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Clarifying the lower incidence of birth defects in the midwest of Western Australia : A study using capture re-capture methodology

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CLARIFYING THE LOWER INCIDENCE OF BIRTH

DEFECTS IN THE MIDWEST OF WESTERN AUSTRALIA,

A STUDY USING CAPTURE RE-CAPTURE

METHODOLOGY

A thesis submitted in fulfilment of the requirements for the degree of

Master of Science – Midwifery

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ABSTRACT

In April 2000 the Western Australian Birth Defects Registry held its 20th Anniversary Scientific Symposium to signify the first 20 years of the Birth Defects Registry in Western Australia. At the Symposium, significant trends and occurrences in birth defects within Western Australia over the past 20 years were addressed. One factor highlighted in the Registry's 1980 – 1999 report was that birth defects were reported less frequently in rural regions compared with metropolitan areas. Data collected from the metropolitan area is considered by the Birth Defects Registry to be the most accurate. Therefore, incidence rates of birth defects in this area are used as a benchmark for determining the incidence of birth defects throughout Western Australia.

The aim of this study was to explore the lower incidence of birth defects in the Midwest of Western Australia compared with that of metropolitan Western Australia. The study investigated if the observed lower incidence of birth defects in the Midwest was due to case under reporting. The study target population included newborns whose mothers were Midwest residents at the time of their birth between the years of 1995 – 1999. A mixed methodological approach, combining capture-recapture and thematic analysis was used.

The study was conducted in two phases. The first phase involved the collection of quantitative data. The potential target population in the Midwest consisted of 5066 births. A sample of 440 newborn case notes were selected randomly from the target population and accessed from client files held at Geraldton Regional Hospital. A record was made of any reportable birth defects. The results provided demographic and descriptive statistics, which were compared with data held by the Birth Defects Registry

regarding the Midwest. This mode of sample collection is known as capture-recapture methodology. Comparison of data from the sample with data from the Registry was used to document the existence of discrepancies and to enable an ascertainment-adjusted analysis to be performed. To address the study's aims, the ascertainment adjusted rate was compared with the rates occurring in the metropolitan area.

In Phase 1, quantitative findings showed the lower incidence of birth defects diagnosed in infants during the newborn period in the Midwest of Western Australia was unlikely to be due to case under reporting. Possible factors contributing to the lower incidence of birth defects in the Midwest compared to the Metropolitan area were discussed. These include the lower maternal age of Midwest mothers, a higher Aboriginal population, study sample bias and case under-reporting beyond the newborn period. Full exploration of these issues, however, was outside the scope of this study and is an area for future research.

The second phase of the study involved collection of qualitative data. Health professionals reporting birth defects in the Midwest were asked to participate in focus group interviews. Three groups of health professionals were targeted: general practitioners; midwives; and child health nurses. Interview responses were transcribed and indexed according to common themes and issues. This analysis provided insight into the cause of empirical findings and discrepancies, allowing for recommendations focused on improving and/or sustaining birth defect notification practices of health professionals in the Midwest of Western Australia. Findings showed that a knowledge deficit regarding the reporting of birth defects exists in targeted Midwest health professionals. Possible reasons for the knowledge deficit included high transfer rates of newborns affected by birth defects, uncertainty regarding the responsibility of birth defect reporting, limited use of birth defect notification cards by health professionals

and the lack of ongoing education and promotion of birth defects in the Midwest Region.

Study recommendations include further research to explore factors contributing to the lower incidence of birth defects in the Midwest, education and promotion regarding birth defects and the role of the Birth Defects Registry, amending and centralising birth defect notification cards, and legislating the reporting of birth defects in Western Australia.

DECLARATION

I certify that this thesis does not, to the best of my knowledge and belief:

(i)   incorporate without acknowledgement any material previously submitted for a degree or diploma in any institution of higher education;

(ii)   contain any material previously published or written by another person except where the reference is made in the text; or

(iii)   contain any defamatory material.

Signature: ___████████___

Donna Baker

November 20th, 2003

ACKNOWLEDGEMENTS

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CHAPTER 1

INTRODUCTION

Advances in perinatal, neonatal and infant care have resulted in a decline in mortality and morbidity caused by congenital malformations. Perinatal death rates in Australia due to congenital malformation has declined dramatically from 35.9 per 10000 births in 1973, to 15.6 per 10000 births in 1996 (National Perinatal Statistics Unit, 2002a). Infant death rates due to congenital malformation also declined from 28.8 per 10000 live births in 1980, to 14.8 per 10000 live births in 1996 (National Perinatal Statistics Unit, 2002a). Many factors have contributed to the declining rate of congenital malformations within our community. These include improvements in antenatal care, women's health and advances in ultrasound technology. However despite these declining incidence rates, congenital malformations still contribute significantly to perinatal and infant mortality and morbidity. Congenital malformations represent the second leading cause of death, surpassed only by Sudden Infant Death Syndrome (Lynberg & Edmonds, 1993). Much remains to be learned about congenital malformation. While 20% of malformations can be attributed to known causes, the causes for the remaining 80% are still unknown (Lynberg & Edmonds, 1993).

Societies require the capacity to monitor population trends in maternal and child health in order to facilitate further study of the major causes of disease in pregnancy and childhood, in addition to evaluating medical intervention and public health policies. These activities could be more easily accomplished if vital population based statistics in the form of databases were readily available (Stanley,

Croft, Gibbins, & Read, 1994). Surveillance systems provide much of the data used to compile database information that forms the basis for public health initiatives (LePorte et al., 1995).

The foundation for further improvement in rates of congenital malformation or birth defects, (both terms will be used interchangeably throughout the study), relies upon implementation of an accurate and comprehensive birth defects surveillance system and database. Surveillance is a critical component in the effort to further reduce the impact of birth defects on public health. Birth defect surveillance systems are responsible for collecting, collating and analysing information relating to birth defects (Lynberg & Edmonds, 1993). Surveillance provides the information necessary to detect the occurrence of birth defects, to investigate potential etiologic agents, to plan and evaluate the effects of interventions, and to ensure appropriate care for people in need of services as a result of birth defect related conditions (Lynberg & Edmonds, 1993).

In Western Australia the Birth Defects Registry is responsible for monitoring congenital malformations. The Birth Defect Registry maintains a database of all newborns and children born in Western Australia who have birth defects diagnosed before the age of six years (Bower, Rudy, Ryan, & Cosgrove, 2000). Registry database information is used to:

- establish the incidence and prevalence of birth defects in Western Australia;
- generate research into the causes and prevention of birth defects;
- investigate changes in the frequency of birth defects;
- provide health professionals with statistical information;
- evaluate screening programs, prevention and treatment strategies of birth defects, to assist with planning of health care facilities; and
- to increase general knowledge about birth defects.

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To effectively achieve these tasks, it is crucial that Birth Defect Registry data are both accurate and complete. In April 2000, the Western Australian Birth Defects Registry held its Twentieth Anniversary Scientific Symposium, to signify the 20th anniversary of the Registry's establishment in Western Australia (Bower et al., 2000). Trends and occurrences in birth defects in Western Australia during 1980 – 1999 were presented at this symposium. A significant factor highlighted in the report was evidence that indicated a lower incidence of birth defects in rural areas, compared with that of the metropolitan area. Furthermore, the report suggested the differing incidence of birth defects was possibly due to the under ascertainment of case numbers rather than a real difference (Bower et al., 2000).

1.1 Purpose

The purpose of this research was two fold:

1.   To determine if the observed lower incidence of birth defects in the Midwest are due to under reporting.

2.   To identify and make recommendations that will sustain or improve upon current birth defect case ascertainment methods.

1.2 Research Questions

The following research questions were explored:

1.   Have birth defects occurred in the Midwest between the years 1995-1999 that have not been recorded by the Birth Defects Registry?

2.   Is the lower incidence of birth defects in the Midwest due to case under reporting?

3.   What recommendations can be made to sustain or improve upon the current case ascertainment methods of the Birth Defects Registry?

1.3 Background

The Western Australian Birth Defects Registry was established in 1980 with the aim of creating a comprehensive database. It's purpose is to facilitate research into the etiology of birth defects, provide direction toward implementing and evaluating preventative health strategies, as well as the ongoing treatment and management of persons with birth defects (National Perinatal Statistics Unit, 2002a), (Bower & Stanley, 1983).

A birth defect is defined by the Registry as an abnormality (probably of prenatal origin) including structural, biochemical, genetic and chromosomal defects (Bower et al., 2000). Most minor malformations that do not cause disfigurement or require treatment are excluded. Appendix 1 contains the Registry's list of excluded birth defects.

A number of information sources are used by the Registry to collect data, and include statutory and non-statutory sources. Statutory sources include midwives' notification data, as well as hospital morbidity and mortality data. Collection of these forms of data is a statutory requirement, and information is provided to the Department of Health in Western Australia (HDWA), as well as the Registrar of Births, Deaths and Marriages. The Birth Defects Registry accesses this information directly from the HDWA. Additionally, Registry information is collected via voluntary notification of birth defect cases from a number of sources. As the name implies, notification of birth defects from these sources holds no statutory

requirement and occurs purely on a voluntary basis. Sources of voluntary notification include the following (Bower et al., 2000):

- paediatric hospital and paediatric hospital speciality departments;
- obstetric hospitals and obstetric hospital speciality departments;
- other hospitals;
- cytogenetic services;
- pathology services;
- genetic services;
- special services;
- private-practitioners;
- child and community health nurses and doctors; and
- the Rural Paediatric Service.

Voluntary notifiers are requested to complete predistributed notification cards that are available at their place of work. Completed cards are either forwarded to, or collected periodically by Registry staff. Registry staff visit metropolitan hospitals regularly, on average eight times per year. The frequency in which Registry staff attend rural hospitals to collect notifications cards is not predetermined and is usually coordinated with visits for other purposes. These visits range from several times a year to not at all. A copy of this notification card has been included in Appendix 2.

This study used data collected from the 1995-1999 birth cohort. Comparatively, the metropolitan regions of Western Australia have a higher incidence of birth defects than rural areas. The Midwest and Southwest regions of Western Australia have the lowest reported incidence of birth defects in the state. This data will be further examined in the next chapter. This study was limited to the Midwest region of Western Australia due to the constraints of a Masters thesis, study support available to the researcher, and the low incidence of birth defects in the Midwest. The

researcher was offered support consisting of access to an office, computer, transport and accommodation by the Combined Universities Centre for Rural Health (CUCRH), which is located in Geraldton (the main health administrative center for the Midwest).

In summary, the aim of the study was to clarify the lower incidence of birth defects in the Midwest of Western Australia and determine if the observed lower incidence is due to case under reporting. In addition, the study identified and made recommendations designed to sustain or improve upon current birth defect case ascertainment methods. The following section will discuss further background to the study and factors relevant to the collection of Midwest data.

CHAPTER 2

THE MIDWEST PERSPECTIVE

2.1 Description of the Midwest Region

The Midwest health zone/region is located 400km north of Perth (the capital city) in Western Australia (WA). The coastal centres of Geraldton and Carnarvon are the two largest townships within the region. The majority of the region's population is located in Geraldton (Figure 1). Geraldton is the third largest population centre in WA with 24 361 residents. The major forms of industry in the region include grain, sheep, mineral sands, agriculture and fishing.

Data collected by the Birth Defects Registry are analysed and then compared according to geographical distribution within the state. This is based upon the mothers' residential postcode at the time of giving birth. Maternal residences are grouped into health zones according to their geographical regions. The classification of Health Zones used by the Registry is displayed in Figure 1.

Health services in the Midwest Region are provided mainly by the Geraldton Regional Hospital, or the St John of God Private Hospital. General medical, surgical, paediatric and maternity services are available at both hospitals. Complicated or high-risk patients are transferred to tertiary referral centres in Perth for care. In addition, there is a smaller hospital located at Carnarvon, as well as nursing posts scattered throughout the area. Available community services include general practitioners, community and child health nurses, and a small contingent of private allied health professionals.
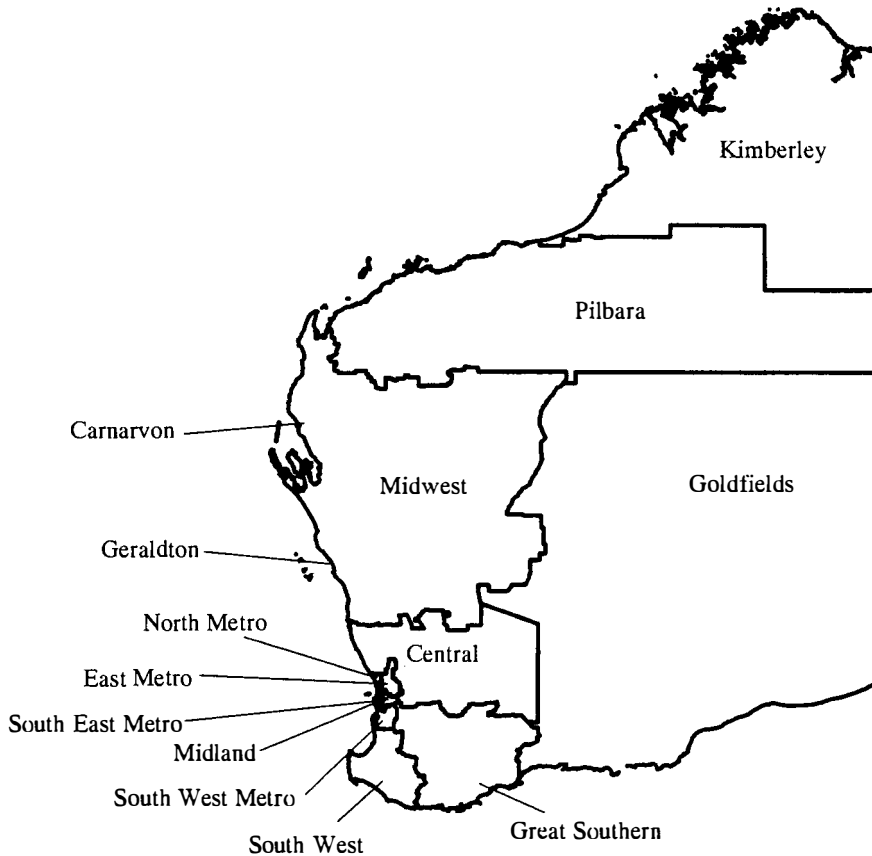
Figure 1. Classification of Health Zones/Regions within Western Australia.

A few specialist practitioners reside in the area, however, most originate from the Perth metropolitan region and travel to the Midwest to provide services at visiting clinics. Notification of birth defects to the Registry could potentially be made by any of the above mentioned health professionals.

The proportion of birth defects occurring in each health zone can be calculated as a percentage of the number of newborns born each year. A breakdown of percentages for the years 1995 – 1999 in each health zone appears in Table 1. This table shows evidence of lower birth defect incidence rates in rural health zones, including the Midwest, Goldfields, Great Southern and Southwest. Of note, the lowest incidences of all rural areas occurred in the Midwest and Southwest regions, and were 4.0% and 3.9% respectively.

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Table 1

Percentages of birth defects in each Western Australian health zone/region, for 1995 – 1999

Year of Birth	East Metro	North Metro	South Metro	Midland	Midwest	Goldfields	Great Southern	South West	Pilbara	Kimberley
1995	4.9	6.6	8.4	5.5	4.9	5.3	4.6	4.2	5.2	4.0
1996	4.8	6.9	8.7	5.4	4.2	5.3	5.3	3.0	4.6	4.6
1997	6.9	6.6	6.5	NA	2.8	4.1	3.9	4.6	4.9	4.5
1998	5.9	5.3	5.5	4.5	4.6	3.6	4.8	4.9	4.6	6.2
1999	5.9	5.9	5.2	5.6	3.4	4.2	5.1	3.2	4.5	4.1
Average	5.6	6.3	6.8	5.2	4.0	4.5	4.7	3.9	4.76	4.6

2.2 Description of the Midwest Population

To explore the observed lower incidence of birth defects in the Midwest it is useful to examine the Midwest population in terms of overall birth rates, gender, Aboriginality and maternal age. Midwest statistics can then be compared with similar incidences occurring in the Metropolitan and other Rural Regions.

Table 2 presents information in terms of maternal age and the proportion of births occurring in metropolitan and rural areas of WA between 1995-1999. For comparison purposes the health zones outlined in Table 1 have been combined to represent the Metropolitan, Midwest and Other Rural Regions. The Metropolitan Region consists of the North, East, South East, Midland and South West Metropolitan health zones. A combination of the remaining health zones, with the exception of the Midwest Region, is represented as Other Rural Regions.

Table 2.

Percentage of Births according to Region and Maternal Age Group between 1995 –
1999, in WA.

Maternal Age (years)	Metropolitan n(%)	Midwest n(%)	Other Rural n(%)
< 20	4671(5)	467(9)	2433(8)
20 - 24	15434(17)	1140(22)	6391(21)
25 - 29	29401(32)	1677(33)	10336(34)
30 - 34	28284(31)	1252(25)	7894(26)
35- 39	12368(13)	462(9)	2882(9)
40 +	2011(3)	56(1)	433(1)

Proportionately, birth rates of mothers less than 30 years of age in WA are
higher in the Midwest and Other Rural Regions, compared with metropolitan
mothers. In contrast, proportionately higher rates of births occurred in the
Metropolitan Region to mothers aged 30 years and above, compared with mothers
from the Midwest and Other Rural Regions.

The demographics of the Midwest can also be examined according to
Aboriginality and non-Aboriginality. Table 3 presents information according to the
total number of births in WA and the percentage of Aboriginal and non-Aboriginal
origin. The total number of WA births represented in Table 3 are only for the years
1996 –1999, this accounts for the discrepancy between totals shown in Table 2 and
Table 3.

Table 3.

WA Births according to Region, Aboriginality and non-Aboriginality, between
 1996 – 1999.

Regions	Total Births	Aboriginal Births	non Aboriginal Births
	N	n(%)	n(%)
Midwest	4184	710(17)	3474(83)
Other Rural	27125	3496(26)	23629(82)
Metro	70560	195(3)	68649(97)

The Midwest and other Rural regions have a significantly higher percentage of Aboriginal births compared with the Metropolitan region. The percentage of total births in these two regions of Aboriginal origin was 17% and 26% respectively, compared with 3% in the Metropolitan region. The Midwest, Metropolitan and Other Rural regions can also be examined according to gender. The gender of all births in Metropolitan, Midwest and Other Rural regions of WA area listed in Table 4. Similar proportions of male and female births occurred in each of the regions with a small male preponderance across all regions (approximately 51% male and 49% female).

Table 4.

WA Births according to Region and Gender in WA between 1996-1999.

Region	Total Births	Males	Females
	N	n(%)	n(%)
Midwest	4184	2114(50)	2070(49)
Other Rural	27125	13972(50)	13149(48)
Metro	70560	36174(51)	34423(49)
Total	101869	52260(51)	49642(49)

2.3 Description of Birth Defects in the Midwest Population

Previously discussed population demographics regarding maternal age, Aboriginality and gender can be further examined in terms of birth defects. Table 5 shows the total rate of birth defects according to maternal age group in the Midwest, Metropolitan and Other Rural regions. Overall, higher rates of birth defects occurred in the Metropolitan region than in Other Rural and Midwest regions. Higher rates of birth defects were seen in older mothers in all regions. With the exception of the 40+ age group mothers in the Midwest (based on only 8 cases), all age specific rates for Midwest mothers were less than those for Metropolitan mothers.

Table 5.

Total Birth Numbers and Rates of Birth Defects according to Maternal Age and Region in WA between 1995-1999

Maternal Age	Metro Births	Metro Birth Defects	Midwest Births	Midwest Birth Defects	Other Rural Births	Other Rural Birth Defects
	N	n(%)	N	n(%)	N	n(%)
<20	4671	232(5)	467	14(3)	2433	100(4)
20-24	15434	802(5)	1140	42(4)	6393	285(4)
25-29	29401	1727(6)	1677	74(4)	10336	501(5)
30-34	28384	1729(6)	1252	57(4)	7894	425(5)
35-39	12368	836(7)	462	18(4)	2882	141(5)
40+	2011	139(7)	56	8(14)	433	27(6)
Total	92269	5465	5054	213	30371	1479

Table 5 shows the rates of birth defects per 1000 of all births in each maternal age group and region. Overall, there were higher rates of birth defects in the Metropolitan region compared with Other Rural and Midwest regions.

Essentially, these findings concur with Australian statistics regarding the incidence of birth defects and maternal age. The Australian National Perinatal Statistics Unit reports that during the period 1993 to 1996 the national malformation rate in mothers aged 40 years and over was almost double that found in mothers aged 20-24 years (National Perinatal Statistics Unit, 2002b). In addition, chromosomal abnormalities were more likely to occur in advanced maternal age, with women aged 40 years and over being 13 times more likely than women aged 20-24 years to give birth to an infant with chromosomal abnormalities (National Perinatal Statistics Unit, 2002b). Australian statistics (National Perinatal Statistics Unit, 2002b) show an increasing number of women are deferring childbearing. In 1999, almost 1 in 10 mothers (9.5%) were having their first baby at the age of 35 years or older (National Perinatal Statistics Unit, 2002b).

The occurrence of birth defects within the Midwest region can be considered in terms of Aboriginality together with gender. As previously mentioned, the Birth Defect Registry 1980-2000 report findings (Bower et al., 2000) highlighted a lower incidence of birth defects in the rural regions of WA compared with the metropolitan area. In addition, birth defects were reported less frequently in Aboriginal newborns compared with non-Aboriginal newborns and in general occurred more commonly in male newborns and multiple pregnancies (Bower et al., 2000). To further explore these statements it is useful to review the total number of births in each region according to gender and Aboriginality. Table 6 shows the numbers and rates of birth

defects in terms of gender and aboriginality in Midwest, Metropolitan and Other Rural areas, between 1995-1999.

Table 6 shows the occurrence of birth defects in males is higher than females in all regions. In addition, the birth defect rate is lower in the Aboriginal population in all regions compared with the non-Aboriginal population. These statistics are consistent with Birth Defect Registry 1980-2000 Report findings that suggest birth defects are higher in males and reported less frequently in Aboriginal newborns (Bower et al., 2000). In view of this finding, the likelihood that the proportionately higher percentage of Aboriginal births in the Midwest region may in part, be responsible for the lower incidence of birth defects in the Midwest should be considered. Midwest trends will be reviewed in conjunction with study findings in the discussion chapter.

Table 6.

Birth Defect Numbers and Rates according to Gender, Aboriginality and Region in WA between 1996-1999

Region	Total Births	Birth Defects	Male Birth Defects	Female Birth Defects	Non-Aboriginal Birth Defects	Aboriginal Birth Defects
	N	n(%)	n(%)	n(%)	n(%)	n(%)
Midwest	4184	148(35)	84(40)	63(30)	127(37)	21(30)
Other Rural	27125	1016(38)	597(43)	406(31)	886(38)	130(32)
Metro	70560	4154(59)	2267(63)	1826(53)	4067(59)	87(44)
Total	102225	5349(52)	2948(54)	2295(46)	5080(53)	238(39)

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Figure 2 shows birth defects by type and region within WA between 1996-1999 (Bower, 2002). A larger proportion of all types of birth defects occurred in children whose mothers resided in the Metropolitan region. Central Nervous System (CNS) defects occur twice as often in the Metropolitan population (13.0) compared with the Midwest population (7.0). Gastrointestinal (GIT) defects were the only defects that occurred in similar proportions in the Metropolitan (7.0), Midwest (5.5) and Other Rural (5.5) regions. As previously mentioned these findings will be considered in the discussion chapter.
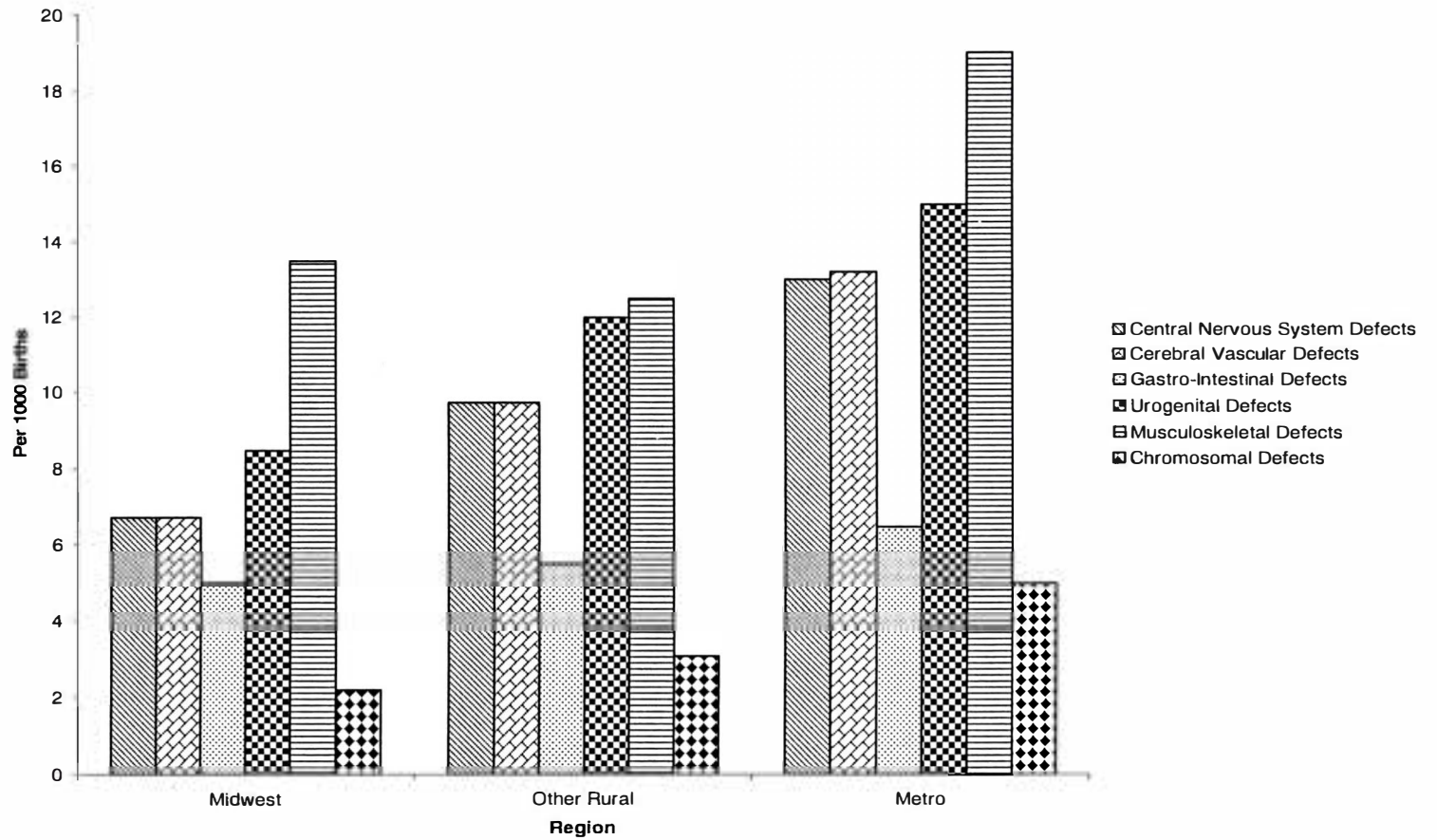
Figure 2. Birth Defects by Type and Region in WA between 1996 – 1999 (Bower, 2002).

2.4 Notification of Midwest Birth Defects to the Birth Defects Registry

During the period of 1995 – 1999 in the Midwest, a total of 451 notifications were reported to the Birth Defects Registry. The notifications related to 213 actual birth defect cases. This represents an average of two notifications per case (Bower, 2001). Multiple notifications of the same case were counted only once. Of the 451 notified cases of birth defects, 213 were confirmed. Furthermore, of the 213 confirmed birth defect cases, only 124 notifications came from Midwest sources, with the remainder of notifications originating from sources in the metropolitan area. The Midwest sources of birth defect data notification and the frequency in which these sources reported birth defect cases are shown in Table 7 (Bower, 2001). Midwife and Hospital Morbidity data were the most frequent sources of Midwest birth defect notification. In contrast rural hospitals and doctors notified the least number of birth defect cases to the Birth Defects Registry.

Table 7 illustrates the discrepancy in the number of birth defect cases notified by Midwest sources compared with the number of cases known to exist. This shortfall suggests possible under reporting of birth defect cases by Midwest notifiers to the Birth Defects Registry. Furthermore, such a discrepancy also poses questions regarding the completeness of data held by the Registry. This discrepancy further supports and justifies the aim of this study.

Table 7.

Rural Notifiers of Births with Birth Defects in the Midwest Region, 1995 – 1999.

Sources of Notification	Cases notified
	n
Midwest midwives	48
Death certificates	9
Midwest Hospital morbidity data	33
Rural hospitals	4
Rural doctors	5
Rural child health nurses	14
Rural paediatric service	12
Total	124
Total cases 1995 –1999	213

In summary, the description of births and birth defects within the Midwest population between 1995 – 1999 indicates the following:

1.   The incidence of birth defects is lower in the Midwest region compared with the Metropolitan region of WA.

2.   There is a higher percentage of births in mothers aged less than 30 years in the Midwest region compared with the Metropolitan region.

3.   The rate of birth defects increases with increasing maternal age in all regions.

4.   The percentage of Aboriginal births is higher in the Midwest and Other Rural regions compared with the Metropolitan region.

5. The rate of birth defects in all maternal age groups, male infants, female infants, Aboriginals and non-Aboriginals is lower in the Midwest region than the Metropolitan region.

6. Birth defects rates of all types are higher in the Metropolitan region than the Midwest and Other Rural regions.

7. Few notifications to the Birth Defects Registry regarding birth defects occurring in Midwest births originated from Midwest case reporting sources.

The incidence of birth defects and their associated complications are a health concern for newborns and children worldwide. Methods of detection, surveillance and prevention of birth defects varies worldwide. In order to provide further background to this study and its methodology it is beneficial to review birth defect surveillance from an international perspective. The following section will review the literature regarding to the surveillance and evaluation of birth defects, as well as literature relating to the study methodology.

CHAPTER 3

LITERATURE REVIEW

Birth defects can potentially affect all newborns regardless of race, culture, religion or socio-economic status, although some birth defects appear more commonly among certain racial groups. Many types of congenital abnormalities exist, each varying in incidence and degree of associated mortality and/or morbidity (Bromberg Bar-Yam, 1999). Birth defects can be caused by genetic mutation that may be the result of maternal illness, maternal drug use and/or environmental factors. However, the cause of a significant portion of congenital abnormalities remains unknown. Effective birth defect surveillance is essential, in order to understand more fully the causes of birth defects, the effect they have on public health, and the steps required to further reduce their incidence (Bromberg Bar-Yam, 1999).

Surveillance is defined as the process of monitoring the occurrence of a condition over time in a population of a specified geographic area. Surveillance data are compiled by a registry. Development of modern registries is usually based on one or more of the following objectives: to conduct surveillance, to develop etiological studies, and/or to determine health service needs (Cordero, 1992). Cordero (1992) cited examples of specific applications for data collected by registries of birth defects and genetic diseases. For clarity, these have been included in Figure 3.

Surveillance

Calculate baseline rates

Study trends over time

Identify geographic clusters

Analytic Studies

Conduct epidemiologic studies to search causes and risk factors

Test hypothesis of potential teratogenic risk

Health Services Research and Evaluation

Identify children with special health needs

Determine needs for health care services

Evaluate access and availability of medical services

Figure 3. Examples of Applications for Data Collected by Registries of Birth Defects and Genetic Diseases.

Information derived from population surveillance provides the basis for many public health initiatives such as evaluating and improving health care services and facilities, in addition to facilitating health promotion and prevention programs. To be effective these initiatives must be generated from complete and accurate population surveillance information. The first step in determining the efficiency of this

information is to assess its completeness (LePorte et al., 1995). A popular approach for assessing population and source completeness used by epidemiologists is known as capture-recapture methodology (Brenner, Stegmaier, & Ziegler, 1994). Capture-recapture methods have been primarily used in epidemiology to estimate the degree of undercounting in the data ascertained by surveillance/registry systems. These methods can also be used to adjust for population undercounting by calculating ascertainment-corrected rates (International Working Group for Disease Monitoring and Forecasting, 1995a).

This chapter will first examine the attributes of an ideal birth defects surveillance system, and then review current birth defect surveillance methods from a global, international and national perspective. Second, various studies relating to birth defects and population surveillance will be outlined, with particular attention to aspects of effective surveillance systems and how this impacts upon the completeness and quality of information that is collected. This information will then be compared with the structure of the Western Australian Birth Defects Registry. Finally, the chapter will address the issue of surveillance system data completeness focusing on the development, principles and application of capture-recapture methodology.

3.1 Definition and Classification of a Birth Defect

Definition

The Western Australian Birth Defects Registry defines a birth defect as an abnormality, probably of prenatal origin, that is present at birth and may include structural, biochemical, genetic and chromosomal defects (Bower et al., 2000). Most countries including Australia use the International Classification of Diseases ninth edition (ICD-9) index to classify and code data on identified birth defects. This enables uniform classification, categorisation and analysis of birth defect information from multiple sources. Most, but not all defects are included in the range of 740.0 to 759.9 of the ICD-9 codes. Conditions within this category include the following heterogeneous group of outcomes with differing morphogenesis (Lynberg & Edmonds, 1992b):

1. Malformations, involving poor tissue formation, e.g. cleft lip/palate;

2. Deformations, involving unusual forces on normal tissue, e.g. strictures caused by oligohydramnios.

3. Disruptions, involving the breakdown of normal tissue, e.g. trisomy 21, Hirschprung's disease.

Recently, ICD-9 codes were reviewed and updated and released for use by classification systems in the form of ICD-10 codes. ICD-10 codes have recently been introduced in Western Australia and are currently in use in Western Australian hospitals. As this study is retrospective, the ICD-9 system was still in use and appeared in the case notes reviewed by this study.

Classification

The majority of birth defects occur in isolation. However, in approximately 20 to 30 percent of newborns with a congenital abnormality, multiple defects are

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involved. It has been possible to classify the incidence of some multiple defects into biologically meaningful categories that are useful in identifying their etiologic and pathogenic mechanisms. Examples of such categories include syndromes and sequences. A syndrome is defined as a series of signs and symptoms resulting from a common cause, or appearing in combination, to present a clinical picture of disease or abnormality (Anderson, 1990). Some syndromes are caused by chromosomal abnormalities and known teratogens. Trisomy 21 and Turners syndrome are examples of syndromes (Lynberg & Edmonds, 1992b).

Alternatively, sequences are defined as conditions/defects that are the result of a disease (Anderson, 1990). An example of a sequence is talipes which are caused by pregnancy complications such as oligohydramnios (Lynberg & Edmonds, 1992b).

The majority of multiple defects have no known underlying pathogenic or etiologic mechanism. Subsequently, these are grouped according to the occurrence of associated defects. For example, VATER is a syndrome with the following associated defects: vertebral anomalies, anal atresia, tracho-esophageal fistula and a renal anomaly (Lynberg & Edmonds, 1992b).

Birth defects are also classified as either major or minor. Major defects include those that affect survival and result in marked physiological and psychological impairment that requires intensive medical care. Examples of major birth defects include coarctation of the aorta, exomphalos, and diaphragmatic hernia (Bower et al., 2000).

Minor malformations include those thought to cause little or no disability. The Birth Defects Registry of Western Australia does not collect data on most minor malformations unless they require treatment or are disfiguring (Bower et al., 2000). Examples of these are clicky hips, inguinal hernia, Perthe's disease, epilepsy and cerebral palsy. Appendix 1 contains a complete list of birth defects not requiring notification to the Western Australian Birth Defect Registry (Bower et al., 2000).

3.2 An Ideal Birth Defects Surveillance System

The main objective of a birth defect surveillance system is to characterise as accurately as possible, the incidence of birth defects within a population. Surveillance systems can be used to develop baseline population data, provide timely incidence rates and identify geographic areas of concern, prompting cluster and ecological investigation (Lynberg & Edmonds, 1992b). In addition, birth defect surveillance data provides the basis for further etiologic investigation, evaluation of economic impact and long term follow-up studies. This information can then be used to evaluate support services and programs for individuals affected by birth defects, and promote public health initiatives such as prevention and intervention strategies (Lynberg & Edmonds, 1992b).

Lynberg and Edmonds (1992b) outlines the important attributes of an ideal birth defects surveillance system, these include a system that is population based, timely and has a comprehensive case ascertainment system. A surveillance system that is population based, has the ability to obtain information concerning all birth defects cases diagnosed within the population of interest. This ensures the information collected provides the most complete and unbiased reflection of birth defects within that population (Holtzman & Khoury, 1986; Kirby, 2000; Lynberg & Edmonds, 1992b).

An ideal birth defects surveillance system should report population based information in a timely manner. The reporting of all affected births within a given population will provide the most rapid indication of a change in the rate of occurrence (Holtzman & Khoury, 1986), thereby, providing timely recognition of a birth defect epidemic such as that caused by the introduction of a new teratogen (Lynberg & Edmonds, 1992b). Examples of a new teratogen may include maternal exposure to environmental pollutants or drugs. However, timeliness is a attribute that

is frequently difficult to combine with completeness and accuracy, because while trying to ensure completeness of cases ascertained, timeliness is often compromised (Sekhobo & Druschel, 2001).

3.3 Components of a Birth Defect Surveillance System

Several components are required to facilitate an ideal birth defect surveillance and data generation. These include uniform case definitions, adequate case sources and methods of case ascertainment, as well as sufficient data collection, analysis, follow-up and dissemination (Lynberg & Edmonds, 1992b).

3.3.1 Case Definition

Uniform definitions and standardised methods of coding birth defect cases within a population are crucial if accurate data and rate generation are to be achieved. This requires the formation of inclusion criteria that specify birth defect diagnosis, type of birth/delivery, age and weight of a fetus or infant, and delineates between major and minor defects. Use of a standardised method of coding such as the ICD – 9 coding system, as previously discussed, is recommended (Lynberg & Edmonds, 1992b).

3.3.2 Case Sources

To enable a birth defect surveillance system to be population based and timely, its methods and source of data collection must focus upon complete case ascertainment. Multiple source case ascertainment provides the best potential for complete case finding and minimising underreporting. Multiple sources of data usually include vital records (birth and death certificates), newborn and/or hospital discharge summaries, hospital records and data from pathology and cytogenetic laboratories (Lynberg & Edmonds, 1992b). In addition, the inclusion of personal

identifiers on notifications from health personnel enables researchers to link infant, maternal and paternal information. According to Lynberg and Edmonds (1992b) the strengths of multiple source case ascertainment include improvement of the following:

- the timeliness or speed of a surveillance system;

- the relative completeness of recording;

- the accurateness of diagnosis;

- the relative ease of case follow-up studies by researchers; and

- the availability of maternal and infant information.

Conversely, the weaknesses of multiple source case ascertainment includes the inherent proportionately high cost. Lynberg and Edmonds (1993) further recommend that a birth defect surveillance system should also include the following characteristics:

- accurate and precise diagnostic criteria;

- etiologically and pathogenically meaningful classification schemes;

- a large database, permitting rate comparison and analysis of trends in the birth prevalence of a relatively rare birth defect;

- the capability to analyse the occurrence of multiple malformations;

- the ability to conduct meaningful and timely analysis; and

- a system to disseminate data in a timely manner.

3.3.3 Methods of Case Ascertainment

As previously discussed, an ideal birth defect surveillance system supports data collection that is derived from multiple source case ascertainment, as this provides the best potential for complete case finding and minimizing underreporting. Edmonds (1997) described case finding in terms of two main categories: active case

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identification and passive case identification. Active case identification involves trained surveillance personnel actively seeking cases in hospitals, clinics and other facilities by systematically reviewing records and questioning informed sources. Edmonds (1997) also suggested this form of case ascertainment provides for complete and accurate data collection, allowing for follow-up studies to be conducted where indicated. The most notable limitation of active case ascertainment is the cost of maintaining such a system.

The alternative method of case ascertainment is known as passive case identification. This technique is dependent upon the identification of cases from vital records and databases in addition to reports submitted to the surveillance programs from hospitals, clinics and health personal (Edmonds, 1997). Edmonds (1997) outlined the strengths of such a method as low cost, with the ability to cover large populations. Weaknesses include slow reporting, lack of control over the quality of data and under reporting of cases. Edmonds (1997) further suggested supplemental approaches such as linking data sets and conducting audits at selected hospitals where reporting rates appear to be low. These approaches, however, may blur the distinction between passive and active case ascertainment.

3.3.4 Data Collection

Ideally, a birth defect surveillance system should have well established parameters outlining the type of data that should be collected on each case. This will enhance the completeness of data collected and ensure it can be appropriately applied to assess the status of a birth defect, relevant population needs, research, and case follow-up. Optimally, the data collected should include precise descriptions of all birth defects, including the identification of syndromes, demographic data, pregnancy history and other birth related data, cytogenetic and laboratory data, family history and etiologic information (Lynberg & Edmonds, 1992b).

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3.3.5 Data Analysis, Follow-up and Dissemination

The monitoring and collection of birth defect surveillance data allows for the analysis and identification of birth defect rates within a population. In addition, researchers are able to determine birth defect rates and identify changes from various geographical locations within a population.

Minimum analysis of surveillance data requires that expected birth defect numbers be obtained from baseline incidence data. The data is then analysed by "statistically evaluating the difference between observed and expected numbers of specific defects or defect combinations for a specified time in a specified area" (Lynberg & Edmonds, 1992b, p157). These comparisons may lead to the identification of birth defect clusters, the investigation of which may reveal useful etiological information that would require follow-up (Lynberg & Edmonds, 1992b).

Finally, the importance of routine data dissemination to health professionals, hospital, state and local officials is important and should be highlighted. Information regarding birth defect rates and reports of changing trends provides the basis for birth defect related practice, prevention strategies and support services to be evaluated and improved. In addition, review of this information encourages continued case notification by health professionals and stimulates follow-up studies (Lynberg & Edmonds, 1992b).

3.4 A Global Perspective – Surveillance Systems

The monitoring of birth defects first became a worldwide issue in the 1960s when the prenatal use of thalidomide by women was associated with the birth of newborns with severe limb deformities. This tragedy led researchers to understand that unidentified teratogens could cause fetal malformations and suggested the

importance of examining other possible environmental risks and causes of birth defects (Bromberg Bar-Yam, 1999). However, it wasn't until the mid and early 1980s that the establishment of organisations committed to the formalised monitoring of birth defects occurred. Since then, interest in birth defect surveillance has continued to grow, with programs currently monitoring outcomes at both national and international levels (Lynberg & Edmonds, 1992b).

The International Clearinghouse for Birth Defect Monitoring Systems (ICBDMS) was founded in 1974 with the aim of communicating birth defect data from surveillance systems all over the world. The ICBDMS is an independent, non-profit organisation affiliated with the World Health Organisation (WHO) and represents more than thirty malformation monitoring programs worldwide (Bromberg Bar-Yam, 1999).

The mission of the ICBDMS is to help registries of congenital malformations identify and prevent birth defects, and to serve as an early warning system to reduce the incidence of congenital malformations (International Clearinghouse for Birth Defect Monitoring Systems, 2001). ICBDMS membership is voluntary and members include countries such as Australia, Canada, China, France, Italy, New Zealand and America. Member countries are required to ensure that birth defect monitoring systems function at a specified level of competency. This requires that countries contribute sufficient funds to ensure development of a high quality monitoring system (International Clearinghouse for Birth Defect Monitoring Systems, 2001). This process has contributed to the elimination of third world or developing countries from the ICBDMS member list, therefore, it is important to note that ICBDMS data represents only a fraction of the world's births each year (Lynberg & Edmonds, 1992a). Importantly, ICBDMS membership does not presume homogeneity of monitoring systems between members. There are significant differences in terms of

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population, number of surveillance systems, legislation, funding and sources of data ascertainment. Differences in the criteria and protocols of the various monitoring systems make collective interpretation of ICBDMS data difficult. Hence, the primary purpose of the ICBDMS is to function as an early warning system, that can alert epidemiologists to changes in birth defect rates. For this purpose, the completeness of monitoring system ascertainment procedures is of secondary importance, provided ascertainment procedures remain stable over time (Lynberg & Edmonds, 1992a).

Birth Defect surveillance systems in the United Kingdom, Europe, USA, developing countries, Australia and Western Australia will now be described.

3.5 United Kingdom Perspective – Surveillance Systems

The Congenital Malformation Monitoring Programme of England and Wales was established in 1964. The surveillance program covers all births in England and Wales, and includes stillbirths and births of 24 weeks gestation or more. The programme uses voluntary case reports prepared by birth attendants (either doctors or midwives), in addition to supplementary reports from neonatal intensive care units. Between 1998 and 2000 reporting was commenced in four additional regions within England and Wales (International Clearinghouse for Birth Defect Monitoring Systems, 2001). These registers cover 26% of all births and use several sources of ascertainment. Information is available from birth certificates on all births (International Clearinghouse for Birth Defect Monitoring Systems, 2001).

3.6 A European Perspective – Surveillance Systems

The European Register of Congenital Abnormalities and Twins (EUROCAT) was officially established in 1979 by the European Economic Community. The aim of EUROCAT was to collect data on all cases of birth defects, chromosomal

abnormalities and metabolic disorders among live births, stillbirths and induced abortions from within its regions (Bromberg Bar-Yam, 1999). EUROCAT designed a prototype for European surveillance, aimed at assessing the feasibility of pooling data across national boundaries in terms of standardised definitions, diagnosis, terminology and confidentiality (Bromberg Bar-Yam, 1999; European Register of Congenital Abnormalities and Twins, 2001). This design recognises that congenital abnormalities are relatively rare and that the exhaustive collection of good quality data is expensive and difficult. A standardised European registry system would allow countries to pool their data for studies and exploit differences when comparing data (Lechat & Dolk, 1993).

By 1991 EUROCAT had achieved its aim by creating a network of twenty coordinated regional registries from differing countries and regions of Europe that could be adjusted and validly pooled for analysis and surveillance (European Register of Congenital Abnormalities and Twins, 2001). EUROCAT maintains homogeneity by ensuring members adhere to specific criteria. The EUROCAT registries were established according to a number of general principals, these were described by Lechat and Dolk (1993) as follows:

1.   Information collected was to be population based, using the mother's residence in order to avoid bias due to hospital selection. The aim was to produce data that was inclusive of congenital malformations in live births, stillbirths and induced abortions following prenatal diagnosis. For the purpose of population based information the WHO definition was adopted which includes fetuses greater than 500gm or 22 weeks gestation, live or still born.

2.   Multiple sources of information and active case finding methods were to be used in order to achieve more complete case ascertainment. Active case

finding methods provide a more accurate case description than a system dependent entirely upon voluntary notification of cases.

3. Registration was to include all cases diagnosed after the neonatal period, in order to allow for information to be collected on late manifesting abnormalities, most notably, cardiac anomalies.

4. Core information was to be reported using the same coding system with participating registries using the British Paediatric Association Classification of Diseases (BPAD).

5. Confidentially was paramount and strict safeguards were implemented to protect an individual's privacy and unauthorised access to registry records.

3.7 The USA Perspective – Surveillance Systems

Birth defects are the leading cause of infant mortality in the United States of America (USA), where 1 in 33 babies are born birth defects (Birth Defects Monitoring Program, 2002; National Birth Defects Prevention Network, 2002). Despite these figures only thirty five states have or are implementing birth defect surveillance programs. Twenty seven states have passed legislation mandating, protecting and/or funding such programs. This is vital if a birth defect surveillance program is to be maintained (Bromberg Bar-Yam, 1999).

The Californian Birth Defects Monitoring Program (CBDMP) is the largest source of newborn data in the USA. Existing newborn hospital discharge databases are accessed to collect data. Although the program includes data collected from many hospitals in California, not all hospitals are included, hence, the program is unable to provide a representative sample of all births in the USA (Bromberg Bar-Yam, 1999).

An alternative surveillance system is the Metropolitan Atlanta Congenital Defects Program (MACDP). Established in 1967, the MACDP was the nation's first population based active ascertainment birth defects surveillance program. This involves actively reviewing hospital and clinic records on all births, as opposed to waiting for a voluntary notification of a defect by a hospital or clinic. Information is collected on all liveborn and stillborn births (of 20 or more weeks gestation) diagnosed with at least one major birth defect within the first year of life, or ascertained in the first five years. MACDP monitors five counties in the metropolitan Atlanta area that have an estimated population of 2.9 million and 50000 births annually. In addition, MACDP has been adopted as the "gold standard", and acts as a model for state based programs, also as a resource for the development of uniform birth defects surveillance methods in the USA (Metropolitan Atlanta Congenital Defects Program, 2002).

On a different scale is the National Birth Defects Prevention Network (NBDPN). The NBDPN was founded in 1997 during an informal meeting of health professionals who were concerned with the importance of birth defect surveillance. The NBDPN operates at national, state and local levels of birth defect surveillance and acts as a forum for the exchange of ideas, development of uniform methods of surveillance and research, and to provide technical support to state and local programs (Bromberg Bar-Yam, 1999; National Birth Defects Prevention Network, 2002).

Birth defect surveillance is a critical component in the effort to reduce the impact of birth defects on public health. The most frequently published information regarding birth defect monitoring systems originates from the USA. Literature from the USA supports the development of accurate birth defect databases as an initial step in reducing the effects of birth defects (Bromberg Bar-Yam, 1999).

Sekhobo and Drushel (2001) evaluated the surveillance of congenital malformations in New York State using the Centres for Disease Control and Prevention (CDC) guidelines for evaluating public health surveillance systems. The New York State Malformations Registry (NYSMR) uses passive case ascertainment, relying on reports from hospitals and physicians. State regulations require all physicians and other hospital staff to report major birth defects diagnosed at birth through to 24 months of age. To assist in this process the NYSMR provides written guidelines to physician and hospital case reporting sources describing what and how to report. Case reporters are asked to provide a narrative description of the birth defect. These are then reviewed by NYSMR staff, with non-specific and incomplete reports actively followed-up with the reporting source (Sekhobo & Druschel, 2001). Few reports are received from individual physicians, the majority of reports are obtained from hospitals that provide obstetric and paediatric services. Notification of birth defect cases is a legislated requirement in New York State. Therefore, physicians are required by law to report cases of birth defects to the NYSMR. This mandatory reporting law also assists the NYSMR by providing some leverage when attempting to clarify diagnosis from the reporting physician (Sekhobo & Druschel, 2001).

Almost 100 percent of case reports are ascertained from inpatient hospital records, as malformations diagnosed on an outpatient basis are generally not well reported (Sekhobo & Druschel, 2001). Sekhobo and Druschel (2001) found that accurate clinical recognition of birth defects depended upon the clinical acumen and interest of hospital staff, consequently the identification may vary by area and time. Hospitals in the New York State area providing higher levels of care and therefore, more accurate diagnosis, report cases more completely, and maybe associated with higher birth defect rates. This can contribute to increased rates of birth defects. Conversely, areas with lower numbers of births, and minor variations in the number

of birth defect cases may contribute to large variations in the incidence or prevalence rate (Sekhobo & Druschel, 2001). This is associated with the lower rates of birth defect reporting.

Cases of multiple congenital malformation present a particular challenge to NYSMR staff, who are responsible for isolating sufficient diagnostic information to allow for accurate classification. Complex cases are reviewed by the medical director and a geneticist/dysmorphologist. As a result, many syndromes cannot be diagnosed in the first two years of life and may not be included in NYSMR reports (Sekhobo & Druschel, 2001).

To improve the efficiency and timeliness of reporting, the NYSMR recommended that reporting cards be completed by hospital medical records personnel at the time the client's record is reviewed and coded for billing. However, New York hospitals complained that completing a congenital malformation reporting card was time and resource intensive. In 1997 the New York Commissioner for Health reviewed this requirement and in response, two new reporting systems were developed. The first system extracts birth defect information from hospital discharge data reported to the department of health. The second system is a secure web based reporting system which hospital staff may access directly and forward to the NYCMR website (Sekhobo & Druschel, 2001).

Following classification, a birth defect is coded using ICD – 9 codes and a modified version of the BPAD. The BPAD coding scheme is used by a number of congenital malformation registries and allows for greater specificity than using ICD – 9 codes in isolation (Sekhobo & Druschel, 2001).

To validate the completeness and accuracy of hospital reports the NYSMR developed a system for monitoring reports. New York's Department of Health Statewide Planning and Research Cooperative System (SPARAC), used the hospital discharge database to match NYCMR data. This method improved the regularity of

hospital audits and increased the number of hospital reports by 30% (Sekhobo & Druschel, 2001). However, the NYCMR has not yet developed a method for systematically reviewing the accuracy of submitted reports.

A study by Honein and Paulozzi (1999) reported the sensitivity of the NYCMR as 86.4%, according to capture-recapture estimates. This sensitivity estimate was similar to that of the CBDMP and MACDP, which use active case ascertainment methods to identify cases (Sekhobo & Druschel, 2001). The major difference between the three registries was the prevalence of neural tube defects. This results from the NYCMR database included information on live births only. The other two databases included information on both stillborn and terminated pregnancies, thus captures the high proportion of neural tube defected pregnancies that are terminated, resulting in an increased prevalence rate (Sekhobo & Druschel, 2001).

In evaluating of the NYMCR, Sekhobo and Druschel (2001) concluded that in general, registry objectives of collecting, organising and distributing information were being meet. The strengths of the NYMCR was the mandatory reporting law, cost effectiveness, collection of narrative diagnosis for specificity and ongoing efforts to track and improve ascertainment completeness. Weaknesses of the NYMCR included the lack of an ongoing system for evaluating the accuracy of reported diagnosis, timeliness and the non-inclusion of terminated cases (Sekhobo & Druschel, 2001).

Olsen, Polan and Cross (1996) used the NYMCR to determine whether expanding case finding to include routine matching of birth certificates had a significant positive effect in identifying unreported birth defect cases. Children born in 1983-1986 who had a congenital abnormality recorded on their birth certificates were matched with registry records. The matched records yielded 1366 new cases of congenital malformation not previously recorded by the NYMCR. When compared

with existing registry cases, the birth certificate cases were more likely to list only one malformation and of these the majority were minor malformations (Olsen et al., 1996). The addition of these cases increased the overall prevalence of major malformations by 1.7% from 416.5 to 423.4 per 10000 live births, however, the prevalence of specific malformations was not measurably altered by the addition of these cases (CORN). The authors concluded that lengthy and continuous follow-up by staff was required to obtain the registry reports on birth certificate information from the hospitals. In light of the small number of additional cases found, the authors believed the amount of resources required to use birth certificate information routinely to augment NYMCR case finding was not justified (Olsen et al., 1996).

Similarly, in 1997 the Council of Regional Networks (CORN) for Genetic Services in Wisconsin conducted a nationwide survey to assess the current status of population based databases concerning genetic services and health outcomes. The study focused on the structure, methodology, data contents and use of birth defect registries. In comparing the case ascertainment methodology of birth defect surveillance systems, the survey used the following standards and definitions (Kirby, 2000):

1.   Active: trained staff visit health care facilities and abstract records on-site.

2.   Passive: reporting sources or case notifiers submit reports on specified surveillance system notification forms. These forms determined if further action was taken on the data, they included:

- Aggressive cases: those where routine follow-up is conducted, with mandatory reporting and penalties for non-compliance.

- Compliant cases: forms are accepted basically as filed, with minimal or no follow-up to ensure completeness or accuracy of the data reported.

3.   Impassive: information is received via automated record linkage from data sources designed for purposes completely unrelated to the surveillance of the disease or health condition.

Kirby (2000) described these definitions as differing from the traditional approach to classifying case finding strategies as outlined by Lynberg and Edmonds (1992b). Kirby (2000) determined these definitions allowed for a more clear delineation of the surveillance program's role in collecting and assessing data. Classification of traditional case finding strategies as 'active' or 'passive' does not reflect whether data was collected for administrative or clinical purpose, and does not reflect the fundamental relationship between a surveillance system and its data collection strategies (Iezzoni, 1997). Particularly important is the difference between surveillance programs that use passive and impassive case finding methods. Those using impassive methods must accept the data as reported, since there is no opportunity for assessing the diagnostic specify of the defect, or if it was appropriately coded. A passive case finding strategy that requires reporting sources to provide text descriptions of the birth defect facilitates greater degrees of data accuracy and case follow-up (Kirby, 2000).

Results from the CORN study showed that in 1997, 30 birth defects surveillance programs existed across the USA. Of these, 10 programs used active methods, 13 programs used passive methods and seven programs used impassive methods of case finding (Kirby, 2000). A variety of definitions, sources, coding methods and age limits were used in each program. Although the standards recommended by the MACDP were more commonly used, there was no evidence of uniformity (Kirby, 2000). Birth defect surveillance data was reported as being used for the planning of state public health genetics programs in only 15 of the 30 states, with specific use of surveillance data involving descriptive statistics and low-level applications. Kirby (2000) recommended that if maternal and child health programs

are to define their populations and provide a range of focused health services, more attention must be paid to developing, maintaining and integrating population based data systems, such as the health aspects of genetic services.

In support of Kirby's (2000) recommendations, Miller (2000) also cited the increasing trend of establishing population based birth defect surveillance in the USA. Similarly, it was shown that only 31 states have such databases. Moreover, only a small number of the 31 states had a comprehensive ongoing system of data collection, with several states using a limited number of data sources (Miller & Kirby, 2000). These were listed as prenatal diagnosis, birth certificates, death certificates, hospital discharge data, obstetric logs and speciality clinics.

Miller's (2000) overview of existing USA birth defect surveillance systems suggests that limiting data sources and the mode by which the information is collected from such sources serves to damage the completeness and therefore the accuracy of the data collected. Another survey conducted by Miller and Kirby (2000) in the USA determined the efficacy of Neural Tube Defect (NTD) surveillance data, and highlighted the potential for incomplete and inaccurate data. It was concluded that as a result of limiting data collection sources and surveillance comprehensiveness, complete and accurate data did not exist to evaluate NTD programs on a national level.

In contrast, Calle and Khoury (1991) evaluated the completeness of using discharge diagnosis as the principal method of case ascertainment. Discharge diagnosis provides an inexpensive tool for the surveillance of birth defects diagnosed within the first few days after birth and is used as a standard form of data collection in many US states. To evaluate the completeness of a discharge diagnosis, Calle and Khoury (1991) compared birth defects recorded in the discharge diagnosis with those recorded anywhere in the hospital birth record. The cohort included 3421 newborns born to US Army veterans between 1966 and 1986. From this cohort 237 birth

defects were documented in hospital birth records. Of the 237 birth defects, 49% of cases had been missed in the discharge diagnosis. Significant predictors of a missed defect were the presence of a multiple defect, female gender and western regions of birth. Calle and Khoury (1991) concluded by suggesting the underascertainment of defects in discharge diagnosis should be considered in the development and operation of surveillance systems using this source of data (Calle & Khoury, 1991).

Calle and Khoury (1991) and Hexter and Harris (1991) also cautioned that congenital malformation data recorded on the birth certificates was both incomplete and biased. Similar to the discharge diagnosis, birth certificate information is viewed as form of passive surveillance, since it relies on physicians and/or other hospital staff to record and report birth defect data. Hexter & Harris (1991) compared the CBDMP case registry (based on active hospital surveillance) with birth certificates and hospital discharge index information (Hexter & Harris, 1991). Findings revealed that discharge information and birth certificates were poor sources of data on congenital malformations. Discharge information was shown to have many false "positives" and "negatives" in almost every category of congenital malformation. As well, birth certificates reported fewer than 25% of the malformations in the CBDMP case registry (Hexter & Harris, 1991)

Hexter and Harris (1991) reported that birth certificates from smaller hospitals with fewer births showed higher rates of congenital malformation than did birth certificates from larger hospitals. Since birth certificates had shown to be such poor sources of malformation surveillance, medical record supervisors at the 42 hospitals in the study were asked how they collected congenital malformation information for the birth certificates. Results showed that hospitals using obstetrician records as the source of information on congenital malformations were less likely to report the presence of a birth defect on the birth certificate, compared with hospitals that used paediatrician or labour and delivery log information. This highlighted the

presence of bias according to information source (Hexter & Harris, 1991). Hexter & Harris (1991) concluded that data collected from smaller hospitals using birth certificates to report congenital malformations was associated with bias, especially where hospitals used obstetrician's medical records as the source of information.

Lynberg and Edmonds (1992a) confirmed the importance of using multiple sources and methods of ascertaining birth defect information. The authors illustrated the effect that using various sources and methods of case ascertainment had on rates of birth defects. In doing so, several of the previously discussed surveillance systems were reviewed, this information is displayed in Table 8 (Lynberg & Edmonds, 1992a). Lynberg and Edmonds (1992a) showed that using exclusive sources of case ascertainment produced varying rates of birth defects within the USA.

Many researchers evaluating birth defect surveillance in the USA have reached conclusions and recommendations for monitoring congenital malformations. These recommendations are in keeping with Lynberg and Edmonds (1992a) outline of the ideal birth defects surveillance system. These include the use of population-based information obtained from a variety of sources, in conjunction with case ascertainment approaches that promote timeliness, comprehension and completeness.

Table 8.

Rates of Major Birth Defects in the USA Using Various Surveillance Methods per 10000 live births.

Method and source	Rate
Birth Certificate (NCHS[1] 1982-83)	88.9
Newborn Hospital Discharge Data (BDMP[2] 1982-85)	282.5
Mandatory Hospital Reporting Data (Nebraska 1982-85)	248.0
Linked Data Sources (Missouri 1980-84)	336.0
Active Hospital Surveillance (MACDP[3] 1982-87)	415.0
(Iowa 1983-86)	549.0
Physical Exam of Infant (CCP[4])	830.0

[1] National Centre for Health Statistics (USA)

[2] Birth Defects Monitoring Program (USA)

[3] Metropolitan Atlanta Congenital Defects Program (Atlanta)

[4] Collaborative Perinatal Project (USA)

A realistic contrast to this purported common belief, however, was proposed by Hanson (1995) who questioned the importance that public agencies and policy makers place on building effective birth defect surveillance programs. Birth defects are an important public health problem. Some birth defects such as fetal alcohol syndrome are preventable, yet adequate research and effective prevention and intervention strategies remain largely unrealised for such defects and many other birth defect categories (Hanson, 1995). Hanson (1995) stated that while complex, the core of the problem is the failure of clinical, research and public health communities to present compelling arguments for the larger investment of resources. Hanson (1995) further described this issue as bound by a circular system of defeat, for without adequate surveillance systems the impact of birth defects and the generation of meaningful estimates of the associated health, educational and human needs cannot occur. Moreover, without compelling arguments, surveillance systems will not be funded and the effectiveness of current intervention will not happen.

3.8 Developing Countries – Surveillance Systems

Lynberg and Edmonds (1992b) suggested that in many developing countries birth defects are not reported as the leading cause of infant morbidity and mortality. However, rates of birth defects are usually higher than those of developed countries. Of further concern, populations in developing countries are likely to be more immunologically vulnerable, malnourished, have older maternal age ranges and suffer from increased exposure to environmental, agricultural and industrial pollutants. These issues are further compounded by the lack of, or failure in health monitoring systems, to highlight and regulate teratogenic exposures (Lynberg & Edmonds, 1992a) such as the introduction of potential environmental pollutants. Lynberg and Edmonds (1992b) suggest developing countries implement surveillance

systems using existing organisations, such as hospitals, that should be expanded as resources permit.

3.9 The Australian Perspective – Surveillance System

The Australian Congenital Malformation Monitoring System (ACMMS) was commenced in 1981. Initially, not all states and territories collected data, however, perinatal data systems have since been introduced throughout Australia (International Clearinghouse for Birth Defect Monitoring Systems, 2001). Data related to birth defects is now collected by birth defect registers and hospitals in each state and territory. Sources of data ascertainment include birth notification, hospital mortality and morbidity data, referral hospitals, pathology departments and cytogenetic laboratories (International Clearinghouse for Birth Defect Monitoring Systems, 2001). Data is then reported to the National Perinatal Statistics Unit (NPSU) at the Australian Institute of Health and Welfare (National Perinatal Statistics Unit, 2002b).

Information reported to the NPSU includes diagnosed cases of congenital malformation of live and stillborn newborns, that are greater than 20 weeks gestation or 400gm in weight. This includes malformations diagnosed prenatally, within the first 28 days of life or terminations of pregnancies in which a congenital malformation was present. The type of data collected includes maternal demographic information (age, parity, country of birth and local government area) and newborn demographic information (gestational age, sex, birth weight, outcome, method of diagnosis), as well as the British Paediatric Association codes for each abnormality and the date of death (National Perinatal Statistics Unit, 2002b).

The major purpose of the perinatal data collection as outlined by the Australian Institute of Health and Welfare (AIHW) is as follows (National Perinatal Statistics Unit, 2002b):

- to describe for all births the demographic and pregnancy characteristics and outcomes of mothers and babies;

- to identify risk factors that contribute to adverse outcomes of pregnancies, mothers and babies;

- to assist in the planning, implementation and evaluation of health services for pregnant women and their babies;

- to facilitate the analysis and comparison of national data;

- to analyse perinatal and infant morbidity and mortality;

- to monitor specific outcomes such as congenital malformations; and

- to conduct epidemiological studies of health concerns among pregnant women and babies.

3.10 The Western Australian Perspective – Surveillance System

The Western Australian Maternal and Child Health Research Database compiles general perinatal statistics from notifications of birth made to the Department of Health (DOH), as well as from birth and perinatal death registries compiled by the Australian Bureau of Statistics (ABS) through record linkage (Stanley et al., 1994). The Western Australian Birth Defects Registry uses information from this database, in addition to case reports from notification sources to compile its data. Data from the following sources are used for record linkage by the Registry:

1. Midwives Notification of Case Attended Form: these forms are completed for every birth in Western Australia (WA) from 20 weeks gestation or 400gms, by the attending midwife (99.9% of all WA births have a midwives form);

2. Birth Registration Form: completed for all births from 20 weeks gestation. Providing parental information such as age, occupation, date of marriage, country of birth and number of previous children;

3. Death Certificates: completed for every death in WA using a multiple cause coding system. The database is currently being expanded to include children up to 15 years; and

4. Hospital Inpatient Morbidity Data: collected from the WA DOH Inpatient Summary Form. Details basic demographic data, length of stay, principal condition treated and other present conditions, operations and procedures preformed and place to which patient was discharged.

These items are all statutory collections that must be completed by law. In addition, data on cerebral palsy and birth defects are also obtained via voluntary notification from referral hospitals, general practitioners, specialist clinics and pathology departments. Voluntary notifications are based upon the good will of practitioners with no statutory regulations requiring practitioners to report birth defects (Stanley et al., 1994). Data relating to cerebral palsy is collected by the WA Cerebral Palsy Register (WACPR). The WACPR records all children with cerebral palsy born and/or living in WA since 1956, data is then updated on these children until five years of age. Data relating to birth defects is collected by the WA Birth Defects Register, which has been previously described in detail (see page 3).

A WA study conducted by Bower, Ryan and Rudy (2001) from the WA Birth Defects Registry, highlighted the importance of data completeness in the

ascertainment of birth defects cases. This study reviewed the effect of adding new data from a source not previously available to the Registry, on prevalence and data completeness. The source was pregnancy termination data for fetal abnormalities less that 20 weeks gestation. Results showed a small but significant increase in the overall incidence of birth defects. The addition of this new data source was found to improve the ascertainment of birth defect cases. Furthermore, it supported the use of Registry data for reliably monitoring the effects of programs aimed at prevention of birth defects in WA (Bower et al., 2001).

Contrary to the recommendations of Lynberg and Edmonds (1992a) outlining the requirements of a comprehensive birth defect notification system, the WA Birth Defects Registry uses extensive passive case identification methods, as well as some active methods. As previously discussed, significant use of passive case ascertainment methods creates potential error in achieving data completeness. This scenario may have significantly affected the accuracy of current incidence rates of birth defects in WA (Bower et al., 2001), particularly in rural areas. This issue is discussed in Chapter 7.

The 2000 Report of the Birth Defects Registry of WA (Bower et al., 2000), highlighted evidence suggesting the possibility of data incompleteness in rural birth defect rates, for which most data sources are passive. The report highlighted a statewide discrepancy in birth defect incidence rates, with lower incidences of birth defects reported in rural compared with the metropolitan area. Bower (2001) suggested a possible explanation for this discrepancy was under ascertainment of case numbers rather than a real difference. In light of the reviewed literature, potential causes for such a discrepancy seem obvious. Examples of such causes include the failure to use active and passive case ascertainment strategies, and the lack of statutory notification requirements. However, before this issue can be objectively explored, the completeness of the WA Birth Defects Registry database

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information must be confirmed. Therefore, the purpose of this study was to determine if the lower incidence rates of birth defects in the rural Midwest region of WA is in fact due to the under reporting of cases.

The following section of the literature review will discuss capture-recapture methodology as an approach to determine data completeness and estimate population size.

3.11 Capture-Recapture Methodology

Routine disease monitoring systems such as congenital malformation registries are continually challenged as efforts to achieve complete case identification increase (Silcocks, Thornton-Jones, & Skeet, 1989). Undercounting is inherent in any monitoring system and contributes to the inability of most monitoring systems to achieve complete case identification (Silcocks et al., 1989). This can be overcome by implementing formal standardised methods to adjust for the degree of underascertainment (International Working Group for Disease Monitoring and Forecasting, 1995a). In recent years there has been a growing trend in epidemiology to correct for underascertainment using capture-recapture methods. This study, therefore, used a capture-recapture methodology.

Hook and Regal (1995) outlined several circumstances in which capture-recapture methodologies can potentially be used to derive population estimates from data. These include:

1.   Estimation when incomplete data are available from two or more sources;

2.   Refinement of prevalence and incidence estimates derived from exhaustive population surveys;

3. Attempted evaluation of source completeness or a registry that receives reports from various sources; and

4. Attempts to derive upper and lower estimates of the total population affected.

An additional benefit to researchers is the interchangeability of circumstances discussed in points one and three. Studies that use a capture-recapture methodology to evaluate the completeness of source ascertainment, in essence, generate data that can also be used to estimate total population size (Hook & Regal, 1995).

Capture-recapture methodology originated in wildlife studies and is generally associated with the estimation of animal populations (Robles, Marrett, Clarke, & Risch, 1988). This method requires an initial capture of a sample of animals from a population of unknown size. The animals are then 'tagged' and released. The same animal population is then re-sampled with the second sample consisting of both 'tagged' and 'untagged' animals. The two samples can then be compared and statistical analysis used to estimate the number of animals in an entire population (Robles et al., 1988).

The most simple capture-recapture model is called the two-sample or two-source model, and is used exclusively to estimate the unknown size of a population. To ensure the validity of this model, several assumptions must be made. These assumptions are as follows (International Working Group for Disease Monitoring and Forecasting, 1995b):

1. There is no change in the population during the investigation period (i.e. the population is closed);

2. These is no loss of tags (i.e. individuals can be matched from capture to recapture);

3. For each sample, each individual has the same chance of being included in the sample; and

4. The two samples are independent.

Application of the first assumption to epidemiology requires the researcher to ensure the study population remains constant, with no additions to the first or second samples during the study period (International Working Group for Disease Monitoring and Forecasting, 1995b).

The second assumption requires matching individuals in the first and second samples. The success of this assumption relies upon the quality of patient records, and the uniqueness of patient code names (International Working Group for Disease Monitoring and Forecasting, 1995b).

The third assumption requires that each individual has the same probability of being captured in the first sample as in the second sample. Unfortunately, this assumption cannot always be assured as patients tend to be heterogenous with regard to being 'caught' (International Working Group for Disease Monitoring and Forecasting, 1995b). For example, newborns with severe defects requiring treatment in tertiary centres may have an increased likelihood of being recorded and are, therefore, easily captured. In contrast, newborns affected by birth defects not requiring referral or extensive treatment may have a lesser chance of being recorded and, therefore, captured. In such cases newborns would not have the same probability of being included in a sample.

The fourth assumption is also problematic since it relies on the two samples being independent. For example, doctors records and hospital admission records could be used individual study samples. If, however, in the study sample doctors referred their patients to the study sample hospitals, sampling of hospital admission records and doctors records would not provide two independent lists. Therefore, the

chance of capturing a patient in the doctors records would not be independent of also capturing that patient in the hospital admission records (International Working Group for Disease Monitoring and Forecasting, 1995b).

Hook and Regal (1995), however, provide reassurance to researchers using capture-recapture methods by recommending that in the event that assumptions are violated, the use of a two-source capture-recapture approach will provide useful estimates with 'considerable practical value' (Hook & Regal, 1995). An extension of the two-sample or two-source capture-recapture methodology is the multiple sample or source approach. This approach was introduced by Schnabel (1938) who recognised that by taking more than two samples from the same population, information could be collected from the multiple recaptures. For each sample, unmarked animals or fish would be given individual marks before being returned to the population, allowing for the capture history of each marked individual to be developed (International Working Group for Disease Monitoring and Forecasting, 1995b).

Schnabel's (1938) theory was further developed by epidemiologists in the 1950s using the same assumptions as the two sample capture-recapture approach. However, it was realised that some of the underlying assumptions of the two-sample approach would not hold true, 'especially the those related to dependence and heterogeneity' (International Working Group for Disease Monitoring and Forecasting, 1995b). Fienberg (1972) developed a solution to this interdependence through the use of log-linear models. However, for the purpose of this study the review of literature will only focus upon the use of two-source capture-recapture approach and it relevant assumptions.

Although, the capture-recapture approach for correcting underascertainment appears straight forward, Brenner (1994) warns this approach has a number of

methodological problems that require careful consideration. Brenner (1994) described the problem of imperfect record linkage as a pertinent problem of population based disease monitoring. This relates to the incorrect linking of notifications on different individuals or the failure to link notifications on the same individual. Problems such as these often arise from a variance in case notification by personal identifiers that may provide incomplete patient information, or due to changes in variables used for record linkage such as last name or address (Brenner, 1994).

Brenner (1994) cited studies that have used capture-recapture methods in estimating the true prevalence of spina bifida, congenital rubella syndrome, cleft lip and plate, mental disability, cancer, drug use, myocardial infarction, Huntington's disease, childhood diabetes and HIV. In addition, the International Working Group for Disease Monitoring and Forecasting (1995a) cited eight birth defect related studies that used capture-recapture techniques to correct for the number of incidence and prevalence cases and reporting system completeness.

A study by LePorte et al. (1995) demonstrated the importance of applying capture-recapture methods to estimate undercount and to evaluate the accuracy of health monitoring systems in general. The study used capture-recapture methods to determine the most efficient and accurate approach to monitoring adolescent injury. The cases of adolescent injury during a three-month school semester in 1991 were ascertained using five sources, including a monthly survey of all physician treated injuries, a review of medical excuses given to the school nurse, a review of daily absentee records, an end of semester student survey to identify all physician treated injuries during the semester, and a teacher survey inquiring whether they were aware of any injuries students in their classes had sustained (LePorte et al., 1995).

A capture-recapture analysis was then undertaken to assess any dependencies between sources, the degree of population under ascertainment, and to evaluate the efficiency of the sources and their combinations. The authors found that by combining sources of injury ascertainment, capture-recapture methods could be used to estimate the number of injuries occurring in the population. In addition, it was noted that the degree of precision with which population estimation was made improved as the level of effort for case finding increased (LePorte et al., 1995). LePorte et al (1995) concluded by recommending the careful and appropriate use of capture-recapture methods as a new approach that could considerably improve disease monitoring.

In contrast, Brenner, Stegmaier and Ziegler (1994) used capture recapture methods to estimate the completeness of population-based cancer registration. Cancer registration is commonly quantified using death certificates or mortality/incidence ratios. The main disadvantage of these measures is their strong dependence on case fatality rates (Brenner et al., 1994). The study reported a high standard of overall completeness of cancer registration, however, this was less satisfactory in some subgroups of patients. Results also emphasised the importance of multiple sources of notification to achieve high levels of completeness (Brenner et al., 1994).

Limitations of using capture-recapture methods to estimate the completeness of cancer registration stemmed from inaccuracies associated with site-specific diagnosis recorded on death certificates. Brenner et al., (1994) attempted to minimise this problem by combining types of cancer into common groups commonly affected by misclassification. In addition, the authors found capture-recapture methods restrictive when used to estimate completeness of a registry that uses death certificates as the basis for notification. This was due to the discrepancy between

patient diagnosis and death, leading to inaccurate and untimely results (Brenner et al., 1994).

Of particular relevance, Brenner et al., (1994) found the use of a three-source capture-recapture method was less sensitive to dependence of ascertainment between sources, compared with a two-source capture-recapture approach. Using a two-source capture-recapture method requires a stronger assumption of independence between the two sources (Brenner et al., 1994). Overall, the study encouraged the frequent and careful use of capture-recapture methods when estimating completeness of cancer registration. In addition, the authors made a strong recommendation regarding the adherence to the underlying assumptions of capture/recapture methods.

Similarly, Robles et al., (1988) used capture-recapture methodology to evaluate the completeness of data reported to an Ontario cancer registry. The registry was responsible for collecting data regarding a number of malignancies from a variety of sources. Of particular importance was the readiness with which capture-recapture methods could be applied to the data. Unlike many other registries, the Ontario cancer registry had distinguished the multiple sources of each case reported and could therefore accurately match the reports to their different sources (Robles et al., 1988). The study reported at least 90% completeness in the information received. As previously stated by Hook and Regal (1995), studies ascertaining source completeness also generate information from which estimates of total population size can derived, although this was not undertaken by Robles et al., (1988).

Roberts and Scragg (1994) used a two source capture-recapture method to assess the completeness of ascertainment between child pedestrian injuries in public hospital discharge statistics, and the active injury surveillance system. The study found that almost half of all non-traffic child pedestrian injuries discharges were misclassified in the public hospital discharge statistics. The authors suggested that

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poor documentation of the circumstances of injury in the clinical case notes was a significant factor in explaining the failure of hospital discharge databases to record pedestrian injury. Similarly, this may also have contributed to the failure of the active surveillance system to record the cases. The result of this positive dependence was an underestimation of the ascertainment adjusted number of pedestrian injury cases (Roberts & Scragg, 1994). Roberts and Scragg (1994) suggested that if three sources of case ascertainment had been available, it would have been possible to use log-linear modelling to assess the extent of source dependency on the estimates obtained. In conclusion, the authors recommended improvement in the recording of injury circumstance in hospital case notes. Additionally, in the presence of multiple over-lapping registries of injury, capture-recapture methods were promoted as a cost effective means for monitoring childhood injuries (Roberts & Scragg, 1994).

A study by Egeland, Perham-Hester and Hook (1995) used capture-recapture methods when estimating the prevalence of fetal alcohol syndrome among Alaska natives born between 1982-1989. Potential cases were identified through Indian Health Service case files, paediatric practice case files and Medicaid claims from private physicians. In applying the assumptions of capture-recapture approach to fetal alcohol syndrome the authors encountered a number of methodological problems. The authors found it difficult to ensure the assumptions of true case matching between the first and second samples, and sample independence when applying the technique to fetal alcohol syndrome (Egeland et al., 1995). Match identification was difficult because many children may have undergone name changes due to adoption or long-term foster care. Egeland et al., (1995) recognised that failure to achieve matching would result in an overestimate of case numbers, however, in the study the authors believed that all matches were identified, as no non-matched cases had the same gender and birth date as another non-matched case.

In addition, the authors had difficultly in assuring the accuracy of case identification or diagnosis by each data source. A fetal alcohol syndrome case definition based upon the identification of five criteria was used to assist diagnosis and eliminate subjectivity. However, since medical charting and documentation tended to underreport the information needed to confirm the case status, syndrome classification was likely to represent a minimum observed prevalence (Egeland et al., 1995). Furthermore, Egeland et al., (1995) discovered that examining the rates of fetal alcohol syndrome across all age groups was not consistent, since newborns, children and adolescence exhibit differing features of the disease as they grow. In an attempt to control for this factor the authors stratified age groups prior to evaluating case ascertainment.

Egeland et al., (1995) discovered the non-adherence to the assumption of random sampling. A previous state-wide screening program had failed to fulfil ongoing active screening and referral, and use of passive reporting via a native health scheme had caused time and location inconsistencies. To overcome this, capture-recapture analysis was performed separately on differing age group stratifications (Egeland et al., 1995).

Finally, Egeland et al., (1995) addressed the assumption of independence between data sources and stated that "negative dependence leads to overestimation of population size and positive dependence leads to underestimation of population size" (Egeland et al., 1995, p337). The authors presumed that data obtained from the Indian Health Service and private physicians would be negatively dependent, as children seeing private physicians would be less likely to be attending the Indian Health Service. This leads to a negative dependence and likely overestimation of prevalence rates (Egeland et al., 1995). Egeland et al., (1995) tackled this problem by suggesting that if all sources of bias are in the "same negative direction, then capture-

recapture methods could be used to provide an upper limit on the expected prevalence rate of fetal alcohol syndrome" (Egeland et al., 1995, p337). The authors recommended the use of capture-recapture methods in fetal alcohol syndrome surveillance, however, they stressed the continued importance of active screening programs coupled with accurate diagnosis in assuring the delivery of essential services to affected children (Egeland et al., 1995).

Bower, Ryan and Rudy (2001) used a capture-recapture methodology to determine the effect on data completeness when adding a new source of data to information already held by the WA Birth Defects Registry. Advances in antenatal screening procedures and an increasing trend to conduct mid-trimester terminations in non-metropolitan hospitals, prompted the authors to question the completeness of notifications regarding terminated pregnancies to the Registry.

The information already held by the Registry was used as the 'tagged' or captured sample of the birth defect population. New data regarding terminated pregnancies was added to the Registry. This constituted the recaptured sample and consisted of 'tagged' and the new 'untagged' cases (Bower et al., 2001). The difference between the two populations were analysed statistically using an ascertainment-adjusted analysis to determine the 'true' birth defect population prevalence rates. The additional new source of information made a small but statistically significant increase in the overall prevalence of birth defect and chromosomal defects (Bower et al., 2001). Bower et al., (2001) stressed the importance of accurate monitoring of birth defect conditions, particularly when evaluating preventive strategies, such as the evaluation of periconceptional folate and rubella vaccination.

Capture-recapture methodology is used to produce quantitative research findings. Qualitative research alone however, does not provide the researcher with insight or explanation regarding empirical evidence. The following section will

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explore the use of focus group interviews as a means of expanding upon the quantitative data collected by in this study.

3.12 Focus Group Interviews

The focus group interview is a qualitative research technique used to determine the feelings and opinions of a small group of participants regarding an identified topic (Dilorio, Hockenberry-Eaton, Maibach, & Rivero, 1994; Nyamathi & Shuler, 1990). Focus group interviews were historically used by advertising and marketing professionals to develop strategies for influencing consumer buying habits, by gaining their opinions regarding products. More recently, this technique has been adopted by social and health researchers to gather group responses about health related issues (Dilorio et al., 1994).

Dilorio et al. (1994) suggested that a focus group interview should ideally follow specific preparation in terms of problem formation, interview guidance, participant selection, interview setting and the use of a moderator and observer. Dilorio et al. (1994) recommends that prior to conducting a focus group interview, individuals involved in the implementation, analysis or interpretation of the interview data should be involved in formulation discussions. These discussions define the purpose of the research, type of information required and the desired outcomes of the interviews (Dilorio et al., 1994). This will enable the researcher to construct an interview guide consisting of an introductory section, six to twelve interview questions and a conclusion (Dilorio et al., 1994).

The first questions of a focus group interview should be related to the topic but be unstructured and aimed at generating discussion and facilitating rapport amongst group members and the researcher (Dilorio et al., 1994; Nyamathi & Shuler,

1990). Furthermore, it is recommended that final interview questions be broad and aimed at summarising ideas (Dilorio et al., 1994).

In designing a focus group interview Dilorio et al. (1994) suggests that the appropriate choice of interviewees will maximise the likelihood of obtaining desired research outcomes. It is further suggested that the group share common experiences and be relatively homogenous in terms of personal characteristics such as age education, social status and income (Dilorio et al., 1994). Homogeneity, however, should be balanced with some diversity, as it is the purpose of a group interview to elicit a range of responses, opinions and ideas (Dilorio et al., 1994).

In addition, Dilorio et al. (1994) refers to the importance of the physical setting in which a focus group interview is conducted. It is suggested that a focus group interview be conducted in a familiar, easily accessible setting, as this will promote group comfort and enhance interaction.

Dilorio et al. (1994), describes the process of focus group interview analysis as dependent upon the reason for conducting the interviews. The goal of analysis, however, is the same and should begin with structuring the interview data for clear communication to others (Dilorio et al., 1994). It is recommended that interview data should first be structured using the research and interview questions for clear communication to others. Second, data should be reduced and information classified into ideas or themes. Themes can be derived from ideas expressed as words, sentences or paragraphs and may refer to specific content or affective details of an interview (Dilorio et al., 1994; Nyamathi & Shuler, 1990). Dolorio et al. (1994) further recommends the researcher identifies major themes and validates this classification with supporting examples and interpretive statements.

3.13 Summary

The review of the literature has highlighted the significance of birth defects and their associated complications, identifying them as a health concern for newborns, children and their families worldwide. Also highlighted, were the differences in surveillance methods used throughout the world. Identified discrepancies in birth defect surveillance methods include the use of multiple data sources and type used, and passive and active case ascertainment methods.

The review of the literature also confirms the need for national and international systems for monitoring birth defects. In addition, case under ascertainment was found to be an issue for all monitoring systems and one that should be regularly evaluated and adjusted for. Capture-recapture methodology appears to provide a logical, realistic and cost effective approach to assessing the completeness of data ascertainment and estimating population size.

In addition, the focus group interview design offers a logical format for eliciting the opinion of a group regarding health related issues in a sensitive manner.

In order to determine the incidence rates of birth defects, accurate and complete data is required. This study has addressed the need to ensure accuracy and completeness Birth Defect Registry data from the Midwest region of WA where there is an identified lower reported incidence of birth defects compared with the metropolitan area. The following chapter will detail the conceptual framework which was used to examine the study aim ideologically and provide directives from which the study method was developed.

CHAPTER 4

CONCEPTUAL FRAMEWORK

The aim of this study was to determine if the observed lower incidence of birth defects in the Midwest of Western Australia is due to case under reporting. In the process of determining the study aim, the researcher examined the method and effectiveness of Birth Defect Registry case ascertainment sources. The following section outlines the relationship of case ascertainment sources used by the Registry. These sources were considered as variables within the conceptual framework of this study.

The conceptual framework guiding this study was developed from the literature review. Research by Lynberg and Edmonds (1992b) suggests the ideal birth defects surveillance system is determined by a comprehensive case ascertainment system that provides timely population based information. Multiple sources of case ascertainment provide the best potential for complete case finding and minimizing under reporting.

The Birth Defects Registry of WA is primarily responsible for the collection, correlation and analysis of birth defects in WA (Bower et al., 2000). As previously mentioned, a number of information sources are used by the Registry to collect data, these include statutory and non-statutory sources. Statutory sources include midwives notification data, hospital morbidity and mortality data. Collection of these forms of data is a statutory requirement, with information collected by hospitals and the Health Department of WA (HDWA). The Birth Defects Registry can access this information directly from HDWA. Alternatively, Registry information is collected

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via voluntary notification of birth defect cases from various sources. As the name implies, notification of birth defects from these sources holds no statutory requirement and occurs purely on a voluntary basis. The Birth Defects Registry lists the following as voluntary case reporting sources: paediatric hospital and paediatric hospital speciality departments, obstetric hospitals and obstetric hospital speciality departments, other hospitals, cytogenetic services, pathology services, genetic services, special services, private practitioners, child and community health nurses and doctors and rural paediatric service. General practitioners, midwives, child health nurses and Perth based tertiary hospitals were considered as variables within the conceptual framework of this study. Relationships between the Birth Defects Registry and the remaining methods of case ascertainment are not mentioned as variables, exploring their association was beyond the scope of this study.

Statutory sources of case ascertainment used by the Registry are derived from vital records such as midwives birth notification forms and hospital morbidity and mortality data (death certificates). Lynberg and Edmonds (1992b) describe the strengths of vital records as complete population coverage, availability of medical and some parental data, low cost and the potential for follow-up. The weaknesses of vital record use include the lack of timeliness in reporting, lack of specificity of most birth defects and difficultly in defining the population base (Lynberg & Edmonds, 1992a).

In addition, Edmonds (1997) describes case ascertainment in terms of two main categories, active case identification and passive case identification. Active case identification involves trained surveillance personnel actively seeking cases in hospitals, clinics and other facilities by systematically reviewing records and questioning informed sources (Edmonds, 1997). Edmonds (1997) suggested this form of case ascertainment provides for more complete and accurate data collection,

allowing for follow-up studies to be conducted where indicated. The most notable limitation of active case ascertainment is the cost of maintaining such a system.

The alternative method is known as passive case identification. This technique depends upon the identification of cases from vital records and databases in addition to reports submitted to the surveillance programs from hospitals, clinics and health personal (Edmonds, 1997). Edmonds (1997) outlines the strengths of such a method as low cost, with the ability to cover large populations. Weaknesses include slow reporting, lack of control over the quality of data and under reporting of cases.

The Birth Defects Registry predominantly uses passive forms of case ascertainment. Active case ascertainment occurs on an irregular basis in metropolitan hospitals, and is very infrequent in rural hospitals were it is usually circumstantial and associated with a staff visit for another reason (Bower, 2002).

The conceptual framework including the association between variables is illustrated in Figure 4. The outer box represents the total number of birth defects within the population. The inner portion of the model is composed of statutory notification sources, with the solid arrows representing the potential flow of case reports to the Birth Defects Registry. The flow of information along these arrows will determine the effectiveness of the Registry. Subsequently, the resulting incidence of birth defects reported by the Birth Defects Registry is contained within the central box.

The study explored if the lower incidence of birth defects in the Midwest is due to case under reporting. This is represented in the conceptual framework as the relationship between the total number of birth defects (outer square) and the incidence of birth defects reported by the Birth Defects Registry (inner square). The completeness of these squares is determined by the efficiency and accuracy of information that flows along the arrows from the notification sources. The outcomes

of these relationships will be determined by the empirical evidence and presented in the discussion chapter.


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                    ┌─────────────────────────────────┐
                    │  Voluntary Notification Sources │
                    └─────────────────────────────────┘

                 ┌──────────┐   ┌──────────────┐
                 │ Midwives │   │ Child Health │
                 │          │   │    Nurses    │
   ┌───────────┐ └──────────┘   └──────────────┘  ┌──────────────┐
   │ Perth     │                                  │ General      │
   │ Tertiary  │                                  │ Practitioners│
   │ Hospitals │                                  └──────────────┘
   └───────────┘
              ┌─────────────────────────────────┐
              │      Birth Defects Registry     │
              │   Incidence of Birth Defects    │
              │        in the Midwest.          │
              └─────────────────────────────────┘

   ┌───────────────────────┐   ┌───────────────────────┐
   │ Midwives Notification │   │  Hospital Morbidity   │
   │        Forms          │   │  and Mortality Data   │
   └───────────────────────┘   └───────────────────────┘

              ┌─────────────────────────────────┐
              │  Statutory Notification Sources │
              └─────────────────────────────────┘

   ┌──────────────────────────────────────────────────────────┐
   │ Total Population of Birth Defects in the Midwest Population│
   └──────────────────────────────────────────────────────────┘
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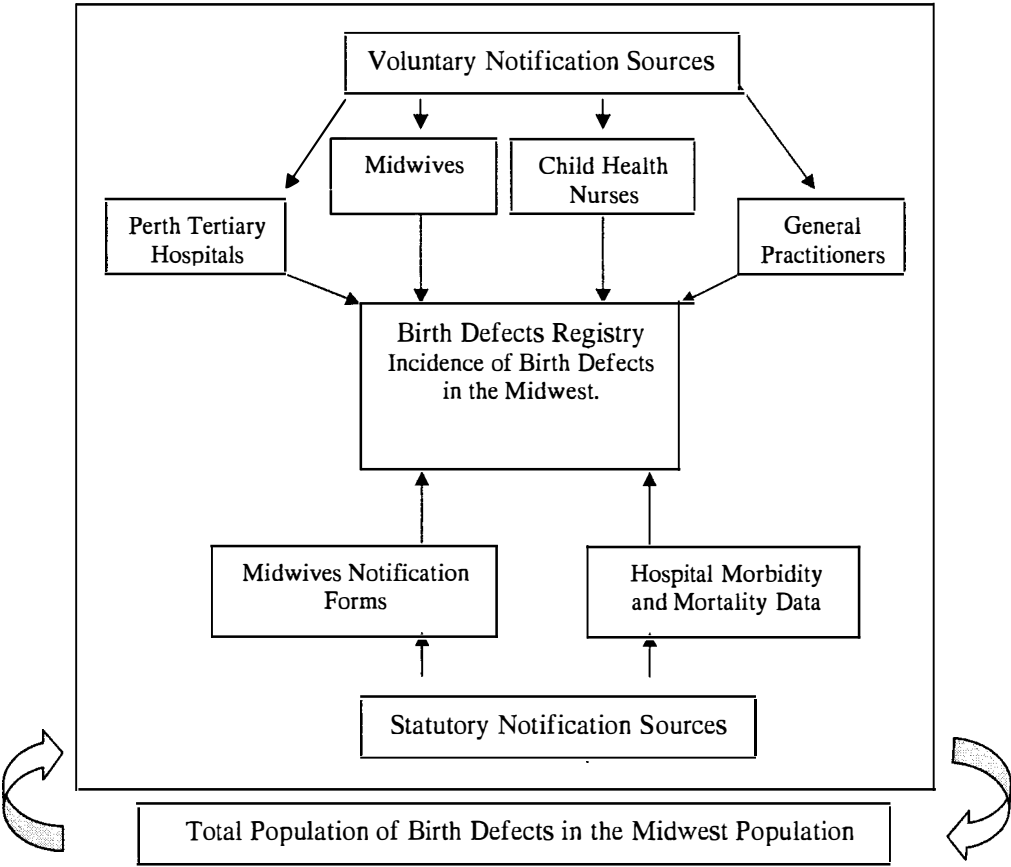
Figure 4. Conceptual Framework – Birth Defect Registry case notification and data flow.

CHAPTER 5

METHOD

In this chapter the methods and procedures used to conduct the study will be discussed. The chapter will describe the quantitative and qualitative phases of the study respectively. The first section of the chapter will describe the methods and procedures relating to the quantitative phase. Similarly the second section of the chapter will discuss the method and procedures relating to the qualitative phase of the study.

5.1 Phase 1: The Quantitative Phase

5.1.1 Design

The quantitative phase of this study used a two sample capture-recapture methodology, which allows estimation of the extent of incomplete ascertainment using information from overlapping lists of cases from distinct sources (Hook & Regal, 1995).

In this study the first sample or "capture", consisted of the cases of birth defects born to women resident in the Midwest between 1995 - 1999, diagnosed at birth, that have already been recorded by the Birth Defects Registry. The second sample, or "re-capture", was a random sample of births to Midwest mothers in 1995 - 1999 to gain information regarding the presence of birth defects in Midwest hospital birth records. Birth defects included in the study sample were required to meet the definition of a birth defect as outlined by the Birth Defects Registry. An

ascertainment-adjusted analysis was then calculated to achieve an estimate of birth defects in the Midwest population.

5.1.2 Setting

The quantitative phase of data collection was undertaken at the Geraldton Regional Hospital located in the Midwest of Western Australia. The hospital offers general medical, surgical, paediatric and obstetric facilities. Additional information was also collected from the Geraldton Community and Child Health Centre, located on the same site as the hospital. An initial visit was made to the Midwest to ascertain the feasibility of the proposed study and resulted in some changes to the initial proposed study methodology.

5.1.3 Sample

The sample was selected from a defined target population. The target population consisted of newborns born between the years 1995 – 1999 and whose mothers resided in the Midwest at the time of their birth. The target population was accessed via maternal confinement case notes held by Geraldton Regional Hospital. A total of 440 cases were sampled. Additional information, supplemental to the random hospital sample was collected from the Community and Child Health Centre and as well as information provided by participating parents. Case notes held by St John of God Geraldton (the private obstetric hospital) were not made available to the researcher, this issue is discussed under ethical considerations in this chapter.

5.1.4 Sampling Method

The target population in this study was accessed through the Geraldton Regional Hospital. The Director of Nursing (DON) at the Geraldton Regional Hospital and the Child Health Manager were initially approached through an

introductory letter and information sheet (Appendix 3 & 4) that outlined the purpose of the research and requested a meeting during a planned forthcoming visit by the researcher. Two weeks following receipt of the introductory letter and information sheet, the DON and Child Health Manager were contacted by the researcher via telephone phone to arrange an initial meeting to discuss the study, as well as their potential participation.

The researcher visited Geraldton on the 15th of February 2002. During this visit the DON and the Child Health Manager were familiarised with the study aims and possible outcomes. The assistance of staff at the hospital and child health centre in data ascertainment was requested and the most efficient and least disruptive method of undertaking this process was discussed. The study proposal was submitted to the DON and Hospital Ethics Committee for review, prior to granting approval and access to hospital case notes. The researcher was informed of ethical clearance and approval to access hospital case notes by correspondence (Appendix 5). Upon receipt of this letter the research contacted the DON by telephone to arrange an appropriate date for data collection to proceed.

The researcher and an experienced research assistant from the Birth Defects Registry undertook data collection at the Geraldton Regional Hospital and Child and Community Health Centre on the 17th of May 2002. The Manager of Medical Records at Geraldton Regional Hospital was provided with the unit record numbers of the case notes that were required for sampling. To facilitate the review of case notes medical records staff pulled case files prior to the arrival of the researcher and research assistant. A room containing desk, chairs and the required case files were provided so that files could be reviewed and information collected confidentially. Case notes of each sampled newborn were read and information regarding the diagnosis of a birth defect was recorded.

5.1.5 Sampling Parameters

Quantitative data was ascertained from newborn hospital birth case notes. These case notes were contained within the maternal confinement record for that birth and were therefore accessed in the mother's file. Information gathered from child health and parental sources could not be randomly selected, thus did not fulfil the assumptions for a two-source capture-recapture model. Therefore, child health and parental information was used to clarify and expand upon the findings of the hospital birth case notes.

As previously discussed, a two-source capture-recapture model requires the application of four assumptions. The researcher considered these assumptions prior to selecting the parameters from which the sample population would be obtained. This resulted in obtaining the study sample from birth case notes only.

To ensure that the assumptions of a capture-recapture model were fulfilled it was important to apply them to both the first and second samples. Application of the assumptions to the first sample or capture, obtained from data held by the Birth Defects Registry, was addressed by firstly considering the assumption of a closed population. Diagnosis of a birth defect can be made up until the age of six years. Therefore, during this six-year time frame it is possible that an infant or child maybe newly diagnosed or have additional diagnosis reported to the Registry. This violates the assumption of a closed population and presents difficulties in accessing a complete sample source from which data for the second sample can be obtained. To close the sample population the researcher choose to limit it to newborns diagnosed with birth defects at birth only. Information relating to the newborn's first days of life is recorded in the birth case notes. In addition, this also presented a complete and comparable recapture from which the second sample could be obtained.

Furthermore, limiting the sample parameters to newborns diagnosed with a birth defect at birth provided a comparable data set, and fulfilled assumptions three and four. This was achieved by ensuring that newborns had the same chance of being captured in the first sample as in the second. As well, data could be collected from the first sample independent of the second sample and visa versa. Ensuring that the assumptions of equal sampling probability and independence are fulfilled.

5.1.6 Sampling Procedure

Sampling of data from hospital birth case notes and collecting information from child health records and parents occurred as follows:

1.   Hospital Birth Case Note Sampling

Hospital birth case notes of clients within the target population were randomly sampled from hospital records. The random sample consisted of births that occurred at Geraldton Regional Hospital between the years 1995 – 1999. The sample was generated from the midwives birth notification records held on the Maternal and Child Health Research Database, in the form of hospital unit record numbers. Hospital unit record numbers were then forwarded to Geraldton Regional Hospital coding staff, who accessed the records from hospital archives on behalf of the researcher.

It was not possible to examine records of all births to Midwest mothers in the study time frame due to logistic and time management issues. During the years 1995-1999, Midwest mothers gave birth to 5054 newborns in Midwest hospitals. The time frame required to review 5054 case notes and the demands on the coding staff to access these records was not within the scope of this study. Furthermore, case notes held by St John of God Geraldton (the private obstetric hospital) were not made

available to the researcher. This contributed to the researcher's inability to sample all births occurring in the 1995-1999 cohort.

A randomly generated sample from the target population was taken as a representation of all births occurring in 1995-1999. Calculation of the required sample size was based upon the 5054 Midwest births that occurred within the five-year study period. Sample size estimation was based upon an expected frequency of 4% birth defects diagnosed at birth, and the farthest from expected rate that was acceptable, being 2.3% birth defects, with 95% confidence. Achieving an expected frequency of at least 4% is based on the percentage of birth defects diagnosed at birth in the metropolitan population, which at present averages 4%. The statistical epidemiological computer package EPIINFO (Centre for Disease Control and Prevention, 1996) was used to calculate the required representative sample size. The sample size calculated was 464. To allow for potential non availability of some records, 500 records were selected in the random sample.

The number of records made available for the sample by the hospital was 440. The 60 outstanding case files could not be found by the medical records staff. Infant birth case notes at Geraldton Regional Hospital are filed together with the mother's obstetric admission. Infant birth case notes do not have their own unit record numbers and were accessed via the mother's unit record number. Subsequent review of the birth case notes found 141 cases where multiple newborns had been born to the same mother within the study's target population. This presented difficulty in deciding which newborn to select for inclusion in the study. Fortunately, none of the case files in which this occurred were found to have a birth defect, therefore, any potential complication due to this occurrence was avoided. These additional newborns were not added to the sample size, each sampled case file was counted once only.

Data analysis required the comparison of data held by the Birth Defects Registry for the same cohort. Therefore, client details were initially retained and recorded on a master list so that this record matching could occur. Following the initial data matching, client details were replaced by a numerical code to assure anonymity.

2.   Child Health Records

The availability and nature of child health records varied between child health nurses, and therefore, access to specific sample numbers was not guaranteed. This variability included the format of documentation, the method of filing and completeness of records. Therefore, child health records were sampled according to convenience, and their sample numbers not factored into the calculated sample size of 500. Information obtained from review of child health records was, therefore, additional to the data obtained from review of hospital birth records. Child Health Assessment records were provided by two child health nurses. The assessments had been undertaken on five-year old children during the years 2000 and 2001. This equates to the child's birth year of 1995 and 1996. The records were not complete because not all children attended their Child Health Clinic for a five-year old health check. In all, 242 records were sampled. Of the 242 records sampled, three children with birth defects were found.

3.   Parent Provided Information

Privacy legislation (Federal Privacy Commissioner, 2001) introduced after the implementation of this study impacted upon the availability and subsequent release of client details by any health practitioner. Guidelines issued by the Office of the Federal Privacy Commissioner pertaining to National Privacy Principles 1 & 10 (Federal Privacy Commissioner, 2001), outlines the Privacy legislation in detail. As a result of this legislation, health practitioners are no longer permitted to authorise the

release of client information without prior client consent. Given that this study was planned prior to the introduction of the legislation, it was not feasible for the researcher to directly contact clients and request their consent to access health practitioner case notes for the purpose of the study. As an alternative, the researcher attempted to access client information by approaching the public directly. This sampling procedure was referred to as parent provided information and occurred according to the following two methods:

- A publicity campaign involving extensive community advertisement of the research was undertaken. The campaign focused upon facilitating maximum exposure to parents with children within the target population. A newspaper article was published in the Geraldton Guardian Community Newspaper (22/02/02) outlining the study and asking parents to contact the researcher for further information. In addition, information flyers advertising the study were displayed at the Child and Community Health Centre and distributed for display at the four main medical practices, the majority of local primary schools, nursery schools, day-care centres and Nursing Mothers Association (Appendix 6).

- Child health nurses were approached via a covering letter and information sheet (Appendix 3 & 4) outlining the study and potential importance of their assistance. Child health nurses were approached and asked to provide parents with information regarding the study in the course of their daily practice for a period of three months. Only parents of children who met the target population criteria were approached. Child health nurses were provided with parent information sheets and consent forms (Appendix 7 & 8) for parents. Parents wishing to participate were requested to complete the consent form and return it directly to the child health nurse, or keep the information and forward the completed consent form (in a reply paid envelope) to the researcher at their convenience. Completion of the parental consent form enabled the release of client contact

details by the child health nurse to the researcher. Parents were then contacted directly by the researcher, via telephone or a home visit, and information regarding their child was subsequently obtained from them.

The community advertisement was released to coincide with the collection of hospital and child health data. The aim of this approach was to enable the researcher to contact and visit parents whilst in Geraldton for hospital data collection. Parent recruitment via the child health nurses was conducted over a three-month period following this date and concluded on the 17th of August 2002. This time frame allowed child health nurses to approach as many parents as possible and in turn give these parents ample opportunity to respond to the researcher.

Study participation from parent provided information sources resulted in the recruitment of only two parents. In both instances the parents were informed of the study via the newspaper article. One parent provided the researcher with study information via email, and the other parent visited the hospital during the data collection visit to provide study details.

Information collected from child health records and provided by parents was clarified via cross-referencing with Birth Defect Registry data. If no record of that child existed, registry letters were forwarded to the relevant general practitioners to clarify the child's condition and diagnosis of a birth defect. Four letters were forwarded to practitioners to clarify birth defects or suggested conditions listed in child health records. No further diagnosis of birth defects were confirmed by practitioners.

4.   Ward Birth Register Search

Supplementary to the random sample of hospital unit record numbers, the researcher actively sought possible additional cases of birth defects from the labour ward delivery register. The labour ward delivery registry is commenced annually on

the 1st of January and is used as a ward based record of all the births that occur within a calendar year. Information regarding a birth defect should be recorded by the midwife in the 'comments section' of each birth record. Use of this section appears to be subjective and incomplete, relying on the discretion of the midwife. Hence, data obtained from this source was considered to be inconsistent and incomplete. The case files of mothers whose newborn infants were recorded as having a birth defect were then accessed. Data were recorded on newborns diagnosed as having a birth defect or a condition suggestive of a birth defect. Ten possible cases of birth defects were listed in the hospital ward birth register.

5.1.7 Instruments and Materials

1.   A random sample of 500 unit record numbers was generated from the Maternal and Child Health Research Data Base for births at Geraldton Regional Hospital during 1995-1999. The unit record numbers were then used to access the hospital case notes.

2.   Birth Defect Registry notification cards (Appendix 2) were used to record study data from the above sources regarding newborns found to have birth defects or conditions suggestive of the same. Information recorded included infant's demographic details and type of birth defect or condition suggesting a possible existence of a defect, but requiring further investigation.

3.   An Additional Question Form (Appendix 9) was completed for each newborn recorded on a notification card as having a birth defect. The additional question form listed the answers to the following questions asked the data:

3.1 Was the defect coded by the Hospital? If *YES,* how was it coded?

3.2 Where in the notes was the information about the birth defect documented?

3.3 Was the infant referred to other departments or health professionals for follow-up?

3.4 Was the child transferred to Perth for treatment?

3.5 Was the birth defect diagnosed prior to birth? If *YES,* by whom and what technique was used to confirm diagnosis?

4.   A data record sheet (Appendix 10) was used to record the unit record number of each file reviewed. In addition, the number of eligible newborns from within the target population filed under the same unit record number was also recorded. For example if a mother had given birth to two children consistent with the target population both these were sampled then a x 2 was placed by her number. Indicating to the researcher that two newborns were sampled from one mothers unit record number.

5.   A data record sheet (Appendix 10) was also used to list the number of child health records applicable to the target population that were reviewed. These were divided into groups according to the year the assessment occurred.

5.1.8 Statistical Technique – Ascertainment Adjusted Analysis, Prevalence, Sample Difference and Confidence Levels

In this study a list of newborns with birth defects 'captured' or recorded by the Birth Defects Registry was compared to the list of newborns 'recaptured' independently from hospital case records.

The following formula for assessing the completeness of ascertainment was used (Bower, Ryan & Rudy, 2001; Stephen, 1996) and is known as an ascertainment-adjusted analysis. The aim of the analysis is to generate N, which equals an estimation of the total population size. The formula for an ascertainment-adjusted analysis is shown as:

$N = [(M+1) (n+1) / (m+1)] - 1$

Where:

N = estimate of population size.

M = subjects in sample 1 (cases recorded by the Birth Defects Registry (BDR)).

n = subjects in sample 2 (cases of birth defects collected in the research data).

m = all subjects common to M and n.

The estimate generated by N was then converted into an ascertainment adjusted prevalence rate (AAPR). A prevalence rate is the estimated number of cases of a disease or occurrence during a particular period of time. Prevalence is expressed as a ratio of events over the population at risk (Anderson, 1990). The formula for calculating a prevalence rate is shown as:

Prevalence rate = the estimated number of birth defects in the sampled population

Sampled population size

This rate provides an estimated percentage of the number of birth defects likely to have occurred within the 1995-1999 study cohort. The AAPR was then compared to the Birth Defects Registry's prevalence rate (BDRPR) for the same sample. Ultimately, an AAPR greater than the BDRPR would indicate that the Birth Defect Registry data are incomplete. This suggests that birth defects have occurred within the sample population and in all probability the entire 1995-1999 cohort that have not been reported to the Birth Defects Registry.

The statistical significance of any findings was considered by calculating 95% confidence intervals around the AAPR and BDRPR (Armitage, 1971) and around the difference between the APPR and the BDRPR (Shott, 1990).

The calculation for the difference allows for the AAPR and BDRPR to be compared and inferences made about this comparison. This calculation is performed

in two stages. Firstly, the sample difference or p1 − p2 is determined. The formula for calculating the standard error of the sample difference (Shott, 1990, p72) is shown as:

$$S\ diff\ (p1\text{-}p2) = \sqrt{\frac{p1\ (1-p1)}{n1} + \frac{p2\ (1-p2)}{n2}}$$

Where:

p1 = AAPR (Ascertainment-adjusted Prevalence Rate).

p2 = BDRPR (Birth Defect Registry Prevalence Rate).

n1 = Sample size of the Study Population.

n2 = Sample size of the Birth Defect Registry Population.

If no difference exists between p1 and p2 then the expected sample difference would be zero. A 95% confidence interval can be calculated around the estimated sample difference (p1-p2) to determine the likelihood that p1- p2 is different from 0. If the confidence interval around the estimated sample difference (p1-p2) includes zero, then the sample difference is not statistically different from zero at the 95% confidence level. If the confidence interval does not include zero, then we can conclude that at, the 95% confidence level, the AAPR is significantly different from the BDRPR. The formula for calculating a 95% confidence interval around the difference (Shott, 1990) is shown as:

95% Confidence Level = [(p1 − p2) − (zα/2)(*s diff*)], (p1 − p2) + (zα/2)(*s diff*)]

Where:

(zα/2) = 1.96

The formula for calculating the confidence intervals around the AAPR and the BDRPR (Armitage, 1971) is shown as:

p-1.96 sq root(p(1-p/n); p+ 1.96 sq root(p(1-p)/n)

where p = AAPR (or BDRPR) and n = study (or BDR) population.

5.2 Phase 2: The Qualitative Phase

5.2.1 Design

This study used an exploratory qualitative methodology. The primary aim of the qualitative phase was to determine recommendations that could improve the current case ascertainment methods of the Birth Defects Registry The focus group interview is a qualitative research technique used to determine the feelings and opinions of a small group of participants (Dilorio et al., 1994). A series of focus groups were used in this study to explore and clarify the research aims.

The study's qualitative phase was designed to use a semi-structured interview format to collect information from specific groups of Midwest clinician's regarding the birth defect reporting process. Participating clinicians are (referred to as respondents) were asked to provide feedback based on a set of interview questions (Appendix 11). The purpose of the interviews was to determine the current practice of reporting birth defects in the Midwest, and respondent feedback on how the

collection of birth defect information could best be achieved and/or improved. Information collected from focus interviews was designed to give depth to the existence of any quantitative discrepancies and provide suggestions that will enable the maintenance or improvement of data collection by the Birth Defects Registry. The interview questions were intentionally open-ended to promote further discussion relevant to the topic. Interviews questions were designed to identify strengths and weaknesses of current birth defect reporting practices, as well as providing information that could be used to make recommendations that would sustain or improve upon current birth defect case ascertainment methods.

5.2.2 Setting

The qualitative phase of data collection was undertaken at the Geraldton Regional Hospital. Focus group interviews with midwives were conducted on the maternity ward, interviews with child health nurses were conducted at the Community and Child Health Centre, and a group discussion occurred with doctors during a medical seminar morning. The interview setting was considered by the relevant staff members as most appropriate to their needs and work constraints.

5.2.3 Sample

This phase used the three major groups of Midwest case reporting sources to conduct focus group interviews: general practitioners, child health nurses and midwives working in the Geraldton Region.

5.2.4 Sampling Method and Procedure

The approach used to recruit the case reporting sources proceeded as follows:

- All possible case reporting sources were approached by an introductory letter and information sheet. The introductory letter (Appendix 3) outlined the purpose of the research and requested a meeting during a forthcoming visit. The information sheet (Appendix 4) outlined study expectations, guidelines, consent and confidentiality.

- Within two weeks of mailing the introductory letter and information sheet, all case reporting sources were contacted by phone or letter to discuss possible study participation. The director and associate professor at CUCRH was asked to provide names and details of all case reporting sources. The staff at CUCRH have a working knowledge of all practising case reporting sources in this area. Access to this information is also readily available in the public domain. Due to time constraints of the clinicians, individual meetings and interviews were not possible. The researcher found it more efficient for them to conduct focus group interviews during their scheduled meeting or work times.

- The researcher made appointments to meet with the groups of general practitioners, midwives and child health nurses at times convenient to them. Three focus group interviews were conducted. At the first, seven general practitioners and an obstetrician were present. The second interview consisted of six midwives, and five child health nurses attended the third interview. The interviews were conducted by the researcher and followed the format outlined by the interview questions (Appendix 11). Each interview lasted approximately twenty minutes with respondents contributing freely to the discussion with both answers and suggestions. The researcher documented information in notation form, during the interview. This allowed for later review and expansion upon the concepts and ideas that were discussed.

- In addition a telephone interview was conducted with the visiting Geraldton paediatrician at his Perth office. The paediatrician stated that he was unable to offer any information to the study.

5.2.5 Data Collection Procedure

The researcher initially proposed that the study would collect qualitative information via personal tape-recorded interviews with each study respondent. However, due to the time constraints of the respondents, individual interview's could be not be conducted. Subsequently, because of the study setting and the expressed need for a "non-threatening" conversational format in focus group interviews, the researcher considered that tape-recording discussions would not be the most efficient means of recording respondent information in this particular situation. When the researcher meet with potential focus groups participants it was clear that tape-recording of the focus group interviews would serve to hinder the flow of respondent information and would not affect the later thematic analysis of responses. Respondent interview information was, therefore, recorded in the form of researcher interview notes. While the recording of data did not follow the usual method prescribed by Dilorio et al. (1994), the researcher considered it appropriate to the needs of the target population, and the environment in which these health professionals worked. The researcher considered the depth of the researcher recorded notes and post interview reflection sufficiently captured the issues discussed, as well as, the depth of participant comment.

As previously stated, health professionals were assigned to one of three focus groups: medical practitioners, midwives or child health nurses. A separate discussion session was carried out with each of these groups and lasted approximately twenty minutes. Focus group interviews were promoted as discussion and information sharing sessions during which interview questions regarding the reporting of birth

defects could be addressed. Medical practitioners were interviewed whilst attending a monthly education breakfast. Midwives and child health nurses were invited to attend a workplace discussion addressing the interview questions, this invitation was relayed to them via their ward/unit managers. Only those midwives and child health nurses working a rostered shift at the time of the discussion chose to participate.

To validate the data collected during focus group interviews, the researcher emailed a copy of the interview record to a member of each focus group for clarification and authentication. The individuals were asked to carefully read the interview data. If focus group members agreed the information was a true and correct record of the interview, they were asked to sign an attached Authentication of Record Form (Appendix 12) and return it via mail to the researcher. To ensure confidentiality, the names of the individual focus group respondents have not been included in this document. Authentication of Record Forms are stored in accordance with NHMRC guidelines.

5.2.6 Data Analysis Procedure

The study's interview questions were used as the structure for presenting the interview data. Interview data was also analysed in terms of the research questions it was directed at answering. In accordance with Dilorios' et al. (1994) recommendations, analysis and reduction of focus group interview data was undertaken, followed by the classification of information into ideas or themes. Themes were derived from ideas expressed as words, sentences or paragraphs and that referred to specific content or affective details of an interview. Further validation of themes with supporting examples and interpretive statements was also undertaken. Data reduction was achieved via a process known as coding which is a means of classifying data into meaningful categories. Burns and Groves (1997) describes three types of codes as follows:

- Descriptive codes classify the data using terms that describe how the researcher is organising the data. These codes remain close to the terminology used by the participants, and provide a means of sorting interview statements.

- Interpretive codes move beyond the sorting of statements, using the participant's terminology to attach meanings to the statements.

- Explanatory codes are part of the researchers attempt to unravel the meanings inherent in the interviews.

The researcher used this coding method as the basis for reducing and analysing focus group interview data, in addition to the suggestions by Dolorio et al., (1994). Interview data was organised and documented according to focus group and interview question. Focus group interview data appears in the Results Chapter. The researcher then examined the data from each focus group for content and respondent attitude. Key statements used by each focus group to answer the interview questions were highlighted and listed as descriptive codes under the relevant interview question.

The main themes from each of these statements were described as interpretative codes. Interpretive codes were derived from commonalities in focus group interview statements. These codes appear as a term that describes the main theme behind the combined focus group response to each interview question.

Finally, the explanatory coding phase used by the researcher to explain the meanings behind the interpretive codes attempted to explore the answers to the interview and research questions. Data was used specifically to answer research question three. Subsequent analysis of qualitative information in conjunction with quantitative findings did, however, provide some additional data for answering research questions one and two.

5.2.7 Instruments and Materials

1. The interview questions detailed in Appendix 11 were used to guide focus group interview responses.

2. A Focus Group Interview Data Collection Sheet (Appendix 13) was used by the researcher to make notations during the interview.

5.2.8 Ethical Considerations

There were no risks to children or mothers involved in the study either in the random hospital sample, child health assessment or parent provided information. The researcher, research assistant and supervisors were the only people to have access to client data. Following data matching with information held by the Birth Defects Registry client names were removed from all records and replaced with a number to ensure confidentiality. All case note reviews were conducted in private. All coded surveys and records are being kept in a locked filing cabinet located in the Associate Supervisor's office (Birth Defects Registry). In accordance with National Health and Medical Research Councils (NHMRC) guidelines, all records will be destroyed five years after publication of the findings. Data will not be accessible by any other people.

Parent provided information was provided voluntarily. Parents were given the opportunity to ask questions at anytime and have them answered satisfactorily, as stated in the consent form. Withdrawal of consent to continue in the study at anytime, and without penalty, was an option for all parents. Approval to conduct the study was obtained from the Human Research Ethics Committee at Edith Cowan University and the Confidentiality of Health Information Committee (CHIC) at the Western Australian Department of Health. Geraldton Regional Hospital ethics approval was granted following hospital review and under the provision of Edith

Cowan University and CHIC approval. Copies of these ethical approvals are included as Appendix 14 & 15.

The Geraldton region supports two hospitals: Geraldton Regional Hospital and a private hospital named St John of God Hospital – Geraldton. Both of these hospitals provide a maternity service with approximately 70% and 30% of the region's births occurring respectively at each hospital. Ethical approval was sought from St John of God Hospital to review 150 client case notes (30% of the study sample size) and Geraldton Regional Hospital to review 350 client case notes (70% of the study sample).

The government introduced new private sector amendments to the Privacy Act, after commencement of this study. These amendments presented unexplored issues and concern for ethics committees, because the requirements of the Act were not specifically defined and largely open to interpretation. This study was the first to request approval to access client notes from St John of God Hospital since implementation of the new Privacy Act amendments and, therefore, presented as a test case. The researcher applied for ethical approval from St John of God Hospital on two occasions. The second application included requested amendments and clarification of the study's proposal aimed at assuring the committee that the methodology would not breach the privacy legislation. Consequently, ethical approval was declined on the basis that members of the committee were uncertain whether the study would breach amendments to the Privacy Act. Therefore, the researcher was unable to include St John of God Hospital as a sample source and the study sample size of 500 was obtained from Geraldton Regional Hospital only.

As stated above, Geraldton Regional Hospital represents by far the largest proportion of births in the Midwest region, therefore, the omission of St John of God Hospital records did not impact on obtaining a sufficient or representative sample size from the Midwest Region.

There were no risks to respondents participating in the focus group discussions. Approval to approach hospital staff regarding participation in the discussion was obtained from the DON. The discussion was commenced with a reiteration of the information sheet detail that respondents had previously received. This ensured that respondents were fully aware of the study and their decision to participate was informed. In addition respondents were asked to sign a consent form (Appendix 16) regarding the same.

CHAPTER 6

RESULTS

This chapter will outline the statistical techniques and results of data collected in Phase 1 (Quantitative) and Phase 2 (Qualitative) of this study. For clarity the first part of this chapter will describe statistical techniques and results for Phase 1. Similarly, the second part of the chapter will describe statistical techniques and results related to Phase 2.

6.1 Data Analysis – Phase 1: Quantitative

Data analysis was undertaken in two parts. Initially a description of the overall population sample was performed, followed by description of the demographic data.

The second part of data analysis involved the comparison of birth defect cases in the sampled population with birth defects recorded in the Birth Defect Registry, using an ascertainment adjusted analysis. In addition, child health data, parent information and other births to mothers in the hospital random sample were also collected. These data are presented and compared with information held by the Birth Defects Registry

6.1.1 Demographic Data

The study's random sample of hospital case notes (n=440) found 13 newborns with birth defects. The 13 newborns with birth defects consisted of eight males and five females. Of the eight male newborns, seven were of non-Aboriginal origin and

one was of Aboriginal origin, while, the group of five females consisted of four newborns of non-Aboriginal origin and one of Aboriginal origin. Of the sampled 440, 84 were Aboriginal and 356 were non-Aboriginal. Thus, the prevalence of birth defects in Aboriginal infants was 2.4%, and 3.1% in the non-Aboriginal group.

6.1.2 Additional Data Questions Related to the Diagnosis, Coding and Referral of Newborns found to have Birth Defects

Additional data questions were asked of the sampled cases found to have birth defects. Answers to the additional questions relating to the sampled data appear in Table 9.

Table 9

Additional Data Questions and Answers of Sampled Cases found to have a Birth Defect.

Questions	Answers
1. Was the defect coded by the hospital?	**Yes**: In 8 cases, birth defects were coded, of these 1 was incorrectly coded. **No**: In 5 cases birth defects were not coded, a record of the newborns birth defect was in the case notes only.
2. **Where in the notes was information about the defect documented?** All defects were recorded using at least one of these methods of documentation. Of particular note was the lack of	Midwives Notification Form; Child Health Book Summary of Birth; Progress Notes; Neonatal History and Assessment; Obstetric discharge Summary; Letters of correspondence; Obstetric Booking

documentation or failure to utilise the Neonatal History and Assessment Form and the Obstetric Discharge Summary.	Form; Ultrasound Examination Reports; Maternity Attendance Form; Princess Margaret Hospital Discharge Summary.
3. Was the infant referred to other departments? Newborns were referred to major metropolitan hospitals, other doctors, x-ray departments and pathology labs.	**Yes**: In 7 cases a newborn with a birth defect was referred to other hospitals or departments **No**: In 6 cases the newborn was not referred or there was no record of referral.
4. Was the child transferred to Perth for treatment?	**Yes**: In 5 cases the newborn was transferred to Princess Margaret or King Edward Memorial Hospital. **No**: In 7 cases there was no record of transfer during that admission. **No**: In 1 case the newborn was stillborn with no record of transfer.
5. Was the defect diagnosed prior to birth?	**Yes**: In 2 cases the defects were diagnosed prior to birth using ultrasound techniques. In 1 of these cases the newborn was stillborn. **No**: In 11 cases the birth defect was not diagnosed prior to birth.

6.1.3 Results of the Randomly Sampled Hospital Case Note Data compared with Birth Defect Registry Data

Cases within the study's randomly sampled hospital data found to have birth defects were compared with the Birth Defect Registry's list of records pertaining to the same sample. This comparison showed a record of 14 cases of birth defects held by the Birth Defect Registry (BDR), in contrast to the 13 sample cases found by the study to have a birth defect. Twelve cases were found to be common to both samples (i.e. of the 14 cases of birth defects known by the BDR, 12 were also found by the study and two were not). The two missed cases included an infant with vascular scalp malformation and an infant with ear abnormalities and hydrops fetalis. The case identified by the study but not by the Registry was an infant with multiple reportable birth marks. These results are shown in Figure 5.
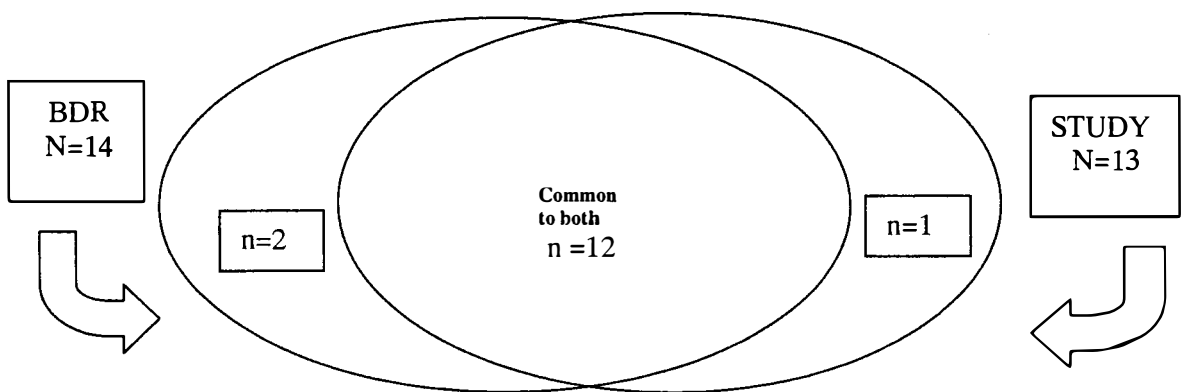
Figure 5. Results of the Randomly Sampled Hospital Case Note Data compared with Birth Defect Registry Data

The diagnosed birth defects found in the study's sample population and in BDR records have been listed in Table 10.

Table 10

<u>Identified Birth Defects in the 12 Cases Common to the Birth Defect Registry and</u>
<u>the Study.</u>

Case Number	Birth Defects
Case 1	Duodenal atresia
Case 2	Trisomy 21, atrial septal defect, multiple ventricular defects
Case 3	Sixth digit
Case 4	Rudimentary third toe
Case 5	Hypospadias
Case 6	Cleft lip and Palate
Case 7	Multiple abnormalities (requiring chromosomal studies)
Case 8	Bilateral talipes (requiring treatment)
Case 9	Bilateral six toes on feet
Case 10	Congenital adrenal hyperplasia
Case11	Cleft palate, Pierre Robin Syndrome
Case 12	Patent ductus arteriosus, Coloboma of the eye, Sixth nerve palsy, missing kidney, tracheal stricture, abdominal hernia, hypospadias

6.1.4 Ascertainment-Adjusted Analysis and Prevalence

The ascertainment-adjusted analysis was calculated as follows:

M= 14 (Sample 1: the number of sample subjects recorded by the BDR)

n= 13 (Sample 2: the number of sample subjects from hospital case note data found by the study to have a birth defect).

m=12 (the subjects common to both M and n).

Therefore;

N= [(14+1) (13+1) / (12+1)] – 1

N= [(15) (14) / (13)] – 1

N=[(210) / (13)] – 1

N=[(16.15)] – 1

N= 15.15

The ascertainment-adjusted analysis shows the estimated number of birth defects in the sampled population is 15.15. The ascertainment adjusted prevalence rate (AAPR) was calculated as follows:

Estimated number of birth defects in the sampled population = 15.15.

Sampled Population Size = 440.

Then;

AAPR = 15.15 x 100

      440

AAPR = 0.0344 x 100

AAPR = **3.44% (95% CI: 1.74 – 5.14)**

The ascertainment adjusted prevalence rate estimates showed that for births occurring in the Midwest between 1995–1999 3.44% of newborns were diagnosed with a birth defect. The corresponding rate based on data recorded on the Birth Defects Registry (BDR) is as follows:

Number of Birth Defect cases recorded by the BDR = 14

BDR Population Size = 440

Then;

BDRPR = $\underline{14.00}$ x 100

       440

BDRPR = 0.0318 x 100

BDRPR = **3.18% (95% CI 1.46 – 4.90)**

This prevalence rate indicates that of births occurring in the Midwest between 1995-1999, 3.18% were recorded by the BDR as having a birth defect.

    The difference between these population proportions is calculated as follows:

p1 = AAPR.

p2 = BDRPR.

Then;

p1 – p2 = 0.0344 – 0.0318

p1 – p2 = 0.0026

$$s\ diff\ =\ \sqrt{\frac{(0.0344)(1 - 0.0344)}{440} + \frac{(0.0318)(1 - 0.0318)}{440}}$$

$$s\ diff\ =\ \sqrt{1.45^{-0}4}$$

$$s\ diff\ =\ \underline{\mathbf{0.012}}$$

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Confidence Intervals were calculated as follows:

Confidence Interval = (0.0026 - (1.96) (0.012), (0.0026 + (1.96) (0.012)

Confidence Interval = **(-0.021, 0.026)**

Thus, the difference between the study prevalence rate and that obtained from the BDR data is small (0.26%) and, as the 95% confidence interval around this difference (-2.1%, 2.6%) includes zero, the difference is not statistically significantly different from zero.

6.1.5 Description of Additional Data collected from Parent Provided Information, Child Health Records and Ward Birth Register Information

In addition to the study's random sample of hospital case notes the researcher was able to collect data regarding the occurrence of birth defects from parents, child health records and the hospital ward birth register. This was not intended for inclusion in the study's capture – recapture methodology, however, it provides clarification and further credence to the outcome of this methodology. Table 11 shows these additional data.

Some of the birth defects discovered during the additional data collection had also been included in the random sample. All birth defects identified were cross-matched with Birth Defect Registry records, the Birth Defect Registry held records of all 15 birth defects, no unreported birth defects were found.

Table 11

Description of Additional Data collected from Parent Provided Information, Child Health Records and Ward Birth Register Information.

Origin	Sample Type	Sample Size	No. Defects
		n	n
Parent provided information	Convenience sample	2	2
Child health record information	Convenience sample		
	Year 2000	150	
	Year 2001	133	3
Ward birth register	Convenience sample		
	Years 1995 – 1999	3382	10

6.2 Data Analysis - Phase 2: Qualitative Phase

The following section will present the qualitative data provided by respondents during focus group interviews. For clarity, this section is divided into the three study focus groups, that is, medical practitioners, midwives and child health nurses. Each focus group is further divided according interview question and the associated responses. Information from each of the focus group interviews was later analysed for thematic content and compared to the other focus group responses.

1. Medical Practitioners

This focus group was undertaken with medical practitioners at a monthly education breakfast. Unfortunately, the researcher was allocated a strict time slot of 20 minutes only. First, a brief overview the study was presented. The researcher then began the interview. The group consisted of seven general practitioners and the resident specialist obstetrician/gynaecologist. Respondent input was variable, with questions receiving responses from three members of this focus group only. However, all interview questions were addressed. The questions, followed by the responses identified from the transcripts are as follows:

Question 1. What are the current procedures for reporting birth defects and how are they defined?

- No standard reporting procedures exist for the reporting of birth defects at the surgery's of the general practitioners (GP).

- Birth defects are normally reported by the hospital or the paediatrician.

Question 2. What is the process of referral for antenatal and unexpected birth defects occurring in the Midwest?

- Women pregnant with fetuses suffering from a known birth defect are referred by the GP to the obstetrician/gynaecologist or the client's care is shared between the two practitioners.

- If the birth defect is considered as life threatening to the infant, transfer maybe warranted in the antenatal period, ensuring birth of the baby at a tertiary perinatal hospital that has the expertise and resources to manage the newborn's condition.

- If the defect is not compatible with life (depending upon the gestation), referral outside the Midwest may not be required. A termination could be conducted at the local hospital.

Question 3. How do you feel the current reporting practices could be improved?

- A general lack of awareness regarding the reporting process of birth defects was expressed. One GP commented, 'Is it that the orange form?'.
- Suggestions to improve the process of reporting included; adding a page to the child health book, as this would make reporting more accessible to doctors and child health nurses and would also advertise the need to report a birth defect to health professionals; a pad of reporting forms with tear off pages, similar to a script pad that practitioners could have on their desks; maybe something could be included on the Medicare rebate form that could then be accessed via a central database.

Question 4. Who is best situated to assume the responsibility of reporting defects?

- The general consensus suggested that child health nurses and the specialist paediatrician involved with review and ongoing care of the child would be best suited to report birth defects.

Question 5. If you were setting up your own Birth Defects Registry what reporting procedures would you put into place?

- No further suggestions were made at this point as responses similar to those in question three were provided.

2. Midwives' Focus Group

The midwives focus group consisted of a group of six midwives who were rostered on shift at the time of the researcher's visit to the ward. No time limit was placed on the length of this interview, however, the time midwives were able to spend was variable, as they were still responsible for client care. The interview took approximately twenty minutes and was conducted in the labour ward office. Prior to commencing the discussion, a summary of the study aims and background was presented. The questions were then asked, the data from respondent transcripts are listed below.

Question 1. What are the current procedures for reporting birth defects and how are they defined?

- The hospital nursing procedure manual contained a procedure entitled congenital malformations. This procedure defined a malformation, listed the expected outcome, outlined a process for reporting and listed those defects included and excluded from reporting. The expected outcome was 'that all congenital abnormalities are notified to the Western Australian Congenital Malformation Register' (Geraldton Health Service, 1992, Reviewed 4/1996). The procedure directs notification of congenital abnormalities by completion of a birth MR15 (mandatory midwives notification of birth/case attended form) for early diagnosis, and completion of a birth defects registry notification card for late diagnosis (Geraldton Health Service, 1992, Reviewed 4/1996).

- Midwives indicated they completed the Midwives Notification Form at the birth of each case attended. Only one of the midwives indicated knowledge of the birth defects registry notification card, however this midwife was unsure who would collect the card or how to forward it to the Birth Defects Registry.

Question 2. What is the process of referral for antenatal and unexpected birth defects occurring in the Midwest?

- Inpatient antenatal women having pregnancies with major anomalies are referred by the GP to the specialist obstetrician/gynaecologist. These women are usually then referred to tertiary hospitals in Perth.

- Unexpected birth defects requiring treatment are usually transferred to Perth. If an infant with a defect that has associated high-risk complications, then the infant is transferred to Perth using the tertiary hospital infant retrieval team. More often than not this occurs, as there is no resident paediatrician in the Midwest to care for these newborns. The visiting paediatrician may be involved in follow-up care.

Question 3. How do you feel current reporting procedures could be improved upon?

- Midwives suggested that knowledge regarding the role and function of the Birth Defect registry was lacking. Some form of ongoing education/promotion sessions would be beneficial in highlighting birth defects in Western Australia, what defects need to be reported and how.

- This was seen to be particularly critical in rural areas, as hospital staff tend to be transient.

- Midwives suggested that because birth defects are so few and the need to report them infrequent, reporting cards need to be implemented with other documentation to ensure it did not get missed.

- Of the six midwives present, only one reported ever having seen a Birth Defect Registry notification card. When asked where the cards were kept on the ward the midwives were not sure and replied "somewhere in the filing cabinet".

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Question 4. Who would be best suited to assume the responsibility of reporting birth defects?

- Midwives suggested that the diagnosing doctor would be best suited to complete the notification cards, as they were the ones reviewing the infant and following them up at home. However midwives said they needed more clear direction and reminding as to what they are required to do regarding birth defect reporting. It was noted that midwives complete the birth notification form on all births and birth defects were included on that form. It was suggested that this form was likely to miss defects diagnosed after birth as the paper work had already been completed and amending this form to reflect the diagnosis often would be forgotten.

- Midwives also suggested that child health nurses would well suited to follow-up and notify the Registry of newborns and children diagnosed with birth defects after the birth period. However they appreciated the difficulty this task may present, as documentation provided to child health nurses in the form of completed child health books in their opinion were poor filled out. It was suggested that this occurred mainly because GPs frequently did not complete the medical neonatal physical assessment records in these books, either at birth or upon discharge.

Question 5. If you were setting up your own Birth Defects Registry what reporting procedure would you put into place?

- Midwives stated that because birth defects are few, the need to report them is infrequent. Therefore notification cards are filed in cabinet drawers and people forget about them. It was suggested that if notification cards were centrally

located, such as in the child health books it would serve to remind people of the need to notify and make sure notification cards were not lost. It was further recommended that the notification cards include an explanation on how to complete cards and where to forward completed cards.

3. Child Health Nurse Focus Group

Five child health nurses attended the group interview. These nurses were on duty and present in the office at the time the interview was being conducted. Prior to commencing the interview the study background and aims were outlined briefly. The interview was presented as an information sharing discussion that would take approximately twenty minutes. All the child health nurses were of senior status and had been working in the area of child health for several years. The interview took place in the staff lounge at that child health centre and took approximately twenty minutes The interview questions and responses identified from the data are listed below.

Question 1. What are your current procedures for reporting birth defects and how are these defined?

- It was stated that no formal procedure existed regarding the reporting of children with diagnosed birth defects to the Birth Defects Registry. This is usually performed by the child's GP. However, all physical and developmental assessments attended by the Child Health Nurses (CHNs) on these children are documented in the child health book.

- On occasions CHNs are contacted by GPs and asked to follow-up a child with a birth defect or assist the co-ordination of their care. This, however, only occurs in extreme cases.

- CHNs also receive discharge information regarding children with birth defects treated at the tertiary hospital in Perth and discharged home.

- CHNs stressed that child health books were under utilised as an ongoing record of the child's health status and as a form of communication between health providers. CHNs reported that many pages of the books were not completed. Parents also are often not familiar with what information is in the child health books and why it is an important record.

Question 2. What is the process of hospital referral for antenatal and unexpected birth defects occurring in the Midwest?

This question was not applicable to CHNs and, therefore, was not asked.

Question 3. How do you feel current reporting procedures could be improved upon?

- CHNs expressed that they would benefit from education and promotion by the Birth Defects Registry regarding what needs to be notified and how.

Question 4. Who would be best suited to assume the responsibility of reporting birth defects?

- CHNs suggested that GPs would be best suited to report birth defects as they often continue as a child's primary health care giver. Child health attendance is not 100%, and is not necessarily ongoing, it depends upon the parent. Parents attend child health clinics with their children for routine health checks on a voluntary basis.

- The decrease in CHN-provided immunisation clinics was also suggested as a contributing factor in reducing the contact that CHNs have with many families.

- CHNs conduct health assessments on all five-year old school children, during school hours. This was suggested as the only guaranteed contact CHNs are likely to have with a child. CHN's are already aware of children with serious birth defects by this time and are following them up.

- CHNs expressed knowledge regarding the Birth Defect Registry notification cards but had not been required to use them. When asked where these cards were kept in the child health office, the response was 'somewhere'.

- CHNs suggested that the promotion of the role and function of the Birth Defects Registry to community health workers would be an important consideration, as they would find it useful.

- CHNs recommended a centralised system of reporting where by notification cards could be accessed by all health professionals coming into contact with a child. Inclusion of these in the child health book was suggested, with two or three notification forms being included to allow for ongoing case notification.

- CHNs stressed that presently that child health books as a resource and health record are very under utilised.

The following section will present the thematic analysis of the focus group interview data.

6.2.2 Thematic Analysis of Focus Group Interview Data

To enable the identification and clarification of themes, data needs to be reduced and analysed in a concise and logical format (Burns & Groves, 1997). Data reduction can be achieved via a process known as coding. Coding is a means of classifying data into meaningful categories. Burns and Groves (1997) describes three types of

codes, these are descriptive, interpretive and explanatory codes. The researcher used this coding method in conjunction with Dilorio et al. (1994) suggestion for analysing focus group interview data. Key statements used by each focus group to answer the interview questions were highlighted and listed as descriptive codes under the relevant interview question. The main themes from each of these statements were described as interpretative codes. Interpretive codes were derived from commonalities in focus group interview statements. These codes appear as a term that describes the main theme behind the combined focus group response to each interview question. Lastly, the explanatory coding phase is used by the researcher to explain the meanings behind the interpretive codes and is the researchers attempt to explore the answers to the interview question and research questions.

The following section will present the major themes arising from each interview question, as suggested this will include interpretative and descriptive codes with supporting examples from which the major themes were developed. Information will be listed under the heading of the corresponding interview question from which it was gained.

Question1: What are the current procedures for reporting birth defects and how are they defined?

Descriptive Codes: Focus group respondents reported the following:

GPs: no standard/formal birth defect reporting procedure;

Midwives: reporting procedure present, but knowledge of it among midwives;

CHNs: no standard/formal birth defect reporting procedure.

Interpretive Code or Theme: Knowledge Deficit

Explanatory Code: All groups expressed knowledge of the Birth Defects Registry but were not clear on the procedures or whose responsibility it was for notifying birth defects. Midwives and CHN identified that they were uncertain exactly what defects

needed reporting, in addition to where completed notification cards should be sent. There was no consensus as to who should be reporting defects. Interview data suggests that a knowledge deficit exists regarding what defects should be reported, by who they should be reported, and who the completed notification cards should be sent.

Question 2: What is the process of referral for antenatal and unexpected birth defects occurring in the Midwest?

Descriptive Codes: Focus group respondents reported the following;

GPs: major abnormalities are referred to the obstetrician and transferred during the antenatal period to the tertiary Perth hospital;

Midwives: newborns born with an unexpected birth defect are retrieved by the Western Australian Neonatal Transport Service and transferred to a Perth tertiary hospital, lack of resident paediatrician makes the need to transfer a routine practice.

Interpretive Code or Theme: High transfer rates of compromised neonates.

Explanatory Code: This feedback is consistent with the information gained from the additional questions asked of the sampled hospital case notes. The lack of a resident paediatrician appears to have resulted in a limited availability of resources to care for newborns compromised by birth defect related conditions. Therefore, the retrieval and transfer rates of these newborns is high to tertiary level hospitals is high. Limited resources may also be a factor in diminishing the ability to transfer these newborns back to the Midwest for lower level care and follow-up. This results therefore in limited exposure of health professionals to birth defects and related conditions.

Question 3: How do you feel the current reporting practices could be improved upon?

<u>Descriptive Codes:</u> Focus group respondents reported the following;

GPs: a general awareness of the need to notify birth defects to the Birth Defects Registry was expressed, however there were inconsistencies in terms of who, how and with what notification was to occur.

Midwives: expressed knowledge of the Birth Defects Registry. However were unsure of the role and function of the Registry, and suggested they would benefit form some form of ongoing education/promotion. Infrequent exposure to newborns with birth defects has contributed to staff being unfamiliar with the procedure for notifying birth defects. Similar comments were made in response to question 4.

CHNs: Similarly stated they were aware of the Birth Defect Registry's existence, however, expressed they would benefit from education and promotion with regard to the role of the Registry as well as what and how to notify birth defects. Feedback regarding Birth Defect Registry results and findings in the form of a newsletter would be of interest.

<u>Interpretative Code or Theme:</u> Need for ongoing education and promotion

<u>Explanatory Code:</u> An overall awareness of the Birth Defects Registry and possible need to report birth defects was expressed by all groups. Generally interview respondents were not clear as to the role and function of the Registry, in addition to whose responsibility notification of birth defects was, where reporting cards were located and to whom completed cards should be sent. The majority of respondents suggested that they would benefit from ongoing education, promotion and feedback regarding the reporting process and results of Registry findings. Importantly, it was recommended that this be an ongoing process as there is a high turn over of staff, particularly in rural areas.

Question 4: Who would be best suited to assume the responsibility of reporting birth defects?

Descriptive Code: Focus group respondents reported the following:

GPs: suggested that CHNs and specialist paediatricians involved in follow-up of the child would be best suited to reporting birth defects.

Midwives: suggested that the diagnosing doctor would be best to assume the responsibility of notifying cases of birth defects, as they were responsible for initial review and referral. Midwives also suggested that CHN would be well suited to notifying ongoing cases of birth defect diagnosis as they were the next community contact point for newborns after the neonatal period. In addition midwives stated that this task is made more difficult for CHN as documentation of child health books by GPs is poor.

CHNs: suggested GPs would be the best suited to assume responsibility of reporting as they will be the newborn's primary health care provider. Since child health attendance is variable, CHN felt that they would not be best suited to assume primary responsibility for notification of birth defects. CHN reported reviewing a significant proportion of 5 year olds as they started school.

Interpretive Code or Theme: Ownership (unwillingness to assume responsibility for notification of birth defects).

Explanatory Code: Interestingly all groups suggested that another professional group than their own would be best suited to report birth defects. This perhaps reflects the knowledge deficit that exists regarding whose responsibility notification is and what is required when reporting a birth defect. The researcher believes that a distinct non-acceptance of ownership was expressed by respondents due to unfamiliarity with reporting procedures and the voluntary nature of birth defect notification.

Question 5: If you were setting up your own Birth Defects Registry what reporting procedures would you put into place?

Descriptive Code: Focus group respondents reported the following:

GPs: suggested that adding the notification forms to the child health book, would make reporting more accessible to doctors and child health nurses, and would also act as a reminder to health professionals that birth defects needed to be reported. Other suggestions included binding notification cards into pad, similar to a script pad, as these could be kept on the desk and are less likely to be misplaced than a single notification card. Also possible linkage with the Medicare rebating scheme.

Midwives: suggested that the infrequency with birth defects occurred, contributes to notification cards being filed and forgotten about or misplaced. If notification cards were centrally located, such as in child health books, this would serve to remind people of the need to notify and prevent the loss of notification cards.

CHNs: suggested a centralised system of reporting that was accessible to all health professionals involved in the infant/child's care. An option may be to include 2 or 3 notification forms in the child health book as this would allow for ongoing notification. However, CHN reiterated the under use of the child health book as a resource and record and suggested this too was an area that needed to be addressed.

Interpretive Code or Theme: Need for Centralisation of notification cards

Explanatory Code: All respondents suggested centralising of notification cards would increase their accessibility, act as a reminder to health professional that birth defect cases need to be notified to the Registry and prevent notification cards from being misplaced. The primary suggestion was to add notification cards to the child health book, however CHN stressed that currently child health books are under utilised.

In summary, the themes identified from the focus group interviews included: knowledge deficit, high transfer rates, ongoing education and promotion, ownership and centralising notification cards. The following section will discuss these themes and make recommendations that could potentially sustain or improve current birth defect case ascertainment methods.

CHAPTER 7

DISCUSSION

The following chapter will present and explore the major findings in this study, and their relationship to methodological, theoretical and practice issues. The study's strengths and limitations will then be discussed. For clarity the first section of the chapter will address the quantitative findings, followed by the qualitative findings. The final section of the chapter will consider the combined quantitative and qualitative findings and outcomes.

The decision to undertake this study was prompted by a WA Birth Defect Registry report finding that indicated the incidence of birth defects in rural areas of Western Australia are comparatively less than those in metropolitan areas. Authors of the report further suggested ... "this was possibly due to case under-ascertainment rather than a real difference" (Bower et al., 2000, p3).

The aim of this study was two fold. The first aim was to determine if the observed lower incidence of birth defects in the Midwest was due to under reporting. The second aim was to identify and make recommendations that would sustain or improve upon current birth defect case ascertainment methods.

Phase 1, or the quantitative phase of the study focused primarily on research questions related to the first study aim. Capture-recapture methodology was used to determine if the observed lower prevalence of birth defects within the 1995-1999 Midwest cohort was in fact a true finding or the result of case under reporting. The study used a random sample of hospital birth records to obtain a sample of newborns

born between 1995-1999. The researcher reviewed hospital birth records to determine if a diagnosis of a birth defect was present. Using an ascertainment-adjusted analysis a prevalence rate of birth defects in the study sample was determined. This prevalence rate was then compared to the prevalence rate of birth defects recorded by the Birth Defects Registry for the same sample and cohort.

Phase 2, or the qualitative phase of the study focused primarily on the second study aim, and used, focus group interviews with Midwest GPs, midwives and CHNs.

7.1 Summary of Findings Phase 1 – Quantitative Phase

7.1.1 Data Related to Demographic Details obtained from the Random Sample of Hospital Case Notes.

Data were collected from a sample of 440 newborn birth case notes found within the maternal confinement records. Of the 440 records sampled, 13 were found to have record of a birth defect. Birth defects were more common in males than females, and the proportion with birth defects in non-Aboriginal infants was higher than in Aboriginal infants. This finding is consistent with the Birth Defect Registry report finding that birth defects were reported less frequently in Aboriginals compared with non-Aboriginal infants, and are generally more common in males (Bower et al., 2000).

7.1.2 Data Related to Additional Questions asked of the Sampled Information

Data related to the Additional Questions was ascertained from the hospital case notes of the 13 newborns diagnosed with birth defects. These data related to documentation, coding and transfer of infants.

Interestingly, eight of the 13 hospital cases were coded correctly and one case was coded incorrectly. The remaining five cases were not coded, although record of the birth defect was found documented within the case notes. Hospitals assign a code(s) for each inpatient admission. All hospital codes are then forwarded to the HDWA and used to justify and determine a hospital's current and future funding needs. This finding suggests that in almost half the cases, the hospital potentially failed to attract correct funding for treatment and care the newborn may have received as a result of the birth defect. Although this finding does not directly relate to either of the study aims, it does highlight a potential misrepresentation of the actual need for birth defect related care and resources at Geraldton Regional Hospital and the community it serves.

Several methods of documenting the presence of a birth defect in an infant's case notes were observed. All defects, regardless of their coding were recorded using at least one method of documentation. A possible reason for failing to code all birth defects occurring at Geraldton Regional Hospital, may be related to the apparent lack of formalised methods used to document birth defects and related conditions. Of particular note was the lack of documentation or failure to use the Neonatal History and Assessment Form and the Obstetric Discharge Summary Form. Inconsistent methods of documentation would cause difficultly and increase the likelihood of error and missed coding for staff.

In the study by Calle and Khoury (1991), error and missed coding associated with inconsistent methods of documentation were confirmed. The study suggested the under ascertainment of defects in discharge diagnosis should be considered in the development and operation of surveillance systems using this source of data (Calle & Khoury, 1991).

The WA Birth Defects Registry collects information regarding notifiers to the Registry of newly diagnosed cases of birth defects. Registry data indicates that health professionals and specialist departments of large Perth metropolitan and tertiary hospitals are consistently the best at reporting new and updated diagnosis of birth defects. This may be due to the fact that larger metropolitan and tertiary hospitals are equipped with the resources to manage and care for the special needs of newborns and children with birth defects. Consequently, these groups are exposed to, and receive referral of birth defects cases from smaller and rural hospitals routinely. This routine increases the likelihood of birth defects being reported to the Registry.

This finding concurs with that of Sekhobo and Druschel (2001) who found that accurate clinical recognition of birth defects depended upon the clinical acumen and interest of hospital staff, consequently the identification may vary by area and time. Hospitals providing higher levels of care and therefore, more accurate diagnosis, report cases more completely, and may be associated with higher birth defect rates. Conversely, areas with lower numbers of births, and minor variations in the number of birth defect cases may contribute to large variations in the incidence or prevalence rate (Sekhobo & Druschel, 2001). This is associated with lower rates of birth defect reporting.

The 13 cases of birth defects found in the sample consisted of seven cases that were referred to other hospitals or departments and six that were not. The six cases that were not referred included one case of stillbirth. Stillborn infants requiring post-mortem are transferred to Perth. Those infants diagnosed with births defects at post-mortem are reported by pathology departments to the Birth Defects Registry. In addition the Registry data are linked with the Health Department Hospital Morbidity and Mortality database. This database would also have a record of these newborns, assuming they had been coded correctly as having a birth defect in the database.

Most importantly, the six cases in which there was no record of referral to Perth hospitals or departments, correlates with the five cases that were not coded. This suggests that a direct correlation exists between referral to a Perth hospital or departments for treatment of a birth defect or related condition and being coded as having a birth defect. This in part may be associated with the completion of the documentation when referring or transferring infants to Perth hospitals.

As discussed in the literature review, there is a fundamental relationship between a surveillance system and its' data collection strategies (Iezzoni, 1997). Particularly important is the use of passive and active case finding methods. Iezzoni (1997) elaborates on the definition of passive case finding strategies previously discussed and describes passive case finding methods on two levels. The first level refers to data extraction from databases such as hospital mortality and morbidity and birth certification. Iezzoni (1997) refers to this method of case finding as impassive. Surveillance systems using impassive methods must accept the data as reported, since there is no opportunity for assessing the diagnostic specificity of the defect, or if it was appropriately coded. The second level of case finding Iezzoni (1997) refers to as passive. Passive case finding strategies usually require reporting sources to provide text descriptions regarding the manifestation of the birth defect, thus, facilitating a greater degree of data accuracy, coding and case follow-up (Kirby, 2000). The Birth Defects Registry notification cards request the birth defect diagnosis as opposed to a text description. However, as suggested by Sekhobo and Druschel (2001) the required clinical knowledge and awareness of staff regarding birth defects in the Midwest is not necessarily conducive to the recommended passive case finding strategies.

Alternatively, Sekhobo and Druschel (2001) stressed that statutory reporting of birth defects by health professionals was cost effective, and when combined with a

narrative diagnosis improved specificity and ongoing efforts to track and improve ascertainment completeness.

Finally, of the 13 sampled cases of birth defects, 11 were not diagnosed prior to birth. Of the two cases diagnosed prior to birth, one case was stillborn and transfer was not required. In the second case, the birth defect was known prior to birth and the infant was not transferred to a Perth hospital for delivery, (although it is considered standard practice to transfer an infant at risk of complication to a tertiary or specialist hospital prior to delivery) (Western Australian Neonatal Transport Service, 1992). The infant was diagnosed with a bilateral cleft lip and palate, using an ultrasound technique, prior to birth. The reason why this infant was not delivered in a Perth hospital could not be ascertained by the researcher.

7.1.3 Additional Data Collected from Parent Provided Information, Child Health Records and Ward Birth Register Information

A total of 3667 infants and children were sampled from the three data sources and 15 birth defects were found. The BDR was then checked for records of these defects. In all cases the Registry held records of the defects. Child health records were incomplete and could not be used as a comprehensive source from which to collect information. Similarly, recruitment of parents to provide information did not prove successful despite significant publicity efforts. Two parents provided the researcher with information regarding their children with birth defects, both of which were documented in BDR held records. The delivery ward birth register of Geraldton Regional Hospital facilitated access to a large sample size, although the researcher did not consider the information gained from this source to be either complete or reliable. Although completion of this register is hospital policy, the detail recorded by midwives was often incomplete or inconsistent. The presence of a birth defect in a

newborn is recorded in the "comments section" of each register entry and is therefore completed at the discretion of the attending midwife.

7.1.4 Data Related to Ascertainment-adjusted Analysis.

The 13 birth defect cases found in the sampled hospital case notes were compared with the Birth Defect Registry's list of records pertaining to the same sample population. Data matching was under taken by comparing the names from the hospital case notes with those in Birth Defect Registry records. Registry records were located for 12 of the 13 cases of birth defects from the study sample. The remaining case not known to the Registry was a child with a series of birth marks. Notification of birth marks, naevus and haemangioma are not routinely required by the Registry unless they are multiple in number or greater than $4cm^2$ (Bower et al., 2000). The researcher suggests that this case was not notified due to the apparent knowledge deficit regarding notification of defects amongst hospital staff and/or the lack of intervention or follow-up required in treating the defect.

Examination of the Birth Defect Registry records revealed an additional 2 cases of birth defects. These cases not known to the study consisted of one infant with hydrops fetalis and ear anomalies, and one infant with a vascular scalp malformation.

7.1.5 Discussion of Data Related to the Ascertainment Adjusted Analysis

Primarily, capture-recapture methodology aims to generate an estimate of the total population size. A estimate of the size of a population can be made by examining the degree of "overlap between incomplete lists of cases from existing sources" (Stephen, 1996, p263). The list of newborns with birth defects 'captured' or recorded by the Birth Defects Registry were compared to the list of newborns

'recaptured' independently during the quantitative phase of this study. The ascertainment adjusted analysis estimated the number of birth defects in the sampled population at 15.15. This suggested that the total number of birth defects in the sampled population should be 15. This value was confirmed when cases known and unknown to the study and the BDR were considered within the randomly sampled population. The study reported a total of 13 birth defects. Two additional birth defects were missed by the study, however, were known to the BDR. This equates to a total of 15 birth defects known to exist within the randomly sampled population. Similarly, the BDR reported a total of 14 birth defects. One additional birth defect was not recorded by the BDR but found by the study. This also equates to a total of 15 birth defects known to exist within the randomly sampled population. However, prior to making any inferences regarding this result the prevalence and significance of these values needs to be explored.

The Ascertainment Adjusted Prevalence Rate (AAPR) was calculated as 3.44% and the Birth Defect Registry Prevalence Rate was calculated as 3.18%. Both values are slightly lower than the average rate of 4.00% birth defects reported in the Midwest between, 1995 –1999 by the Birth Defects Registry (Bower et al., 2000). The later figure includes birth defects diagnosed up to six years of age, while the study was only able to include birth defects diagnosed at birth. However, the difference between AAPR and the BDRPR is small (0.26%) and not statistically significant.

Findings from the quantitative analysis enabled the researcher to address the following research questions:

1. Have birth defects occurred in the Midwest between the years 1995-1999 that have not been recorded by the Birth Defects Registry?

2. Is the lower incidence of birth defects in the Midwest due to case under reporting?

Based on the study findings, the degree of under-reporting of birth defects diagnosed at the time of birth in Geraldton Regional Hospital was small and unlikely to be of significance.

The researcher believes this study confirmed that a lower incidence of birth defects existed in the Midwest of Western Australia compared with the Metropolitan Region between, 1995 - 1999. This lower incidence is not due to the under reporting of birth defect cases to the BDR as previously proposed. However, as birth defect cases could only be ascertained from birth case notes, this statement is true for birth defects diagnosed at Geraldton Regional Hospital at the time of birth only. Births occurred at other hospitals within the Midwest and births will have been diagnosed after birth that may produce different results.

Ascertainment of birth defect cases from birth case notes was necessary to ensure the third assumption of the capture-recapture methodology was not violated. Capture-recapture methodology assumes that each individual has the same probability of being captured in the first and second sample (International Working Group for Disease Monitoring and Forecasting, 1995b). For example, children with severe defects requiring treatment in tertiary centres may have an increased likelihood of being recorded and, therefore, captured. In contrast, children affected by birth defects not requiring referral or extensive treatment may have a less chance of being recorded as such and, therefore, captured. In such a case children would not have the same probability of being included in a sample. It was, therefore, necessary to balance the probability of being 'caught' by limiting the target population to birth defects diagnosed antenatally or within the first week of life.

It is possible that under reporting of birth defects beyond the newborn period exists. To explore this issue, the number of birth defects diagnosed antenatally or in the first week of life in the Metropolitan region between 1995-1999 was ascertained from BDR records (n = 3729). Using the total number of births in the Metropolitan region during this timeframe the proportion of births diagnosed antenatally or in the first week of life is 4%. The 95% confidence intervals around AAPR and BDRPR both encompass 4%. Thus, the incidence of birth defects diagnosed at birth are not significantly different. Given the difference between the Midwest and Metropolitan for birth defects diagnosed up to six years of age, it suggests that this may be the result of reduced ascertainment beyond the newborn period.

7.1.6 Factors Contributing to the Lower Incidence of Defects in the Midwest of Western Australia

Full exploration and determination of the causes for the lower incidence of birth defects in the Midwest are vast and beyond the scope of this study. The following section will discuss some possibilities for the lower incidence of birth defects previously explored during the review of literature for this study.

Current Australian statistics reflect that maternal age contributes significantly to higher rates of birth defects (National Perinatal Statistics Unit, 2002b). The Australian National Perinatal Statistics Unit reports that for the period 1993 to 1996, the malformation rate in mothers aged 40 years and over was almost double that of birth rates in mothers aged 20-24 years of age (National Perinatal Statistics Unit, 2002b). Birth rates of mothers less than 30 years of age are higher in the Midwest and Other Rural Regions when compared with Metropolitan mothers. In contrast, significantly higher rates of births occurred in Metropolitan mothers in the age group of 30 years and above, than in mothers from the Midwest and Other Rural Regions.

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Therefore, the younger maternal age of mothers in the Midwest is likely to be a contributing factor in the lower incidence of birth defects in the Midwest compared with the Metropolitan Region.

BDR findings suggest that birth defects are higher in males, and reported less frequently in Aboriginal newborns (Bower et al., 2000). Current ABS statistics indicated that Midwest and Other Rural Western Australian Regions have a significantly higher percentage of Aboriginal births compared with the Metropolitan Region (Bower, 2002). The proportionately higher percentage of Aboriginal births in the Midwest region may, in part, be responsible for the lower incidence of birth defects in the Midwest, since, as suggested by the BDR birth defects are reported less frequently in Aboriginal infants than non-Aboriginal infants. The study's data provides additional support to this idea, with the rate in Aboriginal newborns being lower than non-Aboriginal newborns.

7.2 Summary of Findings Phase 2 – Qualitative Phase

Three focus groups interviews were undertaken, consisting of GPs, midwives and CHNs. Prior to discussing the results of the focus interview data the components of a focus group and their application to the study will be discussed.

7.2.1 Appling the Components of the Focus Group Interview

7.2.1.1 Problem Formation and Interview Guide

This study initially proposed qualitative data collection via individual taped interviews with Midwest case reporting source. Changes to this methodology resulted in the formation of the study's research and interview questions without focus group interviews as the mode of data collection. It is, therefore, important to explore this issue and consider the impact that using focus group interviews had on the study data.

Dilorio et al (1994) recommends that prior to conducting a focus group interview, individuals involved in the implementation, analysis or interpretation of the interview data, be involved in formulation discussions. This initial process was undertaken whilst considering development of interview and research questions designed for individual interviews. In conjunction with the research supervisors, the research objectives were considered and the questions that were considered best able to achieve these objectives were formulated. In addition, the nature of the interview questions, time constraints of participants in attending such interviews, as well as the setting and opportunity for individuals to respond freely to questions were considered.

Prior to conducting a focus group interview Dilorio et al. (1994) further recommends construction of an interview guide, consisting of an introductory section, six to twelve interview questions and a conclusion. This process was adhered to by the researcher, ensuring that the study aims, purpose of the interviews and use of interview data were clearly outlined before interviews were commenced. The open-ended nature of the interview questions lent itself well to a group interview setting, serving as a guide to discussion and allowing the researcher to direct and maintain the interview focus to ensure all the research questions were addressed.

Additionally, Dilorio et al (1994) suggests that the first interview questions should be related to the topic but be unstructured and aimed at generating discussion and facilitating rapport amongst group members and the researcher. This was not a specific aim of the researcher, given that the focus group consisted of health professionals who were familiar with discussing work related activities as a group. In addition, none of the interview questions were of a personal nature, therefore, pre-emptive introduction or comforting aimed at encouraging the open sharing of such ideas was not required.

Finally, Dilorio et al. (1994) recommends final interview questions be broad and aimed at summarising ideas. Conclusion of the study's focus group interviews were consistent with these recommendations, and were designed to encourage group members to consider previous responses and offer overall suggestions for improving current processes. Overall, these steps met the recommendations for the conduct of a focus group interview outlined by Dilorio et al (1994).

7.2.1.2 Participants and Setting

In designing a focus group interview Dilorio et al. (1994) recommends that the appropriate choice of interviewees and a balance of homogeneity will maximise the likelihood of obtaining desired research outcomes. Participation in the study's

focus group interviews was offered to all members of the GP, midwife and CHN communities. Only individuals on a rostered shift at the time focus group interviews were conducted chose to attend. The volume and range of respondent feedback may have been greater if more individuals had attended the interviews, however, due to the homogeneity of the group the researcher believes this to be unlikely.

In addition, Dilorio et al. (1994) refers to the importance of the conducting interviews in familiar, easily accessible settings, that will promote group comfort and enhance interaction. The study achieved this by conducting interviews at the respondent's place of work, which was not only familiar, but appropriate to the research aims. This setting also ensured group homogeneity and a certain level of interview attendance that may have not been achieved if conducted off site and outside of usual working hours. Considering the lack of participant response to attending individual interviews, this seemed even more relevant. However, conducting interviews in the workplace could be associated with respondent distraction as their time was not their own and they had work commitments that would soon need to be attended. It also meant that the researcher was unable to ensure an optional setting, such as size of the room, lighting, temperature and seating arrangements were not negotiable. Nevertheless, respondents appeared comfortable, relaxed and open to discussion. The researcher believes that the aims of qualitative data collection of this study were met despite changes to the study's initial methodology.

7.2.2 Conclusions of Thematic Analysis

Overall, the majority of infants with major birth defects are referred to Perth tertiary hospitals for delivery or shortly after birth. Interview respondents and review of data collected from hospital case notes supported this statement. Several factors arising from focus group interviews may explain this trend. First, birth defect cases

are transferred to Perth tertiary hospitals as a matter of standard practice. This seems relevant as Perth tertiary hospitals are better equipped with staff and structural resources for treating, care and diagnosis of newborns/children affected with birth defects. Second, at the time of the study there was no resident paediatrician in the Geraldton region, therefore, first line and ongoing specialist care for newborns/children affected by birth defects and related conditions, was not available. These factors have resulted in lowering the exposure of Midwest GPs, Midwives and CHNs to birth defects and related conditions. As such, a knowledge deficit regarding the role of the Birth Defects Registry and notification procedures exists. This has been compounded by the voluntary nature of the birth defect reporting process, which has also contributed to failure of any one health professional group to assume responsibility for case notification to the Registry. In addition, the knowledge deficit is enhanced by the lack of available ongoing education and health promotion.

Nevertheless, quantitative data suggests the lower incidence of birth defects in the Midwest is not due to case under reporting at the time of birth. In addition, review of the study's conceptual framework, in light of the empirical evidence, indicates that current methods of ascertaining birth defects at the time of birth are adequate. Therefore, current reporting practices appear to be effective in attaining notification of birth defects at the time of birth in the Midwest. This result, however, maybe associated with the high transfer rate of birth defect cases to Perth tertiary hospitals. Due to high transfer rates, a significant proportion of the Midwest cases of birth defects are treated in Perth tertiary hospitals. These hospitals represent the original and only source of notification for many cases of birth defects (Bower, 2001). In the study cohort 124 (61%) birth defect cases from a total number of 213 were notified by Midwest sources. Therefore, notification of 79 birth defect cases originated exclusively from Perth tertiary hospitals. The 124 cases notified by

Midwest sources included overlapping notification of the same case from any number of Midwest notifiers. Consequently, the 124 instances of notification does not necessary correlate with the total number of birth defects and is potentially an over representation of cases notified by Midwest sources.

Additionally, of the 124 notified cases 67 (54%) were directly notified from Midwest practitioners, that is, GPs, midwives and CHNs. Notification of the remaining 57 (46%) of notifications were derived from linkage to other databases, such as hospital mortality and morbidity data, and the visiting rural paediatric service. Data presented suggest that notification of birth defects diagnosed at the time of birth from Midwest sources alone is not efficient, and has implications for notifications beyond the newborn period.

7.3 Comparing the Conceptual Framework with Empirical Evidence

The aim of this study was to determine if the observed lower incidence of birth defects in the Midwest of Western Australia is due to case under reporting. Research findings indicate that the lower incidence of birth defects in the Midwest is not due to case under reporting at the time of birth. The process of determining this aim required the researcher to review the method and effectiveness of BDR case ascertainment sources. Birth defect case ascertainment sources were considered as variables within the conceptual framework of this study.

The conceptual framework guiding this study was developed from the literature review. Research by Lynberg and Edmonds (1992b) suggested the ideal birth defects surveillance system is determined by a comprehensive case ascertainment system that provides timely population based information. Multiple sources of case ascertainment using active and passive case finding methods provide

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the best potential for complete case finding and minimizing under reporting (Lynberg & Edmonds, 1992a).

The WA BDR predominately uses passive forms of case ascertainment. Active case ascertainment occurs on a regular basis in metropolitan hospitals, but is very infrequent in rural hospitals where it is usually circumstantial and associated with a staff visit for another reason (Bower, 2002).

Quantitative data from this study suggest, that the lower incidence of birth defects in the Midwest is not due to the under reporting of cases at birth. Therefore, despite the BDR predominant use of passive case reporting in rural areas, the flow of information from all variables in the conceptual framework to the BDR appears to be sufficient. However, as highlighted by qualitative data this result may be associated with the high transfer rate of birth defect cases to Perth tertiary hospitals. Due to high transfer rates of compromised infants, a significant proportion of the Midwest cases of birth defects are treated in Perth tertiary hospitals. These hospitals represent the original and only source of notification for many cases (Bower, 2001).

The WA BDR system of case ascertainment uses multiple statutory and voluntary sources to ascertain cases of birth defects using both passive and active methods. However, the flow of information from each of the sources to the BDR is not balanced. Research data indicates that the majority of case notifications are originating from statutory sources such as Midwives Birth Notification Data and Hospital Mortality and Morbidity Data. All Midwest case notification sources other than Perth Tertiary Hospitals, were found to report cases of birth defects to the BDR significantly less. Despite the completeness of BDR data, the potential flow of case reports to the BDR (Figure 4) is not balanced, with passive voluntary sources of case reporting displaying a significant deficit. Therefore, application of the study's conceptual framework to describe the flow of Midwest case reporting to the BDR,

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indicates a heavy reliance on statutory notification and reports from metropolitan sources. Coupled with minimal active case ascertainment of Midwest birth defects by the BDR, the Midwest system is reliant upon passive forms of ascertainment. Edmonds (1997) outlined the strengths of passive voluntary case reporting as low cost, with the ability to cover large populations, and weaknesses including slow reporting, lack of control over the quality of data and under reporting of cases. This lack of control over data quality and completeness may contribute to the lower incidence of reported birth defects in the Midwest.

In addition, study findings indicate that the flow of data to the BDR fluctuates over time. The study found that the rate of birth defects diagnosed in Midwest and Metropolitan newborns was not significantly different, however, the Midwest birth defect rate over the five-year study period was lower than the Metropolitan birth defect rate. The study data only reflects the rate of birth defects diagnosed at birth, suggesting the possibility of under reporting of Midwest birth defect cases diagnosed after birth.

7.4 Limitations and Strengths

7.4.1 Limitations

The primary limitation of the study relates to the sample used. In an effort to ascertain a random sample that was representative of the total population, the researcher used hospital birth case notes. Using the case notes of neonates born in Geraldton Regional Hospital was the only available method of randomising and accessing a guaranteed comparable sample population size. However, the BDR statistics are based upon birth defect cases recorded on newborns and children up to six years of age. Therefore, cases diagnosed after the initial neonatal period were not captured by the study sample. To overcome this limitation alternative avenues for accessing cases were explored, such, as reviewing hospital case notes on various types of paediatric surgery performed on birth defect related conditions. This proved to be associated with further error and bias, due to the type of surgery performed, the way a surgical procedure was coded and the likelihood that most of these cases would have been treated at a Perth tertiary hospital.

In order to access the range of age groups included within the study cohort, the researcher considered the use of the five-year old health checks performed on children by CHNs when commencing school. This also presented inconsistent data, as there was no guarantee that all children born in the Midwest were still residing in the region at school commencement. In addition, the completeness, access and filing of five-year old health assessment records varied amongst CHNs, and hence access to a sample population that was sufficiently large and representative was not available.

Furthermore, the researcher initially proposed accessing GP records to capture newborns and children diagnosed following the initial birth related admission. Changes to privacy legislation in Western Australia after the commencement of the study made access to GP records unachievable within the scope of the study. The researcher's attempt to access this information in the form of parent provided information was also unsuccessful. Thus the conclusions reached by the study are limited to birth defects diagnosed in the newborn period only.

In addition, conducting focus group rather than individual interviews may have contributed to limiting data obtained in the qualitative phase of the study. However, the researcher believes that in view of respondents' ambivalence to participate, individual interviews would not have been less productive and generated data of questionable significance.

7.4.2 Strengths

Study limitations were balanced by a number of strengths. First, the study was based upon well referenced, sound methodological principles.

Second, the study aims were founded from statistical evidence generated by the BDR, a credible Western Australian database. This gives further credence to value and application of study outcomes.

Third, the use of a combined quantitative and qualitative approach enabled the researcher to further explore in depth the study's major findings, and establish the strength of quantitative outcomes.

CHAPTER 8

CONCLUSIONS, IMPLICATIONS AND RECOMMENDATIONS

This study revealed findings of both theoretical and practical significance. The study was comprehensive in its collection and interpretation of quantitative and qualitative data and clearly outlined the practical implications and recommendations resulting from its findings. The research consisted of two phases that addressed the research questions reflecting the overall aim of the study. Phase 1 (quantitative phase) used capture-recapture methodology. The study methodology met the required criteria for undertaking a two-source capture-recapture study. The study aim was to determine if the observed lower incidence of birth defects in the Midwest is due to case under reporting. Study findings revealed that the lower incidence of birth defects in the Midwest was not due to the under reporting of birth defects diagnosed at birth, between 1995 - 1999. Determining the possible factors contributing to the lower incidence of birth defects in the Midwest requires further investigation that was outside the scope of this study. This research did, however, suggest factors such as lower maternal age, study bias, underascertainment of birth defects beyond the newborn period and the large Aboriginal population in the Midwest as factors possibly contributing to the lower incidence of birth defects in the Midwest.

Phase 2 (qualitative phase) used focus group interviews to explore the views of case reporting sources regarding the BDR. Phase 2 aimed to identify and make recommendations that could sustain or improve upon current birth defect case ascertainment methods. Analysis of focus group interview data identified several themes. Themes included knowledge deficit, high transfer rates of compromised

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neonates, need for ongoing education and promotion, uncertainty regarding ownership and the need for centralisation of notification cards. Recommendations addressing these themes suggested changes to current methods of birth defect case reporting and improvements in education and promotion of birth defects to case reporting sources, particularly in rural areas.

In addition, comparison of the conceptual framework with the empirical evidence revealed an imbalance related to case notification sources and the number of notifications that are made. The study found that birth defect notifications regarding infants and children in the Midwest between 1995 – 1999 were ascertained primarily from statutory notification sources and metropolitan tertiary hospitals. Overall, the completeness of voluntary sources of Midwest birth defect notification was found to be inconsistent and, therefore, unreliable.

In conclusion, this study revealed that only a small number of birth defects diagnosed at birth occurred in the Midwest between the years 1995-1999 had not been recorded by the Birth Defects Registry hence, the lower incidence of birth defects in the Midwest was not found to be due to the under reporting of cases at birth. Several recommendations that could potentially change the notification of birth defects to the BDR and improve the collaborative reporting from all case reporting sources were derived from the qualitative data. In considering the suggestions of focus group respondents and thematic analysis of the interview data, the researcher addressed the following research question:

1. What recommendations can be made to sustain or improve upon the current case ascertainment methods of the Birth Defects Registry?

The following recommendations have been developed in accordance with themes identified in the qualitative analysis. Recommendations are as follows:

- Knowledge Deficit/Ongoing education and promotion: annual educational visits to the Midwest by Registry staff to promote the role of the Birth Defects Registry, and to reinforce the importance and process of birth defect reporting. This should include a review of what birth defects require notification and how to do this.

- Deficit/ownership: educational visits should include reinforcement regarding the responsibility of notifying cases of birth defects to the Registry. Promoting the value in overlapping notifications in ensuring new cases or newly diagnosed conditions in old cases are registered.

- Centralising Notification Cards/Knowledge deficit: incorporating notification cards into a form of documentation that is frequently used and easily accessed would ensure that cards would not be misplaced and may act as a reminder to notify birth defects. Child health books are received by each mother following birth of a baby, and are used as a record of that child's development, immunisation and health status. Adding notification cards to child health books was a popular suggestion for centralising notification cards proposed by all focus groups. However, CHNs and midwives did comment on the current under utilisation of child health books by Midwest GPs. Therefore, perhaps promoting the general use of such books should also be considered by the Western Australian Department of Health.

It is further recommended that notification of birth defects to the BDR become a legislated statutory requirement for all case reporting sources. In addition, notification cards should be amended to include instructions that direct notifying sources with regarding to the forwarding or collection of completed cards.

Overall, study findings add valuable knowledge to the epidemiological aspects of birth defect surveillance in WA. In particular, the uniqueness of the Midwest population and its differences with the metropolitan population has been highlighted.

8.1 Summary of Recommendations

8.1.1 The Practice of Case Notification

1. Educational visits to the Midwest by the BDR staff to improve the knowledge of health professional regarding birth defects, the purpose of the Registry and the requirements of case notification.

2. Incorporation of birth defect notification cards into Child Health Books as a means of centralising birth defect notification.

3. Amendment of birth defect notification cards to include the BDR forwarding address and when and how to forward notification cards.

4. Legislating the statutory reporting of birth defects by all case reporting sources.

5. Consideration of the possibility of a web based birth defect notification system.

8.1.2 Future Research

1. Determine if under reporting of birth defects in the Midwest exists beyond the newborn period.

2. Explore factors that may contribute to a lower incidence of birth defects in the Midwest.

8.1.3 Education

1. Visits by BDR staff to case reporting sources throughout Western Australia with the aim of promoting the BDR and, improving knowledge regarding birth defects, the importance of case notification, a health professionals responsibility to notify birth defect cases.

2. In collaboration with the addition of birth defect notification cards to Child Health Books, case reporting sources should be re-educated regarding the function and importance of completing Child Health Book Information.

3. Geraldton Regional Hospital to review the use of their Neonatal Assessment Forms as these are under utilised.

REFERENCES

Anderson, L. E. (1990). <u>Mosby's dictionary of medical, nursing and allied health</u> (3rd ed.). St Louis: The C.V. Mosby Company.

Armitage, P. (1971). <u>Statistical methods in medical research.</u> Oxford: Blackwell Scientific Publications.

Birth Defects Monitoring Program. (2002, August, 2002). <u>California birth defects monitoring program: preventing birth defects through research.</u> Retrieved, November, 2002, from the World Wide Web: www.cbdmp.org/pbd

Bower, C. (2001). Unpublished data from the Birth Defects Registry. In D. Baker (Ed.). Perth: Unpublished.

Bower, C. (2002). Unpublished data from the Birth Defects Registry. In D. Baker (Ed.). Perth: Unpublished.

Bower, C., Rudy, E., Ryan, A., & Cosgrove, P. (2000). <u>Report of the Birth Defects Registry of Western Australia, 1980 - 1999.</u> Perth: King Edward Memorial Hospital and Centre for Womens Health.

Bower, C., Ryan, A., & Rudy, E. (2001). Ascertainment of pregnancies terminated because of birth defects: effect on completeness of adding a new source of data. <u>Teratology, 63,</u> 23 - 25.

Bower, C., & Stanley, F. J. (1983). Western Australian Congenital Malformations Register. <u>Medical Journal of Australia, 2,</u> 189 - 191.

Brenner, H. (1994). Application of capture-recapture methods for disease monitoring: potential effects of imperfect record linkage. <u>Methods of Information in Medicine, 33(5),</u> 502 - 506.

Brenner, H., Stegmaier, C., & Ziegler, H. (1994). Estimating the completeness of cancer registration in Saarland/Germany with capture-recapture methods. <u>European Journal of Cancer, 30A(11),</u> 1659 - 1663.

Bromberg Bar-Yam, N. (1999). Birth defects surveillance registries in United States and Europe. <u>International Journal of Childbirth Education, March,</u> 1 - 7.

Burns, N., & Groves, S. K. (1997). Qualitative research methodology, the practice of nursing research: conduct, critique and utilization (3rd Ed.). New York: W.B. Saunders and Co.

Calle, E. E., & Khoury, M. J. (1991). Completeness of the discharge diagnosis as a measure of birth defects recorded in the hospital birth record. American Journal of Epidemiology, 134(1), 69 - 76.

Centre for Disease Control and Prevention. (1996). EPIINFO (Version 6.04a). Atlanta.

Cordero, J. F. (1992). Registries of birth defects and genetic diseases. Paediatric Clinics North America, 39, 65 - 77.

Dilorio, C., Hockenberry-Eaton, M., Maibach, E., & Rivero, T. (1994). Focus groups: and interview method for nursing research. Journal of Neuroscience in Nursing, 26(3), 175 - 180.

Edmonds, L. D. (1997). Birth defect surveillance at the state and local level. Teratology, 56(1/2), 5 - 7.

Egeland, G. M., Perham, K. A., & Hook, E. B. (1995). Use of capture-recapture analysis in fetal alcohol syndrome surveillance in Alaska. American Journal of Epidemiology, 141(4), 335 - 341.

European Register of Congenital Abnormalities and Twins. (2001). EUROCAT. Retrieved, 2002, from the World Wide Web: www. lshtm.ac.uk/php/eeu/eurocat

Federal Privacy Commissioner. (2001). Privacy Act: Office of the Federal Privacy Commissioner. Paper presented at the National Privacy Principals.

Fienberg, S. E. (1972). The multiple recapture census for closed populations incomplete 2K contingency tables. Biometrika, 59, 591 - 603.

Geraldton Health Service. (1992, Reviewed 4/1996). Nursing procedure manual: congenital malformations.

Hanson, J. W. (1995). Birth defects surveillance and the future of public health. Public Health Reports, 110(6), 698 - 700.

Hexter, A. C., & Harris, J. A. (1991). Bias in congenital malformation information from the birth certificate. Tetatology, 44, 177 - 180.

Holtzman, N. A., & Khoury, M. J. (1986). Monitoring for congenital malformations. Annual Review of Public Health, 7, 237 - 266.

Honein, M. A., & Paulozzi, L. J. (1999). Birth defects surveillance: assessing the "gold standard". American Journal of Public Health, 89(8), 1238 - 1240.

Hook, E. B., & Regal, R. R. (1995). Capture-recapture methods in epidemiology: methods and limitations. Epidemiological Review, 17(6), 243 - 264.

Iezzoni, L. I. (1997). Assessing quality using administrative data. Annual of Intern Medicine, 127, 666 - 674.

International Clearinghouse for Birth Defect Monitoring Systems. (2001, August 7th, 2001). Worldwide Members (Web Site). Retrieved, 2001, from the World Wide Web: www.icbd.org/who

International Working Group for Disease Monitoring and Forecasting. (1995a). Capture-Recapture and multiple record estimation II: applications in human disease monitoring. American Journal of Epidemiology, 142(10), 1059 - 1069.

International Working Group for Disease Monitoring and Forecasting. (1995b). Capture-recapture and multiple-record systems estimation I: history and theoretical development. American Journal of Epidemiology, 142(10), 1047 - 1058.

Kirby, R. S. (2000). Analytical resources for assessment of clinical genetics services in public health: current status and future prospects. Tetatology, 61, 9 -16.

Lechat, M. F., & Dolk, H. (1993). Registries of congenital abnormalities: EUROCAT. Environmental Health Perspective Supplements, 101(Suppl 2), 153 - 157.

LePorte, R. E., Dearwater, S. R., Chang, Y.-F., Songer, T. J., Aaron, D. J., Anderson, R. L., & Olsen, T. (1995). Efficiency and accuracy of disease monitoring systems: application of capture-recapture methods to injury monitoring. American Journal of Epidemiology, 142(10), 1069 - 1077.

Lynberg, M. C., & Edmonds, L. D. (1992a). Public Health Surveillance. In R. Monson (Ed.), Surveillance of Birth Defects (pp. 157 - 171). New York: Wiley and Sons.

Lynberg, M. C., & Edmonds, L. D. (1992b). Surveillance of birth defects. In R. R. Monson (Ed.), Public Health Surveillance (Vol. Chapt. 12, pp. 157 - 177): Van Nostrand Reinhold.

Lynberg, M. C., & Edmonds, L. D. (1993). State use of birth defects surveillance. From data to action: CDC's public health surveillance for women, infants and children, 217-229.

Metropolitan Atlanta Congenital Defects Program. (2002, August 2002). Metropolitan Atlanta congenital defects program. Retrieved, 2002, from the World Wide Web: www.cdc.gov/ncbddd/bd/documents/MACDP2002

Miller, L. A., & Kirby, R. S. (2000). Neural tube defects surveillance: a national survey. Teratology, 61, 28 - 32.

National Birth Defects Prevention Network. (2002). National birth defects prevention network. Retrieved, 2001, from the World Wide Web: www.nbdpn.org/NBDPN/who.html

National Perinatal Statistics Unit. (2002a). Congenital Malformations, Australia, 1995 and 1996 [Electronic report]. Australian Institute of Health and Wefare. Retrieved 15/10/02, 2002, from the World Wide Web: www.npsu.unsw.edu.au

National Perinatal Statistics Unit. (2002b). National perinatal statistics unit - perinatal data systems. Australian Institute of Health and Welfare. Retrieved, 2002, from the World Wide Web: www.nspu.unsw.edu.au

Nyamathi, A., & Shuler, P. (1990). A research technique for informed nursing practice. Journal of Advanced Nursing, 15, 1281 - 1288.

Olsen, C. L., Polan, A. K., & Cross, P. K. (1996). Case ascertainment for state-based birth defects registries: characteristic of unreported infants ascertained through birth certificates and their impact on registry statistics in the New York State. Paediatric and Perinatal Epidemiology, 10, 161 - 174.

Roberts, I., & Scragg, R. (1994). Application of capture-recapture methodology to estimate the completeness of child injury surveillance. Journal of Paediatric Child Health, 30(6), 513 - 514.

Robles, S. C., Marrett, L. D., Clarke, E. A., & Risch, H. A. (1988). An application of capture-recapture methods to the estimation of completeness of cancer registration. Journal of Clinical Epidemiology, 41(5), 495 - 501.

Schnabel, Z. E. (1938). The estimation of the total fish population of a lake. Amateur Math Monthly, 45, 348 - 352.

Sekhobo, J. P., & Druschel, C. M. (2001). An evaluation of congenital malformations surveillance in New York State: and application of centres of disease control and prevention (CDC) guidelines for evaluating surveillance systems. Public Health Reports, 116(4), 296 - 305.

Shott, S. (1990). Statistics for health professionals. Philadelphia: W.B. Saunders Company.

Silcocks, P. B. S., Thornton-Jones, H., & Skeet, R. G. (1989). Can we achieve 100% ascertainment in cancer registration. Public Health, 103, 23 - 30.

Stanley, F. J., Croft, M. L., Gibbins, J., & Read, A. W. (1994). A population database for maternal and child health research in Western Australia using record linkage. Paediatric and Perinatal Epidemiology, 8, 433-447.

Stephen, C. (1996). Capture-recapture methods in epidemiological studies. Infection Control and Hospital Epidemiology, 17, 262 - 266.

Western Australian Neonatal Transport Service. (1992). Stabilisation and transport of newborn infants and at risk pregnancies (1st ed.). Perth, Western Australia: Western Australian Neonatal Transport Association.

Appendix 1: Birth Defects Registry Exclusion List

Accessory nipples

Laryngeal stridor

Balanced translocation

Laryngomalacia

(in normal individual without malformation(s)

Blocked tear duct

Low birth weight

Birth mark, naevus, haemangioma

(unless multiple or >4cm – give size)

Meconium ileus

Broncho-pulmonary dysplasia

Mental retardation

Clicky hips

Metatarsus adductus (unless splinted)

Congenital infection (if no birth defects)

Mongolian blue spot

Congenital pneumonia

Motor impairment

Cerebral palsy

Oesophageal reflux

Delayed milestones

Paraoxysmal atrial tachycardia

Deviated nasal septum

Patent foramen ovale

Ear abnormalities – minor

Persistent fetal circulation

Epigastric hernia

Perthe's disease

Epilepsy

Pilonidal sinus

Failure to thrive

Posit/postural foot deformity

Galactosaemia – duarte variant

Sacral dimple

Hiatus hernia

Sacral sinus (unrelated to occult spinal dysraphism)

Hydatid of morgagni

Single palmar crease

Hydrocele testis

Single umbilical artery

Hydrops fetalis – immune (include if non-immune hydrops)

Skin tag

Hypoglycaemia

Strabismus

Imperforate hymen

Thalassemia minor

Inguinal hernia

Toe anomalies - minor

Interuterine growth retardation

Tongue tie

Intussusception

Trigger finger/thumb (unless treated)

Labial adhesion or fusion

Umbilical hernia

Large fontanelles

Undescended testis (unless treated)

Webbing of 2nd & 3rd toes

Wide suture lines

Appendix 2: Birth Defect Registry Notification Card

KING EDWARD MEMORIAL HOSPITAL **BIRTH DEFECTS REGISTRY NOTIFICATION CARD**
Bagot Road, Subiaco 6008
9340 2735   Confidential

SURNAME:
FORENAMES:
ADDRESS:
                                        POSTCODE:

Mother's
Surname
Mother's
Forenames
Maiden
Name
Father's
Surname
Father's
Forenames

DATE OF BIRTH: UNIT RECORD No
SEX: M [ ]   F [ ]   LIVEBIRTH [ ]   STILLBIRTH [ ]   OTHER [ ]
RACE: ABORIGINAL [ ]   NON ABORIGINAL [ ]   PLURALITY   SINGLE BIRTH [ ]   MULTIPLE BIRTH [ ]
HOSPITAL OF BIRTH: GESTATION [ ]   BIRTH WEIGHT
IF DECEASED, DATE OF BIRTH   POSTMORTEM EXAMINATION YES [ ] NO [ ]   IF YES, WHERE
PLEASE LIST ALL MALFORMATIONS

BIRTH DEFECTS	AGE AT DIAGNOSIS	OFFICE USE ONLY					
1.							
2.							
3.							
4.							
5.							
6.							
7.							
8.							
9.							
10.							

DOCTOR(S) IN CHARGE OF CARE
NOTIFIED BY   ADDRESS
DATE
KE1/8   P.T.O

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Appendix 3: Study Introduction Letter

Source Address

Mrs Donna Baker (Masters of Midwifery – Student)

Edith Cowan University

Pearson Street Churchlands WA 6018

email: █████████████

Date

To whom this may concern,

My name is Donna Baker and I'm currently studying a Master of Science (Midwifery) degree by research at Edith Cowan University. I am working in conjunction with Dr Carol Bower, Principal Research Fellow, Division of Population Sciences, TVW Telethon Institute for Child Health Research. My research involves investigating an increased trend in the discrepancy between rates of birth defects in the metropolitan and rural areas. The highest proportions of birth defects are evident in the metropolitan regions of Perth, with significantly lower percentages evident in rural areas. Please see attached graph taken from the latest report of the Birth Defects Registry of Western Australia, for representation of these discrepancies.

Support for fieldwork and data collection has been kindly offered by the Combined Universities Centre for Rural Health in Geraldton. Therefore I would like use the Geraldton population to investigate these discrepancies and am writing to request your vital assistance in data collection.

I would like to request access to client records and information that would identify those that have been reported on and/or treated for a notifiable birth defect between the years 1996 – 2000. For accuracy in determining distribution, cases will include only those infants born in the Geraldton region or whose mother's postcode of residence was the Geraldton region at the time of their birth.

Any assistance you can offer in this matter would be greatly appreciated, with any established causality and recommendations ultimately benefiting the infants and health care providers in the Western Australian community. I would like to meet with all parties prior to data collection to introduce myself and discuss a methodology for data collection that would cause minimal disruption to your practices, work schedules and regular duties.

The conduct of this research project is within the parameters of Edith Cowan University policies and procedures and under direct supervision of my supervisors;

Carol Thorogood

Co-ordinator Midwifery Programs

School of Nursing and Public Health

Faculty of Communications, Health and Science

Edith Cowan University

ph: (08) 9273 8623

email: c.thorogood@ecu.edu.au

Dr Leanne Monterosso

Post Doctoral Fellow

School of Nursing and Public Health

Edith Cowan University

ph: (08) 9273 8621

email: l.monterosso@ecu.edu.au

Dr Carol Bower

Principal Research Fellow

Division of Population Sciences

TVW Telethon Institute for Child Health

ph: (08) 94897751

email: caroline.bower@health.wa.gov.au

Application for ethical clearance will be made via Edith Cowan University Faculty Research and Higher Degrees Committee and the Health Department. This is with complete assurance that the methodology used will protect the confidentiality and privacy of clients, families and practitioners.

Once again please let me stress the vital importance of your collaboration in the process of data collection and my willingness to be completely flexible to ensure minimal disruption to your schedules.

I plan to visit Geraldton during the first week in April to establish means for data collection and would gratefully welcome your assistance in this pursuit. Should you require any further information regarding this project please do not hesitate to contact either of the above supervisors.

Yours faithfully

Donna Baker.

Appendix 4: Study Information Sheet

Clarifying the lower incidence of Birth defects in the Midwest Region.

Information Sheet for Clinicians

Current data indicates a lower incidence of birth defects in rural areas of Western Australia in comparison to the Metropolitan area. Of particular interest are the Southwest and Midwest regions, with the lowest rates of birth defects. This finding is based upon information collected by the Birth Defects Registry of Western Australia. Anecdotal evidence exists to suggest that this information is inaccurate. The accurate collection of data is vital to ensure the continuation of a high quality of population based information, so that precise investigation and evaluation of rural concerns and needs occurs. Data collected by the Birth Defects Registry.is used to:

- establish the prevalence of birth defects in Western Australia;

- generate research into the causes and prevention of birth defects;

- investigate changes in the frequency of birth defects;

- provide health professionals with statistical information;

- evaluate screening programs, prevention and treatment strategies of birth defects, to assist with planning of health care facilities; and

- to increase general knowledge about birth defects.

What is the aim of this study?

Anecdotal evidence suggests that under ascertainment maybe responsible for the observed lower incidence of birth defects in the Midwest and Southwest regions of Western Australia. This issue warrants further investigation. However, due to limitations in the possible scope of this research project, in addition to research support received, this study will be limited to the Midwest. The aim of this study is to determine if the observed lower incidence of birth defects in the Midwest is due to under ascertainment of birth defect cases. To achieve this goal this study will combine interview data, to ascertain clinicians' opinions with statistical data, in order to establish the numbers of existing birth defects in the Midwest.

Who is doing this study?

The study is a Masters thesis initiated by the Birth Defects Registry and being conducted by:

Mrs Donna Baker (Masters Student)

Edith Cowan University

Pearson Street Churchlands WA 6018

email: ███████████ ▪

Study supervision and direction is being provided by the Edith Cowan University and Dr Bower of the Birth Defects Registry of Western Australia. The study supervisors are Dr Carol Thorogood (Co-ordinator of Midwifery Programs at Edith Cowan University), Dr Carol Bower (Clinical Associate Professor at the Birth Defects Registry) and Dr Leanne Monterosso (Post Doctoral Fellow, School of Nursing and Public Health at Edith Cowan University).

What will be expected of you during the study?

Part 1

All health professionals involved in the care of infants and children with potential birth defects will be asked if they are willing to participate in the study. If you agree you are asked to take part in an interview, taking approximately 20 minutes to determine your attitudes toward current reporting practices of birth defects. The researcher will take notes during the interview for later referral and analysis.

Part 2

To determine the accuracy of data held by the Birth Defects Registry the researcher is required to collate the number of birth defects occurring in Midwest infants and children between 1995 – 1999, independent of information held by the Birth Defects Registry. To do this case notes of infants and children born to mothers that resided in the Midwest during 1995 – 1999 will be sampled. Case notes will be accessed from records held by the Geraldton Region Hospital and St. John of God Geraldton, proportionately to the number of births during the sample period. Case note information will be used to identify the existence of reportable birth defects. The researcher and a research assistant, Edwina Rudy, who is employed by the Birth Defects Registry and very familiar with this type of study will be responsible for collecting the case note information. This will take place after prior negotiation and ethics clearance with the hospital administrations. Data collected in parts one and two of the study will be later analysed and used to make recommendations that will maintain or improve upon the current reporting practices of birth defects

Please be aware should you wish only to be involved in one part of this study, that is, only in the interview or case note provision, your participation is still valued and encouraged.

How will your privacy be protected?

The researcher, research assistant and supervisors will be the only people to have access to client data and interview information. Following participation in an interview your name will be replaced by a numerical code. Client details will also be replaced by a code following comparison of client data with information held by the Birth Defects Registry. All interviews and case note reviews will be conducted in private. No name identifying data will be used in any reports or publications written as a result of this study and your participation will remain anonymous. All coded surveys and records will be kept in a locked filing cabinet located in the Principal Supervisor's office (Edith Cowan University). These records will be destroyed after five years. No one will have access to this documentation.

Voluntary participation and your right to refuse?

Participation in this study is completely voluntary. You are free to ask questions at anytime and have a right to have them answered to your satisfaction. You are free to withdraw consent to continue in the study at anytime, and without penalty, simply by contacting the principal researcher or an above named supervisor.

Are there risks involved in this study?

There are no known risks to you or your clients if you take part in this study.

Who can you contact if you have questions about this study?

Any questions concerning the projected entitled Clarifying the lower incidence of birth defects in the Midwest Region, can be directed to Mrs Donna Baker or ▮▮▮▮▮ ▮▮▮▮▮▮ (Masters Student, School of Nursing and Public Health, Edith Cowan University).

If you have any concerns about the project or would like to talk to an independent person, you may contact Dr Carol Thorogood (089273 8623).

Who has given permission for this study to proceed?

The Human Research Ethics Committee at Edith Cowan University and the Confidentiality of Human Information Committee at the Health Department of Western Australia.

<u>Thank you for taking time to read this information sheet</u>

Appendix 5: Ethics Approval – Geraldton Regional Hospital

HEALTH WEST

DEPARTMENT OF HEALTH

GERALDTON HEALTH SERVICE
ABN 74 255 316 169

Mrs Donna Baker

███████████
███████████

Dear Donna,

RE: Clarifying the lower incidence of birth defects in the Midwest.

Further to your request of May 1st 2002 and our subsequent telephone conversations. Approval has been granted for you to access hospital records to complete your research.

Yours sincerely

███████████

Beth Anderson
DIRECTOR OF NURSING
GERALDTON HEALTH SERVICE
13th June 2002
020613BA2:JC

Birth Defects

Does your Child have one?

There are lower numbers of recorded
birth defects in rural areas.
Is this true?

A study is being conducted in Geraldton to determine the number of
children born between 1995 - 1999 with birth defects.

If you would like more information about the study or would like to
be involved, please contact the Principle Researcher Mrs. Donna
Baker on

Appendix 7: Information Sheet for Parents

Clarifying the lower incidence of Birth defects in the Midwest Region.

Information Sheet for Parents

The Birth Defect Registry is a government funded database located at King Edward Memorial Hospital for Women. It is responsible for collecting and collating information about birth defects within our state. Information about children with birth defects is usually reported to the Registry by a health professional around the time of diagnosis.

Current data indicates there is a lower number of birth defects in rural areas of Western Australia in comparison to the Metropolitan area. Of particular interest are the Southwest and Midwest regions, which have the lowest rates of birth defects. This finding is based upon information collected by the Birth Defects Registry of Western Australia. Evidence exists to suggest that this finding is inaccurate. The accurate collection of information is vital to ensuring that rural community needs and concerns receive attention and adequate allocation of resources and monies.

Information collected during this study will benefit both you and the Birth Defects Registry by:

- determining accurate rates of birth defects in rural areas;

- generating research into the causes and prevention of birth defects;

- investigating changes in the frequency of birth defects;

- providing health professionals with statistical information;

- evaluate screening programs, prevention and treatment strategies of birth defects, to assist with planning of health care facilities and assure the adequate allocation of monies and resources in rural areas; and

- increasing general knowledge about birth defects.

What is the aim of this study?

There is evidence to suggest that the lower number of birth defects in rural areas is due to a number of cases not being reported to the Birth Defects Registry. The aim of this study is to determine if the lower incidence of birth defects in the Midwest is due to the under reporting of cases. To achieve this goal the researcher will combine information provided by parents, hospital records and interviews with health professionals to determine the true incidence of birth defects in the Midwest.

Who is doing this study?

The study is a Masters thesis initiated by the Birth Defects Registry and being conducted by:

Mrs Donna Baker (Masters Student)

Edith Cowan University

Pearson Street Churchlands WA 6018

email: ███████████

Study supervision and direction is being provided by the Edith Cowan University and Dr Bower of the Birth Defects Registry of Western Australia. The study supervisors are Dr Carol Thorogood (Co-ordinator of Midwifery Programs at Edith Cowan University), Dr Carol Bower (Clinical Associate Professor at the Birth Defects Registry) and Dr Leanne Monterosso (Post Doctoral Fellow, School of Nursing and Public Health at Edith Cowan University).

What will be expected of you during the study, should you choose to participate?

Should you choose to participate in the study your contact details will forwarded to the researcher who will contact you. If your child has a reportable birth defect you will be asked by the researcher to provide details regarding the type of defect, the child's name, date and place of birth.

How will your privacy be protected?

The researcher, research assistant and supervisors will be the only people to have access to the information you provide about your child. Information regarding your child will be compared with data held by the Birth Defect Registry to determine if they have been recorded, your child's details will then be replaced by a numerical code. This means your child will no longer be able to be identified within the study. No name identifying data will be used in any reports or publications written as a result of this study and your participation will remain anonymous. All coded records will be kept in a locked filing cabinet located in the Principal Supervisor's office (Edith Cowan University). These records will be destroyed after five years. No one will have access to this documentation.

Voluntary participation and your right to refuse?

Participation in this study is completely voluntary. You are free to ask questions at anytime and have a right to have them answered to your satisfaction. You are free to withdraw consent to continue in the study at anytime, and without penalty, simply by contacting the principal researcher or an above named supervisor.

Are there risks involved in this study?

There are no known risks to you or your child if you take part in this study.

Who can you contact if you have questions about this study?

Any questions concerning the projected entitled Clarifying the lower incidence of birth defects in the Midwest Region, can be directed to Mrs Donna Baker on (08) 9273 8333 (Masters Student, School of Nursing and Public Health, Edith Cowan University).If you have any concerns about the project or would like to talk to an independent person, you may contact Dr Carol Thorogood (08 9273 8623).

Who has given permission for this study to proceed?

The Human Research Ethics Committee at Edith Cowan University and the Confidentiality of Human Information Committee at the Health Department of Western Australia.

Thank you for taking time to read this information sheet

Appendix 8: Consent Form for Parents

CLARIFYING THE LOWER INCIDENCE OF BIRTH DEFECTS IN THE MIDWEST REGION

PARTICIPANT FORM OF CONSENT

I... ... ...have read

         Given Names   Surname

the information sheet explaining the study entitled **Clarifying the lower incidence of birth defects in the Midwest Region.**

I have read and understood the information given to me. Any questions I have asked have been answered to my satisfaction.

I agree that research data gathered from the results of this study maybe published, provided that names are not used and I am not identifiable.

I am aware that I may withdraw from this study at anytime without penalty or prejudice.

I have provided my contact details below so that they maybe forwarded to the principal researcher to enable then to contact me for further participation in the study.

Dated ... ... day of 2002

Participant's Signature ... ...

Name ... ...

(BLOCKLETTERS)

Address ... ...

Phone (wk) ... ...

(hm) ... ...

Investigator's Signature ... ...

Appendix 9: Additional Question Record Form

Study ID number:
Was the defect coded by the hospital? If Yes, how was it coded?
Where in the notes was the information about the birth defect documented?
Was the infant referred to other hospitals, departments or health professionals for follow-up?
Was the child transferred to Perth for treatment?
Was the birth defect diagnosed prior to birth? If Yes, by whom and what technique was used to confirm diagnosis?

Appendix 10: Master Coding Sheet

Unit Record Number	Study ID Number

Appendix 11: Focus Group Interview Questions

1. What are your current procedures for reporting birth defects and how are these defined?

2. What is the process of hospital referral for antenatal and unexpected birth defects occurring in the Midwest?

3. How do you feel current reporting procedures could be improved upon?

4. Who would be best situated to assume the responsibility of reporting birth defects?

5. If you were setting up a your own Birth Defects Registry what reporting procedures would you put into place?

Appendix 12: Authentication Record Form

Edith Cowan Australian University

Faculty of Communications, Health and Science

CLARIFYING THE LOWER INCIDENCE OF BIRTH DEFECTS IN THE MIDWEST REGION

AUTHENTICATION OF DATA COLLECTION

I... ... ... participated

       Given Names   Surname

In a focus group interview on the ... ... (date) as a part of the qualitative

data collection for the study entitled **Clarifying the lower incidence of birth defects**

in the Midwest Region.

I have read and understood the data transcript of the interview given to me and

concur that it is a true record of the information discussed at the interview. Any

questions I have asked have been answered to my satisfaction.

I agree that research data gathered from the results of this study maybe published,

provided that names are not used and I am not identifiable.

I am aware that I may withdraw from this study at anytime without penalty or

prejudice.

180

I have provided my contact details below so that they maybe forwarded to the principal researcher to enable then to contact me for further participation in the study.

Dated... ... ...day of... 2002

Participant's Signature... ... ...

Name... ... ...
(BLOCKLETTERS)

Address... ... ...

Phone (wk)... ... ...

(hm)... ... ...

Investigator's Signature... ... ...

Appendix 13: Focus Group Interview Data Collection Form

Focus Group... ... ...Time... ... ...Date... ... ...

Question 1.
Question 2.

Question 3.

Question 4.

Question 5.

Appendix 14: Ethics Approval – Confidentiality of Health Information Committee (CHIC)

CONFIDENTIALITY OF HEALTH INFORMATION COMMITTEE (CHIC)
An Independent Committee appointed by the Minister for Health in Western Australia

Please address all correspondence to:-
Project Officer - CHIC
Health Information Centre
1ˢᵗ Floor 'C' Block
189 Royal Street
EAST PERTH WA 6004

ph: (08) 9222 4194
fax: (08) 9222 4236

Mrs Donna Baker

██████████

Dear Mrs Baker

#200202   CLARIFYING THE LOWER INCIDENCE OF BIRTH DEFECTS IN THE MIDWEST OF WESTERN AUSTRALIA

Date of commencement	15/03/2002
Date of completion	30/09/2002
Researchers accessing identifiable data	Peter Cosgrove Carol Bower Aandra Ryan Edwina Rudy
Databases to be accessed	Birth Defects Registry
Ethics approval	ECU Human Research Ethics Committee 25/01/2002 (until 31 December 2002)

Thank you for your letter dated 19 June 2002 enclosing copies of the ethics approval given by Edith Cowan University and Geraldton Regional Hospital.

Please continue to keep us informed of any changes. We wish you well with your project.
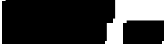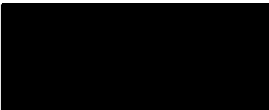
Yours sincerely

██████████   ██████████

Dr David Blackledge   Ms Alison Daly
CHAIRPERSON   **DEPARTMENT OF HEALTH REPRESENTATIVE**
CONFIDENTIALITY OF HEALTH INFORMATION COMMITTEE

10 July 2002

Appendix 15: Ethics Approval – Edith Cowan University

ECU
EDITH COWAN UNIVERSITY

CHURCHLANDS CAMPUS

Pearson Street, Churchlands
Western Australia 6018
Telephone (08) 9273 8333
Facsimile (08) 92738661

13th June 2002   Human Research Ethics Committee   ABN 54 361 485 361

2002
futures

Ms Donna Baker (Student # 301 1849)

Dear Ms Baker

Code: 01-218
Project Title: *Clarifying the lower incidence of birth defects in the Midwest region of Western Australia*

Thank you for advising the Ethics office of your revised methodology. The matter has been considered by the Human Research Ethics Committee and approval has been given for your new methodology.

Please ensure that you provide the Ethics Office with a copy of the ethics approval from the Confidentiality of Health Information Committee. Health Department of Western Australia before commencing your work.

Period of approval: **From**   12th June 2002   **To**   30th April 2003

With best wishes for success in your work.

Yours sincerely

EXECUTIVE OFFICER
Phone 9273 8170
Fax: 9273 8661
Email: m.beresford@cowan.edu.au

Attachment: Conditions of Approval

cc.   Dr L Menterosso, Supervisor
       Ms J Knight, Manager, Graduate School
       Ms R Treloar Cook, Administrative Officer HDC

Appendix 16: Interview Participation Consent Form

<div align="center">

Edith Cowan University Western Australia

Faculty of Communications, Health and Science

</div>

<div align="center">

CLARIFYING THE LOWER INCIDENCE OF BIRTH DEFECTS IN THE MIDWEST REGION

PARTICIPANT FORM OF CONSENT

</div>

I... ... ... have read

        Given Names   Surname

the information sheet explaining the study entitled **Clarifying the lower incidence of birth defects in the Midwest Region.**

I have read and understood the information given to me. Any questions I have asked have been answered to my satisfaction.

I agree that research data gathered from the results of this study may be published, provided that names are not used and I am not identifiable.

I am aware that I may withdraw from this study at anytime without penalty or prejudice.

Dated ... ... day of 2002

Participant's Signature ... ...

Investigator's Signature ... ...