

Population prevalence rates of birth defects: a data management and epidemiological perspective

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Abstract

The Victorian Birth Defects Register (VBDR) is a population-based surveillance system with a primary function of monitoring trends in birth defects. This paper outlines the processes undertaken in Victoria, Australia, to obtain population prevalence rates of birth defects and investigates the effect on the prevalence rates of variations in collection and processing tasks. It includes all birth defects that were notified to the VBDR by 31 December 2004.

The overall prevalence rate of birth defects in Victoria for 2003 was 4.0%, with an overall accuracy rate of 88%. However, this proportion varied according to what birth defects were included, the age by which birth defects were diagnosed, changes to sources of ascertainment, inclusion of terminations of pregnancy, or reporting by cases rate (infants affected) or birth defect rate (individual birth defects). Taking all of these factors into consideration, we are confident that 4.0% is an accurate population prevalence rate of birth defects in Victoria for 2003.

Key words: *Birth defects; congenital malformations; prevalence*

Introduction

In 2003, birth defects were the major cause of all perinatal deaths in Victoria (Consultative Council on Obstetric and Paediatric Mortality and Morbidity 2004) with 23% of all stillbirths and neonatal deaths attributable to a congenital anomaly. With the decline in childhood mortality from other causes (such as infections), this makes the monitoring of population rates of birth defects a very important public health issue.

The Victorian Birth Defects Register (VBDR) was established in 1982 as a direct response to an epidemiological investigation — the Yarram Inquiry of 1978 (Consultative Council on Congenital Abnormalities in the Yarram District 1978). A possible cluster of birth defects was investigated to determine if there was any association with certain agricultural products. Whilst no association was evident during this investigation, the Inquiry concluded that 'Victoria lacks organised research on the epidemiology of birth defects and lacks an adequate system of surveillance of defects which are not rapidly lethal' (Consultative Council on Congenital Abnormalities in the Yarram District 1978). The Victorian Perinatal Data Collection Unit (VPDCU) was therefore established in 1982 under the auspices of the Consultative Council on Obstetric and Paediatric Mortality and Morbidity (*Health Act 1958*)¹, and one of its primary functions was to establish and maintain a birth defects register.

The VBDR is a population-based surveillance system of birth defects with the primary functions of:

- monitoring trends in birth defects (prevalence and survival data)
- provision of data to organisations responsible for providing health care services to people with birth defects
- providing information for epidemiological research
- assessing the effectiveness of primary prevention and screening programs

- responding to community concerns about perceived clusters of birth defects (Riley Halliday 2004).

Given these purposes, it is a fundamental objective of the VBDR to obtain birth defect prevalence data that is as accurate and complete as possible.

Investigation into factors that may affect local population prevalence rates are very important, especially when comparisons may be made between geographical areas (e.g. states of Australia) or when data from different geographical areas may be pooled to provide one global or national figure. For example, in Australia each state cites its own birth defect population prevalence rates. These figures range from 2.0% (in 2002) of all births in New South Wales to 4.0% (in 2003) of all pregnancies in Victoria (Centre for Epidemiology and Research 2004), to 5.0% (in 2002) of all pregnancies in Western Australia (Bower et al. 2004). Obvious questions arise as to why there is such variation in figures between the states, and which, if any, is the 'true' population prevalence rate of birth defects.

The aims of this paper are:

- to outline the process undertaken by Health Information Managers (HIMs) in Victoria, Australia, to obtain population prevalence rates of birth defects, and
- to investigate the effect on the prevalence rates of variations in collection and processing tasks.

Methodology

The VBDR collects data on all birth defects diagnosed before 15 years of age in all Victorian live births, stillbirths, neonatal deaths and terminations of pregnancy (TOPs) for a birth defect, both before and after 20 weeks gestation, occurring since 1 January 1982. Because age at diagnosis may occur at any time from birth to 15 years, the VBDR is continually being updated and new cases are added each year. The data presented in this paper include all birth defects for

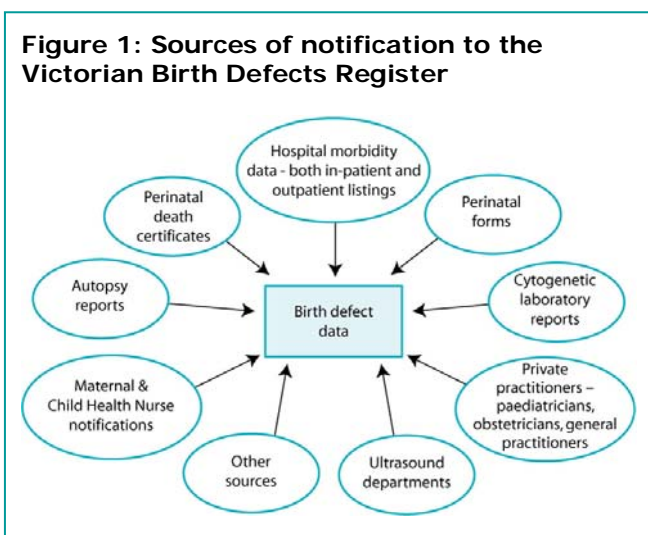
¹ Part 9B of the Act relates to the Consultative Council on Obstetric and Paediatric Mortality and Morbidity.

children born between 1983-2003 notified to the VBDR by 31 December 2004.

A birth defect is defined as '... structural defects or chromosomal abnormalities that are present at birth but not necessarily diagnosed at birth' (Riley & Halliday 2004). Information is also collected on inborn errors of metabolism, haematological disorders, congenital infections, neoplasms and developmental delay, if of probable prenatal origin.

The VBDR is a voluntary reporting system that is maintained by the VPDCU, a mandatory reporting system that collects demographic, obstetric and paediatric information on all births in Victoria on a prescribed form known as the Perinatal Morbidity Statistics Form ('perinatal form').

Information for the VBDR is obtained from multiple sources (Figure 1). Since 1990, the VPDCU actively has followed up each specific source to ensure that data are obtained from the same sources each year, if possible.



These notifications come in the form of electronic listings or hard paper copies. Each electronic listing is printed out and all cases reviewed by a Health Information Manager to determine which case should be included (excluded cases include non-notifiable birth defects and interstate or overseas births). Vague, non-specific or questionable diagnoses from any source are followed up with the notifier or treating paediatrician for more details.

Data from all sources (excluding TOPs for a birth defect before 20 weeks gestation) are linked to a perinatal form, if possible. There are three levels of data linkage attempted, dependent upon the information provided:

1. Mother's full name (if provided), child's date of birth, sex of infant.
2. Mother's given name (if provided), child's date of birth, sex of infant, postcode.
3. Child's surname, date of birth, sex, postcode.

Linkage is also attempted for major anomalies that cannot be linked to a perinatal form using the above three combinations, using the child's date of birth, sex and diagnosis. Here the VBDR is searched to determine whether or not these variables match any cases that have been previously notified but do not match

on name. Because birth defects are rare events, it is sometimes possible to identify cases based upon diagnosis, date of birth and sex alone.

After linkage, all cases are coded by HIMs using the British Paediatric Association Classification of Diseases ICD-9 Supplement (British Paediatric Association Classification of Diseases 1979). Difficult cases are referred to the VPDCU's consultant paediatrician for classification. In the case of syndromes, it is the practice of the VPDCU to code all manifestations of the syndrome along with the primary diagnosis, because not all cases of a particular syndrome reveal exactly the same manifestations. However, for conditions with a standard set of defects in all cases, such as Tetralogy of Fallot, only the one condition is coded, not each of the manifestations.

Data is then entered into an ACCESS database, and a hard copy of all of the data entered is printed for subsequent checking. In cases where there is more than one notification with different diagnoses, a 'case summary' is created based upon the best composite of the diagnoses. If there is any uncertainty about any of the information that should be included, then the case is referred to the consultant paediatrician.

At the end of each calendar year, when processing has been completed, a combined file of all years from the VBDR is compiled, data validation checks performed and reports produced. This combined file remains the active file from which all research is derived for each particular year, until the file is again updated the following year.

Analysis is annually undertaken on 28 sentinel defects (Riley & Halliday 2004), including such things as trends in overall prevalence (including all birth outcomes), birth prevalence (excluding TOPs before 20 weeks gestation) and live birth prevalence (excluding TOPs before 20 weeks gestation and stillbirths). Rates reported can differ depending on whether or not results are presented according to:

- birth defect cases (case rate) — the number of infants affected by at least one defect or,
- individual defect (defect rate) — the number of occurrences of a particular birth defect.

Time trend analysis for the 28 selected defects is undertaken using chi-square linear trend analysis. Selected infant and maternal characteristics are also investigated for significant associations.

Results

In 2003, there were 3544 notifications to the VBDR from all sources (Table 1). The number of notifications exceeds the number of cases due to multiple reporting of cases from different sources. These notifications referred to 2709 cases, approximating to 1.3 notifications per case.

There were 2205 babies born at, or after 20 weeks gestation with a birth defect, and 339 TOPs before 20 weeks gestation with a birth defect. In 2003, there were 63 551 births in Victoria (including late TOPs at 20 weeks or more for any reason). This gives an overall birth defect prevalence rate of 400/10 000 or 4.0%. There were also an additional 165 notifications of conditions that are collected by the VBDR but not reported in our routine publications (e.g., un-

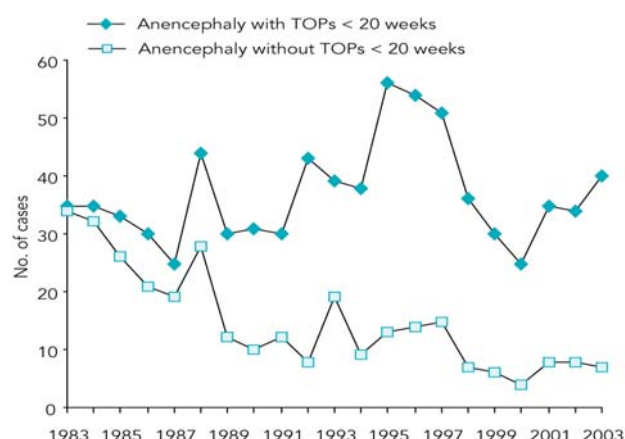
Table 1: Sources of notification, 2003

Notification source	Percent (%)
Perinatal Data Collection Unit forms	52.8
Hospital listings	26.5
Cytogenetics reports	9.5
Perinatal death certificate/autopsy reports	7.6
Maternal and child health nurses	3.3
Other	0.3
Total	100.0

descended testes (UDT) \geq 37 weeks, vesicoureteric reflux [VUR]). (Centre for Epidemiology and Research 2004). If these conditions are included then the overall birth defect prevalence rate for 2003 is 420/10 000 or 4.2%.

Table 2 shows the birth defect prevalence rate from 1983 to 2003, incorporating all notifications (excluding UDT, VUR and other excluded minor anomalies) received by 31 December 2004. This table shows an increase in the overall birth defect prevalence rate from a minimum of 2.6% in 1985 to a maximum of 4.6% in 1999.

Figure 2: Prevalence of anencephaly with and without terminations of pregnancy before 20 weeks gestation, 1983-2003



Presented in Table 3 are variations in overall prevalence rates and birth prevalence rates for selected conditions when TOPs before 20 weeks are included and excluded respectively.

Figure 2 shows graphically the differences in the overall prevalence rate and birth defect prevalence rate for anencephaly when TOPs before 20 weeks are included.

Table 2: Birth defects by year, 1983–2003

Year	Total births 20 weeks and later	Defects 20 weeks and later	Defects before 20 weeks (terminations)	N/10 000 pregnancies (including terminations)	Percentage (%)
1983	60 628	1675	2	276.3	2.8
1984	60 737	1714	19	283.7	2.8
1985	61 189	1599	18	264.3	2.6
1986	61 253	1624	80	278.2	2.8
1987	61 566	1638	55	275.0	2.8
1988	63 666	1886	103	312.4	3.1
1989	64 255	1987	123	327.8	3.3
1990	66 878	2205	132	348.8	3.5
1991	65 248	2294	140	372.2	3.7
1992	66 305	2344	152	375.6	3.8
1993	64 737	2278	203	382.0	3.8
1994	64 932	2327	250	395.4	4.0
1995	63 717	2496	255	430.0	4.3
1996	62 951	2231	272	395.9	4.0
1997	62 308	2338	298	421.0	4.2
1998	62 091	2369	276	424.1	4.2
1999	62 689	2598	296	459.5	4.6
2000	62 564	2545	288	450.7	4.5
2001	62 148	2283	300	413.6	4.1
2002	63 072	2294	300	409.3	4.1
2003	63 551	2205	339	398.2	4.0
Total	1 326 485	44 930	3 901	368.1	3.7

Table 3: Overall prevalence and birth prevalence for 28 selected defects, 2003.
Rates are cases per 10 000 births

Defect	Overall prevalence (including all outcomes)	Birth prevalence (excluding TOPs before 20 weeks)
Nervous system		
Anencephaly	6.3	1.1
Spina bifida	5.3	3.1
Encephalocele	1.6	0.8
Microcephalus	1.6	1.4
Hydrocephalus	10.3	8.2
Cardiovascular system		
Transposition of great arteries	4.5	4.5
Tetralogy of Fallot	4.2	4.1
Ventricular septal defect	29.3	28.3
Hypoplastic left heart syndrome	3.4	2.7
Coarctation of aorta	4.1	3.9
Digestive system		
Cleft palate	10.5	10.2
Cleft lip	3.9	3.9
Cleft lip and palate	6.6	6.0
Oesophageal atresia and/or stenosis	3.1	3.1
Atresia of the small intestine	3.1	3.1
Genitourinary system		
Anorectal atresia and/or stenosis	4.2	3.9
Hypospadias	28.0	28.0
Renal agenesis and dysgenesis	7.4	6.5
Cystic kidney disease	5.2	5.0
Obstructive defects of renal pelvis	35.4	35.1
Musculoskeletal system		
Congenital dislocation of hip	24.9	24.9
Limb reduction defects	3.9	3.6
Diaphragmatic hernia	3.3	2.8
Exomphalos	3.3	1.4
Gastroschisis	2.0	1.9
Chromosomal anomalies		
Trisomy 21	26.6	8.5
Trisomy 13	2.7	0.9
Trisomy 18	8.9	2.8

Neural tube defects (NTDs), which are major structural abnormalities, continue to be a focal point of interest in birth defect monitoring. In 2003, there were 79 cases of NTDs (case rate) reported to the VBDR, giving an overall prevalence rate of 12.4/10 000 pregnancies. Of these 79 cases, five had two NTDs (i.e., anencephaly with spina bifida, or spina bifida with encephalocele). If each condition rather than each case is counted separately, then there were a total of 84 NTDs in Victoria in 2003, giving a birth defect rate of 13.1/10 000.

Of the 28 selected defects which are routinely reported (Centre for Epidemiology and Research 2004), the following significant trends were observed between 1997–2003:

- increase in overall prevalence (including terminations) — Trisomy 21
- decrease in overall prevalence (including terminations) — spina bifida, oesophageal atresia and/or stenosis, anorectal atresia and/or stenosis and limb reduction defects.

However, if the period over which trends are monitored is changed from 1997–2003 to 1983–2003, then the following significant trends are observed:

- increase in overall prevalence (including terminations) — hydrocephalus, hypospadias, cystic kidney, obstructive defects of the renal pelvis, Trisomy 21, Trisomy 13, Trisomy 18
- decrease in overall prevalence (including terminations) — coarctation of aorta.

Discussion

Determining prevalence rates of disease is fundamental to all epidemiological endeavours. However, interpreting these rates can often be complex, as with the case of determining population prevalence rates of birth defects in Victoria.

Impact of variations in inclusion criteria (i.e., birth defects included or excluded)

In 2003, Victoria reported a birth defect prevalence rate of 4.0%. This figure excludes cases with UDTs \geq 37 weeks or VUR. If these cases are included then the prevalence rate increases to 4.2%. From this example, it is apparent that the inclusion of these two conditions substantially increases the birth defect prevalence rate. When we note that other states such as Western Australia report a higher birth defect prevalence rate, we must immediately consider whether or not they are including other conditions which Victoria has excluded. Conversely, when considering the lower New South Wales birth defect prevalence rate, we must consider whether or not Victoria has included conditions which they have excluded.

Impact of variations in reporting period

Not only do we need to consider whether or not the conditions included by each state birth defects register are the same, but also whether or not the reporting covers the same 'type' of population and reporting period. Victoria includes birth defects diagnosed before 15 years of age in all live births, stillbirths, neonatal deaths and TOPs. Western Australia, on the other hand, includes birth defects diagnosed before 5 years of age, and New South Wales includes cases diagnosed within the first year of life. These differences could give rise to variations in birth defect prevalence rates between the states.

Impact of variations in ascertainment

Variations between the states can occur because of inclusion or exclusion criteria; they can also occur within a single state over long time periods where there may be variations in either the collection methodology or conditions reported. Table 2 shows that the prevalence of birth defects in Victoria varied from a low of 2.6% in 1985 to a high of 4.6% in 1999. Either more pregnancies are now being affected by birth defects or there has been some other reason for the increase in Victorian birth defects.

A validation of the VBDR in 1986 (Kilkenny, Riley & Lumley 1995) showed that the register was receiving notifications for 43.5% of babies with birth defects treated as inpatients at two paediatric teaching hospitals. A repeat of this exercise in 1993 and 2001 showed that the notification rate had increased to 73.5% and 87.8% respectively (Riley, Phyland & Halliday 2004; Hennekens & Buring 1987). This shows that reporting, or ascertainment, has been the major reason for the increase in the birth defect prevalence rate in Victoria. In 1988–1989, the VPDCU began to receive inpatient listings from two paediatric teaching hospitals for all children who were admitted to these hospitals with a birth defect. This accounts for an increase from 2.8% to 3.1% over this time period. In the mid 1990s, the VPDCU also began to receive listings from certain outpatient clinics that also treated children with birth defects that did not require hospitalisation. This could account for an increase from 3.5% in 1990 to 4.0% in 1994. The continued rise to 4.6% in 1999 can probably be related to the practice of receiving notifications of children up to the age of 5 years who have been admitted, or attended, a paediatric teaching hospital in any single year. Sources of notification have now substantially remained the same for 10 years, so variations in birth prevalence rates will need to look to other sources for an explanation.

Impact of inclusion of terminations of pregnancy (TOPs) before 20 weeks gestation

The inclusion of TOPs before 20 weeks gestation for a birth defect can greatly affect the reported prevalence rate, as shown in Table 3 and Figure 2. For some conditions, such as hypospadias or congenital dislocated hip, where very few of the cases are terminated, exclusion of TOPs has very little impact on the prevalence rates, with the overall prevalence rate for the latter being 24.9/10 000 and the birth prevalence rate of 24.9/10 000. However, for conditions where a large proportion of the cases are terminated, exclusion of these cases dramatically affects the overall prevalence rate. This can be seen in Figure 2 where the number of cases of anencephaly are presented both with and without terminations. The birth prevalence of anencephaly in 2003 was only 1.1/10 000 births compared with an overall prevalence of 6.3/10 000 pregnancies.

The most appropriate prevalence rate to cite is dependent upon the issue of interest. For instance, if a group were responsible for planning health services for children with Down Syndrome, then their area of interest is the live birth prevalence rate. However, genetic counsellors, wishing to advise clients on the risk of recurrence of Down Syndrome would be more interested in the overall prevalence rate (i.e., how many pregnancies are affected by Trisomy 21). The issue is not so much determining what is the true prevalence, but rather clearly indicating the type of prevalence which is being quoted.

Impact of case rate versus birth defect rate

Another factor which affects the reporting of a birth defect prevalence rate is whether or not reporting is

concerned with how many infants are affected by a particular condition (case rate), or rather with the individual birth defects (birth defect rate). As indicated in the results, there were 79 cases affected by NTDs in Victoria in 2003, but there were 84 individual birth defects. Since the effectiveness of the periconceptional use of folic acid in the primary prevention of NTDs is monitored through prevalence figures of NTDs, reporting by defect rate rather than case rate could alter conclusions drawn about the effectiveness of the program.

Impact of variations in trend analyses

We have also shown in the results that trend analysis is another complex area that can be affected by the time period over which the trend is reported. For the 7-year period between 1997–2003, only one of the 28 selected defects showed a significant increase in overall prevalence — Trisomy 21. However, if the trend analysis covered 21 years, from 1983–2003, then a significant increase in prevalence was also observed for six other birth defects. This indicates that careful and meaningful consideration needs to be given when attempting such analyses. We have already discussed changes in sources of notification that have contributed to the increase in the state-wide birth defect prevalence rate. These same concerns come into consideration here. To obtain the most meaningful trends in prevalence we need to choose the time period over which the birth defects have been consistently collected, that is, 1993–2003.

Given all of the factors discussed above — issues of inclusion criteria of conditions and reporting periods, ascertainment of cases, overall prevalence versus birth prevalence, case defect rates versus birth defect rates, and variations in trend analyses over time — the question remains: Is there such a thing as a true population prevalence rate for birth defects? As Hennekens states, 'epidemiology is the study of the distribution and determinants of disease frequency'. The merit of the prevalence rate reported is directly proportional to the quality and accuracy of the data obtained.

In Victoria, the population birth defect prevalence rate in 2003 was 4.0%, with an accuracy rate from our latest validation study of 88% (Riley, Phyland & Halliday 2004). We cannot extrapolate this figure to other areas of Australia without extensive comparison between the collection methodologies; however, we

can be confident of this figure as the population birth defect rate in Victoria.

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