed to determine whether the cases are registrable. The data are then loaded into the HSR system and assigned a medical code. Cases are reviewed for potential consulting services.

The system is capable of identifying rates of anomalies in the health regions. Information on selected conditions is mailed out to each region every three months so they can monitor trends.

For the years 1980-1998, anomalies as a whole have shown a slight downward trend and a recent upward trend. Rates of anencephaly, cleft palate, cleft lip, clubfoot, and limb reductions are all down. Down syndrome is going up significantly.

The Health Act does not include provision for collection of data on pregnancies that are terminated for fetal anomalies; however, data comes informally from two cytogenetics laboratories. In 150 reports, 36 were for terminated pregnancies, of which 21 were for Down syndrome and 10 for trisomy 18. Most cases were for mothers 40-44 years of age.

Plans for enhancing the HSR include:

< expanding reporting sources

< improving data quality

< participating in research projects

< releasing reports on a website

< receiving cases of medically terminated pregnancies due to congenital anomalies

< upgrading the system to Oracle.

Northwest Territories

Penny Sutcliffe, a Regional Medical Health Officer, reported that there is as yet no surveillance of congenital anomalies in the NWT. There is interest in maternal blood serum screening but the widely dispersed population makes it very difficult. A disease registry system exists that could be used as a basis for surveillance. Dr. Sutcliffe reported that in neighbouring Yukon, legislation will be passed soon making the reporting of FAS mandatory.

Other Related Canadian Activities

Judy Lee, of the Canadian Institute for Health Information (CIHI), reported on the status of abortion databases. CIHI has a national therapeutic abortion database which originated in 1970 when Statistics Canada was mandated to collect abortion information. When the abortion law was struck down in 1988, the collection of data was no longer mandated and some jurisdictions stopped submitting data. The data now collected by CIHI are from hospitals, clinics, and Canadian terminations in 15 U.S. clinics. CIHI does the data collection and Statistics Canada reports on and releases the data. There are approximately 100,000 abortion records annually. The most current data is for 1998 for some jurisdictions.

Most hospitals across Canada submit data electronically from their Discharge Abstracts Database (DAD) of clinical information. Some data are still not on DAD. However, with the revised DAD, collection of data on abortion is mandatory and gestational period is also captured. The revised DAD excludes marital status but includes age. Software applications for submissions from abortion clinics are being developed. Currently CIHI receives primarily counts with no other supporting information.

Discussion on the revised DAD included the following key points:

- In April, 2001, CIHI will be able to accept ICD-10-CA codes but not all jurisdictions will be on ICD-10 by then. Most therapeutic abortions for congenital anomalies occur in hospitals and most hospitals complete the DAD. If information on the reason for the abortion is on the chart, coders will be allowed to collect it. However, hospitals do not have to provide the specific reason for the abortion.
- The CA in ICD-10-CA stands for “Canadian”. The codes have been enhanced to expand the possible diagnoses. The Canadianization of the codes has been completed.
- The ICD-10 codes do not include all the necessary specifications for fetal anomaly diagnoses. Some improvements have been made, particularly around NTD anomalies in terminated pregnancies.
- Varying definitions of stillbirth and abortion are still an issue for coders around the 20-22 gestational week period.

3. Lessons from International Experience

Introduction

Three guests provided their perspectives on congenital anomaly surveillance based on years of diverse experience in the field. Elisabeth Robert is Vice Chair of the ICBD and presented information about the France Central-East Registry of Congenital Malformations in Lyon, France. Larry Edmonds of the Centers for Disease Control in Atlanta, Georgia reported on his work with the National Birth Defect Prevention Network and the Centers for Birth Defects Research and Prevention in the U.S. Charlotte Hobbs reported on her work as Director of one of these U.S. Centers located in Little Rock, Arkansas.

Brief summaries of the presentations are provided below. The three presenters also contributed to discussions throughout the workshop sessions.

Elisabeth Robert, Lyon , France

The France Central-East (FCE) Registry of Congenital Malformations was created in Lyon in 1976 primarily for surveillance to avoid another thalidomide-type crisis. It was the first registry of morbidity in France and was initiated by a motivated clinician as a regional project. By 1990, the registry had expanded to include more than 100,000 annual births from 16 hospital departments in the region.

Financial support for the registry came originally from non-profit associations involved in screening programs, e.g., PKU. When this funding ceased, the project was supported with private funding from an insurance company. Recently, public funding has increased.

The 1981 identification of a cluster of malformations by a clinical observation related to use of the anticonvulsant, valproic acid, was confirmed by the registry and eventually led to recognition by the national health authorities and a health warning in 1982. However, it was not until 1997 that financial support was received from the National Committee of Registries which had been formed in 1986.

The FCE registry was a founding member of the International Clearinghouse for Birth Defects Monitoring Systems (ICBDMS) which has 30 member countries on 5 continents. Membership in ICBDMS has been important to FCE because they have learned from the experience of others, including methods for data analysis, and have collaborated in a number of joint studies.

The purpose of malformation surveillance is to detect a sudden epidemic of a special type of malformation or a long-term, gradual increase or decrease in malformations. Rarely are registries responsible for identifying a teratogen. However, if a clinical observation suggests a problem, a registry can help to confirm a problem and thus reduce the time between the occurrence and recognition of the effect and action by the medical community. In the case of valproic acid, confirmation of a problem was due to a combination of factors including the existence of the registry, the special interest in spina bifida at the time, the use of a questionnaire for mothers with a routine question about epilepsy, and the fact that the registry was a member of ICBD.

FCE receives reports from various systems, each with its strengths and weakness. For example, birth certificates give good ascertainment but poor descriptive quality whereas special notification forms (e.g., fetal pathology reports) provide a higher descriptive quality but do not give complete coverage of all cases of malformations. When weighing the benefits of various methods for malformation surveillance, it is important to consider the level of precision in describing and diagnosing anomalies as well as the completeness of coverage.

In determining what malformations to monitor, some people question the usefulness of data on minor malformations because of the variability in their reporting. However, findings of minor malformations may be more significant than a cluster of a major malformation. For example, identifying malformations as resulting from pesticides needs these minor diagnoses. In addition, most known teratogens produce multiple malformation complexes so specific associations of common malformations should be monitored. A study showed that larger registries tend to register fewer multiple malformations than the smaller registries.

The coding system selected for reporting malformations should be precise enough to permit the description of unusual cases, but even then, no code can replace an authorized look at the actual case description.

In terms of trends in specific malformations, the FCE registry has found the following:

< Gastroschisis has increased significantly since the late 1980s.

< Obstructive gastrointestinal malformations have shown a sharp increase.

< Anencephaly is down.

< The number of Downs cases at birth has stayed consistent in spite of increased abortions for Down syndrome, probably due to increasing rates of births to women over 35 years of age.

Studies conducted in collaboration with ICBD include:

< a study of genetic anomalies related to distance from a nuclear plant

< molecular epidemiology studies looking at the molecular level of risk factor relationships, e.g., relationship between facial clefts and nutrition and genetic susceptibility to teratogens

< risk associated with a hazardous landfill site

< congenital defects in twins

< trends in NTDs.

Larry Edmonds, Atlanta, Georgia, U.S.A.

In the U.S., the public health importance of birth defects is exemplified by the following:

< 120,000 to 160,000 children are born with major birth defects each year.

< Birth defects are responsible for 30% of admissions to pediatric hospitals.

< The 17 most significant birth defects cost the U.S. $8 billion annually.

< Birth defects are a leading cause of infant mortality.

< Some cases are entirely preventable.

The number of state birth defect surveillance programs has increased from 3 in 1974 to 40 in 1999. The greatest number of these programs operate in maternal and child health units in hospitals, followed by epidemioliogical and environmental health departments, universities, and vital statistics registries. The Birth Defects Prevention Act of 1998 changed the role of CDC somewhat by giving it a mandate to collect data, operate regional centres for epidemiological research, and provide information and education to the public on prevention of birth defects.

The Birth Defects and Pediatric Genetics Branch at CDC conducts research and offers technical assistance to states and local regions in the areas of birth defects surveillance, research, and prevention. The Branch also drafts legislation, promotes relationships with advocacy groups, and awards grants and cooperative agreements. Most recently, the purpose of birth defects surveillance in the U.S. has shifted to evaluating the need for, and guiding and assessing the progress of, intervention and prevention, and education and advocacy.

The characteristics of a good surveillance system include:

< comprehensive review of multiple data sources

- accurate and precise diagnostic criteria
- appropriate and meaningful classification schemes
- large database
- meaningful and timely analysis
- timely dissemination of data
- personal identifiers for follow-up and data linkage
- confidentiality of patient records.

In the various state surveillance systems, case ascertainment methods differ and, consequently, rates of major birth defects determined from different data sources vary significantly. CDC is working with the jurisdictions to agree on standard case ascertainment methods. The active hospital surveillance system now has a very strict definition of a major defect. However, a mandatory reporting law has not been very successful in improving physician reporting but does give a mandate to surveillance programs to follow up with physicians to collect missing information. Vital statistics registries in the U.S. are reducing the number of defect codes. (Canadian vital statistics registries have none.)

Currently the challenges for surveillance include:

- consistent diagnosis
- timeliness of data capture, analysis and reporting (especially now for NTDs)
- prenatal diagnoses and elective terminations.

CDC has awarded cooperative agreements to seven states to operate Centers for Birth Defect Research and Prevention. The purpose of the agreements is to bolster ongoing surveillance activities, conduct local research studies, and to collaborate in the National Birth Defects Prevention Study. The emphasis is on using surveillance and research to improve services for children with birth defects and their families. NTDs are a major focus for surveillance and research at this time.

CDC and the Centers have a strong research agenda and various research groups have been started, e.g., hearts, clefts, micronutrients, NTDs. Additional proposed research activities include the following:

- environmental - pesticides, heat exposures, drinking water
- diet - folate, zinc, fat
- maternal - obesity, fever, alcohol/smoking
- genetic - candidate genes, genetic markers
- medication - OTCs, HIV retrovirals, anti-epileptic
- other - costs, quality of care.

In addition to working with the Centers, CDC collaborates with twenty-nine state surveillance programs in the National Birth Defects Prevention Network. The Network is incorporated as a not-for-profit organization and covers approximately 58% of annual U.S. births. The Network provides a forum for exchanging ideas, has a regular newsletter and other publications, offers a website at www.nbdpn.org/nbdpn, meets annually, and collaborates on the development of

surveillance guidelines and standards, and on rapid ascertainment of NTDs and folic acid education. The Network has produced a manual of surveillance guidelines and standards to share with states where surveillance is under development.

The CDC Birth Defects and Pediatric Genetics Branch has forty FTEs, and fifteen contractors. The budget for the Network is $200,000 USD and members do enormous work for nothing. Eighteen state cooperative agreements exist, each for $100,000 USD per year for three years and the seven Centers receive $1 million USD each. The Branch's top priorities for the next five years are to expand and improve state surveillance programs and the national Network, to make the Centers fully functional, and to expand and evaluate the folic acid campaign. Support for work on birth defects has been achieved in large part through the lobbying of congress by the March of Dimes.

Charlotte Hobbs, Little Rock, Arkansas, U.S.A.

Charlotte Hobbs is Director of the Arkansas Center for Birth Defects Research and Prevention, one of the seven Centers supported by CDC.

Arkansas is one of the smallest and poorest states. It has a population of 2.5 million, with approximately 37,000 births annually and an infant death rate of 9 per 1000 births. The surveillance budget is $200,000 annually. The program has one full-time clinician plus 18 other staff to do both surveillance and research. The Center also receives the $1 million annually from CDC as well as research grants.

Surveillance of birth defects in Arkansas originated at a 1979 conference on prevention of mental retardation. In1985, the state governor signed an Act to support the Arkansas Reproductive Health Monitoring System, broad legislation for access to all reproductive end points. Up until 1995, the purpose of surveillance was to collect baseline data on birth defects and monitor trends; to identify cases for etiological studies; and prevention, advocacy and education.

To be included in the surveillance data, a case has to be a major defect identified from terminations, fetal deaths, stillbirths, live births, with the mother a resident of Arkansas. (Fetal deaths are below 20 weeks; stillbirths are more than 20 weeks. Both are reported with death certificates.) Both chromosomal and physical defects are included. Each is coded and then may be eligible for special study. With the 1995 cooperative agreement with CDC, the program went state-wide. The relationship of the Center with the Arkansas State Department of Health has been critical in facilitating access to birth data.

The mission of the Arkansas Center is to evaluate data and to improve the timeliness of reporting on conditions with an emphasis on NTDs, oral clefts, gastroschisis and Down syndrome. Timeliness has improved—the mean age of infants for whom a report is available has decreased. Timely follow up with mothers results in more reliable information from them and a greater chance of preventing an NTD recurrence for that mother.

The Center collaborates with the CDC and other Centers in the National Birth Defects Prevention Study. It also conducts local research studies in the following areas:

< epidemiological and surveillance

< multidisciplinary health services research and community interventions, e.g., to determine the level of knowledge about folic acid among women in a certain region

< gene-environment interactions, e.g., NTD and obstructive heart defects; micronutrient interactions (several studies suggest taking multiple vitamins with folic may protect against the heart defects).

The Center produces reports on surveillance and research, written in lay language and disseminated to health care planners, health care providers, health information management departments, legislators, researchers, and the public.

4. Emerging Issues for Surveillance

Introduction

Each workshop participant was assigned to one of three topics for small group discussion (facilitator in brackets):

- cluster management (Elisabeth Robert)
- prenatal diagnoses (Shia Salem)
- NTD surveillance (Charlotte Hobbs).

Summaries of the small group discussions are provided below as reported in plenary by the rapporteurs from the groups. In addition, a summary is provided of ad hoc plenary discussions about the growing interest in monitoring cases of FAS.

Cluster Management

Richard Stanwick reported on his small group's discussion about cluster management. Elisabeth Roberts had presented briefly about work with clusters in Europe. Richard recommended that everyone read the paper by Philippe De Wals on cluster management.

It was highlighted that each cluster is unique and, although common themes exist among them, each requires uniquely different teams of expertise to study them. In clear cut examples of clusters for which the etiology is irrefutable, it is a local observation that spawns the investigation. However, explaining clusters is usually not that simple. If abortion data is included in conjunction with data on live births, it can increase the cluster. Media can dictate whether a cluster has taken place. The problem is people not appreciating true cause and effect. Lifestyle factors may be a more powerful explanation of a cluster, but it is common for the community to point to external factors.

Participants in the small group commented about the Canadian situation. Canada has the capacity to recognize clusters but there was doubt about the capacity to respond. Detecting a cluster initially falls to public health. The two phases, recognition and then investigation, should not necessarily fall to the same team. Teams are multidisciplinary and each member brings his or her own approach. There tends to be an overemphasis of epidemiology over clinical expertise. Investigation of clusters needs a more collaborative model.

The difficulties determining etiology are rooted in time and space. Determining the significance of the geographical dispersion of a cluster is difficult—is it more important if it is limited to one street or scattered around a province? In addition, if a jurisdiction wants to study clusters, they have to go back to individual families. Timing is critical—it is pointless to ask what someone

had for breakfast 18 months ago. Also whether or not an etiological hypothesis exists affects the nature of the investigation. Almost all "real" clusters were first identified by a clinician.

In terms of the role the federal government now plays, the emphasis appears to be on epidemiology. The group challenged the appropriateness of general anomaly surveillance. The major function should be to confirm or rule out clusters suggested by others rather than to discover clusters. For example, the media may stumble on two cases, further investigation may find two more, but surveillance data reveals that a normal rate is six. It is important to have quality data available to be able to make these statements.

When additional expertise, e.g., genetic or clinical, is required for investigation of a cluster, the federal government needs a more formal process for bringing in experts with the appropriate specific talents. It was suggested that the government call upon the College of Geneticists to recommend a geneticist for each investigation depending on the expertise required.

Emerging issues related to privacy of information are affecting surveillance significantly. Increasingly the approval of an ethics committee is being required for capture of certain data or for case follow-up.

Prenatal Diagnoses

Rob Liston reported on his small group's discussion about prenatal diagnoses. Recent technological developments have permitted screening and diagnosis in early to mid-second trimester. These earlier and quicker diagnoses are resulting in earlier interventions (terminations) which are posing challenges for ascertainment. Sources of information about prenatally diagnosed anomalies include genetic, pathology, and ultrasound laboratories, families, and the hospitals and clinics where terminations are performed.

Having information on prenatal diagnoses would have an impact on quality of care. The difficulty so far has been transferring the information to surveillance systems. It would help if the collection and transfer of this information were legislated. The small group discussion included suggestions for ways in which the information could be obtained from the various sources. Genetics laboratories could report through the Canadian College of Medical Geneticists. Genetics information from laboratories could be linked to perinatal databases provincially and territorially. Capturing information on defects from ultrasound laboratories was seen as more complex. Regardless of whether they are hospital-based or not, all ultrasound labs must be accredited and the process for accreditation could include mandatory reporting. CIHI is working to enhance the reporting about terminations performed in hospitals and free-standing clinics. Reports from hospital pathology labs would also be needed.

Related to legislating the transfer of information, some people felt that it was important to have a clear guiding principle to get all of the provinces on board. A parallel was drawn to the list of reportable communicable diseases for which it is mandatory for laboratories and physicians to report. Manitoba is considering adding congenital anomalies to its mandatory reportable list.

An important consideration is the ability to link prenatal diagnoses to the pregnancy outcomes, i.e., live births, stillbirths, fetal deaths and elective terminations. Individual identifiers are essential for linking information from ultrasound and genetics labs (prenatal diagnoses) to information from hospitals, clinics and pathology labs (pregnancy outcomes). Another consideration is being able to link to diagnoses after birth, e.g., reports from radiologist labs on limb anomalies.

Most activity around prenatal diagnoses is centred in primary care centres with satellites. For example, in Nova Scotia, one person has the responsibility of travelling to each of the centres to bring all of the information together. The group felt that it was feasible to achieve good capture of data on prenatally diagnosed defects. The emphasis for promoting capture of this data should be a better understanding of what is happening in order to improve and enhance care. The group also noted that surveillance systems can be used to evaluate diagnostic techniques.

Increasingly early diagnoses followed by interventions such as dilatation and suction are resulting in a loss of material that could permit a physical diagnosis by pathology labs. Physical diagnoses are also important for confirming ultrasound diagnoses especially from level one labs—the number of unsubstantiated ultrasounds is high. Patient reporting was considered an unlikely data source. However, involving families of prenatally identified cases in follow-up was seen as important to enhancing prevention.

NTD Surveillance

Rebecca Attenborough reported on her small group's discussion about NTD surveillance. Based on Charlotte Hobbs' description of the Center in Little Rock, Arkansas, Rebecca stressed the importance of the following for effective surveillance:

< the right approach and attitude to get surveillance programs going

< the sensitive and competent qualities of the key individuals involved

< setting achievable goals

< accessing multiple sources of data

< enabling legislation.

The group characterized the current level of NTD surveillance in Canada as variable, from comprehensive to non-existent. Timeliness is an issue for those systems which are in place, including CCASS. Barriers to NTD surveillance include:

< regional differences in ascertainment and capture of termination data

< regional sensitivities to termination data

< the general weakening of the public health system

< differences in capacity to recognize and investigate clusters of NTD cases

< "provincial schizophrenia", i.e., they want support from the federal government but do not want to be told what to do

< limitations of CCASS in terms of coding issues related to hospital data.

The suggestion was made that sensitivities to termination data could be avoided by focusing on prevention, i.e., primary prevention which would prevent the need for terminations.

Suggestions for how to enhance NTD surveillance in Canada included the following:

< Work with what we already have and what we can get.

< Establish a voluntary network of the provinces/territories and regions with an active role in NTD surveillance. Reach agreements at the provincial/territorial Department of Health level,

as well as with the local people involved, to lay out the responsibilities and benefits of participation for all network participants including Health Canada.

- Work with a limited number of variables and reach for more.
- CCASS should focus on population-based information.
- Provinces/territories and regions should work from their individual strengths. They should collaborate with multiple groups—their public health systems, screening services, level two ultrasound laboratories, hospitals, etc.

International trends in NTDs are also important. Information from the ICBD should be readily available. Anyone interested in receiving a copy of the ICBD annual report should contact I.D. Rusen.

Discussion in plenary following the small group report raised key issues which participants felt required more lengthy discussion and could be addressed by a Canadian network on congenital anomaly surveillance:

- Would the network limit itself to NTDs? One participant felt it might be easier to get cooperation in her province if the focus was on NTDs because of their mandatory public health program to reduce NTDs. Others felt that it would be possible to build on any success with NTDs to address other preventable anomalies.
- Agreeing on a list of five or six preventable anomalies is not as simple as it seems. If preventable means preventing the birth of an infant with an anomaly, i.e., through pregnancy termination, then it is easy to list the anomalies. It is somewhat more difficult if it means primary prevention. If the purpose of surveillance is service, then it is hard to say which birth defects are the most important.
- If this meeting were to recommend that prenatally diagnosed fetal anomalies should be tracked in every jurisdiction, then the issue of inconsistency in the data would have to be addressed. Agreement would have to be reached on minimal criteria and a minimal data set. Attention would have to be paid to regional priorities to achieve their buy-in.

FAS

Health Canada has declared FAS an important issue and allocated $11 million over the next 3 years, and $5 million ongoing thereafter. The Bureau of Reproductive and Child Health would like to provide evidence to help determine where best to direct the government's efforts. Facts about FAS prevalence and incidence are not easy to obtain.

One representative provided an example of why the issue can be so clouded. An explosion in diagnoses of FAS in British Columbia could be attributed to the fact that the education system has designated FAS as one of ten categories of special needs which are eligible for the services of a classroom teaching aid.

FAS is a large problem. Children with FAS often act out as teens and many die before the age of 19. Resources put into tertiary treatment appear to have no impact. Resources need to be in prevention. Capturing data on FAS is not easy because of the difficulty of diagnosing the condition before the age of one year. The Developmental Clinic at the Alberta Children's Hospital in Calgary has sent staff to Seattle for training in a new diagnostic methodology which tries to provide an objective system for diagnosing FAS. The Newfoundland representative noted that they collect information on FAS on a paper registry.

5. A Vision for Surveillance of Congenital Anomalies in Canada

Introduction

The workshop included a brainstorming session for participants to express their vision for surveillance of congenital anomalies in Canada. The intent was to identify an ideal collection of activities which would include surveillance at various levels throughout the country. The points made in the vision session have been grouped into categories and summarized below under two headings—objectives and characteristics for success.

The brainstorming was followed up with discussion of roles and responsibilities, and how the identified objectives could be supported within Health Canada's mandate for congenital anomaly surveillance and other related activities at the federal level.

Objectives

These objectives are summarized from points made in the vision/brainstorming session and are presented here as draft objectives only. Further discussions will be needed to ensure that the objectives are complete and that the wording clearly represents the intent of each objective. The draft objectives are accompanied by synopses of workshop discussion related to each objective.

To prevent birth defects. The focus of all surveillance activities should be on prevention not on data collection. Prevention is intended to include primary prevention as well as early detection and intervention.

To accurately describe the burden of congenital anomalies (trends) at national, provincial/territorial, regional and local levels. Standardization of definitions would be needed for major and minor anomalies and for diagnoses criteria in order to ensure consistency across the country.

To identify cases for etiological studies. An etiological study is not possible without following up with family members. Timely data to support the follow-up are critical.

To conduct research on prevention. Apart from its value in increasing knowledge about congenital anomalies, building in a solid research component was viewed as one way in which surveillance programs can outlive the short-term legislative priorities of politicians.

To provide tools for prevention, education, advocacy and promotion. Education must be user-friendly and accessible as a resource to the public. Participants distinguished between advocacy and promotion as follows: advocacy is action to make the system work. Advocates might be parents, families, organizations, etc. Promotion is enhancing awareness about the importance of the issue of birth defects. Often surveillance programs work with partners, e.g., March of Dimes in the U.S., to keep the issue of birth defects highly visible.

To evaluate community concerns about suspected clusters of anomalies. The emphasis here was on timely data of high quality to respond adequately to community concerns about clusters. Trying to identify clusters through the study of trends was questioned as not very productive.

To exclude teratogens and provide support to front-line public health professionals for the investigation of clusters. The media are very good at identifying teratogens. A surveillance

system should have the capacity to exclude suggested teratogens and provide technical support to the front-line public health professionals who must answer questions from the of public and media.

To facilitate linkages between public health and clinical services. The intent here was to create closer links between public health professionals and clinicians in terms of their interest in identifying cases, noting trends and preventing birth defects.

To integrate special health care services for children and other services for their families and improve access to these services.

To facilitate closer links and long-term follow-up with affected families. The intent is to enhance planning for the future needs of affected children and their families for services and medical equipment.

To evaluate interventions designed to prevent congenital anomalies.

To serve as a quality assurance tool. To evaluate various aspects of clinical care related to congenital anomalies, such as prenatal diagnostic technology.

To provide training opportunities in the field of congenital anomaly surveillance. The intent was to promote, strengthen, and encourage training in broad epidemiology to advance knowledge in the field.

Characteristics for Success

These characteristics for success are summarized from points made in the vision/brainstorming session as well as other discussions during the workshop. The characteristics apply to an ideal system based on surveillance activities at the local, regional, provincial/territorial, and national levels. Activities at all levels would be linked in some way but the emphasis at each level would vary.

The characteristics discussed included the following:

- Each province/territory or region should build its surveillance program on its individual strengths, where capacity and interest already exist—start small, do it well and grow from there.
- A central theme to garner support for all surveillance activities should be the concerns of mothers—the clinical care aspects (service to children and families, prevention). A continuum of accessible services should be available to families.
- The systems should be built on more than one reason to maintain the support of a variety of stakeholders.
- A support base should be built within the health field by finding out what data they need and supplying them with it.
- Agreement is needed on a few core birth defects to be monitored across the country. (Birth registries do not capture things like retarded growth, impaired cognitive function, etc. Hearing impairment, blindness, mental retardation require a whole different data source.)
- Agreement is needed on the minimum data to be collected and definitions of terms.
- Surveillance across the country should be characterized by consistent, accurate, and precise diagnoses.

- Detailed individual case data should be kept at the regional or provincial/territorial level. Investigations involving rarer conditions may benefit from several jurisdictions combining their case data.
- Many partners are needed, i.e., sources of data, service providers for affected children and families, regional and national advocates, scientific expertise, etc. All levels need to identify partners with the reputation and expertise to perform certain roles so that they are not reinventing the wheel.
- One role for a federal program should be to encourage those provincial/territorial systems which respect existing regional systems and serve families. Because larger provinces experience geographic limitations and regional sensitivities, this may mean that the federal program would collaborate with individual regional systems with the agreement of the province/territory.

Unique Roles for Various Levels

The objectives and characteristics above represent a preliminary framework for activities in surveillance of congenital anomalies at all levels across the country. Health Canada, the provinces/territories, and regions each have a unique role to play in meeting the objectives of this framework. Discussion took place about how Health Canada could contribute to the achievement of the identified objectives within their mandate for congenital anomaly surveillance and related activities.

The overall goal for Health Canada's work on congenital anomalies was presented as follows:

- ***To improve the health of the population through the prevention of congenital anomalies.***

  The population health focus of Health Canada and other governments includes attention to genetic determinants of health which supports the work of surveillance of birth defects.

  Within this overall goal, the following were presented as three proposed Health Canada activities:

  - ***Enhance CCASS, the current national surveillance system,*** through:
    - attempts to include termination information
    - attempts to increase the timeliness of the available data
    - increased reporting on congenital anomalies in Canada.

  - ***Support a formal network of provinces, territories and other stakeholders to enhance congenital anomaly surveillance at the national, provincial, territorial and local levels.*** This network would be under the umbrella of the CPSS and its activities may include:
    - determination of a core minimum set of anomalies for surveillance
    - recommendations on surveillance standards and approaches, e.g., inclusion of prenatal diagnoses
    - others as identified by the network.

- < ***Increase the access to and availability of technical support for investigation of clusters of congenital anomalies*** by:
  - establishment of a more formal mechanism for responding to local and provincial/territorial concerns
  - working with the Canadian College of Medical Geneticists to establish a process to ensure the availability of appropriate expertise.

Discussion about how the framework objectives could be fulfilled within these three activities included the following key points:

- < Some objectives would not be addressed directly by these Health Canada activities but could be indirectly addressed, e.g., the proposed network could strengthen and increase local, regional and provincial/territorial programs which may be able to achieve the objectives.
- < Other recommended activities for the network included the following:
  - sharing information and ideas (e.g., a newsletter featuring provincial activities, selected projects, international news, announcement of events; working groups on various disorders).
  - identifying needs of families and intervention priorities
  - identifying advocacy opportunities
  - addressing common issues (e.g., understanding and working within privacy legislation, coding ethnicity).
  - collaborating on specific research questions, e.g., NTD data.
- < The network should raise awareness of the importance of prevention and surveillance of congenital anomalies. The developing network could fulfill this advocacy function through various channels, including federal-provincial-territorial channels.
- < An attempt at a national collaboration was made in 1967 but it gradually dwindled from lack of leadership. The participants are looking to Health Canada for leadership to move this issue forward. A large number of provinces are willing to contribute their efforts without additional funding.
- < Two commitments are needed, one from the provinces/territories or regions that they will do the work and the second from Health Canada that they will assign a person and some funding to support these efforts. Interest and support must be present in the provinces/territories before funding can have the greatest impact.
- < The existing budget of CCASS is $30,000. An additional $300,000 per year has been proposed for related activities. A substantial portion of these funds are already committed to the Newfoundland fortification study.
- < A Canadian network could learn from the experiences of the U.S. Network but will differ somewhat because of differences in legislated responsibilities at both the federal and state or provincial/territorial levels. For example, the public health system in Canada may not be in a position to do substantive research and case control studies on congenital anomalies as is done by the members in the U.S. Network. This research may be more appropriately funded by the Canadian Institute of Health Research (CIHR).

- International expertise and information on international trends are needed.
- Silos of research are a real concern. Various professional organizations carry out research on congenital anomalies. An inventory of existing perinatal activities would be useful. Linking clinicians across all sub-specialties is important.

1. Next Steps and Wrap-Up

The first concrete next step following this workshop will be the production and dissemination of this workshop report to participants and other interested stakeholders.

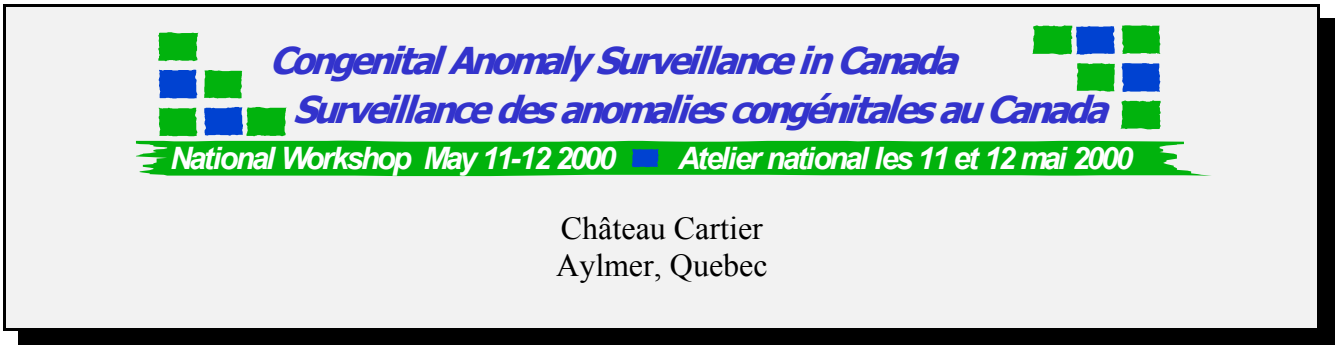
One participant suggested that everyone take advantage of opportunities to raise the profile of birth defects in Canada, e.g., two upcoming conferences were mentioned, one in June on community genetics and the other in November sponsored by geneticists.

I.D. Rusen reported on the Bureau's planned follow-up to the workshop. The Bureau plans to commit staff to develop and support the proposed network and to work toward a first meeting in the fall. Plans for follow-up will take into account the feedback received from participants in their evaluation forms for this workshop. I.D. will keep the participants up-to-date on the progress of the network initiative.

I.D. thanked the participants for contributing their knowledge and views at the workshop and expressed enthusiasm for working with the participants in the future.

Appendix A

List of Participants



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Appendix B

Agenda

Congenital Anomaly Surveillance in Canada
Surveillance des Anomalies Congénitales au Canada
A National Workshop May 11-12, 2000 ■ Un atelier national le 11-12 mai 2000

Agenda

Thursday, May 11

8:00 - 8:30 am	Continental breakfast	
8:30 - 8:40 am	**Welcoming remarks**	
	Dr. Paul Gully, Deputy Director General, LCDC Dr. Catherine McCourt, Director, Bureau of Reproductive and Child Health	
8:40 - 9:00 am	**Objectives of workshop** **National congenital anomaly surveillance activities**	
	Dr. I.D. Rusen, Bureau of Reproductive and Child Health	
9:00 - 10:00 am	**Existing surveillance activities in Canada**	
	Dr. Brian Lowry, Alberta Congenital Anomaly Surveillance System Ms. Rebecca Attenborough, Nova Scotia Reproductive Care Program Mr Soo-Hong Uh, British Columbia Vital Statistics	
10:00 - 10:30 am	Coffee	
10:30 - noon	**Current status of provincial and territorial activities:** **A round table update**	
noon	Lunch	
1:00 - 2:30 pm	**Congenital anomaly surveillance:** **International perspectives**	
	Dr. Elisabeth Robert Dr. Larry Edmonds Dr. Charlotte Hobbs	Lyon, France Atlanta, Georgia Little Rock, Arkansas
2:30 - 3:00 pm	Coffee	

3:00 - 5:00 pm	**An ideal system: Vision for congenital anomaly surveillance in Canada** **Roles and responsibilities**
Friday, May 12	
8:00 - 8:30 am	**Continental breakfast**
8:30 - 9:15 am	**Breakout sessions** **- cluster management** **- prenatal diagnosis** **- NTD surveillance**
9:15 - 10:00 am	**Summary of breakout sessions**
10:00 - 10:30	**Coffee**
10:30 - noon	**Summary of breakout sessions (continued)**
noon	**Lunch**
1:00 - 2:30 pm	**Conclusions, recommendations & next steps**
2:30 pm	**Adjourn workshop**

All sessions will take place at:

Chateau Cartier Hotel
1170 Aylmer Road
Aylmer, Quebec

Appendix C

Organizational Chart of the Bureau of Reproductive and Child Health

Appendix C
Organizational Chart