

Recommendations for development of a new Australian Birth Anomalies System

**A review of the National Congenital
Malformations and Birth Defects Data Collection**

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Recommendations for development of a new Australian Birth Anomalies System

**A review of the National Congenital
Malformations and Birth Defects Data Collection**

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Foreword

This review of the Congenital Malformations and Birth Defects Data Collection was commissioned by the Australian Institute of Health and Welfare following the review of the AIHW National Perinatal Statistics Unit in 2001 which highlighted concerns about data quality and comparability in this collection.

For Australia to have a comprehensive perinatal data system, a robust national birth anomalies data collection is needed. The first phase in developing a new data system was to examine the strengths and limitations of the existing system. The second phase involved consulting with stakeholders and identifying best practice.

This report sets initial parameters for a new Australian Birth Anomalies System, and further development is required. The Institute considers the further development of the Australian Birth Anomalies System to be a priority, as birth anomalies have a long-term impact on the quality and length of life of those affected.

Richard Madden

Director

Executive summary

Birth anomalies remain a significant public health problem in Australia. They often result in disabilities and are a major reason for hospitalisation in infancy and childhood and are a leading cause of infant mortality. Despite this, we do not have quality national data on birth anomalies in Australia.

The national collation and reporting of birth anomalies data has been suspended in recent years due to concerns about data quality and comparability. In response to the AIHW's continuing commitment to birth anomalies as a core data collection, a review was conducted over the last year which was aimed at assessing the utility and scope of the National Congenital Malformations and Birth Defects Data Collection and making recommendations for its future.

The key findings of the review were that there is a lack of national consistency in birth anomalies data and that this affects the quality and utility of the national collection. There is variability among the states and territories in the scope of their birth anomalies data collections, the sources of birth anomalies notifications, the definitions and classifications used, the method of data collection and the available resources. There is also variability among the states and territories in the timing and method of the provision of birth anomalies data to the AIHW National Perinatal Statistics Unit (NPSU) for national collation and reporting.

In order to achieve national consistency and to develop a high quality national birth anomalies data collection, it was recommended that initially the scope of the new Australian Birth Anomalies System should be data for birth anomalies detected up to 1 year of age, including data on terminations of pregnancies with birth anomalies, regardless of gestational age (i.e. including <20 weeks gestation). This scope was considered to be better practice by the review and internationally. Therefore the system will initially be based on data from the states able to detect birth anomalies at least up to 1 year of age, with a view to further extending the period of detection in the future. These states are New South Wales, Victoria, Western Australia and South Australia. It was recommended that a timeframe for the other jurisdictions to progress their collections be developed.

In addition, a significant program of data development will need to be undertaken to achieve national standardisation of the data and clinical definitions used and the classification of birth anomalies, with the objective of the development of a National Minimum Data Set. Work to identify the conditions to be included in the collection will also be undertaken.

A critical outcome of this review was the reinforcement of the need to enhance partnerships with data providers, experts in the field, governments and consumers. This will be pivotal in developing the structure and content of the new national report. It is intended that this revised report will be based on data for 1998–2001 and will be released in June 2005. The timing reflects factors such as multiple sources of notification of birth anomalies and the need to enhance data quality and completeness of the collection.

A technical steering committee will be convened to assist with this work.

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Ainsley Morrissey coordinated the printing and publication process.

Abbreviations

ABAS	Australian Birth Anomalies System
ACT	Australian Capital Territory
AGSA	Association of Genetic Support of Australasia Inc
AHMAC	Australian Health Ministers' Advisory Council
AIHW	Australian Institute of Health and Welfare
ANZARD	Australia and New Zealand Assisted Reproduction Database
APSU	Australian Paediatric Surveillance Unit
ART	Assisted Reproduction Technology
CDC	Centers for Disease Control
CM&BDNDRC	Congenital Malformations and Birth Defects National Data Review Committee
DoHA	Commonwealth Department of Health and Ageing
EUROCAT	European Concerted Action on Congenital Anomalies and Twins
FACS	Commonwealth Department of Family and Community Services
FBE	Full Blood Examination
FTE	Full-time equivalent
ICBDMS	International Clearinghouse for Birth Defects Monitoring Systems
ICD-9	International Classification of Diseases, 9th Revision
ICD-9-BPA	International Classification of Diseases, 9th Revision, British Paediatric Association
ICD-9-CM	International Classification of Diseases, 9th Revision, Clinical Modification
ICD-10-AM	International Statistical Classification of Diseases and Related Health Problems, 10th Revision, Australian Modification
ICF	International Classification of Functioning and Disability
ICSI	Intracytoplasmic Sperm Injection
IVF	In-vitro Fertilisation
MSS	Maternal Serum Screening
NBASC	National Birth Anomalies Steering Committee
NCC	National Coding Centre
NCCH	National Centre for Classification in Health
NHIA	National Health Information Agreement
NHIG	National Health Information Group
NMDS	National Minimum Data Set
NPHP	National Public Health Partnership
NPSU	AIHW National Perinatal Statistics Unit

NSW	New South Wales
NT	Northern Territory
Qld	Queensland
SA	South Australia
SIMC	Statistical Information Management Committee
Tas	Tasmania
TMS	Tandem Mass Spectroscopy
TOR	Terms of reference
Vic	Victoria
WA	Western Australia
WHO	World Health Organization
n.a.	Not available
..	Not applicable

Recommendations

Based on the findings of the review, the following recommendations are made:

- That the National Congenital Malformations and Birth Defects Data Collection continue and that it be referred to as the Australian Birth Anomalies System (ABAS).
- That the term 'birth anomalies' be used for the national collection and national reporting.
- That the ABAS be the national data repository for data from jurisdictional birth anomalies data collections and other identified suitable data collections. For example, from the Human Genetics Society of Australasia (Newborn Screening).
- That the ABAS be supplemented by data on birth anomalies from other sources, for example, data collected by the Australian Paediatric Surveillance Unit (APSU).
- That the custodian of the ABAS be the AIHW National Perinatal Statistics Unit (NPSU).
- That data for the ABAS be provided to the NPSU on an annual basis according to agreed specifications and an agreed timetable. There should be an agreed policy for updating data at the national level.
- That a National Birth Anomalies Steering Committee (NBASC) be established to provide expert advice on the scope, development and implementation of the ABAS. The steering committee should comprise 2–3 state and territory representatives, a representative from the Department of Health and Ageing (DoHA), a representative from the Department of Family and Community Services (FaCS), and other relevant stakeholders, including from clinical, epidemiological and technical backgrounds. The steering committee should meet on a regular basis, including one annual face-to-face meeting.
- That funding be identified for the support of the NBASC.
- That the NPSU in consultation with relevant stakeholders develop a nationally consistent birth anomalies definition.
- That the initial scope of the ABAS be data for birth anomalies detected up to 1 year of age with a view to extending the period of detection as or when appropriate.
- That the scope of the ABAS include terminations of pregnancies with birth anomalies data regardless of gestational age (i.e. including <20 weeks gestation).
- That the ABAS initially be based on data from those states able to detect birth anomalies up to 1 year of age, including terminations of pregnancies with birth anomalies data. These states are New South Wales, Victoria, Western Australia and South Australia.
- That a timeframe for national consistency be developed.

- That the development of partnerships between the more capable states and the less capable jurisdictions to achieve national consistency be explored.
- That the NPSU should report reliable and valid national birth anomalies statistics in the public domain.
- That linkage with other data sets (e.g. assisted conception) be undertaken, subject to appropriate ethics approval, to improve data quality, to improve the utility of the data and to facilitate research.
- That information on maternal and paternal risk factors be included in the ABAS.
- That the NPSU develop collaborations with organisations collecting data on screening and diagnostic testing in the first year of life.
- That the structure and content of the current national Birth Defects Series of reports be revised with advice from the NBASC.
- That the first revised national report should include birth anomalies data for births from 1998 to 2001. Data from collaborations with organisations collecting data on screening and diagnostic testing in the first year of life should also be included.
- That data be collected by year of notification and reported by birth cohort.
- That the NPSU coordinate access to ABAS data for national and international reporting and collaborative research, subject to appropriate ethics approval.
- That this review report be referred to the Statistics Information Management Committee (SIMC) to provide information about the lack of a national birth anomalies system, the inability of some jurisdictions to provide data because of funding and other resource constraints, and the need for data development.

Introduction

Birth anomalies are a key component of the necessary parts of a developed country's infant health information system. Although birth anomalies are rare, national reporting of them is seen as a core responsibility of the Australian Institute of Health and Welfare (AIHW).

In recent years the national collation and reporting of birth anomalies data has been suspended due to concerns about data quality and comparability. The last national report was published in 2001 with data for 1981-97.

The aim of this review was to assess the utility and scope of the National Congenital Malformations and Birth Defects Data Collection which comprises the Congenital Malformations Australia data collection and the Birth Defects series of reports. The review was an outcome of the Australian Institute of Health and Welfare's (AIHW) review of the AIHW National Perinatal Statistics Unit (NPSU) conducted in 2001. It was undertaken by the NPSU in consultation with the Congenital Malformations and Birth Defects National Data Review Committee (CM&BDNDRC) which was convened by the AIHW to advise the NPSU during the review. In addition to consultation with the committee, consultation included a survey of state and territory birth anomalies data collections and a 2-day workshop held with members of the committee and other key stakeholders.

The findings of this review are considered and show a path forward for the development of a best-practice national collection on birth anomalies.

For consistency, the term 'birth anomalies' will be used throughout the report to describe congenital malformations and birth defects. This terminology was agreed to by the Congenital Malformations and Birth Defects National Data Review Committee.

This report

Chapter 1 describes the National Congenital Malformations and Birth Defects Data Collection and outlines the purpose of the review.

Chapter 2 describes the methodology used for the review.

Chapter 3 presents background information related to the utility and scope of birth anomalies monitoring systems.

Chapter 4 presents the findings of the survey of state and territory birth anomalies collections.

Chapter 5 presents the outcomes and key recommendations of the Congenital Malformations and Birth Defects National Data Review Workshop.

Chapter 6 presents the recommendations of the Congenital Malformations and Birth Defects National Data Review Committee against the terms of reference of the committee.

1 The National Congenital Malformations and Birth Defects Data Collection 1981–2001

The NPSU was established in 1979 following concerns about claims of increased incidence of birth anomalies in relation to the use of herbicides. The Congenital Malformations Australia data collection began in 1981 with data from four states, and all states and territories provided data from 1986. The last request for data was made in 2000 and related to the 1997 birth cohort. The last national report in the Birth Defects Series was published in 2001 and included data for 1981–97.

Objectives 1986–2001

The principal objectives of the National Congenital Malformations and Birth Defects Data Collection when it was established were:

- to determine the prevalence of major birth anomalies;
- to monitor trends in the reported prevalence of major birth anomalies;
- to detect new drug and environmental teratogens;
- to use the database for research (aetiology, descriptive epidemiology and evaluation studies) on birth anomalies; and
- to meet national and international reporting requirements.

Data sources

The National Congenital Malformations and Birth Defects Data Collection is a compilation of notifications of major birth anomalies from state and territory birth defects registers and perinatal data systems. Birth anomalies are mainly notified from data collected as part of perinatal collections. Other sources of data include perinatal death certificates, cytogenetic or pathology reports, inpatient data, hospital lists, maternal and child health nurses, and medical officers.

The data are provided to the NPSU by the states and territories for national collation. The data are provided on mothers and babies and include information on demographics, diagnosis, method of prenatal diagnosis, sources of diagnosis, birth outcome, plurality and birth order, birth weight, and previous pregnancies and outcomes.

The data are specified in an agreed minimum data set which is based mainly on data elements in the Perinatal National Minimum Data Set. However, some items requested (e.g. Mode of separation) are not consistent with those in the National Health Data Dictionary, or are not included in the National Health Data Dictionary.

Definitions and scope

For the purposes of the national collection, birth anomalies are defined as anatomical defects or chromosomal abnormalities that are present at birth.

Data are requested for birth anomalies diagnosed in live born infants in the first 28 days, or in stillbirths of at least 20 weeks gestation or 400g birth weight; and terminations of pregnancy that have occurred at gestational ages of 20 weeks or more. Data are requested for chromosomal abnormalities diagnosed up to 1 year of age.

Birth anomalies and chromosomal abnormalities listed in the chapter on Congenital anomalies in the International Classification of Diseases, 9th Revision, Clinical Modification (ICD-9-CM) are included.

Classification of birth anomalies

The NPSU developed a classification based on the Royal College of Paediatrics and Child Health's (formerly, the British Paediatric Association (BPA)) Classification of Diseases. The BPA codes for malformation syndromes, limb reduction defects and other selected birth anomalies were modified to enable more specific classification of these birth anomalies.

The NPSU recoded the data sent by the states and territories using this classification. Recoding was last undertaken for the 1997 birth cohort and will not be undertaken in the future.

The BPA Classification of Diseases was based on ICD-9-CM. The International Statistical Classification of Diseases and Related Health Problems, 10th Revision, Australian Modification (ICD-10-AM) was introduced in July 1998 and the specificity for birth anomalies has been enhanced in the third edition (introduced in July 2002) and the fourth edition (to be introduced in July 2004).

There is variation among the states and territories in the classifications used to code birth anomalies. The classifications used are ICD-9-BPA, ICD-9-BPA extension and ICD-10-AM (second and third editions).

National reporting

Prior to 1992, data were published in quarterly congenital malformation monitoring reports. The first report of the Birth Defects Series was published in 1995 and presented data for births occurring between 1981 and 1992. Subsequently, two biannual reports in the series (1993–1994 and 1995–1996) were published in 1997 and 1999 respectively. The last report was published in 2001 and was for births that occurred in 1997. It was published in an electronic format only.

International reporting

The International Centre for Birth Defects publishes annual reports from the International Clearinghouse for Birth Defects Monitoring Systems (ICBDMS). The NPSU has been a member of the ICBDMS since 1982 and has provided national birth anomalies data to the ICBDMS over this period. The most recent national data submitted to the ICBDMS were the 1997 data for use in the 2000 ICBDMS annual report.

Purpose of the review

There is a lack of consistency and comparability among the states and territories in the scope of their birth anomalies data collections, the sources of birth anomalies notifications, the definitions and classifications used, the method of data collection, the available resources and the publication of data. There is also variability among the states and territories in the timing and method of the provision of birth anomalies data to the NPSU for national collation and reporting.

The purpose of this review is to assess the utility and scope of the National Congenital Malformations and Birth Defects Data Collection and to make recommendations regarding its future.

2 Review process

The review process involved: the compilation of background information; a survey of state and territory birth anomalies data collections; a 2-day workshop; and the formulation of recommendations for the future of the National Congenital Malformations and Birth Defects Data Collection.

Background

The purpose of the background information was to examine the utility and scope of birth anomalies monitoring systems. This was intended to provide information to the review about the functions of birth anomalies monitoring systems, including surveillance, research, informing policy and decision making, and evaluation of interventions. It was also intended to provide information relating to scope and to provide justification for continuing a national birth anomalies monitoring system.

Survey of state and territory birth anomalies data collections

In order to review the national collection it was important to develop an understanding of the data collections that underlie it. The states and territories provide the NPSU with data for the National Congenital Malformations and Birth Defects Data Collection. However, there are differences among the states and territories including in the scope of their collections, the definitions and classifications used, and in the collection methods.

In order to describe the state and territory collections, the NPSU consulted with data providers through a survey designed for this purpose.

The questions in the survey sought information on the scope of the collections, the definitions used, periods of detection of birth anomalies, notification processes, funding and resources, linkages with other collections, number of years of data held, latest data, reporting, stated purpose of the collection, content and use. The respondents were also asked whether they thought there was any information that is not currently collected that should be and a section was included for general comments.

The survey was sent to the managers of the respective state and territory collections in April 2003 and additional questions were sent in June 2003. All managers returned completed responses.

A copy of the survey is included in Appendix 1. The results of the survey were presented at the workshop.

Workshop

In order to facilitate wide consultation and input from experts from diverse disciplines, a workshop was held in May 2003. Participants included members of the Congenital Malformations and Birth Defects National Data Review Committee (CM&BDNDRC) and other key stakeholders. The aims of the workshop were to develop an agreed set of objectives for the national collection, determine the scope of the collection, develop a proposal for a National Minimum Data Set (NMDS) for birth anomalies and list any issues for further work.

Tasmania, the Australian Capital Territory and the Northern Territory were not represented at the workshop. Separate teleconferences were conducted with these jurisdictions following the workshop to ensure that their input was included.

Recommendations

The recommendations were developed through consultation with the CM&BDNDRC, the survey and the workshop.

3 Background

Introduction

Birth anomalies remain a significant public health problem in Australia and are a major reason for admission to hospital during infancy and childhood. They often result in disabilities and handicaps and, in some cases, death (AIHW 2002). An estimated 5% of all Australian births and terminations of pregnancy have a major birth anomaly (Stanley 1995) and birth anomalies account for about 13% of the disease burden for children aged 0–14 years (AIHW 1999). Birth anomalies are also a leading cause of infant mortality in Australia, with 25% of infant deaths in 2000 caused by birth anomalies (Al-Yaman et al. 2002).

Birth anomalies are caused by genetic (including chromosomal), environmental and unknown factors, or combinations of these factors. It is estimated that the cause of about 65–75% of birth anomalies is unknown. About 15–25% of birth anomalies have a genetic cause and about 10% have an environmental cause (Brent 2001).

The utility of birth anomalies monitoring systems

Introduction

Birth anomalies monitoring systems enable the surveillance of birth anomalies through monitoring of prevalence within a population and over time and space. The data from these systems can be used to estimate the burden of birth anomalies; to determine the causes of and risk factors associated with particular birth anomalies; to identify high risk groups; and to inform planning, implementation and evaluation of programs and services used in the prevention and treatment of these conditions.

Birth anomalies monitoring systems maintained over a number of years enable a large sample size to be achieved which is useful for the study of less prevalent birth anomalies. International collaboration for the surveillance of birth anomalies also increases sample size and provides diversity of populations and environments.

Surveillance and monitoring of conditions

Surveillance is the process by which the occurrence of a condition within a population is monitored over time (Cordero 1992; Castillo et al. 1986). Birth anomalies monitoring systems enable trends in the prevalence of birth anomalies to be monitored. These systems can provide early warning of changes or unusual patterns in the prevalence of birth anomalies over time or geographical location which may indicate factors including:

- exposure to a teratogen (a factor that has an adverse effect on an embryo or fetus between fertilisation and birth, for example drugs, infections, chemical or physical agents);

- a genetic cause;
- a change in the population, for example a change in the pattern of maternal behaviour such as peri-conceptual use of folic acid (Wald 1991);
- geographical variation, for example, an increased risk of cardiovascular anomalies in Ostergotland county compared to the rest of Sweden (Blomberg et al. 2000);
- variation in the use of or provision of diagnostic services;
- variation in the collection of data, for example increasing rates of renal agenesis due to changes in nosologic coding (Stroup et al. 1990);
- unknown reasons or chance.

Combinations of these factors may be indicated.

Many countries established population-based birth anomalies monitoring systems to detect new teratogens after thousands of babies were born with limb deficiencies, due to their mothers taking the drug thalidomide during pregnancy to alleviate symptoms of nausea (Blomberg et al. 2000; Correa-Villasenor et al. 2003; Holtzman & Khoury 1986). In Australia, the National Perinatal Statistics Unit was established in 1979 to monitor birth anomalies in response to concerns about claims of increased incidence of birth anomalies in relation to the use of herbicides. The resulting Congenital Malformations and Birth Defects Data Collection is the subject of this review.

Surveillance and monitoring of programs

Public health programs

Data from birth anomalies monitoring systems can be used to monitor and assess the effectiveness of public health programs such as peri-conceptual folic acid use.

Peri-conceptual supplementation of folic acid has been shown in randomised trials to be effective in reducing the risk of having a baby with a neural tube defect (Wald 1991; Czeizel & Dudas 1992). Data from birth anomalies monitoring systems have been used to assess the effectiveness of peri-conceptual folic acid use as a primary prevention strategy (Castilla et al. 2003; Halliday & Riley 2000; Martinez de Villarreal et al. 2002). In these studies, fortification of food with folic acid (Castilla et al. 2003) was found to be associated with decreased prevalence of neural tube defects as was peri-conceptual supplementation of folic acid (Martinez de Villarreal et al. 2002).

Screening programs and diagnostic testing

Birth anomalies monitoring systems can be used to collect data on prenatal screening programs and diagnostic testing and to evaluate the impact of these interventions.

Screening programs differ from diagnostic testing in that screening programs are conducted at a population level and, in the context of birth anomalies, are used to identify pregnant women that may be at risk of having a baby with a birth anomaly. Screening programs may be targeted at 'high risk' individuals, for example pregnant women aged 37 years and over. Diagnostic testing is conducted at the individual level to diagnose or confirm a birth anomaly. Indications for diagnostic testing include advanced maternal age (37 years and over), abnormal screening results, history of a birth anomaly and exposure to a teratogen (Muggli & Halliday 2003).

The aim of prenatal screening and diagnostic testing is the early detection of birth anomalies. This enables strategies for prevention and treatment to be developed early on, thus reducing the impact of the disease.

Prenatal screening tests include: Full blood examinations (FBE); First and second trimester Maternal Serum Screening (MSS) (this is a biochemical test which is usually done in the second trimester, but is being increasingly performed in the first trimester to coincide with the measurement of nuchal translucency conducted by ultrasound); Ultrasound including nuchal translucency (this is performed at 8–12 weeks to determine gestational age and at 10–14 weeks to determine the nuchal translucency which can indicate a risk of chromosomal abnormality). Ultrasound performed in the second trimester can be used to screen for structural birth anomalies (NPHP 2002).

Prenatal diagnostic tests include: amniocentesis, which involves the collection of amniotic fluid from the amniotic cavity in the uterus. The cells from the amniotic fluid are grown in culture and chromosomal, biochemical and molecular biological analyses are undertaken to detect various birth anomalies and certain genetic diseases. This test is performed at

14–18 weeks of pregnancy; Chorionic Villus Sampling, which involves the collection of tissue from the villi of the chorion (part of the placenta) and testing for birth anomalies and genetic diseases. This test is performed at 10–12 weeks of pregnancy, so can be performed earlier than amniocentesis.

Assessing prenatal screening programs and diagnostic testing

Birth anomalies monitoring systems can be used to assess prenatal screening programs and diagnostic testing. For example, in Europe, many birth anomalies registers provide information such as the proportion of cases with birth anomalies that were diagnosed prenatally, which may give an indication of the coverage of screening programs and/or how well they detect birth anomalies, and may provide some indication of the uptake of prenatal diagnostic testing. Information about the outcome of the pregnancy is also available, for example, the proportion of prenatally diagnosed cases which led to termination of pregnancy (EUROCAT 2002).

Assessing the impact of prenatal screening and diagnostic testing

Data from birth anomalies monitoring systems can be used to assess the impact of prenatal screening and diagnostic testing.

For example, a study using data from the South Australian Birth Defects Register and other data sources assessed the effect of screening using serum alpha fetoprotein concentration and ultrasonography on the birth prevalence of neural tube defects and the effectiveness of screening in detecting neural tube defects (Chan et al. 1993). It was found that the prevalence of neural tube defects remained stable over the study period, but that prenatal screening resulted in a decrease of 84% in the birth prevalence of neural tube defects. It was also found that screening was effective, with 85% of affected pregnancies being detected before 28 weeks gestation.

Another study, examining changes in the prevalence of neural tube defects using data from the Victorian Birth Defects Register linked to data on all births in Victoria showed that the prevalence of neural tube defects remained stable between 1983 and 1997, but the birth prevalence decreased significantly over this period (Owen et al. 2000). The study also showed that the number of terminations of pregnancy because of a diagnosed neural tube defect increased by six-fold over the study period, suggesting that improved early detection through wider screening (i.e. not just women at

increased risk) and termination of pregnancies affected by neural tube defects contributed to the decrease in birth prevalence.

Therefore, in order to determine the prevalence of birth anomalies accurately, data on prenatal diagnostic tests and terminations of pregnancy for birth anomalies should be included in birth anomalies collections.

Epidemiological research

Data from birth anomalies monitoring systems are used in epidemiological studies to investigate the aetiology of and risk factors associated with birth anomalies.

For example, data from seven regional registers in Europe were used to examine the risk of birth anomalies associated with living close to hazardous waste sites (Dolk et al. 1998). The risk of non-chromosomal birth anomalies was 33% greater for women who lived within 3 km of a landfill site than women living between 3 km and 7 km from the site, and the risk decreased with increasing distance from the site.

The teratogenic potential of two trimethoprim-sulfonamide combinations (used to treat infections of the urinary tract, and respiratory and gastrointestinal systems) during pregnancy was examined in a case-control study using data from the Hungarian Congenital Abnormality Registry (HCAR) (Czeizel et al. 2001). The study found that the risk of cardiovascular birth anomalies and multiple birth anomalies may be increased by the use of cotrimoxazole by women during the second and third months of pregnancy, and that the use of trimethoprim-sulfamethazine during pregnancy may also have some association with cardiovascular birth anomalies.

Owen et al. (2000) used data from the Victorian Birth Defects Register to examine potential contributing factors associated with neural tube defects. Infant and maternal characteristics were investigated and it was found that the risk of neural tube defects was significantly higher for epileptic women, multiple births, teenage mothers, and women with three or more previous pregnancies.

Bower et al. (2002) used data from the Western Australian Birth Defects Registry to report on trends in neural tube defects in Western Australia. It was found that there was a 30% decrease in neural tube defects between 1996 and 2000. Increased periconceptional folate intake in response to fortification of selected foods and health promotion campaigns was thought to have led to the fall in neural tube defects.

Parental work as a potential risk factor for birth anomalies was examined using data from the National Birth Defects Register in Singapore (Shi et al. 2002). No association was found between type of parental work and birth anomalies in this study. Blatter et al. (2000) examined the association between spina bifida and parental occupation using data from birth anomalies registers in Sweden, Spain and Hungary. They found an increased risk of spina bifida for women in agricultural occupations in Sweden and in Spain, but not in Hungary. Associations of other occupations with spina bifida were inconsistent.

Inform policy and decision making

Well-informed community discussion and decision making about birth anomalies requires high quality statistics and information. Birth anomalies monitoring systems provide information on the prevalence of birth anomalies and on the characteristics of those affected. This information can be used in the planning and evaluation of services

and interventions aimed at reducing the occurrence of birth anomalies or at reducing their impact.

For example, potential contributing risk factors for neural tube defects were examined using data from the Victorian Birth Defects Register (Owen et al. 2000). This study identified subgroups of the population that were at higher risk for neural tube defects and that would therefore benefit from targeted education and screening programs. The study also produced baseline data that could be used for evaluating the effectiveness of such programs.

Indicators of the health of mothers and babies

The prevalence of birth anomalies is a key indicator of adverse perinatal outcome and of the performance of peri-conceptual counselling and pregnancy care. Birth anomalies monitoring systems that collect nationally consistent, high quality data are critical for providing such information.

Scope of birth anomalies monitoring systems

Introduction

The scope of a birth anomalies monitoring system depends on its purpose and on the available resources (Cordero 1992). It is important to clearly define the scope (Lynberg & Edmonds 2004) because it affects the prevalence of birth anomalies and comparability with other collections. The scope of a birth anomalies monitoring system can be defined by the period of detection of birth anomalies, the sources of notification, the conditions that are included, additional data items that are included (e.g. characteristics of the infant/mother/father; risk factors), and the population.

Period of detection of birth anomalies and sources of notification

The prevalence of birth anomalies is affected by the sources of notification and also by the period within which birth anomalies are diagnosed and subsequently notified. For example, there may be under-ascertainment of some birth anomalies if the scope of the collection is restricted to the perinatal period as birth anomalies diagnosed at a later stage would not be included (EUROCAT 2002). Birth anomalies can be detected prenatally through prenatal diagnostic tests. The prevalence of some birth anomalies would be affected by the availability and use of prenatal screening programs and diagnostic testing services and by whether the results of prenatal diagnostic tests are notified to birth anomalies collections (EUROCAT 2002).

In Australia, the period of detection varies considerably among the state and territory collections and ranges from prenatal diagnosis to detection up to 15 years of age (see Chapter 4). The period of detection also varies internationally. However, over half of the countries reporting to the International Clearinghouse for Birth Defects Monitoring Systems (ICBDMS) have a maximum age at diagnosis of 1 year or more (ICBDMS 2003).

Terminations of pregnancies with birth anomalies

The prevalence of some birth anomalies is also affected by whether terminations of pregnancy following prenatal diagnosis of a birth anomaly are undertaken, and whether these birth anomalies are notified to birth anomalies data collections.

A number of Australian studies have found that prenatal diagnosis of birth anomalies and subsequent terminations of pregnancy have contributed to the decline in the birth prevalence of neural tube defects, while the total prevalence has remained stable over time (Chan et al. 1993; Owen et al. 2000).

Limiting the gestational age at which terminations for birth anomalies are notified can also affect prevalence. Ethan and Canfield (2002) in a study using data from the Texas Birth Defects Registry found that excluding cases with birth anomalies that were electively terminated before 20 weeks gestation resulted in incomplete counts and rates of birth anomalies. This was especially true for conditions that are more commonly detected and terminated before 20 weeks gestation (e.g. neural tube defects). They noted that the exclusion of these cases has implications for prevention and research.

In Australia, terminations of pregnancies with birth anomalies are not notified in all jurisdictions. Of those that receive notifications, not all receive notifications of terminations of pregnancy at <20 weeks gestation (see Chapter 4).

Multiple sources of notification

Multiple sources of notification increase the completeness of the collection and the precision and accuracy of diagnoses, but also increase the resources and time needed to compile the collection (Holtzman & Khoury 1986; Correa-Villasenor et al. 2003; Lynberg & Edmonds 2004). Sources of notification of birth anomalies include forms designed to collect information on birth anomalies, admitted patient records, birth certificates, chromosome laboratories, genetics, cardiac and other specialty clinics, death certificates and pathology reports.

Conditions for inclusion

The scope of the collection is also defined by the conditions that are included. This could be all birth anomalies or a particular set of birth anomalies (Holtzman & Khoury 1986). The selection of the conditions to be included may depend on the period of detection selected because some conditions may only be apparent or may have resolved after the birth episode, for example (Holtzman & Khoury 1986).

Minor birth anomalies are not usually included in birth anomalies monitoring systems, unless they are present with other birth anomalies. EUROCAT has developed a standard list of birth anomalies for exclusion (EUROCAT 2002), as has the Centers for Disease Control (CDC) (Lynberg & Edmonds 2004).

Currently, the conditions included in the National Congenital Malformations and Birth Defects Data Collection in Australia are those listed in the chapter on Congenital anomalies in the International Classification of Diseases, 9th Revision, Clinical Modification (ICD-9-CM).

The International Clearinghouse for Birth Defects Monitoring Systems (ICBDMS) collects information on all birth defects but defined 35 'sentinel' conditions for international reporting in the 2003 Annual Report. The selection of this set of conditions is arbitrary and may change from year to year (ICBDMS 2003).

Classification of conditions

The prevalence of birth anomalies and the utility of birth anomalies monitoring systems can be affected by the accuracy, specificity and completeness of the coding of birth anomalies (Rasmussen & Moore 2001). This can be exacerbated by the rarity of some birth anomalies, whereby a small number of incorrectly coded cases can influence the rates markedly (Rasmussen & Moore 2001).

Many birth anomalies monitoring systems use a coding system based on the International Classification of Diseases (ICD-9) which was developed by the World Health Organization and was designed to enable international comparability for mortality statistics. The ninth revision does not have sufficient detail for most birth anomalies monitoring systems (Rasmussen & Moore 2001). A clinical modification (ICD-9-CM) was subsequently developed and provides further detail on birth anomalies, although some birth anomalies are still not classified with sufficient precision or specificity in ICD-9-CM for use in birth anomalies monitoring systems (Rasmussen & Moore 2001). An Australian version of ICD-9-CM was developed by the National Coding Centre (NCC 1995).

To enable birth anomalies to be classified with greater precision and specificity, the Royal College of Paediatrics and Child Health (formerly, the British Paediatric Association (BPA)) modified ICD-9-CM by adding a fifth character to the fourth character level of ICD-9-CM (Rasmussen & Moore 2001). ICD-9-CM codes mainly have four characters but some have five characters. Therefore, the two classifications are identical at the fourth character level, but where a fifth character level exists in ICD-9-CM, they are generally different.

The NPSU developed modified BPA Classification of Diseases codes for malformations syndromes, limb reduction defects and other selected birth anomalies, which enabled even more specific classification of each birth anomaly. However, this classification is no longer used.

The International Statistical Classification of Diseases and Related Health Problems, 10th Revision, Australian Modification (ICD-10-AM) was introduced in July 1998 and the specificity for birth anomalies has been enhanced in the third edition (introduced in July 2002) and the fourth edition (to be introduced in July 2004). The ICD-10-AM is used by a number of jurisdictions in Australia (see Chapter 4).

Characteristics and risk factors

The aetiology of congenital malformations is multifactorial and in many cases unknown (Brent 2001). The consideration of a number of confounding factors is required in the study of birth anomalies. These factors include: family history, maternal age, weight and height, race, maternal conditions (e.g. diabetes and epilepsy), diet, and smoking and other teratogenic exposure (Milunsky 1996).

Currently, limited information on maternal characteristics and no information on paternal characteristics is requested for the National Congenital Malformations and Birth Defects Data Collection. Collecting more maternal information and some paternal information would increase the utility of the collection.

Maternal Indigenous status is currently requested, but the quality of the data is limited. Paternal Indigenous status is not collected. Since Aboriginal and Torres Strait Islander peoples suffer a greater burden of ill health than other Australians (AIHW 2002), it is important to work towards improving the quality of these data and considering broadening the scope to include paternal Indigenous status.

National birth anomalies monitoring

A study of birth anomalies prevalence rates in Arkansas found that residents along the state border travel to health care facilities outside of the state for obstetric and paediatric care and that the prevalence of birth anomalies is therefore significantly underreported (Mosley et al. 2002). They noted that the exchange of information between birth anomalies surveillance systems in different states is prevented because of inconsistencies in methodologies, the lack of formal agreements for data-sharing and in some cases state legislation that restricts the sharing of data. They suggested that efforts towards developing a national system should be made to enable the true characterisation of birth anomalies.

In Australia, most states and territories are not notified of birth anomalies detected in other jurisdictions, so they cannot determine the prevalence of birth anomalies among their residents. A national birth anomalies system would enable cross-border issues to be addressed. It would also provide better information on rare conditions.

A national birth anomalies monitoring system would encourage standardisation of scope, definitions and classifications, and enable high quality, consistent data to be reported. It would also facilitate the meeting of international reporting requirements.

Collaborations

Introduction

The National Congenital Malformations and Birth Defects Data Collection could be enhanced by developing partnerships with other organisations to integrate the available information on birth anomalies into one reporting system.

Newborn screening

The conditions tested for using newborn screening are not monitored at a national level in Australia (NPHP 2002). It was noted in the National Public Health Partnership's report on public health surveillance of genetic disorders (NPHP 2002) that a national approach to newborn screening would ensure more equitable access to a consistent range of tests and testing processes and would also facilitate consistent reporting and data collection.

Newborn screening began in the 1960s and involves the analysis of a blood sample which has been dried on filter paper (the Guthrie test). The aim is early detection of treatable conditions to enable the prevention or treatment of these conditions (Wilcken 2003). In Australia, all states and territories have a newborn screening program that targets all newborns. All states and territories test for phenylketonuria, congenital

hypothyroidism and cystic fibrosis, and all states and territories except Victoria test for galactosemia (NPHP 2002).

Tandem mass spectrometry was introduced in the 1990s and has enabled over 30 additional disorders to be detected (NPHP 2002). It provides an extended newborn screening program. In Australia, this technology has been introduced in all states and territories except Western Australia and Queensland (NPHP 2002).

Newborn screening tests are conducted at five screening laboratories across Australia. Laboratories are located in New South Wales (covering the Australian Capital Territory), Victoria, Queensland (covering part of the Northern Territory), Western Australia and South Australia (covering Tasmania and part of the Northern Territory) (NPHP 2002).

The four conditions identified under the current newborn screening program are reported to the state birth defects registers and therefore could easily be incorporated into a national reporting system. The extended program could be considered in collaboration with the Joint Human Genetics Society of Australasia/Royal Australian College of Physicians Newborn Screening Committee.

The Australian Paediatric Surveillance Unit

The Australian Paediatric Surveillance Unit (APSU) is a unit of the Royal Australian College of Paediatrics. APSU conducts retrospective national epidemiological surveillance on selected uncommon paediatric conditions. The information is disseminated to health professionals, state and federal governments and consumers.

The monitoring system relies on the cooperation of paediatricians to notify APSU. Selected conditions are studied for a period of 3 years and must satisfy three criteria:

1. The condition studied must be sufficiently infrequent to ensure the APSU surveillance system is not over burdened.
2. The condition studied must require referral to a paediatrician or related specialist.
3. The condition studied must provide information that satisfies the study aims and is not available from another source (Williams & Elliott 1998).

APSU provides information on the aetiology of the condition studied, risk factors, seasonal occurrence, disease severity and effect on the child (Williams & Elliott 1998).

A component of the APSU work program has included the surveillance of rare birth anomalies, including congenital adrenal hyperplasia and congenital and neonatal varicella. Other studies include congenital rubella, congenital cytomegalovirus, fetal alcohol syndrome and adverse effects associated with the use of complementary or alternative medicine (APSU 2004).

It has been proposed that the APSU, state and territory Birth Defects Registers and the NPSU could collaborate better to maximise ascertainment of cases and to validate data collection (see Appendix 2).

Assisted conception

The Australia and New Zealand Assisted Reproduction Database (ANZARD) was implemented in 2002 to monitor Assisted Reproductive Technology (ART) in Australia and New Zealand. The ANZARD is industry-based and is compiled by the NPSU from data provided from participating fertility clinics in Australia and New Zealand.

Assisted conception techniques are commonly available to assist couples to become pregnant who previously had difficulty conceiving naturally.

Using linked data from Western Australian registers on births, births after assisted conception and birth anomalies, Hansen et al. (2002) found that babies conceived with ART were more than twice as likely to have a major birth anomaly diagnosed by 1 year of age compared to babies conceived naturally. The increase in risk was similar for babies conceived after standard IVF and after intracytoplasmic sperm injection (ICSI). The use of ART in Australia continues to increase, so it is important that accurate information is available on the risk of birth anomalies associated with ART.

Information on birth anomalies is collected in the ANZARD; however, the data are of poor quality and have therefore not been reported on. Linkage with the national birth anomalies data collection could provide a mechanism for improving the quality of the available data and of improving the utility of both collections.

International reporting

The International Clearinghouse for Birth Defects Monitoring Systems (ICBDMS) was established in 1974 and since 1986 has had an official relation with the World Health Organization (WHO). The main objectives of the ICBDMS are:

- the exchange of routine information on the prevalence of congenital malformations;
- to undertake collaborative epidemiological research; and
- to provide expert consultation and assistance for existing monitoring systems to investigate outbreaks and for the establishment of new monitoring systems (ICBDMS 2003).

The advantages of international collaboration for the surveillance of birth anomalies include increased sample size and diversity of populations and environments.

Comparison with other registers may help to confirm a change in the prevalence of a birth anomaly (Castillo et al. 1986). International collaboration also allows for the study of birth anomalies across diverse gene pools and environments so may enable genetic and teratogenic factors to be examined more effectively.

Participation in the ICBDMS improves the quality and utility of registers because it encourages standardisation of the definitions and classification systems used and of the conditions monitored. The National Congenital Malformations and Birth Defects Data Collection should aim to have internationally comparable data and to incorporate international standards (e.g. definitions and conditions) where possible.

Over 40 birth anomalies monitoring programs worldwide participate. Australia has three member programs: the National Congenital Malformations and Birth Defects Data Collection, the Victorian Birth Defects Register and the Western Australian Birth Defects Registry. National data have not been contributed to the ICBDMS recently, however, because of concerns related to this review (ICBDMS 2003). The new Australian Birth Anomalies System will provide nationally consistent data that can be used for international reporting.

4 Survey

Introduction

As part of the review process, the NPSU conducted a survey to gain information about the state and territory birth anomalies data collections. The survey was sent to the managers of the respective state and territory collections in April 2003 and additional questions were sent in June 2003. All managers returned completed responses. A copy of the survey is included in Appendix 1.

Results

The results of the survey are summarised below. The results show that there is variation among the states and territories in the scope of their birth anomalies data collections, the sources of birth anomalies notifications, the definitions and classifications used, the method of data collection, the available resources, the publication of data, and the provision of data to the NPSU for national collation, surveillance and reporting.

The utility of the National Congenital Malformations and Birth Defects Data Collection is limited by the variation in birth anomalies data among the states and territories. National consistency in the scope, definitions and classifications used would greatly improve the quality and comparability of these data.

Some of these results were presented at the workshop which was held in May 2003 and is discussed in Chapter 5.

Purpose and utilisation

The states and territories were asked to indicate the purpose of their birth anomalies data collections and also to provide information on the utilisation of the data. The responses indicate that the main purpose of the collections is to monitor the occurrence of birth anomalies. The data were also reported to be used for reporting (at various levels, e.g. area health service, state, national and international), planning of services, evaluation of programs, identification of clusters, education and research. The data were reported to be used by various entities including government organisations, special interest groups, researchers and the general public.

Definitions

Birth anomalies

The NPSU defines birth anomalies as ‘anatomical defects or chromosomal abnormalities that are present at birth’.

The states and territories were asked to provide the definition of ‘congenital malformation’ and/or ‘birth defect’ used in their jurisdiction. Tasmania indicated that these are not defined. Queensland noted that they are reviewing their definition.

The definitions varied among the states and territories in terms of the types of birth anomalies included and the timing of the presentation or detection of birth anomalies. Table 1 shows that six jurisdictions included structural anomalies in their definition and four jurisdictions included biochemical anomalies (i.e. PKU, etc). Chromosomal anomalies were included by four jurisdictions, anatomical anomalies and genetic anomalies were included by two jurisdictions, and functional anomalies were included by one jurisdiction. The types of anomalies included may not be specifically stated in the definition used by each jurisdiction.

Birth anomalies present at birth were included in the definitions for New South Wales, Victoria, Queensland, the Australian Capital Territory and the Northern Territory, but for Queensland and the Northern Territory, only those birth anomalies detected prior to separation (from hospital) were included. Queensland noted that their definition will also include those cases where a prenatal diagnosis was made and the birth anomaly is present at birth. Birth anomalies of prenatal origin were included in the definitions for Victoria, Western Australia and South Australia and birth anomalies detected during pregnancy were included for New South Wales.

Table 1: Types of birth anomalies included in definitions of birth anomalies, by state and territory, 2003

Type of birth anomaly	NSW	Vic	Qld	WA	SA	Tas	ACT	NT	AIHW NPSU
Structural	✓	✓		✓	✓	n.a.	✓	✓	
Anatomical			✓			n.a.	✓		✓
Functional		✓				n.a.			
Chromosomal		✓	✓	✓	✓	n.a.			✓
Genetic				✓	✓	n.a.			
Biochemical	✓	✓		✓	✓	n.a.			

n.a. not available.

The Northern Territory used the National health data dictionary (NHDC 2001) definition of ‘Congenital malformations’.

These results show that there is no nationally consistent definition of birth anomalies.

Data collection

Legislation

Information on whether the notification of birth anomalies to the state and territory collections is covered by legislation was sought. Table 2 shows that all jurisdictions except the Australian Capital Territory and the Northern Territory have legislation which covers notification of birth anomalies. The coverage of this legislation varies among the states and territories, however, and legislation does not cover all sources of data in some jurisdictions. For example, notification of terminations of pregnancies with birth anomalies is not enacted through legislation in some jurisdictions. Therefore, although the states have legislation which covers notification of birth anomalies, notification is voluntary from some sources.

Table 2: Legislation for notification of birth anomalies, by state and territory, 2003

Legislation	NSW	Vic	Qld	WA	SA	Tas	ACT	NT
Yes	✓	✓	✓	✓	✓	✓		
No							✓	✓

Scope

Period of detection

The states and territories were asked to indicate the period of detection within which notifications of birth anomalies are included in their collections. Table 3 shows that this varies considerably among the states and territories.

Birth anomalies detected prenatally were reported to be notified in all jurisdictions except Queensland and Tasmania. Queensland reported that birth anomalies diagnosed prenatally are notified if they are apparent at birth. All jurisdictions reported that birth anomalies detected during the birth episode are notified. However, Queensland noted that birth anomalies are not notified if they are detected during the birth episode and the baby is aged greater than 28 days. For Tasmania, only birth anomalies detected during the birth episode are notified. However, they indicated that they will be able to move to the notification of birth anomalies detected between birth and 28 days in the near future. Birth anomalies are notified if they are detected up to a maximum age of 1 year for New South Wales, the Australian Capital Territory and the Northern Territory, up to a maximum age of 5 years for South Australia, up to a maximum age of 6 years for Western Australia and up to a maximum age of 15 years for Victoria.

New South Wales is the only jurisdiction that collects date of diagnosis. Western Australia reported that age at diagnosis is collected. Therefore, New South Wales and Western Australia are the only states for which data could be reported based on age at diagnosis. Subsequent to the survey, Victoria and South Australia indicated that they intended to include date of diagnosis on their systems in the future. Currently, although they collect data on birth anomalies detected up to a maximum of 15 and 5 years respectively, they cannot easily report the age at diagnosis because this information is not included on their systems.

The extent to which terminations of pregnancy are diagnosed with birth anomalies and the extent to which these are notified to the states and territories is unclear. Only the

Australian Capital Territory and the Northern Territory indicated that there are policies for the disposal of the products of conception following terminations of pregnancy. These policies were reported to be hospital based.

Table 3: Detection period for notification of birth anomalies, by state and territory, 2003

Detection period	NSW	Vic	Qld ^(a)	WA	SA	Tas	ACT	NT
Prenatal	✓	✓	✓ ^(c)	✓	✓ ^(d)		✓	✓
Birth ^(b) to discharge from birth episode	✓	✓	✓	✓	✓	✓	✓	✓
Birth ^(b) to 28 days of age	✓	✓	✓	✓	✓		✓	✓
Birth ^(b) to 1 year of age	✓	✓		✓	✓		✓	✓
Birth ^(b) to 5 years of age		✓		✓	✓			
Birth ^(b) to 6 years of age		✓		✓				
Birth ^(b) to 15 years of age		✓						

(a) If the baby remains in hospital after birth for more than 28 days, the detection period ends at 28 days.

(b) Birth includes stillbirths of at least 20 weeks gestation or 400 g birthweight.

(c) Where a prenatal diagnosis was made and the birth anomaly is present at birth.

(d) Only if termination of pregnancy performed.

Note: Periods are not mutually exclusive.

Period of collection

The states and territories were also asked to indicate the scope of their collections in terms of the collection period. All states and territories indicated that their collections were based on calendar years.

Table 4: Number of years of complete data and latest complete year of data held, by state and territory, 2003

Year	NSW	Vic	Qld	WA ^(a)	SA	Tas	ACT	NT
Number of years of complete data								
None								
1–3 years								
4–5 years						✓		
>5 years	✓	✓	✓	✓	✓		✓	✓
Latest complete year of data								
Pre-2000							✓	
2000	✓		✓					
2001		✓		✓	✓	✓		✓
2002								

(a) WA reported that their latest year of complete data was 1996. This is because they collect data on birth anomalies detected up until the age of 6 years, so 1996 was the latest complete birth cohort. However, the latest full calendar year that they have data for is 2001.

Table 4 shows the number of years of complete data in each state and territory collection and which year the latest complete year of data is held for. All jurisdictions except Tasmania indicated that they held more than 5 years of complete data. Most jurisdictions indicated that their latest year of data was for 2000 or 2001.

Cross-border notifications

All jurisdictions indicated that babies born in their state or territory were notified to their collection regardless of the baby's state or territory of usual residence. Western Australia and the Northern Territory indicated that residents, who were born outside of Western Australia or the Northern Territory, respectively, were notified. Western Australia stated that these notifications were not included in the data provided to the NPSU.

Notification of terminations of pregnancies with birth anomalies

The states and territories were asked to indicate whether terminations of pregnancy following adverse prenatal tests were notified. All jurisdictions except the Australian Capital Territory and the Northern Territory indicated that they were (Table 5).

The states and territories were then asked to indicate the gestational age at which terminations of pregnancy following adverse prenatal tests were notified. All states except Queensland and Tasmania indicated that terminations of pregnancy at <20 weeks following adverse prenatal tests were notified and all states indicated that terminations of pregnancy at ≥20 weeks following adverse prenatal tests were notified (Table 5). The Australian Capital Territory indicated that terminations of pregnancy at ≥20 weeks following adverse prenatal tests would be reported on the mother's record in the admitted patient data and classified as an induction of labour with a birth outcome of either stillbirth or neonatal death.

Table 5: Notification of terminations of pregnancies with birth anomalies, by state and territory, 2003

Notifications		NSW	Vic	Qld	WA	SA	Tas	ACT ^(a)	NT ^(b)
Termination of pregnancy following adverse prenatal screening/diagnostic tests	Yes	✓	✓	✓	✓	✓	✓		
	No							✓	✓
Termination of pregnancy <20 weeks	Yes	✓	✓		✓	✓			
	No			✓			✓	✓	✓
Termination of pregnancy ≥20 weeks	Yes	✓	✓	✓	✓	✓	✓	✓	
	No								✓

(a) The ACT indicated that terminations of pregnancy ≥20 weeks would be reported on the mother's record in the admitted patient care data, and classified as an induction of labour with a birth outcome of either stillbirth or neonatal death.

(b) Information on adverse prenatal screening/diagnostic tests and terminations of pregnancy <20 weeks was collected until 2000.

Trisomies notifications

The states and territories were asked to indicate if a clinical diagnosis of a trisomy has to be confirmed with cytogenetic testing before the case can be included in the collection. New South Wales, Western Australia and South Australia indicated that they do. New South Wales noted that if a trisomy is not confirmed with cytogenetic testing then it is reported using the syndrome name (e.g. Down Syndrome). Western Australia noted that they seek cytogenetic confirmation of trisomies, but if this is not available, then a trisomy would still be included in their collection based on a clinical diagnosis. Queensland noted that if a clinical diagnosis of trisomy is made they do not require laboratory confirmation of the diagnosis. However, any suspected trisomies (as well as other suspected anomalies) are confirmed before inclusion in the data collection.

Confirmed diagnosis

The states and territories were asked to indicate whether there is a set of criteria for the diagnosis of each birth anomaly and if these criteria have to be met to confirm the diagnosis. New South Wales and Western Australia reported that they have specific criteria for the diagnosis of some birth anomalies. Where criteria exist, cases must meet them to be included in the collection.

Follow-up of notifications

The states and territories were asked to provide information on whether follow-up of notifications is conducted. New South Wales and Victoria indicated that follow-up was conducted for confirmation of a prenatal diagnosis. New South Wales also indicated that probable, likely or suspected conditions are confirmed before inclusion on the database. Queensland indicated that they confirm 'suspected' birth anomalies at birth. Western Australia indicated that active follow-up was conducted for confirmation of a prenatal diagnosis and to obtain missing information on the original notification, or to collect details for those anomalies for which specific criteria must be met to be included, and that passive follow-up was conducted for up to 6 years. South Australia indicated that

follow-up was conducted for confirmation of terminations following prenatal diagnosis of birth anomalies and where there was uncertainty about the inclusion of a case. Tasmania and the Australian Capital Territory indicated that no follow-up was conducted and the Northern Territory did not provide an indication.

Sources of notifications

The states and territories were asked to indicate the sources from which birth anomalies are notified to their collections (Table 6).

All states and territories reported that they receive notifications of birth anomalies from perinatal/midwives form data. All jurisdictions except Queensland and Tasmania received notifications from hospital morbidity data, and all jurisdictions except Tasmania, the Australian Capital Territory and the Northern Territory received notifications from medical officers and cytogenetic/pathology reports. Western Australia reported the highest number of sources of notification of birth anomalies.

The states and territories were also asked to indicate if notification of birth anomalies was part of a routine process(es). All states and territories indicated that it was. For most jurisdictions there were a number of routine processes (i.e. associated with different data sources) and the frequency with which birth anomalies were notified differed with each data source (i.e. monthly, quarterly, annually).

Format of notifications

The states and territories were asked to indicate the format(s) in which notifications are made. All jurisdictions except the Northern Territory indicated that notifications are made using state sanctioned notification forms. Most jurisdictions indicated that notifications are also made in other formats, depending on the source of the notification.

Multiple notifications

All states and territories except Tasmania indicated that they sometimes receive multiple notifications of the same individual. They also indicated that they can identify multiple notifications and adjust for them.

Table 6: Sources of notifications, by state and territory, 2003

Sources of notifications	NSW	Vic	Qld	WA	SA	Tas	ACT	NT
Routine data collections								
Hospital morbidity data	✓	✓		✓	✓		✓	✓
Perinatal/midwives form data ^(a)	✓	✓	✓	✓	✓	✓	✓	✓
Death certificates/mortality data		✓	✓	✓	✓			
Termination of pregnancy form data		✓			✓			
Primary healthcare staff								
General practitioners				✓				
Early childhood centre staff		✓		✓				
Rural healthcare workers				✓				
Other								
Disability services staff								
				✓	✓			
Medical officers								
Paediatricians	✓	✓	✓	✓	✓			
Other	✓	✓	✓	✓	✓			
Screening and diagnosis services								
Cytogenetic/pathology reports	✓	✓	✓	✓	✓			
Ultrasound reports				✓				
Newborn/genetic screening/diagnosis reports	✓	✓		✓				

(a) Includes perinatal deaths notified through state-based perinatal/midwives forms.

Linkage with other state and territory based data collections

The states and territories were asked to indicate if their birth anomalies data were linked to other state and territory based data collections. All jurisdictions indicated that their birth anomalies data were either linked to or able to be linked to their perinatal data collection (Table 7). Western Australia indicated that their birth anomalies data were also able to be linked to their assisted conception data collection and that they were linked to a number of other collections, including their hospital morbidity collection. South Australia indicated that their birth anomalies data were able to be linked to their assisted conception data collection and to their Cerebral Palsy Register, and possibly their hospital morbidity collection. The Australian Capital Territory and the Northern Territory indicated that their birth anomalies data were also linked to their hospital morbidity collections.

Table 7: Linkage with other data collections, by state and territory, 2003

Data collections	NSW	Vic^(a)	Qld	WA^(b)	SA^(c)	Tas	ACT	NT^(d)
Perinatal	✓	✓	✓	✓	✓	✓	✓	✓
Assisted conception				✓	✓			
Hospital morbidity				✓	✓		✓	✓

(a) Not routinely linked to the perinatal database, but able to be linked to it for research purposes.

(b) Able to be linked to the assisted conception database. Birth anomalies data are also linked to cerebral palsy database, birth & death registrations, census district data and intellectual disability data.

(c) Able to be linked to the assisted conception database, Cerebral Palsy Register and possibly to the hospital morbidity database.

(d) Not routinely linked to the perinatal database, but able to be linked to it.

Resources

Funding

The states and territories were asked to indicate the source of funding for their birth anomalies data collections and also whether the collection was funded exclusively or as part of another data collection.

Table 8 shows that birth anomalies data collections are funded by state and territory governments in all states and territories. In South Australia, funding for the birth anomalies data collection is provided to the Women's and Children's Hospital. New South Wales, Western Australia and South Australia indicated that their birth anomalies data collections were funded exclusively. The other jurisdictions indicated that their collections were funded as part of their perinatal data collections.

Staffing

The states and territories were asked to indicate the number of full-time equivalent (FTE) staff allocated to their birth anomalies data collections. This varied among the states and territories (Table 8). Queensland, Tasmania and the Northern Territory indicated that there was no specific allocation of staff for the collection of birth anomalies data. The Australian Capital Territory indicated that <1 FTE was allocated. New South Wales, Western Australia and South Australia indicated that 1.4 FTE, 2.4 FTE and 2.0 FTE were allocated respectively. Victoria indicated that about 0.5 FTE is specifically allocated to the birth anomalies data collection but that staff who work on the perinatal data collection also contribute to their Birth Defects Register.

State and territory publications

The states and territories were asked to indicate the year of the most recent state or territory based report published using data from their birth anomalies data collections and the most recent year of data used in that report.

The Australian Capital Territory indicated that they published a report in 2003. Victoria, Queensland, Western Australia and South Australia indicated that they published reports in 2002. New South Wales published a report in 2001 and Tasmania published a report in 1999. The Northern Territory indicated that they have not published any territory-based reports on birth anomalies (Table 9).

The latest year of data used in the reports was 2000 for Victoria, Queensland and South Australia, 2001 for New South Wales and Western Australia, and 1999 for Tasmania and the Australian Capital Territory (Table 9).

Table 8: Source and mode of funding, and staffing of birth anomalies data collections, by state and territory, 2003

	NSW	Vic ^(a)	Qld	WA	SA	Tas	ACT	NT
Source of funding								
State/territory government	✓	✓	✓	✓	✓	✓	✓	✓
Institutional core funding								
Mode of funding								
Exclusively funded	✓			✓	✓			
Part of another data collection ^(b)		✓	✓			✓	✓	✓
Staffing								
No specific allocation			✓			✓		✓
<1 FTE							✓	
1.0–1.4 FTE	✓							
2.0–2.4 FTE				✓	✓			
Other		✓						

(a) Victoria indicated that staffing of the birth defects register is 0.5 FTE, but that staff from the Perinatal Data Collection Unit also contribute.

(b) The perinatal data collection.

Table 9: Year of latest birth anomalies report and latest year of data used in the report, by state and territory, 2003

Year	NSW	Vic	Qld	WA	SA	Tas	ACT	NT
Year of latest report								
1999						✓		n.a.
2000								n.a.
2001	✓							n.a.
2002		✓	✓	✓	✓			n.a.
2003							✓	
Latest year of data								
1999						✓	✓	n.a.
2000		✓	✓		✓			n.a.
2001	✓			✓				n.a.
2002								n.a.

n.a. not available.

Provision of data to the AIHW National Perinatal Statistics Unit

The NPSU requests data on major birth anomalies diagnosed in live born infants in the first 28 days, or in stillbirths of at least 20 weeks gestation or 400 g birth weight (i.e. the perinatal period). Data on terminations of pregnancies with birth anomalies are also requested.

An agreed set of data elements is requested which includes maternal and baby characteristics, and information about birth anomalies. All of the data elements requested are also included in the perinatal data collection except data source (i.e. source of notification), method of prenatal diagnosis, pregnancy outcome, sequential order of the birth anomaly, birth anomaly code, birth anomaly position and birth anomaly description.

Scope of data provided

The states and territories were asked to indicate the period of detection for which they provide birth anomalies data to the NPSU. All states and territories except Queensland and the Northern Territory indicated that they provide data for birth anomalies detected in the perinatal period. Queensland indicated that they provide data for birth anomalies detected between birth and discharge from the birth episode. This is consistent with the National health data dictionary definition of 'Congenital malformations' (NHDC 2001). The Northern Territory indicated that they provide data for birth anomalies detected between birth and 1 year of age.

Classification of birth anomalies

The states and territories were asked to indicate the classification they currently use to code birth anomalies. The classification used varies among the states and territories. Victoria, Western Australia and South Australia use ICD-9-BPA, New South Wales uses an extension of the BPA classification, Tasmania uses the 2nd edition of ICD-10-AM and the other jurisdictions use the 3rd edition of ICD-10-AM (Table 10).

Table 10: Classification used to code birth anomalies, by state and territory, 2003

Classification	NSW	Vic	Qld	WA	SA	Tas	ACT	NT
ICD-9-BPA		✓		✓	✓			
Extension of BPA codes	✓							
ICD-10-AM 1st edition								
ICD-10-AM 2nd edition						✓		
ICD-10-AM 3rd edition			✓				✓	✓

Frequency of provision

All states and territories except Queensland currently provide birth anomalies data to the AIHW National Perinatal Statistics Unit on an annual basis. Queensland currently provides these data quarterly. Two states indicated that they could provide these data more frequently. New South Wales indicated that they could provide these data monthly and Western Australia indicated that they could provide these data quarterly.

Media and format of data provided to the AIHW National Perinatal Statistics Unit

All states and territories except Queensland and South Australia currently provide birth anomalies data to the NPSU electronically (in the format requested by the NPSU) (Table 11). In the past, Queensland and South Australia provided these data to the NPSU using paper-based forms. Queensland have developed an electronic extract to satisfy NPSU requirements but are awaiting the outcome of this review to determine the final format for the extract.

Table 11: Format of provision of data to the AIHW National Perinatal Statistics Unit, by state and territory, 2003

Media	NSW	Vic	Qld	WA	SA ^(a)	Tas	ACT	NT
Paper-based			✓					
Electronic	✓	✓		✓	✓	✓	✓	✓

(a) Data for terminations of pregnancy are provided on paper-based forms.

Final data availability

The states and territories were asked to indicate the timeframe for providing final birth anomalies data after the end of the collection period (Table 12). For birth anomalies detected in the perinatal period, final data could be provided by 18 months after the end of the collection period for most jurisdictions. For birth anomalies detected up to 1 year of age, final data could be provided within 18 months of the end of the collection period for three jurisdictions (New South Wales, Victoria and the Northern Territory). Queensland indicated that they would be unable to provide these data. Subsequent to the survey, Victoria and South Australia indicated that they were unable to easily and accurately report cases with birth anomalies detected up to 1 year of age because they do not have date or age of diagnosis on their systems. They indicated, however, that they would be able to do this in the future.

The reasons for delays in data provision were reported to include variation in the timeframes for the receipt of data from different data sources, linkage with other data collections, follow-up and lack of resources.

All jurisdictions except the Australian Capital Territory and the Northern Territory indicated that updates could be provided to the NPSU.

State and territory comments

The national birth anomalies data collection

The states and territories were asked to comment on whether a national birth anomalies data collection is needed. All states and territories agreed that the national collection should exist.

The states and territories were asked to comment on the purpose of the national collection. Most states and territories indicated that the National Congenital Malformations and Birth Defects Data Collection was not currently useful for monitoring birth anomalies. They indicated that the purposes of the national data collection include: being a national repository for birth anomalies data; surveillance of birth anomalies; state and territory comparisons; report on rare malformations; international reporting; standardisation; planning; epidemiology; and research.

Birth Defects Series

The states and territories were asked to indicate who they thought the audience for the Birth Defects Series of reports was. The jurisdictions indicated that the audience included researchers, service planners, policy makers, health professionals and small sections of the general public. They indicated that reporting should be on an annual basis.

Table 12: Timeframe for availability of final birth anomalies data, by state and territory, 2003

Timeframe	NSW	Vic	Qld ^(a)	WA	SA ^(b)	Tas ^(c)	ACT	NT
Detected in the perinatal period^(d)								
Up to 6 months	✓							
7–12 months			✓		✓	✓		
13–18 months		✓					✓	✓
19–24 months				✓				
>24 months								
Detected up to 1 year of age								
Up to 6 months			n.a.		
7–12 months			n.a.		
13–18 months	✓	✓	n.a.		✓
19–24 months			..	✓	..	n.a.		
>24 months			n.a.	✓	

(a) Queensland indicated that provision of data on birth anomalies detected up to 1 year of age would not be possible.

(b) South Australia indicated that the date of diagnosis/notification is not recorded, although notifications are collected up to the child's 5th birthday.

(c) Tasmania indicated that the timeframe for availability of final birth anomalies data detected up to 1 year of age is not known as this would depend on the volume and source of the notifications.

(d) From birth (livebirths and stillbirths of 20 weeks gestation or more or birth weight >400 g) to 28 days of age.

n.a. not available.

.. not applicable.

5 Workshop

Introduction

A 2-day workshop was held in May 2003 to enable wide consultation and input into the review from experts from diverse disciplines. Participants included members of the Congenital Malformations & Birth Defects National Data Review Committee (CM&BDNDRC) and other key stakeholders. A list of the workshop participants is included in Appendix 2.

The aims of the workshop were to develop an agreed set of objectives for the national birth anomalies data collection, to determine the scope of the collection, to develop a proposal for a National Minimum Data Set (NMDS) for birth anomalies (i.e. the items to be collected) and to list any issues for further work.

Tasmania, the Australian Capital Territory and the Northern Territory were not represented at the workshop. Separate teleconferences were conducted with these jurisdictions following the workshop to ensure that their input was included.

The proceedings of the workshop are included in Appendix 2, so only the major outcomes are reported here.

Structure

The workshop was held over 2 days and was divided into seven sessions.

Day 1

Session 1: Overview of the current status of congenital malformations and birth defects (CMBD) data collection.

Session 2: Should we have a national congenital malformations and birth defects collection in Australia in 2003?

Session 3: Where do we go from here with congenital malformations and birth defects monitoring? Consultation on scope.

Session 4: What functions/activities/programs should a national congenital malformations and birth defects register be able to perform?

Day 2

Session 5: Under what principles should a congenital malformations and birth defects collection operate? What definitions should the congenital malformations and birth defects collection adopt?

Session 6: What are the major barriers to a national congenital malformations and birth defects collection?

Session 7: How should the national system of congenital malformations and birth defects monitoring operate to fulfil its aims and objectives; and to overcome barriers?

Sessions 1, 2 and 3 consisted of presentations by nominated participants. A list of these presentations is included in the proceedings in Appendix 2. Discussion relating to the sessions on the second day of the workshop overlapped, so the sessions are presented together.

Outcomes of the workshop

National Congenital Malformations and Birth Defects Data Collection

Limitations of the current National Congenital Malformations and Birth Defects Data Collection were identified. The limitations of the national collection include inconsistent and incompatible data which do not reflect the true prevalence of birth anomalies due to variation among the states and territories in the scope of their collections, the definitions and classifications used, the method of data collection and the available resources. Other limitations include the lack of timeliness in the provision of data to the NPSU and lack of updating of data which results in the state and territory collections being different to the national collection.

It was agreed there should be a National Congenital Malformations and Birth Defects Data Collection and that the NPSU should be the custodian. It was also agreed that the national collection and reporting of birth anomalies should be referred to as the Australian Birth Anomalies System. Some of the reasons for having a national system included that an Australian Birth Anomalies System will:

- improve ascertainment and quality of data by providing a framework and setting to standardise definitions, classifications and collection methods among the states and territories;
- provide high quality, nationally consistent data for use in policy development and planning, including identifying areas of need and funding requirements;
- enable smaller states and territories to merge with larger collections for more meaningful statistics (e.g. increases the sample size);
- enable national compilation and reporting of conditions (e.g. genetic disorders) not currently compiled and reported at the national level;
- identify the need for and then enable the evaluation of national health promotion activities aimed at preventing birth anomalies;
- identify the need for and then enable the evaluation of prenatal screening and diagnostic testing services at a national level;
- enable research at the national level, including assisting with planning and managing collaborative research;
- facilitate Australia's participation in international reporting and research;
- provide a context for the evaluation of reported clusters of birth anomalies; and
- assist in reducing the burden of disease associated with birth anomalies within the Australian population.

Scope of the new Australian Birth Anomalies System

The scope of the new Australian Birth Anomalies System was not completely agreed during the workshop. Further work to be undertaken by specific working groups was recommended.

Conditions for inclusion

There was discussion about what conditions should be included in the new Australian Birth Anomalies System and proposals included; internationally agreed commonly occurring birth anomalies and genetic disorders; conditions detected through newborn screening and newborn hearing screening; conditions detected through prenatal screening and prenatal diagnostic testing; and other conditions relevant to the Australian context.

It was agreed that a working group be established to identify the conditions to be initially included in the new Australian Birth Anomalies System.

Terminations of pregnancies with birth anomalies

It was agreed that terminations of pregnancies with birth anomalies should be included in the Australian Birth Anomalies System, regardless of gestational age. It was agreed that this is important because of the effect on prevalence of not including them. It was acknowledged that some jurisdictions are not notified of these cases. It was agreed that notification of data on terminations of pregnancies with birth anomalies, especially from private service providers, should be explored and that a working group to address state and territory implementation issues should be convened.

Period of detection for birth anomalies

There was discussion about the variation among the states and territories as to when the period of detection of birth anomalies ends. Currently, the end point of the detection period is notionally 28 days. However, the workshop participants agreed that ending the detection period at 1 year of age would be best practice at this stage.

It was decided that a working group should be convened to determine the benefits of ending the detection period at 1 year of age compared to 28 days of age with respect to case ascertainment.

Classification of birth anomalies

It was noted that there is variation among the states and territories in the classifications used to code birth anomalies. It was agreed that states and territories should work towards using a single classification and that this should be the International Statistical Classification of Diseases and Related Health Problems, 10th Revision, Australian Modification (ICD-10-AM). It was acknowledged that the ICD-10-AM lacks specificity for birth anomalies. Workshop participants agreed that a working group should be convened to address the issue of specificity.

National Minimum Data Set (NMDS)

It was noted that there is no NMDS for birth anomalies. For the current National Congenital Malformations and Birth Defects Data Collection, the NPSU requests an agreed set of data elements. However, the specification for the collection was not agreed through the governance arrangements for health information management and information technology. Some of the data elements requested are specified in the Perinatal NMDS.

Workshop participants acknowledged that there was variation among the states and territories in the scope of their birth anomalies collections, the definitions and classifications used and the method of data collection.

They agreed that a NMDS should be developed for birth anomalies. They agreed that the details of the NMDS were beyond the scope of the workshop and that a working group should be convened to develop it following the process required under the health information management and information technology governance arrangements.

It was also agreed that in the interim a national agreement on a minimum set of data to be reported should be obtained.

Workshop participants discussed whether age at diagnosis should be included in the national collection and agreed that it should be.

Data linkage

Linkage of birth anomalies data with other data sets was discussed. It was acknowledged that this could improve the utility of the data by enabling more accurate case ascertainment and by expanding the research questions that can be answered.

State and territory reporting

There was discussion about the variation in birth anomalies data collections among the states and territories. It was noted that four jurisdictions (New South Wales, Victoria, Western Australia and South Australia) have dedicated Birth Defects Registers and have more comparable data than the other jurisdictions. For example, notifications of birth anomalies are received from a wider variety of sources than for other jurisdictions, including from cytogenetic and pathology reports, notification of birth anomalies detected prenatally and up to 1 year of age are received and notifications of terminations of pregnancies with birth anomalies where the gestational age is <20 weeks are received.

It was noted that the lack of uniformity among the states and territories is problematic for national reporting. Workshop participants considered whether reporting of birth anomalies should be limited to the four jurisdictions with more consistent and comparable data. It was agreed that a capacity building approach towards including all states and territories should be promoted.

Resource implications

It was agreed that the issue of funding was fundamental to the ongoing development of a new Australian Birth Anomalies System, but that this was not in scope for the workshop.

It was agreed, however that recurrent core programmatic funding was needed to sustain birth defects registers at both the state and territory and national levels; and that in the short term additional 'catch-up' funding was needed to develop a nationally consistent birth anomalies system and to support the jurisdictions with less developed birth anomalies data collections.

Key recommendations

- That the National Congenital Malformations and Birth Defects Data Collection continue and that it be referred to as the Australian Birth Anomalies System (ABAS).
- That the custodian of the ABAS be the NPSU.
- That a working group be convened to consider the conditions for initial inclusion in the ABAS. Consideration should be given to including the 35 ICBDMS conditions, four newborn screening conditions (phenylketonuria, congenital hypothyroidism, cystic fibrosis and galactosemia), multiple malformations, other chromosomal abnormalities and any other condition identified as being important in the Australian context.
- That terminations of pregnancies with birth anomalies be included in the ABAS, regardless of gestational age.
- That a working group be convened to address state and territory implementation issues, including consultation with relevant stakeholders to examine access to data on terminations of pregnancies with birth anomalies (public and private providers).
- That birth anomalies detected up to 1 year of age be included in the ABAS in the first instance.
- That a single classification be used for coding birth anomalies and that this should be ICD-10-AM.
- That a working group be convened to address issues of lack of specificity for birth anomalies in ICD-10-AM.
- That a National Minimum Data Set for birth anomalies be developed following the process required under the governance arrangements for health information management and information technology.
- That an interim Minimum Data Set be agreed nationally.
- That recurrent core programmatic funding be identified for birth anomalies collections at the state and territory and national levels.
- That a National Birth Anomalies Steering Committee be established to advise on the scope, development and implementation of the ABAS.

Proposed steering committee and working groups

It was proposed that a National Birth Anomalies Steering Committee be established to provide expert advice on the scope, development and implementation of the Australian Birth Anomalies System. The steering committee should comprise 2–3 state and territory representatives, a representative from the Department of Health and Ageing (DoHA), a representative from the Department of Family and Community Services (FaCS), and other relevant stakeholders, including from clinical, epidemiological and technical backgrounds. The steering committee should meet on a regular basis, including one annual face-to-face meeting.

The formation of a number of working groups to undertake work arising from the workshop was also proposed and members of each group were decided. The Steering Committee should oversee the work of these groups. The proposed working groups were:

Working group 1

Purpose: Identification of the conditions to be initially covered in the Australian Birth Anomalies System (ABAS).

Members: David Tudehope, Liz Elliott, Lee Taylor, Chris Stone, Elizabeth Sullivan, and Carol Bower.

Working group 2

Purpose: Data development including agreeing and defining a National Minimum Data Set (NMDS) for birth anomalies.

Members: Seeta Duvasula, Annabelle Chan, Merrilyn Riley, Elizabeth Elliott, Elizabeth Sullivan and Sue Cornes.

Working group 3

Purpose: Examination of the notification of trisomies data; conduction of an analysis on the different periods of detection of birth anomalies (i.e. birth, 28 days, 1 year of age).

Members: Lee Taylor, Carol Bower, Jane Halliday, Annabelle Chan, Elizabeth Sullivan.

Working group 4

Purpose: Coding in ICD-10-AM and conversion to ICD-10-AM from ICD-9-BPA.

Members: Elizabeth Elliott, Susan Travis, Kerry Innes and John Edward.

Working group 5

Purpose: State and territory implementation issues, including issues related to notification of terminations of pregnancies with birth anomalies.

Members: Lee Taylor, Carol Bower, Jane Halliday, Annabelle Chan, Sue Cornes, Elizabeth Sullivan, Maureen Bourne, Karen Wheeler and Margaret Stewart.

Further work was also proposed in a number of other areas, but working groups were not established to undertake this work. These areas were:

- development of the content and structure of the national report; and
- consideration of ways to broaden the usage of national birth anomalies data (nationally and internationally).

6 Committee

Congenital Malformations and Birth Defects National Data Review Committee

The Congenital Malformations and Birth Defects National Data Review Committee (CM&BDNDRC) was convened by the AIHW to advise the NPSU during the review of the National Congenital Malformations and Birth Defects Data Collection. The committee met on 12–13 May 2003 (workshop), on 28 October 2003 (by teleconference) and on 18 November 2003 (by teleconference).

Committee membership

The members of the committee are:

Dr Richard Madden (Chair) (Director, Australian Institute of Health and Welfare)
Associate Professor Michael Frommer (Workshop Chair) (University of Sydney)
Dr Carol Bower (Telethon Institute for Child Health Research, Western Australia)
Dr Annabelle Chan (Department of Human Services, South Australia)
Ms Sue Cornes (Queensland Health)
Dr Seeta Durvasula (Royal Rehabilitation Centre for Sydney)
Associate Professor Elizabeth Elliott (Australian Paediatric Surveillance Unit)
Dr John Glover (National Public Health Information Working Group)
Associate Professor Jane Halliday (Department of Human Services, Victoria)
Dr Helen Moyle (Australian Institute of Health and Welfare)
Dr Lee Taylor (New South Wales Health)
Professor Michael Peek (Royal Australian College of Obstetricians and Gynaecologists)
Professor Paul Colditz (University of Queensland)
Professor David Tudehope (Australian College of Paediatrics)
Dr Elizabeth Sullivan (Director, AIHW National Perinatal Statistics Unit).

Terms of reference

The terms of reference of the committee are:

1. To advise on the utility of the Congenital Malformations Australia data collection.
2. To advise on the utility of the current national Birth Defects Series of reports.
3. To advise on the scope, funding and capacity of state and territory birth anomalies data collections and systems.

4. To advise on the scope of the collection, including which major and minor birth anomalies and other conditions (e.g. fetal alcohol syndrome, autism, etc) should be separately identified, what information on drugs, environmental teratogens, diagnostic, genetic and other screening tests and terminations of pregnancy should be included.
5. To advise on a national timeframe for collection of data for: diagnostic tests; terminations of pregnancy; all births; and infants (e.g. 28 days, 1 year or 2 years).
6. To make recommendations on the content of a proposed National Minimum Data Set (NMDS).
7. To determine how to broaden the usage of the data collection, including usage in conjunction with other disease databases, such as hospital morbidity and national death and proposed birth indices.
8. To advise on issues around benchmarking with international birth anomalies data collections.
9. To advise on future arrangements to organise birth anomalies NMDS, steering group, use of data (data collection and schedules and form of data transfer from states and territories) and type of reporting.
10. To advise on National Perinatal Statistics Unit role as an advocate for birth anomalies collections, birth anomalies prevention and birth anomalies research.

Workshop

12–13 May 2003.

The outcomes and key recommendations of the workshop are included in Chapter 5 and the proceedings of the workshop are included in Appendix 2.

Teleconference

28 October 2003.

The teleconference was held to conduct a preliminary review of the review report, including the proceedings of the workshop. The teleconference was chaired by Dr Elizabeth Sullivan, and was intended as a working meeting only.

Teleconference

18 November 2003.

The teleconference was held to review and endorse the recommendations of the review arising from the survey and the workshop. The naming of the collection and the schedule for submission of the review report were also discussed.

Recommendations

A summary of the recommendations of the review are presented at the beginning of this report.

The following recommendations were supported by the committee and their relation to the terms of reference (TOR) of the committee is indicated:

- That the national collection of data on birth anomalies continue and that it be referred to as the Australian Birth Anomalies System (ABAS) (TOR 1).
- That the NPSU be the custodian of the ABAS (TOR 1 and 10).
- That the term 'birth anomalies' be used.
- That the ABAS be the repository for data from state and territory birth anomalies data collections and other identified suitable data collections (TOR 4).
- That NPSU reports reliable and valid birth anomalies statistics in the public domain (TOR 2 and 9).
- That subject to the findings of Working Group 1, the 35 sentinel conditions agreed by ICBDMs, four conditions detected by newborn screening (phenylketonuria, congenital hypothyroidism, cystic fibrosis and galactosemia) and other conditions relevant to the Australian context be included in the ABAS (TOR 4).
- That the NPSU should commence national reporting of newborn screening (TOR 4).
- That the scope of the ABAS include birth anomalies detected up to 1 year of age, for jurisdictions capable of providing this (TOR 5).
- That the scope of the ABAS include terminations of pregnancies with birth anomalies data, regardless of gestational age, for jurisdictions capable of providing this. The working group examining state and territory implementation issues should undertake consultation with relevant stakeholders to examine access to terminations of pregnancies with birth anomalies data in the other states and territories (TOR 5).
- That a timeframe be developed for the jurisdictions unable to meet the recommended scope to develop their collections (TOR 3 and 5).
- That the development of partnerships between the more capable states and the less capable jurisdictions to improve compliance with the proposed national standards be explored.
- That a single classification be adopted and that collaboration with the National Centre for Classification in Health (NCCCH) be undertaken for the development of ICD-10-AM for this purpose (TOR 6).
- That the NPSU in consultation with relevant stakeholders develop a nationally consistent definition for birth anomalies (TOR 1 and 6).
- That data for the ABAS be provided to the NPSU on an annual basis according to agreed specifications and an agreed timetable. There should be an agreed policy for updating data at the national level (TOR 9).

- That a national steering committee be established to advise on the scope, development and implementation of the national birth anomalies data collection and that the steering committee have an annual face-to-face meeting (TOR 7 and 9).
- That working groups be established to continue work arising from the workshop and that these working groups be overseen by the steering committee (TOR 9).
- That the structure and content of the current national Birth Defects Series of reports be revised, with advice from the national steering committee (TOR 2).
- That the NPSU develop collaborations with organisations collecting data on screening and diagnostic testing in the first year of life (for example, the Australian Paediatric Surveillance Unit and the Human Genetics Society of Australia) (TOR 7).
- That the NPSU coordinate access to the ABAS for national and international reporting and collaborative research, subject to appropriate ethics approval (TOR 8, 9 and 10).
- That ongoing and sustainable funding be identified for data development related to the ABAS, ongoing support of the collection and support of the steering committee (TOR 9).
- That this review report be referred to the Statistics Information Management Committee (SIMC) to provide information about the lack of a national birth anomalies system and the inability of jurisdictions to provide data because of funding and other resource constraints (TOR 3).

Appendix 1 Survey questions

**CONGENITAL MALFORMATION & BIRTH
DEFECTS**

NATIONAL DATA REVIEW QUESTIONNAIRE

Please fax back to 02 9382 1025 or
e-mail to m.birch@unsw.edu.au

1. Please indicate the **primary** State or Territory you represent.

- ACT
- NSW
- NT
- QLD
- TAS
- VIC
- WA

2. Do you hold the congenital malformation and birth defect register for another state or territory?

YES NO

3. If YES, please indicate the other States or Territories for whom you hold a register?

- ACT
- NSW
- NT
- QLD
- TAS
- VIC
- WA

4. Please state the definition of **congenital malformation** and **birth defect** that you use.

a) Congenital malformation

a) Birth defect

5. Please state the reporting period you use for notification of congenital malformation and birth defects (CMBD) **Please tick all applicable**

- Antenatal (before birth)
- Perinatal (birth to 28 days)
- Birth to 5 years of age
- Older than 5 years to 12 years of age
- Other (*Please specify*) _____

6. How do you deal with notifications received outside the above-mentioned reporting periods?

7. Is notification to your CMBD register part of a 'routine' process? YES NO

8. If YES, briefly describe the routine notification process

9. Is notification to your CMBD register voluntary or enacted through legislation?

- Voluntary
- Enacted through legislation
- Other (*Please specify*) _____

10. Do you have a formal agreement for reporting CMBD at a National level?

YES NO

11. Do you have an agreement with the Australian Health Ministerial Advisory Council (AHMAC) to report CMBD? YES NO

12. Do you have a State or Territory Congenital Malformations and Birth Defects Committee?

YES NO

13. If YES, please name the chair person for the committee _____

WE WOULD LIKE TO KNOW HOW YOUR REGISTER IS FUNDED

14. Please list all current funding sources

Please \checkmark tick all applicable

- Commonwealth Government
 - State / Territory Government
 - Institution core funding (e.g. university)
 - Private / donations
 - None
 - Other (*Please specify*)
-

15. Please state the approximate total amount of annual funding you receives, from all sources, for the purpose of maintaining your register.

\$ _____

16. What human resources do you currently have to maintain your register?

- No specific allocation
 - < 1 Full-time equivalent (FTE)
 - 1 FTE
 - 2 FTE
 - Other (*Please specify*)
-

17. Is your CMBD register funded exclusively or is CMBD register part of another, separately funded, data collection?

- Exclusively funded
 - Part of another data collection
 - Other (*Please specify*)
-

DATA COLLECTION

18. Is your register linked to another database?

Please \checkmark tick all applicable

- Perinatal database
 - Assisted conception database
 - Hospital database (excludes midwives)
 - Midwives data base
 - Other (*Please specify*)
-

19. Please indicate the parameters of your annual collection period

- Calendar year (1st January to 31st December)
 - Financial year (1st July to 31st June)
 - Other (*Please specify*)
-

20. With reference to Q19, how many years of complete data do you hold?

- None
- We only have incomplete data
- 1 year
- 2 - 3 year
- 4 - 5 years
- > 5 years
- Other (*Please specify*) _____

21. When was your register computerized?

- Remains paper based
- Pre 1998
- 1999
- 2000
- 2001
- 2002
- 2003
- Other (*Please specify*) _____

22. Please state the latest complete year of data on your register?

- Pre 1998
- 2000
- 2001
- 2002
- Other (*Please specify*) _____

23. Please state the most recent year a report was published using the data from your register.

- Pre 1998
- 2000
- 2001
- 2002
- Other (*Please specify*) _____

24. With reference to **Q22**, please state the latest year of data used in the publication?

- Pre 1996
- 1997
- 1998
- Other (*Please specify*) _____

25. What is the stated purpose of the State or Territory CMBD register?

Please \surd tick all applicable

- Incidence reporting only
- Ongoing surveillance
- Other (*Please specify*) _____

26. What reporting controls do you use?

Please \surd tick all applicable

- Diagnosis must be confirmed
- Diagnosis can be preliminary
- Other (*Please specify*) _____

27. How long do you conduct follow up?

Please \surd tick all applicable

- No follow-up conducted
- Follow-up for confirmation of diagnosis only
- 12 months later
- Up to 5 years
- Other (*Please specify*) _____

28. Please indicate the sources of notification

Please ✓ tick all applicable

- State hospitals
- Private hospitals
- Midwives
- Pathologists / microbiologist
- Ultrasound Centres
- Cytogenetics perinatal screening
- Early Childhood Centres
- Rural Health Care workers
- Paediatricians
- Other specialist (cardiologists, physiotherapists)
- Other (*Please Specify*)_____

29. How are you notified?

Please ✓ tick all applicable

- Verbal and/or telephone
- Unofficial electronic (e-mail)
- State sanctioned notification form
- Other (*Please specify*)_____

30. Do you receive multiple notifications for the same individual? YES NO

31. Can you identify and adjust for multiple notifications? YES NO

32. Do you receive notifications from other States and Territories? YES NO

33. Do you have a minimum amount of data that must be reported? YES NO

34. Who sets the minimum data requirements?

Please attach a descriptive list of the minimum data set that you collect.

E.g. M_DOB (Maternal date of birth); B_DOB (Baby date of birth)

35. Do you record whether an alpha protein test is performed and the outcome?

- No
 - Yes, testing only
 - Yes, testing and outcome
 - Other (*Please specify*)
-

36. Is the register advised of termination of pregnancy (TOP) following adverse prenatal diagnostic screening tests? YES NO

37. With reference to **Q35**, is the register notified of TOP when the gestational age is < 20 weeks? YES NO

38. With reference to **Q35**, is the register notified of TOP when the gestational age is > 20 weeks? YES NO

39. Do you have a policy for disposal of the products of conception following termination of pregnancy (TOP)? YES NO

If YES, please attach a copy of the policy.

USES OF DATA

40. How is your data utilised?

Please* ✓*tick all applicable

- Results available to general public on request
- Results are available for special interest groups
- Results available upon formal (written) request
- Results supplied to National Regulatory Authorities
- Local Government State reports (intrastate use)
- Local Government publications
- Formal commissioned reports
- Other (*Please specify*)

Nil

41. Is there is any additional information that you are not currently collecting that you believe should be collected?

42. Do you have any comments?

**THANK YOU FOR COMPLETING THIS QUESTIONNAIRE.
PLEASE FAX YOUR COMPLETED QUESTIONNAIRE TO THE NATIONAL PERINATAL
STAISTICS UNIT ON 02 9382 1025 AT YOUR EARLIEST CONVIENCE**

CONGENITAL MALFORMATIONS AND BIRTH DEFECTS ADDITIONAL
QUESTIONS

Q1. Please indicate the state or territory that you represent _____

Q2. Please state the parameters your state or territory uses to define the perinatal period

Birth to discharge from the birth episode

Birth to 28 days

Other (please state)

Q3. Does your state or territory confirm trisomies?

Yes

No

Q4. Does your state or territory confirm trisomies with chromosomal studies?

Yes

No

Q5. Does your state or territory record date birth defect diagnosed?

Yes

No

Q6. Do you record the date of diagnosis, if the diagnosis was made in the antenatal period?

Yes

No

Q7. Do you record the date of diagnosis, if the diagnosis was made between birth and 28 days of age?

Yes

No

Q8. Do you record the date of diagnosis, if the diagnosis was made between 29 days of age and one year of age?

Yes

No

Appendix 2 Proceedings of the Congenital Malformations and Birth Defects National Data Review Workshop

National Perinatal Statistics Unit

Congenital Malformations and Birth Defects National Data Review Workshop

University of New South Wales

12–13 May 2003

Participants

Associate Professor Michael Frommer (Chair) (University of Sydney)
Dr Carol Bower (Telethon Institute for Child Health Research, Western Australia)
Dr Annabelle Chan (Department of Human Services, South Australia)
Ms Sue Cornes (Queensland Health)
Dr Seeta Durvasula (Royal Rehabilitation Centre for Sydney)
Associate Professor Elizabeth Elliott (University of Sydney)
Dr John Glover (Public Health Information Working Group)
Associate Professor Jane Halliday (Department of Human Services, Victoria)
Ms Rosemary Lester (Department of Human Services, Victoria)
Dr Richard Madden (Director, Australian Institute of Health and Welfare)
Dr Helen Moyle (Australian Institute of Health and Welfare)
Professor Michael Peek (University of Sydney)
Mrs Dianne Petrie (Association of Genetic Support of Australasia)
Ms Sheree Gray (National Centre for Classification in Health)
Ms Christine Stone (Department of Human Services, Victoria)
Dr Lee Taylor (New South Wales Health)
Professor David Tudehope (Mater Misericordiae Mothers Hospital, Queensland)
Dr Elizabeth Sullivan (Director, AIHW National Perinatal Statistics Unit)

Dr Deborah Kennedy (Royal Hospital for Women, Randwick, New South Wales)

Workshop support

Ms Mary-Rose Birch (AIHW National Perinatal Statistics Unit)

Dr Siva Sivarajasingam (AIHW National Perinatal Statistics Unit)

Introduction

A 2-day workshop was held in May 2003 to enable wide consultation and input into the review from experts from diverse disciplines. Participants included members of the Congenital Malformations & Birth Defects National Data Review Committee (CM&BDNDRC) and other key stakeholders.

The aims of the workshop were to develop an agreed set of objectives for the national birth anomalies data collection, determine the scope of the collection, develop a proposal for a National Minimum Data Set (NMDS) for birth anomalies (ie the items to be collected) and list any issues for further work.

Tasmania, the Australian Capital Territory and the Northern Territory were not represented at the workshop. Separate teleconferences were conducted with these jurisdictions following the workshop to ensure that their input was included.

Briefing papers

The following briefing papers were provided to workshop participants:

1. National monitoring of congenital malformations and birth defects in Australia 1981–2003.
2. State and territory congenital malformations and birth defects collections 2003.
3. Utility of congenital malformations and birth defects registers.
4. Review of selected international birth defects and congenital malformations reports.
5. Collaborative international registers.
6. EUROCAT Guide 1.2.
7. Definitions used in international registers.
8. Journal articles related to birth anomalies monitoring systems.
9. *An Overview of Public Health Surveillance of Genetic Disorders and Mapping of Current Genetic Screening Services in Australia* (National Public Health Partnership).
10. International Clearinghouse for Birth Defects Monitoring Systems Annual Report 2000 with data for 1998.

In addition, the paper *Beyond the crystal ball: the epidemiology of some genetic disorders in Victoria* (Stone & Lester 2002) was distributed on day 1.

Structure

The workshop was held over 2 days and was divided into seven sessions. These were:

Day 1

Session 1: Overview of the current status of congenital malformations and birth defects (CMBD) data collection.

Session 2: Should we have a national congenital malformations and birth defects collection in Australia in 2003?

Session 3: Where do we go from here with congenital malformations and birth defects monitoring? Consultation on scope.

Session 4: What functions/activities/programs should a national congenital malformations and birth defects register be able to perform?

Day 2

Session 5: Under what principles should a congenital malformations and birth defects collection operate? What definitions should the congenital malformations and birth defects collection adopt?

Session 6: What are the major barriers to a national congenital malformations and birth defects collection?

Session 7: How should the national system of congenital malformations and birth defects monitoring operate to fulfil its aims and objectives; and to overcome barriers?

Sessions 1, 2 and 3 consisted of presentations by nominated participants. Discussion relating to the sessions on the second day of the workshop overlapped, so the sessions are presented together.

Proceedings

Day 1

The Chair welcomed the workshop participants. He then provided an overview of the purpose and structure of the workshop and introduced the first presenter of session 1.

Session 1: Overview of the current status of congenital malformations and birth defects data collection.

This session consisted of four presentations in which international birth anomalies systems were outlined, an international perspective on future directions for the development of a birth anomalies system was provided, the National Congenital Malformations and Birth Defects Data Collection was outlined and the results of the survey of state and territory birth anomalies collections were presented.

Presentation 1

Presenter: Mary-Rose Birch

Title: Profile of reports being used internationally.

Birth anomalies monitoring systems in a number of countries including Australia, Canada, England and Wales, Norway and the United States of America, as well as two of the international monitoring systems – the International Clearinghouse for Birth Defects Monitoring Systems (ICBDMS) and the European Concerted Action on Congenital Anomalies and Twins (EUROCAT) – were outlined in this presentation.

Similarities between these systems and the Australian one, for example that, like Australia, most of these systems have multiple sources of notifications and are used for surveillance of birth anomalies, were identified. Differences among the countries included definitions and classifications used and periods of detection of birth anomalies.

Some of the advantages of national birth anomalies systems were outlined, including that the sample size is increased from that at the sub-national level, which is particularly important when monitoring rare birth anomalies, and that a national system enables better understanding of the prevalence of birth anomalies within the whole population.

Presentation 2

Presenter: Dr Carol Bower

Title: Future directions for the development of a national monitoring system: an international perspective.

Birth anomalies monitoring systems in a number of countries including Canada, England and Wales, Norway and the United States of America, as well as the international monitoring systems – the International Clearinghouse for Birth Defects Monitoring Systems (ICBDMS), the European Concerted Action on Congenital Anomalies and Twins (EUROCAT) and the Latin American Collaborative Study of Congenital Malformations (ECLAMC) – were outlined in this presentation.

Future directions for Australia in developing a national birth anomalies system were identified. These were to determine:

- the purposes for a national collection;
- whether it needs to cover the total population;
- whether it needs to cover all birth anomalies;
- whether it needs to include terminations of pregnancies with birth anomalies data;
- funding;
- whether reporting should be mandatory; and
- whether other state-based systems can be used to inform this (e.g. cancer registers).

Presentation 3

Presenter: Dr Elizabeth Sullivan

Title: National birth anomalies monitoring, Australia 2003.

The current national birth anomalies system and options for the system in the future were outlined in this presentation. It was noted that data quality issues have limited the utility of the national birth anomalies data. The limitations of the data include variation among the states and territories in the scope of their collections, the definitions and classifications used, the method of data collection and the available resources. Other limitations include the lack of timeliness in the provision of data to the NPSU and lack of updating of data which results in the state and territory collections being different to the national collection.

Mechanisms for improving the utility of the national birth anomalies system were proposed. These included: increasing the scope of the collection to include genetic

disorders, prenatal and newborn screening, terminations of pregnancy associated with a diagnosis of a birth anomaly and developmental disabilities; development of consistent definitions; development and agreement of a National Minimum Data Set (NMDS); use of a single classification; development of a standard reporting form; and extension of the detection period to 1 year of age.

Mechanisms for enhancing birth anomalies surveillance were proposed. These included the development of a formal network of states and territories and other stakeholders; the fostering of collaborative research; dissemination of birth anomalies data; and the production of a national birth anomalies report.

Options for the future national birth anomalies monitoring system to overcome the current limitations were outlined. These were to:

- cease national reporting and rely on state and territory reports and studies;
- publish an annual national report on 'sentinel anomalies' based on aggregate data provided by the states and territories;
- publish an annual national report on all birth anomalies based on aggregated data provided by the states and territories; or
- develop and restructure the existing national birth anomalies system, based on unit records for births and terminations of pregnancies with birth anomalies, which would be provided electronically by all states and territories.

The first option was seen as limited because the capacity of the smaller states and territories, in particular, to undertake surveillance of birth anomalies is constrained by the amount of resources available, and because it would not provide national reporting which was thought to be needed.

It was thought that the second and third options were limited because of differences among the states and territories in scope, definitions and data quality and that significant investment in data development would be required.

The third option was also thought to require significant investment in data development. However, it was thought that this option would better enable the data to be used to estimate the burden of birth anomalies in Australia, detect temporal and geographical variation, evaluate public health interventions and screening programs, and provide the evidence base for perinatal and maternal health programs and policy development.

Presentation 4

Presenter: Mary-Rose Birch

Title: Survey results of state and territory congenital malformations and birth defects collections.

This presentation was used to outline the results of the survey of the state and territory birth anomalies data collections. The results of the survey are presented in Chapter 4. The results highlighted marked variation among the state and territory collections.

From the survey results a number of tasks were identified to improve the national birth anomalies system: development of nationally consistent clinical and data definitions; development and agreement of a National Minimum Data Set for birth anomalies; identification and implementation of an agreed classification (e.g. ICD-10-AM); agreement on scope including period of detection of birth anomalies (e.g. up to

1 year of age) and terminations of pregnancies with birth anomalies data; and securing funding to support birth anomalies collections.

Session 2: Should we have a national congenital malformations and birth defects collection in Australia in 2003?

This session consisted of seven presentations in which the issue of whether a national birth anomalies system is required in Australia was addressed.

Presentation 1

Presenter: Dr Helen Moyle

Title: National data collections addressing policy, planning and information needs.

In this presentation the role of national data collections in addressing policy, planning and information needs was discussed. It was noted that birth anomalies data are important in the planning of health, disability, welfare and education services and that a national birth anomalies system was needed in Australia. A major role of the national birth anomalies system would be to produce high quality, nationally consistent birth anomalies data for use in policy development and planning.

In order to achieve this, birth anomalies definitions and indicators would need to be developed and agreement sought through the National Health Information Group, and adequate funding for the development and maintenance of a national birth anomalies monitoring system would be required.

Presentation 2

Presenter: Dr Seeta Durvasula

Title: Developmental disability and extension of congenital malformations and birth defects monitoring.

In this presentation, the extension of the scope of the national birth anomalies monitoring system to include developmental disabilities was proposed. It was noted that developmental disabilities affect approximately 4% of the population, and those affected have greater mortality and morbidity than in the general population. However, developmental disabilities are not identified in routinely collected data sets (e.g. death registers, cancer registers, injury databases).

The utility of a national data collection for developmental disabilities was discussed. It was proposed that a national collection could be used to identify and monitor causes, including genetic causes, monitor trends in incidence, link to population-based registers, and assist in health and other services planning.

Some of the issues for consideration related to including developmental disabilities in the national birth anomalies system. These were the heterogeneity of the conditions, the existence of an acceptable classification, age of ascertainment, sources of ascertainment, links with the International Classification of Functioning and Disability (ICF), links with other registers and the politically sensitive nature of data collection in this population.

Presentation 3

Presenter: Christine Stone

Title: Key findings from the overview of public health surveillance of genetic disorders and mapping of current genetic screening services in Australia.

The Victorian Department of Human Services in collaboration with the National Public Health Partnership, Public Health Genetics Working Group (PHGWG) conducted a project to survey existing genetic services and programs in Australia. The project consisted of two components: (1) the genetic programs and services; and (2) the supporting legislation, including overarching issues such as consent, privacy and ethics. The survey identified the large number of programs that are occurring in each state and territory such as the newborn screening programs and prenatal screening programs. It also identified the variety of services that support these programs such as Genetic Services including diagnostic, community and educational programs, and research to support such programs.

Integral to the provision of these services is the development of appropriate surveillance and monitoring systems, not only for the genetic conditions but also for the monitoring of the impact of the programs themselves. The information is currently in diverse forms, from formal registries (e.g. birth defects, child deaths, cancer registries), data bases (e.g. laboratory, jurisdictional, genetic services, familial cancer services) to regular or ad hoc reports. Other relevant issues identified were the importance of identifying the scope of conditions to be monitored, the programs and the type of collection (temporary/permanent). The recommendations from the survey included a recommendation for a national surveillance system for integration of genetic disorders and programs. Specific attention was drawn to the need for national consistency in data collection, specifically in relation to newborn screening data and birth defects registers. An enhanced national birth anomalies system that included newborn screening could provide this. Newborn screening data are collected by states and territories and could be included in the national birth anomalies collection.

Presentation 4

Presenter: Dr John Glover

Title: Why collect data on birth anomalies?

Reasons for collecting data on birth anomalies were examined in this presentation. A major reason presented for collecting these data is to report the prevalence of birth anomalies. Other reasons presented include determining trends in birth anomalies over time and detecting unusual patterns of birth anomalies following exposure to teratogens; monitoring and surveillance of particular birth anomalies; providing information for planning the provision of services including health, welfare, disability and education services; assessing screening and diagnostic testing; and indicating a need for and then evaluating birth anomalies prevention and education programs.

Presentation 5

Presenter: Dr John Glover

Title: Why collect data at a national level?

Reasons for collecting birth anomalies data at a national level were examined in this presentation. The reasons proposed include: to determine the national prevalence of birth anomalies; to undertake research of the aetiology of specific birth anomalies which occur in isolation or in concert with environmental factors; to analyse the impact of prenatal diagnosis, interventions, and health care delivery at a national level; to contribute to state and national policy on screening for genetic disorders and other public health genetics activities; to conduct comparisons with national collections in other countries and benchmarking against international data collections.

Presentation 6

Presenter: Dr Siva Sivarajasingam

Title: Utility of birth anomalies registers.

The utility of birth anomalies registers was outlined in this presentation. It was noted that birth anomalies registers can be used for monitoring and surveillance (spatial, temporal, and for international comparisons), epidemiology (in relation to genetic and environmental factors) and for public health services planning (e.g. prenatal screening).

Some of the limitations of national birth anomalies registers were outlined. It was noted that data from multiple sources (i.e. states and territories) leads to sampling variance, differences in population size, differences in ascertainment, differences in data and statistical methods used, and differences in timeliness.

Presentation 7

Presenter: Associate Professor Jane Halliday

Title: The Victorian Birth Defects Register.

The Victorian Birth Defects Register was described in this presentation. It was stated that the Victorian Birth Defects Register has collected data on all birth defects since the beginning of 1982. All livebirths, stillbirths and terminations of pregnancy with birth anomalies are included, irrespective of age at diagnosis (up to 15 years of age). It was noted that birth anomalies are notified to the register on a voluntary basis from multiple sources.

Some of the advantages of a national collection were outlined. These were that a national collection:

- enables smaller states and territories to merge with larger collections for more meaningful statistics (i.e. collation of data for larger population statistics);
- provides a framework and setting for development and maintenance of standardised collection and coding;
- facilitates a more effective lobbying force for resources (e.g. mandatory reporting of birth defects and terminations of pregnancy); and
- assists with planning and management of collaborative research.

Examples of where a national approach to research would be advantageous were given. These included investigating in-vitro fertilisation and birth anomalies and the effect of folate on neural tube defects and other birth anomalies.

A number of disadvantages of the current national birth anomalies collection were noted. These were that it is unrepresentative of the true prevalence of birth anomalies because of the collation of complete and incomplete data sets. For

example, the prevalence of birth anomalies using NPSU data is much lower compared to the prevalence when data from the Victorian Birth Defects Register are used; and issues around data ownership.

A number of challenges for the Victorian Birth Defects Register were provided, including providing timely data, raising awareness of those developing policy, service planning or doing research of the existence and potential of the available data, obtaining more resources dedicated to both collaborative and in-house research that utilises the data, and to review the association with the NPSU and the ICBDMS.

Session 3: Where do we go from here? Consultation on scope.

This session consisted of eight presentations in which aspects of scope were discussed, including conditions that could be included in a national collection, terminations of pregnancies with birth anomalies and prenatal diagnosis (i.e. the timing of detection of birth anomalies), and the classification of birth anomalies. A group discussion followed the presentations.

Presentation 1

Presenter: Christine Stone

Title: Surveillance of genetic conditions an emerging issue.

An overview of *Beyond the Crystal Ball: The epidemiology of some genetic conditions in Victoria* was provided in this presentation. This descriptive epidemiology report is a first step in enhancing the surveillance and monitoring of genetic conditions, and the impact of Victorian services. It also recognises that scientific advances in genetics were moving ahead at such a rapid rate. The report describes the prevalence of and the testing for genetic conditions and how they have changed over the last 10 years in Victoria.

Many issues that were faced in the development of the genetic report are relevant to the structure and function of a national birth anomalies system. Firstly, as there is no single registry or equivalent, the sources of information are many and varied. They include expert opinion, the medical literature, testing laboratories data, the Birth Defects Register, Prenatal Diagnostic Testing Report, Victorian Inpatient Data and Health Insurance Commission data. Secondly, genetic conditions are complex and the field of genetics is rapidly expanding with new information and technology. Part of the complexity is inherent in genetic conditions themselves: genotype versus phenotype; single gene or multiple; high or low penetrance; time during the life course when diagnosis is more likely or more appropriate. All have implications for case ascertainment, case definitions and standardisation of genetic testing whether they are for diagnostic, predictive or carrier testing.

A recommendation of the report is the development of a national surveillance system that integrates genetic disorders and programs. The national system should have a monitoring role and would have implications for national policy decisions on screening. Consideration of newborn screening and newborn hearing screening should be made.

Presentation 2

Presenter: Professor David Tudehope

Title: The utilisation of antenatal ultrasound for fetal diagnosis in the management of birth anomalies.

In this presentation the utilisation of antenatal ultrasound for fetal diagnosis in the management of birth anomalies was outlined. It was noted that maternal fetal medicine is the newest and most rapidly developing sub-specialty of Obstetrics.

It was noted that through antenatal diagnosis clinicians can provide information for use by parents to assist them in assessing their options and making decisions about further pregnancy care. Antenatal diagnosis also provides information for optimising the outcome for the baby in terms of medical care. For example, it enables the development of a multidisciplinary care plan.

Factors in decision-making after antenatal diagnosis of a birth anomaly include the antenatal and birth diagnoses, short- and long-term health outcomes, educational outcomes, and social outcomes for the child, the family and the community.

The tertiary referral pattern for fetal anomalies was presented and it was noted that the pattern depends on the provider (maternal-fetal medicine specialist, other clinician, ultrasonographer). There are a number of different practices used for antenatal diagnosis with varying levels of sensitivity and specificity: routine fetal ultrasound and nuchal fold thickness at 17–19 weeks; amniocentesis at 16–18 weeks; chorionic villus sampling with DNA studies at 11–12 weeks; fetal blood sampling; biochemical screening (e.g. alpha feto protein, triple screen).

The advantages of antenatal diagnosis were outlined and include: enables a multidisciplinary approach to diagnosis and management options; enables antenatal therapy (e.g. obstructive uropathy, twin to twin transfusion, Rh isoimmunisation); enables planning of neonatal management (e.g. plan for neonatal surgery for gastroschisis).

Some of the areas where more knowledge is required include fetal lesions, fetal surgery (e.g. hysterotomy) and other fetal therapies (e.g. randomised controlled trials on diaphragmatic hernia).

It was noted that maternal-fetal medicine clinicians have some concerns over: the accuracy of antenatal diagnosis, evaluations of antenatal screening programs, medico-legal implications of missed or wrong antenatal diagnoses, and accountability for Commonwealth expenditure on ultrasound (it was noted that 35% of all obstetric Medicare payments is for ultrasound).

Concerns with definitions of birth defects were raised. The lack of standardisation of clinical definitions of birth anomalies (e.g. renal pelvic dilation – 5 mm v 10 mm) was noted. It was also noted that the definitions used affect the prevalence of birth anomalies. They also affect the accuracy of tests.

The requirements of a birth anomalies collection from a maternal-fetal medicine perspective were given as the development of agreed definitions for fetal anomalies, a central registry for all fetal therapy in Australia and the development of clinical indicators.

It was suggested that the birth anomalies notification form could be enhanced by including questions related to antenatal ultrasound (e.g. was an ultrasound performed? If so, when and where? Was a fetal anomaly detected? If so, was the diagnosis accurate?).

It was noted that this review provides an opportunity to look at the scope of birth anomalies surveillance in Australia, to recommend standardisation of clinical definitions of birth anomalies and provides a forum for the assessment of what conditions should be monitored. It should also look at the role and outcomes of antenatal and newborn screening in birth anomalies surveillance.

Presentation 3

Presenter: Associate Professor Jane Halliday

Title: Prenatal diagnosis in Victoria and linkage to the Birth Defects Register for the most accurate figure on Down syndrome.

In this presentation, data on prenatal diagnosis testing in Victoria was presented. A study on Down syndrome using prenatal diagnostic testing data from various sources linked to the Birth Defects Register to determine whether ascertainment of cases in the Birth Defects Register was accurate was also presented.

It was noted that women with advanced maternal age (≥ 37 years) are 'eligible' for prenatal diagnosis testing. Between 1979 and 2003, passive screening was available for women with advanced maternal age. Active screening of women of all ages has been available since 1997. It was shown that between 1997 and 2002 there was an increase in the number of women undergoing prenatal diagnostic testing for women aged <35 years and for women aged 40+ years. However, there was a decrease for women aged 37–39 years and for women aged 35–36 years.

It was also shown that the number of women undergoing prenatal diagnostic testing as a result of an abnormal ultrasound increased between 1990 and 2001 for women aged <37 years and for women aged ≥ 37 years. Similarly, the number of women undergoing prenatal diagnostic testing as a result of abnormal maternal serum screen increased between 1996 and 2002.

The linkage study was then presented. It used data from the prenatal diagnosis collections, the Birth Defects Register, Genetic Health Services Victoria and obstetricians to determine whether the ascertainment of cases with Down syndrome in the Birth Defects Register was accurate.

The aims were to ensure that all cases where Down syndrome had been diagnosed prenatally were included in the Birth Defects Register; to determine how many of all the recorded cases of Down syndrome are detected prenatally; and to examine demographics related to live births or babies with Down syndrome.

It was shown that the data in the Birth Defects Register were inaccurate. In 1999 there were 158 births with Down syndrome in the Birth Defects Register and 117 of these had been diagnosed prenatally. However, after linkage with the other data and in-depth follow-up it was found that there were 167 births with Down syndrome and that 107 of these had been diagnosed prenatally. Therefore, the prenatal detection rate using the linked data was 65%, not 74% as found using the Birth Defects Register data alone. A similar finding was obtained using the data for 2000 (66% and 73% respectively).

The proportion of live births with Down Syndrome to women aged <35 was presented using these more accurate data. It was shown that, in 2000, live births were more evenly distributed between women <35 years (54%) and ≥ 35 years (46%) compared with earlier years.

These data were also used to show where live births with Down syndrome were occurring. In 2000, almost 25% of all births with Down syndrome were in rural areas.

It was concluded that many considerations are necessary in using Birth Defects Registers data to determine prenatal detection rates of Down syndrome and that linking to separately ascertained databases on prenatal diagnostic tests reveals important adjustments to figures.

Presentation 4

Presenter: Dr Annabelle Chan

Title: Terminations of pregnancy with birth anomalies.

In this presentation, the role of terminations of pregnancy data in birth anomalies monitoring was outlined. Data from South Australia were presented.

It was shown that between 1970 and 1999 the pregnancy rate and the birth rate in South Australia decreased, but the abortion rate increased over this period. The grounds for terminations of pregnancy also changed over time, with pre-existing psychiatric conditions, medical disorders and potential damage to fetus being indicated less in 1990-94 compared to 1970-74.

The most common reasons for termination of pregnancy between 1995 and 2001 were identified as chromosomal abnormality and other identified fetal abnormality (39% and 53% respectively). The least common reason was possibility of damage from maternal rubella or rubella vaccination (0.1%).

The total prevalence and the birth prevalence of neural tube defects between 1966 and 2002 were presented. Total prevalence and birth prevalence were similar until 1977. After this time, the total prevalence remained fairly stable, but the birth prevalence decreased markedly. Similarly, the birth prevalence for Down syndrome decreased markedly between 1988 and 1996, but the total prevalence increased slightly over this period. This shows the effect of prenatal diagnosis and terminations of pregnancy on birth prevalence rates of these birth anomalies.

A folate promotion campaign was introduced in 1995 and the data show that the prevalence was fairly stable between 1988 and 1995, but decreased after 1995, suggesting that the folate promotion campaign affected the prevalence of neural tube defects.

The presentation demonstrated the importance of termination of pregnancy data and in ascertaining the true prevalence of birth anomalies. Information on the indication for termination of pregnancy and on prenatal diagnoses should be collected in birth anomalies monitoring systems.

Presentation 5

Presenter: Sheree Gray

Title: ICD-10-AM.

The International Statistical Classification of Diseases and Related Health Problems, 10th Revision, Australian Modification was described in this presentation. It was noted that the National Centre for Classification in Health (NCCH) was established in 1994 as the National Coding Centre (NCC) and that it is funded by the Department of Health and Ageing under the Casemix Program.

Some of the functions of NCCH were outlined. These were to: develop classifications for Australia – inpatient, community, emergency, outpatients, allied health; publish ICD-10-AM derived products such as the Early Parenting Manual and Mental Health Manual; educate coders and clinicians in coding and casemix; provide advice on classification design; produce quality measurement products such as PICQ and ACBA; and research.

It was noted that prior to 1994 the USA version of ICD-9-CM was used in Australia and annual updates from the USA were used. In 1995, ICD-9-CM was modified to create an Australian version, following the establishment of the National Coding

Centre in Australia. By 1996 there was less reliance on the USA updates and significant changes were made for the Australian version. The first edition of ICD-10-AM was released in July 1998. The ICD-10-AM is updated biennially and the fourth edition will be released in July 2004.

Some of the benefits of using ICD-10-AM were outlined, including that it is a comprehensive classification that uses current Australian medical terminology. The process for updating ICD-10-AM was also outlined and included review of other classifications, public submissions, evaluation by clinical classification and coding groups and approval by the NCCH Coding Standards Advisory Committee.

Some of the limitations of the British Paediatric Association (BPA) Classification of Diseases, which is used by some jurisdictions to classify birth anomalies, were outlined. These included that it is not part of the WHO Family of Classifications, it is outdated, there is no central support system and it does not follow logical classification principles. This classification provides more specificity than ICD-10-AM for birth anomalies.

However, the NCCH has been working with NSW Health to better accommodate concerns about limitations of ICD-10-AM in classifying birth anomalies by incorporating components of the BPA classification.

Some of the advantages of standardised coding were outlined. These included: quality data; benchmarking; ability to compare data nationally; easier training; multiple uses of one coded dataset; and consistent/common language.

Presentation 6

Presenter: Associate Professor Elizabeth Elliott

Title: The role of the Australian Paediatric Surveillance Unit (APSU) in monitoring birth anomalies.

In this presentation, the role of the Australian Paediatric Surveillance Unit (APSU) in monitoring birth anomalies was examined.

The APSU was described as a national unit for the study of rare diseases in children. Surveillance commenced in 1993. Active monthly surveillance of children seen with one of 28 conditions is undertaken by paediatricians or other child health specialists using reply-paid card or e-mail. The APSU is funded by competitive grants from the Department of Health and Ageing and is one of 14 national paediatric surveillance units.

It was noted that a number of birth anomalies have been studied by the APSU, including Congenital Rubella, Congenital Nephrotic Syndrome and Fetal Alcohol Syndrome. A study on Fetal Alcohol Syndrome conducted by the APSU in collaboration with the Telethon Institute of Child Health Research in Western Australia was described.

The general aims of the study were to:

- involve child health specialists in the identification and reporting of Fetal Alcohol Syndrome;
- increase clinician's awareness of Fetal Alcohol Syndrome by providing information on clinical features and diagnostic criteria; and
- conduct a 3-year study to prospectively ascertain newly diagnosed cases of Fetal Alcohol Syndrome by child health specialists in Australia.

The specific aims of the study were to determine:

- incidence of Fetal Alcohol Syndrome;
- epidemiology (gender, age, geography, ethnicity and socio-economic status);
- clinical features, co-morbidity;
- extent of alcohol and other drug exposures in pregnancy (acknowledging study limitations for accurate reporting of alcohol use); and
- use of health services.

The results of the study were presented. The case definition was any child aged <15 years with newly diagnosed Fetal Alcohol Syndrome, suspected Fetal Alcohol Syndrome or partial Fetal Alcohol Syndrome. It was found that there were 69 Fetal Alcohol Syndrome related notifications to APSU in 2001–02. The response rate was 86% and 20 notifications were invalid. There were 17 confirmed cases and 22 probable cases.

General paediatricians notified 9 cases, geneticists notified 6 cases and an endocrinologist and a neonatologist notified 1 case each.

Fetal Alcohol Syndrome was diagnosed in 7 cases and partial Fetal Alcohol Syndrome was diagnosed in 10 cases. No cases had suspected Fetal Alcohol Syndrome.

Confirmed alcohol exposure was reported as high risk (≥ 4 drinks/sitting/week or binge of >5 drinks, during first trimester) for 16 cases. Confirmed alcohol exposure was reported as some risk (<4 drinks/sitting but ≥ 1 /month) for 1 case.

The median age of cases was 2.9 years and ranged from newborn to 12 years. There were slightly more males than females (53% males) and 23.5% of cases were reported as Indigenous, 52.9% were reported as not Indigenous and the Indigenous status of 23.5% of cases was unknown. The mother's birthplace was Australia for all but 1 case. Cases were from New South Wales (52.9%), Queensland (23.5%), South Australia (5.9%), the Australian Capital Territory (11.8%) and the Northern Territory (5.9%). Three cases had other birth anomalies and this was not known for 5 cases.

Some family characteristics of cases were presented. The mother's highest level of education was reported as secondary school for 9 cases and unknown for the other 8. A sibling was also affected for 7 cases and this was not known for 7 cases. The place of residence for cases was reported as the biological parents for 6 cases, the grandparents for 3 cases, adopted for 4 cases and carer and parent for 1 case. The place of residence was not known for 3 cases.

Other substance use in pregnancy was presented. Some children were exposed to more than one drug. The proportion of cases exposed to nicotine was 47.1% and 11.8% were exposed to heroin. Six per cent of cases were exposed to cocaine, glue/solvents and methadone respectively. Other drugs used included marijuana, carbamazepine, benzodiazepines and naltrexone.

Health service usage was also presented. Some children used more than one service. Specialist paediatric services were used by 73.7% of cases, followed by child development/disability services which were used by 68.4% of cases and Department of Community Services services which were used by 52.6% of cases. Remedial education services, respite services and psychological medicine were used by 36.8%, 15.8% and 10.5% of cases respectively.

A comparison between the 17 definite cases and the 22 probable cases showed that high risk use of alcohol was evident for 94% of definite cases and 73% of probable cases. The median age was higher for probable cases (4 years) compared to definite

cases (2.9 years) and the proportion of males in the definite and probable groups was similar (53% and 50% respectively). The proportion of cases reported as Indigenous was markedly higher for probable cases (68%) compared to definite cases (24%). More cases in the probable group had a sibling with Fetal Alcohol Syndrome (55%) compared to cases in the definite group (41%). A higher proportion of definite cases lived with their biological parents (35%) than probable cases (27%).

The birth prevalence of Fetal Alcohol Syndrome in 2001–02 was reported as 0.81 per 100,000 live births for definite cases and 1.82 per 100,000 live births for definite and probable cases.

It was concluded that the APSU currently provides the only national data on Fetal Alcohol Syndrome in Australia and that there is an urgent need for data to underpin interventions for this preventable condition.

It was noted that the strengths of the APSU data are that it is national, prospective, active, cheap, detailed clinical data, timely, high reporting rate, targeted clinicians, only data source, hypothesis generating. The weaknesses are that it is de-identified, so not a disease register, limited data on prognosis, lack of funding, cases seen by paediatric specialists, and does not include terminations of pregnancy or stillbirths.

It was noted that the APSU, Birth Defects Registers and the NPSU could collaborate better to maximise ascertainment and validate data collection. This would require funding, standardised case definitions, cross-reporting and access to data.

Presentation 7

Presenter: Dr Lee Taylor

Title: Perinatal data linkage in New South Wales.

In this presentation, perinatal data linkage in New South Wales was outlined. A number of data sources for the surveillance of population health were noted, including 'census-type' data collections, data from the New South Wales Health Survey Program, data linkage and other data sources (e.g. adverse event monitoring).

It was noted that there are two data linkage programs in New South Wales. One is the annual linkage of data sets which is used as a resource for surveillance purposes, and the other is ad hoc linkage to support research and other projects.

It was reported that the data sets that are linked annually are the NSW Midwives Data Collection (MDC) to the New South Wales Birth Defects Register (BDR), the MDC to the Registry of Births, Deaths and Marriages birth registration data, and the MDC to the Inpatient Statistics Collection (ISC).

A number of examples of ad hoc linkages were provided, including: linkage of the Bankstown–Lidcombe hospital pregnancy ultrasound data with MDC and BDR data to examine false positive and false negative results of prenatal diagnosis with ultrasound; and linkage of the Sydney Obstetric and Gynaecological Ultrasound database with the MDC to examine miscarriage rates following amniocentesis.

It was reported that ethics approval has been granted to carry out linkage of the following data sets for the period 1993–2003: the MDC; the ISC for mothers and infants; and the New South Wales Registry of Births, Deaths and Marriages birth and death registration data. The linked data set will cover mothers, and babies up to 1 year of age.

The legislation and ethics guidelines relating to data linkage in New South Wales were outlined. Data linkage requests that involve internal data sets and are consistent with the original purpose of the collection can be approved by the Chief Executive Officer. Requests for linkage of internal data sets that are not consistent with the original purpose of the collection require institutional ethics committee approval, as do requests for linkage with external data sets.

It was noted that the utility of the data collections, including the New South Wales Birth Defects Register, is greatly enhanced by linkage. Case ascertainment and data quality are improved.

Presentation 8

Presenter: Diane Petrie

Title: The Association of Genetic Support of Australasia Inc. (AGSA).

In this presentation, the Association of Genetic Support of Australasia Inc. (AGSA) was outlined. The AGSA is a peer support consumer organisation and is supported by the New South Wales government.

The aims of AGSA were outlined and include:

- to provide information, support and contacts (support group) for families affected by rare genetic conditions;
- to provide a forum for the exchange of information;
- to educate medical and health professionals; and
- to consult with government bodies for funding.

It was noted that a peer support information officer deals with enquiries and facilitates ongoing support for individuals, families, health professionals and other interested groups.

The AGSA were appreciative of being consulted on the scope of the national birth anomalies system and noted that a comprehensive system of birth anomalies monitoring that includes genetic disorders is important.

Session 3 – Group discussion

Following the presentations, there was general discussion about the need for standardisation in all aspects of the national birth anomalies monitoring system and specifically in all aspects of data development initiatives (including national consistency in data and clinical definitions). There was also support for the adoption of one nationally agreed classification system.

Consultation and participation from stakeholders were seen as being important in the development of the proposed Australian Birth Anomalies System. This includes consultation relating to the inclusion of conditions, for example, internationally agreed commonly occurring birth anomalies and genetic disorders, conditions detected through newborn screening and newborn hearing screening, conditions detected through prenatal screening or diagnostic testing, and other relevant conditions (e.g. resulting from the use of drugs in pregnancy).

There was discussion about the need for adequate funding for the state and territory registers and collections and for the proposed Australian Birth Anomalies System, and how lack of funding limits the ascertainment of cases and the utility of birth anomalies monitoring systems.

It was noted that there was a need to identify key opinion leaders and stakeholders within and external to governments to advocate for birth anomalies monitoring at a national level and to align the activity of the new Australian Birth Anomalies System so that it could provide valuable feedback on the programs and policies of key stakeholders.

Workshop participants agreed that many of the issues identified during the first day needed to be discussed in greater depth and would flow through to discussions held on the second day of the workshop.

Session 4: What functions/activities/programs should a national congenital malformations and birth defects register be able to perform?

The proceedings of the first day of the workshop were reviewed by the Chair in the final session of the day.

A number of issues were then discussed. The first was determining the most important purposes of a national system for the collection of information on birth anomalies. Some of the suggested purposes were to:

- promote the development of an agreed minimum data set to encourage the standardisation of definitions, classifications and collection methods among the states and territories;
- enable the monitoring of rare birth anomalies through the pooling of data from the states and territories;
- enable research on birth anomalies;
- enable the evaluation of national initiatives for primary and secondary prevention of birth anomalies;
- inform planning and policy;
- provide information on the epidemiology of birth anomalies;
- provide a context for the evaluation of reported clusters of birth anomalies; and
- contribute to International reporting and research.

The second issue raised was whether a national system was needed to fulfil these purposes given the existence of state and territory collections. The workshop participants agreed that a national birth anomalies system was required.

The final issue raised considered what the intended scope should be given that a national system was required. The workshop participants agreed that the scope was still to be determined.

The workshop participants also indicated that the NPSU should be the data custodian of a national system.

Day 2

The Chair opened the second day of the workshop and welcomed Richard Madden. The Chair then presented a document that he had prepared after the first day of the workshop which contained four potential models for a national birth anomalies monitoring system. These were:

Model 1 – Universal: The national collection would obtain comprehensive information on all birth anomalies, with a large data set on each notified case. A ‘universal’ system would make use of information collected at birth and during the neonatal period; information collected early in fetal development from abortions

(induced and spontaneous), cytogenetics services, and other diagnostic services; and possibly information collected during infancy and childhood.

Model 2 – Limited: The national system would compile only limited information on birth anomalies. The information could be limited in at least three different ways: (a) the conditions covered by the national system could be limited, e.g. to specified sentinel conditions; (b) the stages in the reproductive process when data are to be collected could be limited, e.g. the national collection could be restricted to birth defects diagnosed at birth (excluding antenatal diagnoses, diagnoses made on aborted tissue, and cytogenetics laboratory diagnoses); and (c) the amount of information assembled at the national level could be restricted to a relatively narrow minimum data set (individual states and territories could collect variables of local interest other than those in the minimum data set).

Model 3 – Support of Jurisdictions: The national system would not necessarily compile case data. Instead, its capacity, resources and expertise would be used to promote birth anomalies data collections in the states and territories, and to develop a nationally consistent standardised collection. While the Australian Government could provide resources for the states and territories, some of the larger states could provide assistance (e.g. with coding) for the smaller jurisdictions.

Model 4 – Selected Jurisdictions: In this model, the national compilation would be based on information provided only by the jurisdictions that had a full capacity to provide it. Over time an increasing number of jurisdictions could be expected to build their capacity to collect and supply data. This model could possibly embrace a staged approach. For example, while a large proportion of jurisdictions might supply data on anomalies diagnosed at birth, national information on conditions diagnosable early in pregnancy (from cytogenetics and aborted tissue) would be provided only by South Australia, Western Australia, and Victoria in the first instance. As other jurisdictions developed the capacity to supply these data, their input would be added to the national compilation.

There was discussion about the four proposed models and their suitability as models for the Australian setting. Agreement was reached by the workshop participants that Model 2 – Limited and Model 3 – Support of Jurisdictions were limited in coverage, utility and quality and were not to be pursued. It was agreed that Model 4 – Selected Jurisdictions was exclusionary and would not be consistent with national data development initiatives, but still needed to be considered. It was felt that some key elements of Model 1 – Universal might be included in a new national model but that the scope of the model did not adequately address the need for development of nationally consistent data and clinical definitions, nor express the need clearly for a uniform classification system. It was noted that Model 1 did not adequately span the conditions and programs that might be included in the national birth anomalies monitoring system.

A suitable model was not developed by the workshop participants in the time available for discussion.

Following this discussion, the three sessions for the second day of the workshop were held. These were:

Session 5: Under what principles should a congenital malformations and birth defects collection operate? What definitions should the congenital malformations and birth defects collection adopt?

Session 6: What are the major barriers to a national congenital malformations and birth defects collection?

Session 7: How should the national system of congenital malformations and birth defects monitoring operate to fulfil its aims and objectives and to overcome barriers?

The sessions are not presented separately as discussion on each overlapped.

National Public Health Partnership

Background information about the National Public Health Partnership's report on public health surveillance of genetic disorders and mapping of current genetic screening services in Australia was presented (NPHP 2002). The need for a national approach to public health genetics which includes adequate surveillance; consistent policies for screening and follow up; ensures access and availability of genetic services at all levels; is systemised to avoid duplication; and fosters partnerships between service providers to promote coordination and delivery of services was identified in the report.

It was noted that the report states that to achieve a national approach special attention is needed to attain national consistency in data collection, specifically around birth defect registers and newborn screening data (NPHP 2002). It was noted that the proposed Australian Birth Anomalies System is consistent with the objectives of the National Public Health Partnership (NPHP) report.

It was also noted that for national monitoring of birth anomalies to attain government and programmatic relevance, partnerships must be developed and strengthened between governments, professional groups and public and private service providers. It was suggested that there needs to be integration of the birth anomalies monitoring system to include prevention services such as screening.

Jurisdictional differences in prenatal screening and prenatal diagnostic testing

It was reported that criteria for and access to prenatal screening and diagnostic testing are determined by each jurisdiction and differ among them.

It was also noted that there are differences in prenatal diagnostic testing for Trisomy 13, 18 and 21 among the jurisdictions. In some jurisdictions (New South Wales, Victoria, South Australia and Western Australia) chromosomal studies to ascertain the karyotype of reported chromosomal abnormalities are undertaken.

Workshop participants acknowledged the differences in trisomy ascertainment, sensitivity of diagnosis and quality assurance among the jurisdictions. They agreed that an analysis of trisomies notifications should be conducted for the jurisdictions which undertake chromosomal studies to determine the proportion of trisomies diagnosed or confirmed by such studies. It was agreed that a working group should be established to undertake the analysis.

Newborn screening program

Information about newborn screening in Australia was presented. It was noted that in Australia all states and territories have a newborn screening program that targets all newborns. All states and territories test for phenylketonuria, congenital hypothyroidism and cystic fibrosis, and all states and territories except Victoria test for galactosaemia (NPHP 2002). It was also noted that newborn screening data are not compiled nationally.

Workshop participants agreed that surveillance of newborn screening programs is an important public health mechanism to inform policy development in relation to genetic conditions. The workshop participants agreed that inclusion of newborn screening data in the proposed Australian Birth Anomalies System should be considered. They agreed in principle that phenylketonuria, congenital hypothyroidism, cystic fibrosis and galactosaemia should be included with a view to extending this to other conditions detected using tandem mass spectroscopy in the future. It was agreed that these proposals should be referred to a working group tasked with determining the conditions to be included in the proposed Australian Birth Anomalies System.

Newborn hearing screening

Background information on newborn hearing screening was presented. It was noted that there is currently no national approach to newborn hearing screening, and that a report on child health screening and surveillance found that there is fair evidence to recommend universal neonatal hearing screening for permanent childhood hearing impairment (NHMRC 2002). It was noted that the report found that hearing screening before discharge for all neonatal intensive care unit neonates and preferably all neonates admitted to special care nurseries for more than 48 hours is now accepted best practice and that the report recommended that this should become a high priority at state level.

Workshop participants agreed that newborn hearing screening was likely to be undertaken nationally in the near future and that permanent childhood hearing impairment detected by newborn hearing screening programs should be included in the proposed Australian Birth Anomalies System in the future.

Early childhood screening

Early childhood screening conducted at 1 year of age was discussed briefly. It was noted that conditions such as cardiac anomalies, cataracts, clicky hips, and Hirschprung's disease can become evident in early childhood. Workshop participants agreed that early childhood screening should not be included in the proposed Australian Birth Anomalies System, but that this could be reviewed in the future.

Sentinel birth anomalies

It was noted that the International Clearinghouse for Birth Defects Monitoring System (ICBDMS) has identified and defined 35 commonly occurring birth anomalies which have been agreed for international reporting. It was noted that New South Wales, Victoria, Western Australia and South Australia are able to report on all of these conditions and that the other jurisdictions are able to report on some of them.

Workshop participants agreed that a working group should be convened to consider the conditions for initial inclusion in the proposed Australian Birth Anomalies System. It was agreed that the 35 ICBDMS conditions and definitions should be used as the starting point. However, it was also agreed that the conditions included should not be limited to these and that some of them may not be relevant to the Australian context.

It was also agreed that the working group should consider the inclusion of four newborn screening conditions (phenylketonuria, congenital hypothyroidism, cystic

fibrosis and galactosaemia), multiple malformations, other chromosomal abnormalities and any other condition identified as being important in the Australian context.

National Minimum Data Set (NMDS)

A NMDS is a core set of data elements agreed by the Statistical Information Management Committee (SIMC) and endorsed by the National Health Information Group (NHIG) for mandatory collection and reporting at a national level. The collection and reporting of an NMDS is undertaken under the auspices of the Australian Health Ministers' Advisory Council through the National Health Information Agreement.

It was noted that there is no NMDS for birth anomalies. For the current National Congenital Malformations and Birth Defects Data Collection, the NPSU requests an agreed set of data elements. However, the specification for the collection was not agreed through the governance arrangements for health information management and information technology described above. Some of the data elements requested are specified in the Perinatal NMDS.

Workshop participants acknowledged that there was variation among the states and territories in the scope of their birth anomalies collections, the definitions and classifications used and the method of data collection.

They agreed that a NMDS should be developed for birth anomalies. They agreed that the details of the NMDS were beyond the scope of the workshop and that a working group should be convened to develop it following the process required under the governance arrangements.

It was also agreed that in the interim a national agreement on a minimum set of data to be reported should be obtained.

Workshop participants discussed whether age at diagnosis should be included in the proposed Australian Birth Anomalies System and agreed that it should be.

Classification of birth anomalies

It was noted that in the survey of state and territory birth anomalies data collections, variation among the states and territories in the classifications used to code birth anomalies was found (see Chapter 4).

Workshop participants agreed that there was a need for states and territories to work towards using a single classification and that this should be the International Statistical Classification of Diseases and Related Health Problems, 10th Revision, Australian Modification (ICD-10-AM).

Workshop participants acknowledged that ICD-10-AM lacks specificity with regard to birth anomalies. However, it was noted that the National Centre for Classification in Health (NCCH) has tried to address this in the fourth edition of ICD-10-AM.

Workshop participants agreed that a working group should be convened to address the issue of specificity and that the working group should include a representative from the NCCH, a clinical coder and auditor with experience working in the area of newborn care, and representatives from jurisdictions which use different classifications.

Period for detecting birth anomalies

In the survey of state and territory birth anomalies data collections, it was found that there is variation in the period of detection of birth anomalies among the states and territories (see Chapter 4). For example, the detection period for Tasmania begins at birth and ends 28 days after birth, but for Victoria, the detection period begins prenatally and ends at 15 years of age.

The workshop participants considered a timeline for detecting birth anomalies. It was noted that if the timeline started at conception then it would include all terminations of pregnancy for fetal abnormality. It was acknowledged that inclusion of terminations of pregnancies with birth anomalies is not currently the practice in all states and territories, but there was general agreement that this would be best practice.

The end point of the period for the detection of birth anomalies was discussed. Currently, the end point of the detection period is notionally 28 days. However, the workshop participants agreed that ending the detection period at 1 year of age would be best practice at this stage.

It was decided that a working group should be convened to determine the benefits of ending the detection period at 1 year of age compared to 28 days of age with respect to case ascertainment.

It was acknowledged that increasing the detection period to 1 year of age could be difficult for some jurisdictions as changes in legislation may be required and there may be resource limitations. It was proposed that if a period of detection of up to 1 year of age was agreed, a staggered implementation could be considered.

It was also recognised that, for a few birth anomalies, case ascertainment and definition could be improved by extending the period of detection beyond 1 year, for example, hypospadias for which surgery is performed at around 12 months of age. The risk of using a shorter time period is that most of these cases would go unrecognised and the cases that are described may be incidental findings associated with other conditions. Cases could be defined by their need for hospital admission and therefore could be identified from inpatient data or hospital surgery lists.

Terminations of pregnancies with birth anomalies

It was noted that terminations of pregnancies with birth anomalies where the gestational age was <20 weeks are not routinely notified to all birth anomalies registers or collections in all states and territories. It was acknowledged that some of the barriers to notification of terminations of pregnancies with birth anomalies were lack of legislation and data ownership issues related to private providers of prenatal diagnostic testing services.

It was noted that South Australia is the only state that is notified of all terminations of pregnancy from all sources.

Workshop participants agreed that, terminations of pregnancies with birth anomalies should be included in the proposed Australian Birth Anomalies System, irrespective of gestational age. It was noted that it is important to include these data because they affect prevalence rates of birth anomalies.

It was proposed and agreed that for national reporting, data on terminations of pregnancies with birth anomalies should be presented in a separate section of the

national report. This would enable the data to be reported separately for jurisdictions that had full, partial or no reporting of terminations of pregnancy data.

It was agreed that notification of data on terminations of pregnancies with birth anomalies, especially from private service providers, should be explored in another forum and that a working group should be convened to address state and territory implementation issues and that this issue should be considered by that working group.

State and territory reporting

It was noted that the survey of state and territory birth anomalies data collections revealed variation among the states and territories. It was noted that four jurisdictions (New South Wales, Victoria, Western Australia and South Australia) have dedicated Birth Defects Registers and that these are of high quality. It was noted that these states have more comparable collections than the other jurisdictions. For example, notifications of birth anomalies are received from a wider variety of sources than for other jurisdictions, including from cytogenetic and pathology reports, notification of birth anomalies detected prenatally and up to 1 year of age are received, and notifications of terminations of pregnancies with birth anomalies where the gestational age is <20 weeks are received.

It was noted that the lack of uniformity among the states and territories is problematic for national reporting. Workshop participants considered whether reporting of birth anomalies should be limited to the four jurisdictions with more consistent and comparable data. It was agreed that a capacity building approach towards including all states and territories should be promoted.

Resource implications

It was noted that resource allocation to Birth Defect Registers and birth anomalies data collections has been limited in all jurisdictions. This has had an effect on the utility and quality of the data systems and on the development of nationally consistent systems.

The workshop participants agreed that the issue of funding was regarded as fundamental to the ongoing development of an Australian Birth Anomalies System, but that the details were not in scope for this workshop.

They agreed, however, that recurrent core programmatic funding was needed to sustain birth defects registers at both the state and territory and national levels; and that in the short term additional 'catch-up' funding was needed to develop a nationally consistent birth anomalies monitoring system and to support the jurisdictions with less developed birth anomalies data collections.

Key recommendations

- That the National Congenital Malformations and Birth Defects Data Collection continue and that it be referred to as the Australian Birth Anomalies System (ABAS).
- That the custodian of the ABAS be the NPSU.
- That a working group be convened to consider the conditions for initial inclusion in the ABAS. Consideration should be given to including the 35 ICBDMs conditions, four newborn screening conditions (phenylketonuria, congenital

hypothyroidism, cystic fibrosis and galactosemia), multiple malformations, other chromosomal abnormalities and any other condition identified as being important in the Australian context.

- That terminations of pregnancies with birth anomalies be included in the ABAS, regardless of gestational age.
- That a working group be convened to address state and territory implementation issues, including consultation with relevant stakeholders to examine access to data on terminations of pregnancies with birth anomalies (public and private providers).
- That birth anomalies detected up to 1 year of age be included in the ABAS in the first instance.
- That a single classification be used for coding birth anomalies and that this should be ICD-10-AM.
- That a working group be convened to address issues of lack of specificity for birth anomalies in ICD-10-AM.
- That a National Minimum Data Set for birth anomalies be developed following the process required under the governance arrangements for health information management and information technology.
- That an interim Minimum Data Set be agreed nationally.
- That recurrent core programmatic funding be identified for birth anomalies collections at the state and territory and national levels.
- That a National Birth Anomalies Steering Committee be established to advise on the scope, development and implementation of the ABAS.

Proposed Steering Committee and Working Groups

It was proposed that a National Birth Anomalies Steering Committee be established to provide expert advice on the scope, development and implementation of the proposed Australian Birth Anomalies System. The steering committee should comprise 2-3 state and territory representatives, a representative of the Department of Health and Ageing (DoHA), a representative of the Department of Family and Community Services (FaCS), and other relevant stakeholders, including from clinical, epidemiological and technical backgrounds. The steering committee should meet on a regular basis, including one annual face-to-face meeting.

The formation of a number of working groups to undertake work arising from the workshop was also proposed and members of each group were decided. The Steering Committee should oversee the work of these groups. The proposed working groups were:

Working group 1

Purpose: Identification of the conditions to be initially covered in the Australian Birth Anomalies System (ABAS)

Members: David Tudehope, Liz Elliott, Lee Taylor, Chris Stone, Elizabeth Sullivan, and Carol Bower

Working group 2

Purpose: Data development including agreeing and defining a National Minimum Data Set (NMDS) for birth anomalies.

Members: Seeta Duvasula, Annabelle Chan, Merrilyn Riley, Elizabeth Elliott, Elizabeth Sullivan and Sue Cornes

Working group 3

Purpose: Examination of the notification of trisomies data; conduction of an analysis on the different periods of detection of birth anomalies (i.e. birth, 28 days, 1 year of age).

Members: Lee Taylor, Carol Bower, Jane Halliday, Annabelle Chan, Elizabeth Sullivan

Working group 4

Purpose: Coding in ICD-10-AM and conversion to ICD-10-AM from ICD-9-BPA.

Members: Elizabeth Elliott, Susan Travis, Kerry Innes and John Edward

Working group 5

Purpose: State and territory implementation issues, including issues related to notification of terminations of pregnancy with birth anomalies.

Members: Lee Taylor, Carol Bower, Jane Halliday, Annabelle Chan, Suzanne Cornes, Elizabeth Sullivan, Maureen Bourne, Karen Wheeler and Margaret Stewart.

Further work was also proposed in a number of other areas, but working groups were not established to undertake this work. These areas were:

- development of the content and structure of the national report; and
- consideration of ways to broaden the usage of Australian Birth Anomalies System data (nationally and internationally).

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