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Congenital Malformations Australia 1995 and 1996

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Requests for data

Any enquiries about data for regions within States and Territories and for individual hospitals should be directed initially to State and Territory health departments. Other enquiries should be made to the AIHW National Perinatal Statistics Unit.

The report may be obtained from:

Government Info Shops, or
AusInfo Mail Order Sales

Call toll-free on 132 447
or visit http://www.ausinfo.gov.au/

Abbreviations

NSW	-	New South Wales
Vic	-	Victoria
Qld	-	Queensland
WA	-	Western Australia
SA	-	South Australia
Tas	-	Tasmania
ACT	-	Australian Capital Territory
NT	-	Northern Territory
ABS	-	Australian Bureau of Statistics
AIHW	-	Australian Institute of Health and Welfare
NPSU	-	National Perinatal Statistics Unit
a/s	-	atresia/stenosis
ASD	-	atrial septal defect
BPA	-	British Paediatric Association
bronch.	-	bronchus
cerv.	-	cervix
dis.	-	disease
dysgen.	-	dysgenesis
grt.	-	great
ICD	-	International Classification of Diseases
L	-	left
musculoskelet.	-	musculoskeletal
NEC	-	not elsewhere classified
NOS	-	not otherwise specified
pat.	-	patent
pulm.	-	pulmonary
spec.	-	specified
st./insuffic.	-	stenosis/insufficiency
synd.	-	syndrome
TOF	-	tracheo-oesophageal fistula
trach.	-	trachea
transpstn.	-	transposition
vent.	-	ventricular

Highlights

- Among 3.88 million births in the period from 1981 to 1996, 62,194 (1.6%) infants with major congenital malformations diagnosed at birth or in the first 28 days were notified to the national monitoring system. These included 4,561 (1.8%) infants born in 1995 and 4,458 (1.7%) born in 1996.

- Perinatal deaths due to congenital malformations declined from 35.9 per 10,000 births in 1973 to 15.6 per 10,000 births in 1996, the lowest level of any year. The perinatal death rate for anencephalus showed the most pronounced decline but there were also substantial falls for spina bifida, hydrocephalus and congenital heart defects. Perinatal death rates due to chromosomal abnormalities increased slightly in the same period. Congenital malformations accounted for 18.3% of perinatal deaths in 1996.

- Infant deaths due to congenital malformations declined from 28.8 per 10,000 live births in 1980 to 14.8 per 10,000 live births in 1996. Congenital malformations were the cause of 27% of infant deaths, and 10% of childhood deaths, in 1995, and 26% of infant and 10% of childhood deaths in 1996.

- In the period from 1993 to 1996, the total malformation rate of 293.5 per 10,000 among births to mothers aged 40 years and over was almost double that of 155.4 per 10,000 among births to mothers aged 20-24 years. Chromosomal abnormalities were more likely with advancing maternal age. Women aged 40 years and over were 13 times more likely than women aged 20-24 years to give birth to an infant with a chromosomal abnormality and more than 40 times more likely to have a termination of pregnancy before 20 weeks' gestation.

- Isolated and multiple malformations were more common in twins and other multiple births than in singleton births.

- The national rate of anencephalus has been declining during recent decades, with slight fluctuations in recent years. Similarly, spina bifida declined to its lowest rate of 3.0 per 10,000 births in 1994 and 1996, with a higher rate of 3.5 per 10,000 births in 1995. In 1995, there were 78 reported terminations of pregnancy for anencephalus and 55 for spina bifida, and in 1996, there were 70 and 66, respectively.

- Increasing rates of ventricular septal defect and obstructive defects of the renal pelvis and ureter are likely to reflect better ascertainment, particularly by prenatal ultrasound screening for the renal defects.

- Following an increase in the early 1990s and then a slight decline to 1.5 per 10,000 births in 1994, the rate of gastroschisis reached its highest level of 2.1 per 10,000 births in 1996. This malformation is much more common among births to younger than to older mothers.

- Notified terminations of pregnancy for fetal malformations increased from 537 in 1992 to 722 in 1994, then fell to 527 in 1996, but notification was incomplete. The main indications for termination of pregnancy were trisomy 21 (Down syndrome), other chromosomal abnormalities, neural tube defects (anencephalus and spina bifida) and cardiac defects. Most terminations of pregnancy for fetal malformations (78.6%) were performed at gestational ages between 16 and 22 weeks.

- Reported terminations of pregnancy for trisomy 21 (Down syndrome) and for trisomy 18 (Edwards syndrome) were higher in 1994 than in any previous year, but then declined to 113 and 45, respectively, in 1996.

1 Introduction

Congenital malformations are a significant public health problem because they are relatively common, they are major reasons for admission to hospital during infancy and childhood, they may frequently lead to disabilities and handicaps, and some types are fatal. In 1995, 20.4% (428/2,094) of all perinatal deaths in Australia, and 27.2% (394/1,449) of infant deaths, were due to congenital malformations; in 1996, 18.3% (398/2,170) of all perinatal deaths, and 25.5% (372/1,460) of infant deaths, were due to congenital malformations (Australian Bureau of Statistics 1996 and 1997).

Data collected in the State and Territory perinatal data systems, by birth defects registers, and from hospitals provide valuable information on the number of children with malformations diagnosed early in life. These data enable analysis of trends over time and variations by region; collaborative studies of the descriptive epidemiology of congenital malformations, both within Australia and internationally; and evaluation of the impact of prenatal diagnosis or other interventions on trends in occurrence. Almost universal prenatal screening of pregnant women by ultrasound in Australia, and the frequent use of amniocentesis or chorionic villus sampling in high-risk women, have increased the likelihood of detecting some types of congenital malformations during fetal life. If a pregnancy is terminated, it is important to collect information about these pregnancies so that the effect on trends can be monitored adequately.

This report contains national data on congenital malformations among births that occurred in the period 1981 to 1996. Data for the years up to 1994 were published previously in *Birth Defects Series Number 2* (Lancaster, Hurst, Day, Huang & Shafir 1997).

1.1 Criteria

Congenital malformations are anatomical defects or chromosomal abnormalities that are present at birth. Major congenital malformations are either lethal or significantly affect the individual's function or appearance. Minor malformations do not have functional or cosmetic importance but may sometimes signify an underlying genetic disorder. Although infants with only minor malformations may be included in State birth defects registers, these infants are not notified to the AIHW National Perinatal Statistics Unit unless major malformations are also present (see Appendix 1 for list of minor malformations).

Major congenital malformations diagnosed in liveborn infants in the first 28 days, or in stillbirths of at least 20 weeks' gestation or 400g birthweight, are included in this report. Some tables on selected malformations also give incomplete data on terminations of pregnancy that followed prenatal diagnosis. If terminations of pregnancy occurred at gestational ages of 20 weeks or more, these fetuses are included in the figures for stillbirths.

1.2 Sources of data

Congenital malformations are notified on forms designed to obtain data on all births occurring in each State and Territory. Ascertainment of congenital malformations is improved by using additional sources of notification including children's hospitals and other referral hospitals, cytogenetics laboratories, perinatal death certificates, autopsy reports and notifications of induced abortions. Four States - New South Wales, Victoria, Western Australia and South Australia - have birth defects registers that include notifications of malformations diagnosed in infants and children after the perinatal period.

Only South Australia has mandatory notification of induced abortions. Ascertainment of terminations of pregnancy that follow prenatal diagnosis of fetal abnormalities is variable in the other States and Territories, except for chromosomal abnormalities which have been reported by cytogenetics laboratories since 1982.

Data for 1981 were incomplete in New South Wales and excluded births in Victoria, Western Australia, the Australian Capital Territory and the Northern Territory. Notifications of congenital malformations in the Northern Territory began in 1986.

1.3 Data items

Demographic, maternal and infant or fetal data are recorded on each notification to the AIHW National Perinatal Statistics Unit (NPSU). The data items are listed in Appendix 2. Some items are incomplete, either because they are not recorded on notification forms or because the information may not be readily available at the data source (e.g. children's hospitals, cytogenetics laboratories).

South Australia is the only State that routinely collects some information on the family history of birth defects or on maternal exposures to drugs and environmental agents. Such information is not coded in the national monitoring system but has been used to review reported associations between specific drugs (e.g. isotretinoin) and congenital malformations.

1.4 Data processing

Data on congenital malformations are coded in each State and Territory by the groups responsible for perinatal data collection and birth defects registers. Major congenital malformations and chromosomal abnormalities listed in the chapter on congenital anomalies in the Ninth Revision of the International Classification of Diseases (ICD) are notified to the NPSU. In due course, ICD-10 will replace ICD-9 for classifying congenital malformations. The NPSU and the States and Territories use the British Paediatric Association (BPA) Classification of Diseases, which is a 5-digit system compatible with ICD at the 4-digit level, to code congenital malformations. The BPA classification enables more specific coding of malformations and, with some modification of codes for malformation syndromes, limb reduction defects and other selected malformations, has been used by the NPSU since 1981. The list of minor malformations that are excluded from the national data are given in Appendix 2. If notification forms contain information on which side of the body is affected by a malformation, this information is coded by the NPSU.

Since 1994, most States have provided data in an agreed format on floppy disk. By 1996, all except two States sent data in this format. Because some States obtain maternal information by linking perinatal or hospital morbidity data to notifications of congenital malformations, there may be delays in forwarding the complete records to the NPSU, particularly when linkage is deferred until all births in a specific year are included.

Whenever possible, coding by State and Territory groups and the NPSU is based on written descriptions of congenital malformations. Further information is requested if diagnoses are doubtful or if key data items such as maternal age are missing. While this report gives widely accepted definitions of selected major congenital malformations, it is not always possible to ensure that notified diagnoses of malformations conform to these definitions.

1.5 Contents of report

Data in all tables, graphs and maps are based on the year of birth, or the year of induced abortion, and on the State or Territory in which the birth or termination of pregnancy occurred. Denominators for calculating rates are based on the State or Territory of registration, obtained from the Australian Bureau of Statistics for the years 1981 to 1990, and are obtained from State and Territory perinatal data collections for the years 1991 to 1996 (ABS 1996; ABS 1997; Day et al. 1997; Day et al. 1999). Malformation rates are expressed per 10,000 total births, including those malformations that are specific to one sex. As the criteria used by the States and Territories for their published reports may differ from those used here, their numbers and rates may differ from those in this report. Also, the criteria and sources of notifications vary among the States and Territories, so differences in malformation rates should be interpreted cautiously. Small numbers of specific types of malformations may also influence variations in rates.

Tables 2.1-2.7 are similar in content to annual tables published previously. Tables 2.1-2.4 give national data, and data for each State and Territory, on the total number of infants and fetuses with major congenital malformations in the years 1981 to 1996. In Tables 2.5-2.7, State/Territory and national data are given on major malformations affecting all anatomical systems. National data are presented separately for 1981-1994, 1995, 1996 and the whole period; data on selected malformations in each State and Territory are given for the same years.

Tables 3.1-3.25 and the accompanying graphs (Figure 3.1-3.25), provide data on national trends of 25 congenital malformations or chromosomal abnormalities that are either lethal, have significant consequences for surviving children and their families, or are relatively common. For each congenital malformation, the proportion occurring as an isolated malformation, in association with one or more other major malformations, or as part of a chromosomal syndrome is given. Further information on the descriptive epidemiology of these malformations was given in the earlier report *Birth Defects Series Number 1* (Lancaster & Pedisich 1995).

As the level of ascertainment of induced abortions at less than 20 weeks' gestation is uncertain in most States and Territories, these abortions have been excluded from the reported rates of the 25 selected malformations. Induced abortions of fetuses less than 20 weeks' gestation, or unstated gestational age, are given under that heading in the tables. Induced abortions at gestational ages of 20 weeks and over are included in the figures for stillbirths, but these terminations of pregnancy had not necessarily been reported as perinatal deaths. The inclusion of stillbirths in these Australian data will affect comparisons with those other countries where data on stillbirths are not available.

Information on induced abortions is recorded twice in the NPSU data, once in the outcome of the pregnancy and again in the outcome of the infant/fetus. This enables induced abortions at gestational ages of 20 weeks or more (which are included in the figures for stillbirths) to be identified. Hence, a pregnancy terminated prior to 20 weeks' gestation should be recorded as an induced abortion for both the pregnancy outcome and infant/fetus outcome, and a pregnancy terminated at 20 weeks' gestation or more, should be recorded as an induced abortion in the pregnancy outcome and as a still birth for the infant/fetus outcome. It must be noted that some States are unable to identify induced abortions of 20 weeks' gestation or more, as they receive such data with the pregnancy outcome recorded as a stillbirth. This results in under-reporting of induced abortions occurring on or after 20 weeks' gestation, so Tables 2.9 to 2.15 should be read with extra caution.

The data reported here on the proportion of stillborn and liveborn infants with selected malformations dying before birth or in the neonatal period (within 28 days of birth) may not always be reliable for several reasons. If a malformed infant dies of some related complication such as an infection or cardiac failure, the congenital malformation may not always be recorded on the perinatal death certificate. On the other hand, if there is incomplete reporting of malformations on birth notifications, the proportion of stillbirths and neonatal deaths may be overestimated because ascertainment of malformations recorded on perinatal death certificates is complete in all States and Territories. Also, because infants with multiple malformations are included in the tables for each type of malformation, there may be relatively high proportions of stillbirths and neonatal deaths, and sometimes induced abortions, included in the figures for some apparently mild malformations.

During the period from 1982 to 1996, the annual number of births increased from 238,684 in 1982 (excluding the Northern Territory) to 260,044 in 1995 and 257,092 in 1996, with some fluctuations in the intervening years (Table 2.19).

1.6 International monitoring of congenital malformations

National, regional or hospital-based monitoring systems similar to the Australian national system operate in numerous other countries around the world. Through the International Clearinghouse for Birth Defects Monitoring Systems, and its International Centre for Birth Defects located in Rome, Australia participates in reporting of congenital malformations and in studies of the epidemiology and causes of congenital malformations. The definitions of selected major congenital malformations in this report are generally those adopted by the International Clearinghouse for Birth Defects Monitoring Systems.

2 Major congenital malformations

2.1 Births

There were 62,194 infants and fetuses with major congenital malformations notified in the years 1981 to 1996 among 3.88 million births, a total rate of 160.1 per 10,000 births, or 1.6%, in this 16-year period (Table 2.1). The total malformation rate of 173.4 per 10,000 births in 1996 was slightly less than the rate of 175.4 per 10,000 in 1995. Overall, 76.5% of infants had malformations affecting a single body system, 7.3% had multiple malformations affecting more than one system, and 16.2% had identifiable chromosomal or other syndromes. In 1995 there was more identifiable chromosomal or other syndromes than in other years (16.1% in 1981-1994, 17.5% in 1995, 16.2% in 1996), with a corresponding decrease in malformations affecting a single body system.

The reported malformation rates in the 6-year period from 1991 to 1996 were highest in Victoria (225.5 per 10,000 births), the Australian Capital Territory (195.4 per 10,000 births), Queensland (193.6 per 10,000 births) and South Australia (188.8 per 10,000 births) and lowest in Tasmania (98.7 per 10,000 births), Western Australia (112.0 per 10,000 births) and the Northern Territory (118.2 per 10,000 births) (Table 2.2). These variations are likely to reflect differences in the sources and ascertainment of malformations rather than real differences in incidence.

Each State and Territory publishes data on congenital malformations, either in separate reports on birth defects or in reports on all births (Bourne & Kee 1998; Bower et al. 1996; Chan et al. 1997; Consultative Council on Obstetric and Paediatric Mortality and Morbidity 1997; Markey et al. 1996; Marsden (ed.) undated; Queensland Health 1998; Taylor et al. 1996). Birth Defect Registers have been established statewide in New South Wales, Victoria, South Australia, and Western Australia; the Northern Territory has a Birth Defect Section in their Department of Health and Community Medicine. Perinatal Data Collection Units, which also collect congenital malformation data, are located in Queensland, Tasmania and the Australian Capital Territory. The malformation rates in this report may differ from rates published by the States and Territories because of differences in the age criteria for inclusion of infants, differences in the criteria for including major and minor malformations and other birth defects, varying sources of data, and occasionally differences in coding practices. Comparisons of some congenital heart defects and other malformations diagnosed beyond the perinatal period are particularly affected by these factors.

The major source of notifications was the perinatal data collected on all births in each State and Territory (Table 2.3). Other important sources were perinatal death certificates and sometimes autopsy reports, and reports of chromosomal abnormalities from cytogenetics laboratories. Notifications from children's hospitals in New South Wales were no longer specified in the data after 1992, accounting for the sharp decline in the proportion of notifications from that source. The data source has not been well reported in electronic data; approximately one-third of all data received in 1996 did not indicate the source of the data.

The main anatomical systems in which major malformations occurred were the musculoskeletal and cardiovascular systems and genital organs (Table 2.4). The specific malformations contributing to these different systems are shown for Australia for births in 1981 to 1994, 1995 and 1996 (Table 2.5) and for each State and Territory (Tables 2.6, 2.7).

Comparison of total malformation rates by year and by State and Territory may be influenced both by the completeness of clinical detection and notification of major malformations and by the extent to which the various sources of notifications are used. The ascertainment of three relatively common malformations - congenital dislocation of the hip, ventricular septal defect, and hypospadias - may vary considerably, affecting total malformation rates. Congenital dislocation of the hip accounted for more

than 60% of musculoskeletal malformations, ventricular septal defect was the most frequently notified congenital heart defect, and 82% of malformations of the genital organs were due to hypospadias.
Most of the difference between the total malformation rates of 157.9 per 10,000 births in 1981-94 and 175.4 per 10,000 births in 1995 and 173.4 per 10,000 in 1996 could be attributed to the following changes in individual malformations rates: the reported rate of patent ductus arteriosus increased by 2.2 per 10,000, ventricular septal defect increased by 2.0 per 10,000, ostium secundum ASD by 2.0 per 10,000, hydronephrosis increased by 2.0 per 10,000 and hypospadias by 1.0 per 10,000; the reported rate of chromosomal abnormalities decreased by 2.5 per 10,000 (trisomy 21 by 1.8 per 10,000 and trisomy 18 by 1.0 per 10,000) between 1995 and 1996 (Table 2.5). These findings emphasise that the variations in total malformation rates, and in the rates of specific malformations known to have varying ascertainment, should be interpreted with caution.

The source of notification of malformations affects the completeness of reporting of various maternal and infant variables. When information is obtained from death certificates, cytogenetics laboratories or children's hospitals, missing data are more likely than when the perinatal data form is the source. The proportion with missing information among births notified to the NPSU varied between 0.5% for infant's sex and 9.5% for the maternal race (Table 2.8). The completeness of reporting of these variables can be improved by linking notifications from other sources with the perinatal data form for each birth.

2.2 Terminations of pregnancy

South Australia is the only State that has a legislative requirement for notification of terminations of pregnancy performed for any indication, including terminations after prenatal diagnosis of congenital malformations (Chan & Taylor 1991). Birth defects registers in New South Wales, Victoria, South Australia and Western Australia obtain information on termination of pregnancy and ascertainment has improved considerably in the past decade. Also, although the other States and Territories do not have birth defects registers, information on some terminations has been provided from cytogenetics laboratories and sometimes from other sources. All States and Territories provide the available information on terminations of pregnancy to the AIHW National Perinatal Statistics Unit, but the level of completeness of national data remains uncertain. By comparing the national trends, and the numbers reported by each State and Territory, some inferences can be drawn about the overall level of reporting.

For this report, terminations of pregnancy for fetal malformations are divided into two main groups - induced abortions performed at less than 20 weeks' gestation (or when gestational age was not stated) and induced births at 20-27 weeks' gestation. The latter group includes fetuses that have reached a gestational age of 20 weeks, at which registration of perinatal deaths and notification in State and Territory perinatal collections is required. If there is information indicating that these terminations occurred after prenatal diagnosis, this is recorded for each notification so that induced births can be distinguished from other non-induced stillbirths occurring at the same gestational age of 20-27 weeks.

In the four-year period from 1993 to 1996, the reported number of induced abortions increased from 455 in 1993 to 588 in 1994, and then declined to 506 in 1996 (Table 2.9). There were relatively fewer induced births, decreasing from 131 in 1993 to 21 in 1996. The induced births accounted for 22.4% of all reported terminations of pregnancy performed for fetal malformations in 1993 and 4.0% in 1996. The apparent fall in the number of induced births may be partly due to an increase in terminations of pregnancy where gestational age was not stated. These terminations are included with the induced abortions.

The most common indications for terminations of pregnancy were trisomy 21, other chromosomal abnormalities, neural tube defects (anencephalus, spina bifida and encephalocele) and cardiac defects (Table 2.9). Fetuses with multiple malformations are enumerated for each specific type of malformation and the numbers of malformed fetuses with single or multiple malformations, or malformation syndromes, are also given.

To show trends in the notified terminations and their relationship to births in the perinatal collections, both induced abortions and induced births are expressed as ratios per 10,000 births. The ratio of notified induced abortions for all fetal malformations varied in the period from 1993 to 1996, increasing from 17.5 per 10,000 births in 1993 to 22.5 per 10,000 in 1994, and then declining again to 19.7 per 10,000 in 1996. Reflecting their relatively smaller numbers, the ratio of notified induced births decreased from 5.0 per 10,000 births in 1993 to 0.8 per 10,000 in 1996. The overall ratio of notified terminations of pregnancy for fetal malformations decreased from 22.5 per 10,000 births in 1993 to 20.5 per 10,000 in 1996, indicating that there was about 1 termination of a malformed fetus for every 488 births in 1996. The reported decrease in induced abortions and births in 1995 and 1996 may reflect incomplete reporting of terminations, and induced births in particular.

In 1995 and 1996, there was a marked decrease in the number of reported terminations at all gestational ages and a considerable increase in those with an unspecified age (Table 2.10, Figure 2.1). The duration of pregnancy was not given for 40.0% of notified terminations in the period from 1993 to 1996, increasing from 13.8% in 1993 to 68.1% in 1996. Among terminations with known gestational ages, three-quarters (78.6%) were performed between 16 and 22 weeks and the modal week was either 18 or 19 weeks in different years.

Chromosomal abnormalities, particularly trisomy 21, accounted for the majority of terminations performed before 16 weeks (Table 2.11, Figure 2.2.). Terminations for anencephalus, spina bifida and other malformations detected by ultrasound or other methods of prenatal diagnosis were more likely to occur just before 20 weeks, when most pregnant women are screened by ultrasound examination.

2.2.1 Notifications of terminations of pregnancy in each State and Territory

As already noted, there is varying ascertainment of induced abortions and induced births in different States and Territories. Also, additional information from autopsy reports provided with notifications of terminations to the NPSU enables coding of prenatal diagnostic methods and the indications for terminations, particularly for those at gestational ages of 20 weeks or more. Active review of hospital records by staff of birth defects registers in some States, particularly in Victoria, is probably an important factor influencing comparisons of terminations between the States where there is no legal requirement to notify terminations.

The reported numbers of terminations are affected by these various factors and should not be regarded as the complete figures for any State or Territory (Table 2.12). Nevertheless, by comparing the reported numbers from the different States and Territories, valuable insights can be obtained for improving ascertainment and for reducing the deficiencies of the national data.

By comparing the ratios of terminations for the most recent year's data (1996), the effect of changing ascertainment on differences in ratios can be avoided to some extent. In 1996, for terminations of pregnancy at less than 20 weeks (including unstated gestational ages), the highest reported ratios were in Victoria (38.3 per 10,000 births), South Australia (31.9 per 10,000 births) and Western Australia (30.1 per 10,000 births) and the lowest were in the Australian Capital Territory (0.0 per 10,000 births), Northern Territory (0.0 per 10,000 births) and Queensland (5.8 per 10,000 births) (Table 2.13). For terminations at 20-27 weeks, the reported ratios in South Australia (7.8 per 10,000 births) were considerably higher than elsewhere. When data on induced abortions and induced births are combined, the highest reported ratios of terminations were in South Australia (39.8 per 10,000 births), Victoria (38.3 per 10,000 births) and Western Australia (30.1 per 10,000 births), and the lowest ratios were in the Northern Territory (2.9 per 10,000 births) and the Australian Capital Territory (0.0 per 10,000 births).

The gestational age distribution of the terminations of pregnancy showed considerable variation in the different States (Table 2.14, Figure 2.3). Because of small numbers, the data for Tasmania, the Australian Capital Territory and the Northern Territory were combined as 'other'.

As the forms used to obtain information about terminations of pregnancy often have limited data on maternal characteristics, there are substantial deficiencies in some variables such as maternal country of birth and Indigenous status. These variables may be important in analysing differences in malformation rates between the various population groups. In all States and Territories, a high proportion of notified induced abortions at less than 20 weeks lacked this information (Table 2.15). Also, the gestational age was not reported for 47.1% of these terminations. This information is needed to analyse the impact of different methods of prenatal diagnosis. For example, chorionic villus sampling is usually performed at an earlier stage of pregnancy than amniocentesis in screening for chromosomal abnormalities. Variations in the relative use of these two tests and the gestational ages at which they are done will affect interpretation of differences in rates.

2.3 Congenital malformations by maternal age

In the four-year period from 1993 to 1996, malformation rates among births were generally higher for younger and older mothers (Table 2.16). There was more variation by maternal age among infants with multiple malformations than for those with an isolated malformation. There was a pronounced association between advancing maternal age and an increasing rate of chromosomal abnormalities, ranging from 10.1 per 10,000 births for infants of mothers aged less than 20 years to 133.0 per 10,000 births for infants whose mothers were 40 years and over. There was much less variation by maternal age for other non-chromosomal syndromes. An earlier report on *Congenital Malformations Australia 1981-1992* (Lancaster & Pedisich 1995) gave data on the maternal age distribution of 25 selected major malformations.

For terminations at gestational ages of less than 20 weeks, the ratio of syndromes due to chromosomal abnormalities increased with advancing maternal age. Terminations for isolated malformations were also more likely with advancing maternal age. Terminations of fetuses with multiple malformations were slightly more likely for younger and older mothers.

2.4 Congenital malformations in singleton and multiple births

In 1993 to 1996, there were higher rates of isolated and multiple malformations in twins and other multiple births than in singleton births (Table 2.17). Singleton infants and twins had similar rates of chromosomal abnormalities, but there were higher rates of non-chromosomal syndromes in twins than in singleton births. More than twice as many multiple malformations were present in twins and other multiple births as in singleton births. Relatively few terminations for fetal malformations were performed in multiple pregnancies, so little is gained by comparing them with singleton pregnancies.

Figure 2.1: Terminations of pregnancy for fetal malformations, by gestational age, Australia, 1993-1996

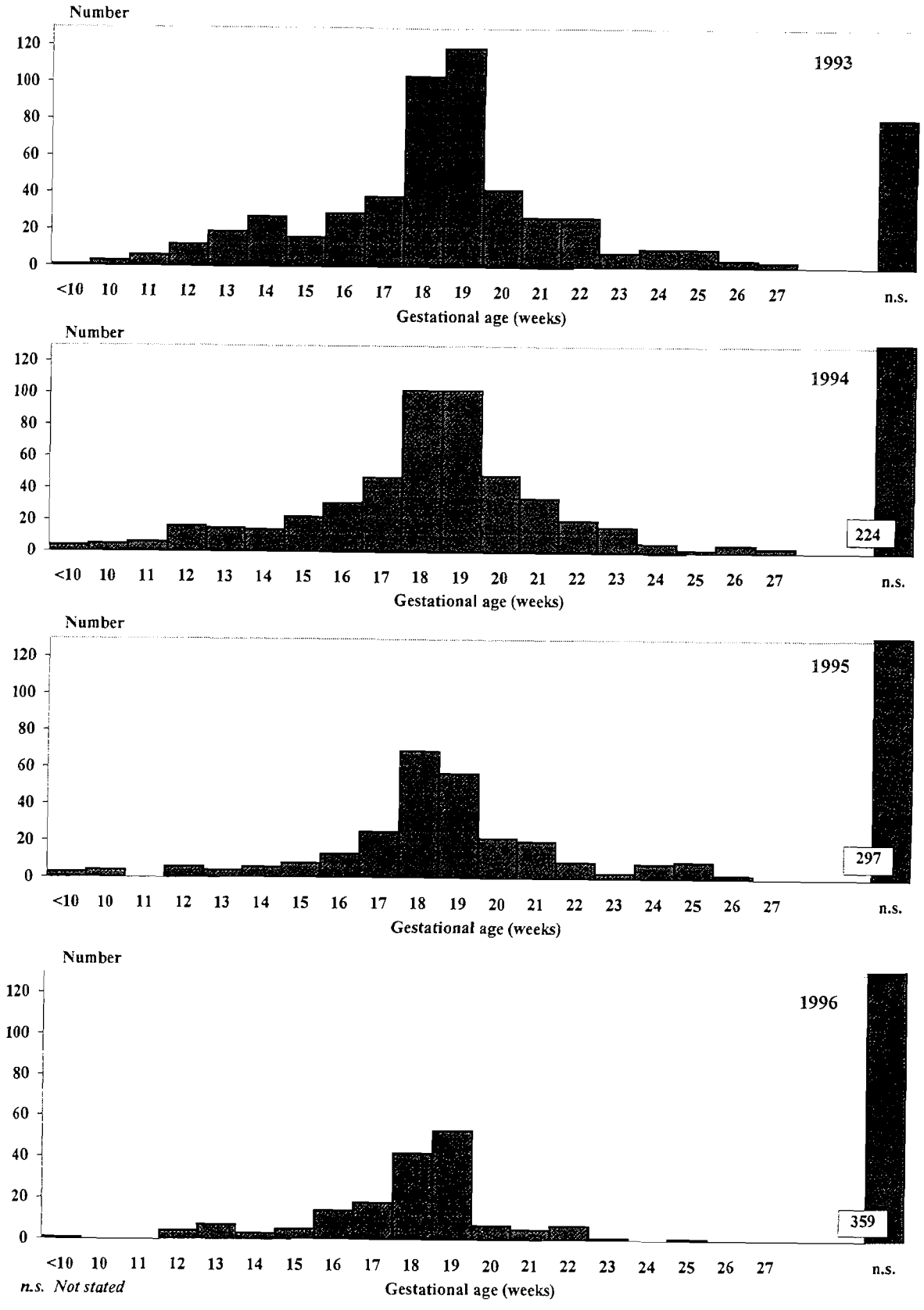
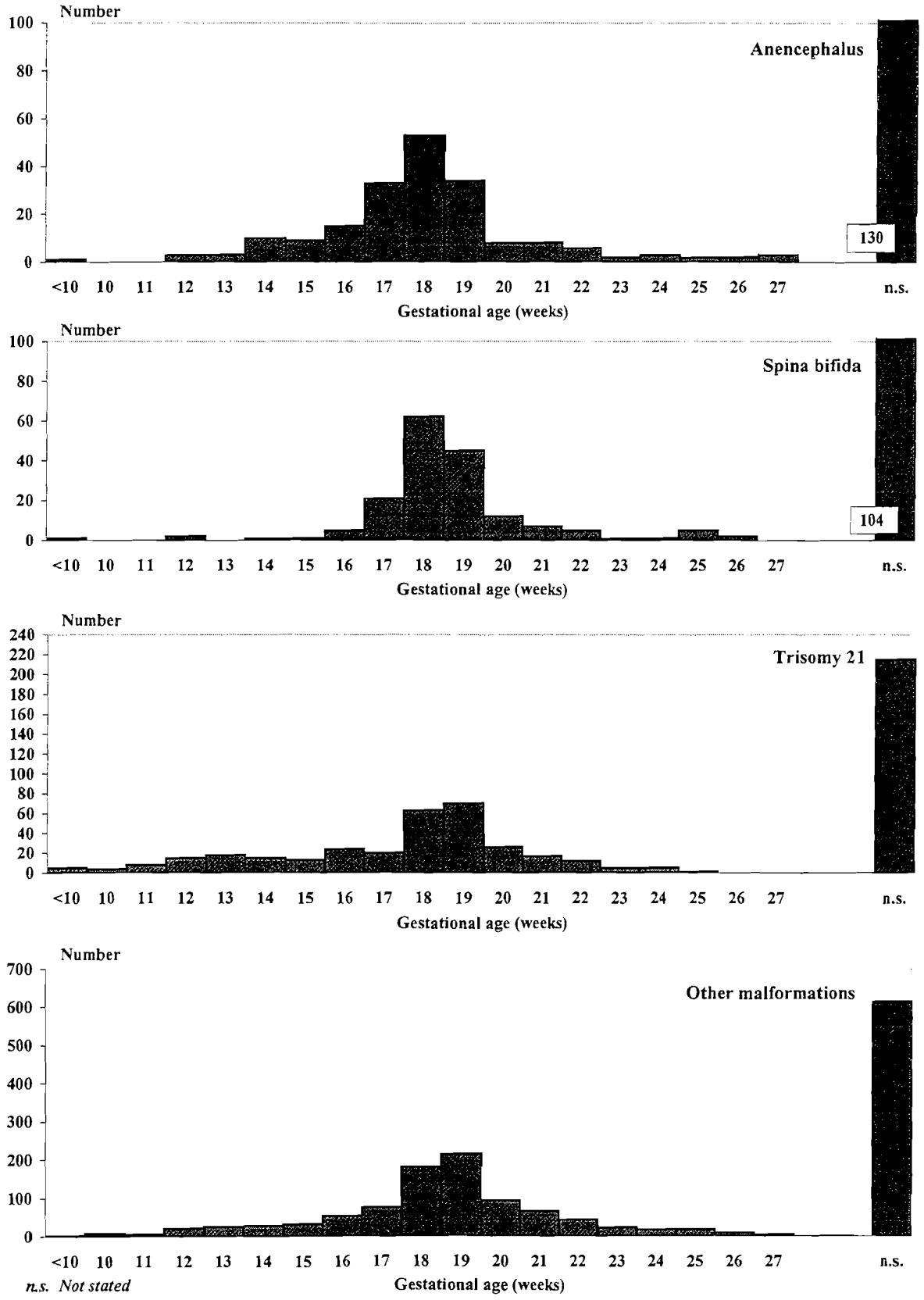
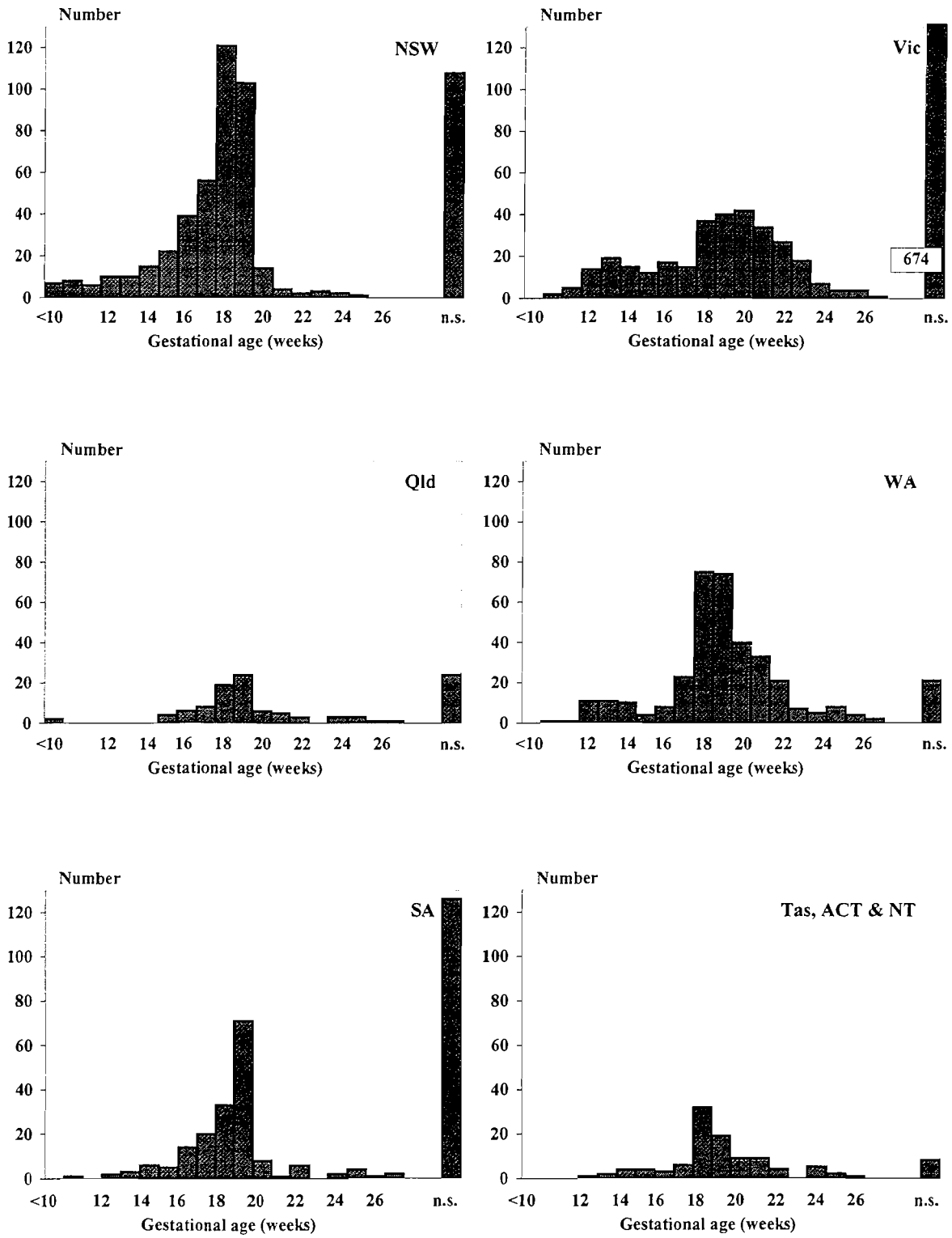


Figure 2.2: Terminations of pregnancy for fetal malformations, by type of malformation, Australia, 1993-1996



n.s. Not stated

Figure 2.3: Terminations of pregnancy for fetal malformations, by gestational age, selected States, 1993-1996



n.s. Not stated

Table 2.1: Single and multiple congenital malformations, Australia, 1981-1996

Type of malformation	1981-1994	1995	1996	1981-96	1981-1994	1995	1996	1981-96
	Number				Rate per 10,000 births			
All types	53,175	4,561	4,458	62,194	157.9	175.4	173.4	160.1
Single system	40,694	3,457	3,427	47,578	120.9	132.9	133.3	122.5
Multiple systems	3,902	304	308	4,514	11.6	11.7	12.0	11.6
- 2 systems	2,649	210	242	3,101	7.9	8.1	9.4	8.0
- 3+ systems	1,199	88	66	1,353	3.6	3.4	2.6	3.5
- unknown	54	6	-	60	0.2	0.2	-	0.2
Syndrome	8,579	800	723	10,102	25.5	30.8	28.1	26.0

Note: Data for 1981 exclude Vic, WA, ACT, NT and certain hospital in NSW; data for 1982-1985 exclude NT.

Table 2.2: Single and multiple congenital malformations by State or Territory of birth, 1991-1996

Type of malformation	NSW	Vic	Qld	WA	SA	Tas	ACT	NT	Australia
					Number				
All types	7,565	8,746	5,486	1,704	2,235	407	557	252	26,952
Single system	5,349	6,992	4,495	1,155	1,766	292	461	182	20,692
Multiple systems	569	543	236	167	160	24	16	29	1,744
- 2 systems	389	402	179	114	121	16	15	19	1,255
- 3+ systems	179	132	55	51	39	7	1	10	474
- unknown	1	9	2	2	-	1	-	-	15
Syndrome	1,647	1,211	755	382	309	91	80	41	4,516
					Rate per 10,000 births				
All types	143.9	225.5	193.6	112.0	188.8	98.7	195.4	118.2	172.9
Single system	101.8	180.3	158.6	75.9	149.2	70.8	161.7	85.4	132.8
Multiple systems	10.8	14.0	8.3	11.0	13.5	5.8	5.6	13.6	11.2
- 2 systems	7.4	10.4	6.3	7.5	10.2	3.9	5.3	8.9	8.1
- 3+ systems	3.4	3.4	1.9	3.4	3.3	1.7	0.4	4.7	3.0
- unknown	0.0	0.2	0.1	0.1	-	0.2	-	-	0.1
Syndrome	31.3	31.2	26.6	25.1	26.1	22.1	28.1	19.2	29.0

Table 2.3: Source of notification of congenital malformations, Australia, 1991-1996

Source	1991-94	1995	1996	1991-96	1991-94	1995	1996	1991-96
	Number				Per cent			
All sources	17,933	4,561	4,458	26,952	100.0	100.0	100.0	100.0
Birth notification	16,528	4,094	2,607	23,229	92.2	89.8	58.5	86.2
Referral hospital	2,565	116	297	2,978	14.3	2.5	6.7	11.0
Death certificate	1,101	48	187	1,336	6.1	1.1	4.2	5.0
Pathology report	779	55	65	899	4.3	1.2	1.5	3.3
Cytogenetics report	1,617	410	313	2,340	9.0	9.0	7.0	8.7
Other reports	1,369	149	418	1,936	7.6	3.3	9.4	7.2
Not stated	15	19	1,534	1,568	0.1	0.4	34.4	5.8

Table 2.4: Congenital malformations by major anatomical system, Australia, 1981-1996

Codes	Malformations	1981-94	1995	1996	1981-96	1981-94	1995	1996	1981-96
		Number				Rate per 10,000 births			
	All malformations	53,175	4,561	4,458	62,194	157.9	175.4	173.4	160.1
740-2	Nervous system	6,065	375	332	6,772	18.0	14.4	12.9	17.4
743	Eye	1,010	77	84	1,171	3.0	3.0	3.3	3.0
744	Ear, face & neck	408	60	57	525	1.2	2.3	2.2	1.4
745-6	Heart	9,945	817	906	11,668	29.5	31.4	35.2	30.0
747	Circulatory system	3,735	268	340	4,343	11.1	10.3	13.2	11.2
748	Respiratory system	962	73	84	1,119	2.9	2.8	3.3	2.9
749	Cleft palate/lip	4,982	431	382	5,795	14.8	16.6	14.9	14.9
750-1	Digestive system	3,858	273	271	4,402	11.5	10.5	10.5	11.3
752	Genital organs	8,204	662	685	9,551	24.4	25.5	26.6	24.6
753	Urinary system	4,630	558	566	5,754	13.8	21.5	22.0	14.8
755	Limbs	4,749	431	394	5,574	14.1	16.6	15.3	14.4
754, 6	Other musculoskeletal	11,799	844	818	13,461	35.0	32.5	31.8	34.7
757	Integument	418	102	80	600	1.2	3.9	3.1	1.5
758	Chromosomal	6,690	659	587	7,936	19.9	25.3	22.8	20.4
759	Other & unspecified	1,361	103	113	1,577	4.0	4.0	4.4	4.1
760	Maternal conditions	57	2	2	61	0.2	0.1	0.1	0.2

Note: Data for 1981 exclude Vic, WA, ACT, NT and certain hospital in NSW; data for 1982-1985 exclude NT.

Table 2.5: Selected congenital malformations, Australia, 1981-1996

Codes	Malformations	1981-94	1995	1996	1981-96	1981-94	1995	1996	1981-96
		Number				Rate per 10,000 births			
740	Anencephalus & similar anomalies	1,235	36	42	1,313	3.7	1.4	1.6	3.4
740.0	Anencephalus	1,061	31	35	1,127	3.2	1.2	1.4	2.9
740.1	Craniorachischisis	155	4	7	166	0.5	0.2	0.3	0.4
740.2	Iniencephaly	20	1	-	21	0.1	0.0	-	0.1
741	Spina bifida	2,015	92	77	2,184	6.0	3.5	3.0	5.6
742	Other nervous system	2,906	254	215	3,375	8.6	9.8	8.4	8.7
742.0	Encephalocele	393	19	14	426	1.2	0.7	0.5	1.1
742.1	Microcephalus	592	48	38	678	1.8	1.8	1.5	1.7
742.2	Brain reduction	667	63	55	785	2.0	2.4	2.1	2.0
742.3	Hydrocephalus	1,324	100	84	1,508	3.9	3.8	3.3	3.9
742.4-9	Other	274	55	53	382	0.8	2.1	2.1	1.0
743	Eye	1,010	77	84	1,171	3.0	3.0	3.3	3.0
743.0	Anophthalmos	98	8	3	109	0.3	0.3	0.1	0.3
743.1	Microphthalmos	311	14	10	335	0.9	0.5	0.4	0.9
743.2	Buphthalmos	73	8	6	87	0.2	0.3	0.2	0.2
743.3	Cataract & lens	297	27	28	352	0.9	1.0	1.1	0.9
743.32	Cataract	289	27	26	342	0.9	1.0	1.0	0.9
743.4-9	Other	371	28	44	443	1.1	1.1	1.7	1.1
744	Ear, face & neck	408	60	57	525	1.2	2.3	2.2	1.4
744.0	Ear-affecting hearing	291	31	26	348	0.9	1.2	1.0	0.9
744.00	Auditory canal a/s	245	21	21	287	0.7	0.8	0.8	0.7
744.01	Absent auricle	44	10	5	59	0.1	0.4	0.2	0.2
744.1-3	Other ear	61	23	24	108	0.2	0.9	0.9	0.3
744.4-9	Face & neck	68	6	10	84	0.2	0.2	0.4	0.2
745	Bulbus cordis & cardiac septal closure	7,925	666	729	9,320	23.5	25.6	28.4	24.0
745.0	Common truncus	233	11	16	260	0.7	0.4	0.6	0.7
745.1	Transpstn. grt. vessels	1,206	90	94	1,390	3.6	3.5	3.7	3.6
745.2	Tetralogy of Fallot	548	57	55	660	1.6	2.2	2.1	1.7
745.3	Common ventricle	213	9	15	237	0.6	0.3	0.6	0.6
745.4	Vent. septal defect	5,294	434	480	6,208	15.7	16.7	18.7	16.0
745.5	Ostium secundum ASD	1,550	120	169	1,839	4.6	4.6	6.6	4.7
745.6	Endocardial cushion	630	56	81	767	1.9	2.2	3.2	2.0
745.7-9	Other	45	4	6	55	0.1	0.2	0.2	0.1
746	Other heart	3,494	254	321	4,069	10.4	9.8	12.5	10.5
746.0	Pulmonary valve	1,298	94	102	1,494	3.9	3.6	4.0	3.8
746.00	atresia	437	22	29	488	1.3	0.8	1.1	1.3
746.01	stenosis	717	60	67	844	2.1	2.3	2.6	2.2
746.1	Tricuspid a/s	460	31	37	528	1.4	1.2	1.4	1.4
746.2	Ebstein anomaly	114	9	7	130	0.3	0.3	0.3	0.3
746.3-4	Aortic valve stenosis, insufficiency	417	33	52	502	1.2	1.3	2.0	1.3
746.5-6	Mitral st/insuffic.	262	20	33	315	0.8	0.8	1.3	0.8
746.7	Hypoplastic L heart	775	36	59	870	2.3	1.4	2.3	2.2
746.8	Other specified	546	57	67	670	1.6	2.2	2.6	1.7
746.9	Unspecified	263	21	24	308	0.8	0.8	0.9	0.8

Table 2.5: Selected congenital malformations, Australia, 1981-1996 (cont.)

Codes	Malformations	1981-94	1995	1996	1981-96	1981-94	1995	1996	1981-96
		Number				Rate per 10,000 births			
747	Circulatory	3,735	268	340	4,343	11.1	10.3	13.2	11.2
747.0	Pat. ductus arteriosus	2,319	155	212	2,686	6.9	6.0	8.2	6.9
747.1	Coarctation of aorta	973	69	58	1,100	2.9	2.7	2.3	2.8
747.2	Other aorta	392	28	35	455	1.2	1.1	1.4	1.2
747.3	Pulmonary artery	306	36	55	397	0.9	1.4	2.1	1.0
747.4	Great veins	376	18	16	410	1.1	0.7	0.6	1.1
747.42	Total anomalous pulm. venous return	222	13	13	248	0.7	0.5	0.5	0.6
747.6	Peripheral vascular	131	3	6	140	0.4	0.1	0.2	0.4
747.8	Other specified	40	6	7	53	0.1	0.2	0.3	0.1
747.9	Unspecified	9	1	2	12	0.0	0.0	0.1	0.0
748	Respiratory	962	73	84	1,119	2.9	2.8	3.3	2.9
748.0	Choanal atresia	344	33	21	398	1.0	1.3	0.8	1.0
748.1	Other nose	98	8	10	116	0.3	0.3	0.4	0.3
748.2-3	Larynx/ trach./ bronch.	192	9	19	220	0.6	0.3	0.7	0.6
748.4-6	Lung	345	27	37	409	1.0	1.0	1.4	1.1
748.8-9	Other respiratory	23	-	-	23	0.1	-	-	0.1
749	Cleft palate/ lip	4,982	431	382	5,795	14.8	16.6	14.9	14.9
749.0	Cleft palate	1,911	197	178	2,286	5.7	7.6	6.9	5.9
749.1	Cleft lip	1,052	99	78	1,229	3.1	3.8	3.0	3.2
749.2	Cleft palate + lip	2,017	135	126	2,278	6.0	5.2	4.9	5.9
750	Upper alimentary tract	1,103	80	71	1,254	3.3	3.1	2.8	3.2
750.3	TOF, oesophageal a/s	1,040	73	64	1,177	3.1	2.8	2.5	3.0
750.*	Other	72	9	7	88	0.2	0.3	0.3	0.2
751	Other digestive	2,947	206	209	3,362	8.8	7.9	8.1	8.7
751.1	Small intestine a/s	715	48	46	809	2.1	1.8	1.8	2.1
751.10	Duodenum a/s	437	29	29	495	1.3	1.1	1.1	1.3
751.11	Jejunum a/s	137	6	5	148	0.4	0.2	0.2	0.4
751.12	Ileum a/s	100	11	6	117	0.3	0.4	0.2	0.3
751.19	Unspecified a/s	59	3	6	68	0.2	0.1	0.2	0.2
751.2	Large intestine, rectum, anal canal a/s	1,122	82	82	1,286	3.3	3.2	3.2	3.3
751.20	Large intestine a/s	86	4	5	95	0.3	0.2	0.2	0.2
751.21-2	Rectum a/s	116	3	4	123	0.3	0.1	0.2	0.3
751.23-4	Anus a/s	978	76	74	1,128	2.9	2.9	2.9	2.9
751.3	Hirschsprung dis., etc	401	32	29	462	1.2	1.2	1.1	1.2
751.4	Intestinal fixation	403	19	18	440	1.2	0.7	0.7	1.1
751.5-9	Other digestive	522	37	56	615	1.6	1.4	2.2	1.6

Table 2.5: Selected congenital malformations, Australia, 1981-1996 (cont.)

Codes	Malformations	1981-94	1995	1996	1981-96	1981-94	1995	1996	1981-96
		Number				Rate per 10,000 births			
752	Genital organs	8,204	662	685	9,551	24.4	25.5	26.6	24.6
752.0-1	Ovaries/fallopian, etc	101	4	3	108	0.3	0.2	0.1	0.3
752.2-3	Uterus	189	8	7	204	0.6	0.3	0.3	0.5
752.4	Cerv., vagina, external	179	9	4	192	0.5	0.3	0.2	0.5
752.6	Hypospadias etc	7,052	590	615	8,257	20.9	22.7	23.9	21.3
752.60,3-5	Hypospadias	6,619	584	604	7,807	19.7	22.5	23.5	20.1
752.61	Epispadias	85	6	11	102	0.3	0.2	0.4	0.3
752.62	Chordee	981	-	-	981	2.9	-	-	2.5
752.7	Indeterminate sex, etc	517	47	42	606	1.5	1.8	1.6	1.6
752.74	Ambiguous genitalia	241	11	20	272	0.7	0.4	0.8	0.7
752.79	Indeterminate sex NOS	229	32	19	280	0.7	1.2	0.7	0.7
752.8	Other specified	444	16	27	487	1.3	0.6	1.1	1.3
752.9	Unspecified	22	1	2	25	0.1	0.0	0.1	0.1
753	Urinary	4,630	558	566	5,754	13.8	21.5	22.0	14.8
753.0	Renal agenesis/dysgen.	1,180	95	106	1,381	3.5	3.7	4.1	3.6
753.00	Bilateral	682	34	47	763	2.0	1.3	1.8	2.0
753.01	Unilateral	449	60	57	566	1.3	2.3	2.2	1.5
753.1	Cystic kidney disease	883	84	92	1,059	2.6	3.2	3.6	2.7
753.11-3	Polycystic	365	23	21	409	1.1	0.9	0.8	1.1
753.16	Multicystic	453	49	62	564	1.3	1.9	2.4	1.5
753.2	Obstructive defects renal pelvis/ureter	1,850	290	316	2,456	5.5	11.2	12.3	6.3
753.20	Hydronephrosis	1,171	190	238	1,599	3.5	7.3	9.3	4.1
753.21-9	Other	801	114	122	1,037	2.4	4.4	4.7	2.7
753.3	Other spec. kidney	556	62	68	686	1.7	2.4	2.6	1.8
753.32	Horseshoe kidney, etc	277	19	15	311	0.8	0.7	0.6	0.8
753.4	Other spec. ureter	226	35	23	284	0.7	1.3	0.9	0.7
753.5	Exstrophy of urinary bladder	114	10	6	130	0.3	0.4	0.2	0.3
753.6	Atresia/ stenosis of urethra, bladder neck	293	20	31	344	0.9	0.8	1.2	0.9
753.7	Urachus	33	1	1	35	0.1	0.0	0.0	0.1
753.8	Other bladder/ urethra	155	18	18	191	0.5	0.7	0.7	0.5
753.9	Unspecified	48	5	5	58	0.1	0.2	0.2	0.1
754	Certain musculoskeletal	7,386	481	488	8,355	21.9	18.5	19.0	21.5
754.30	Dislocation of hip	7,216	472	469	8,157	21.4	18.2	18.2	21.0
754.*	Other	178	11	19	208	0.5	0.4	0.7	0.5
755	Limb	4,749	431	394	5,574	14.1	16.6	15.3	14.4
755.0	Polydactyly	2,143	204	182	2,529	6.4	7.8	7.1	6.5
755.1	Syndactyly	846	77	66	989	2.5	3.0	2.6	2.5
755.2	Reduction, upper limb	1,140	94	87	1,321	3.4	3.6	3.4	3.4
755.3	Reduction, lower limb	531	42	35	608	1.6	1.6	1.4	1.6
755.4	Reduction, unspec limb	22	4	1	27	0.1	0.2	0.0	0.1
755.5	Other upper limb	362	37	39	438	1.1	1.4	1.5	1.1
755.6	Other lower limb	252	34	31	317	0.7	1.3	1.2	0.8
755.8	Other specified	244	20	23	287	0.7	0.8	0.9	0.7
755.80	Arthrogryposis multiplex congenita	194	16	20	230	0.6	0.6	0.8	0.6
755.9	Unspecified	12	-	1	13	0.0	-	0.0	0.0

Table 2.5: Selected congenital malformations, Australia, 1981-1996 (cont.)

Codes	Malformations	1981-94	1995	1996	1981-96	1981-94	1995	1996	1981-96
		Number				Rate per 10,000 births			
756	Other musculoskeletal	4,564	368	348	5,280	13.6	14.2	13.5	13.6
756.0	Skull, face & bones	1,000	82	72	1,154	3.0	3.2	2.8	3.0
756.00	Craniosynostosis	476	34	27	537	1.4	1.3	1.1	1.4
756.03	Pierre Robin synd.	310	33	28	371	0.9	1.3	1.1	1.0
756.1	Spine	606	58	52	716	1.8	2.2	2.0	1.8
756.3	Ribs & sternum	260	16	10	286	0.8	0.6	0.4	0.7
756.4	Chondrodystrophy	344	19	26	389	1.0	0.7	1.0	1.0
756.43	Achondroplasia	166	6	11	183	0.5	0.2	0.4	0.5
756.44	Other dwarfing synd.	117	4	7	128	0.3	0.2	0.3	0.3
756.5	Osteodystrophies	215	9	15	239	0.6	0.3	0.6	0.6
756.50	Osteogenesis imperfecta	155	5	10	170	0.5	0.2	0.4	0.4
756.6	Diaphragm	1,074	84	84	1,242	3.2	3.2	3.3	3.2
756.61	Diaphragmatic hernia	949	74	76	1,099	2.8	2.8	3.0	2.8
756.7	Abdominal wall	1,279	102	105	1,486	3.8	3.9	4.1	3.8
756.70	Exomphalos	713	42	42	797	2.1	1.6	1.6	2.1
756.71	Gastroschisis	417	52	55	524	1.2	2.0	2.1	1.3
756.8	Other specified	99	16	10	125	0.3	0.6	0.4	0.3
756.9	Unspecified	18	1	3	22	0.1	0.0	0.1	0.1
757	Integument	418	102	80	600	1.2	3.9	3.1	1.5
757.80	Cystic hygroma	253	32	24	309	0.8	1.2	0.9	0.8
758	Chromosomal	6,690	659	587	7,936	19.9	25.3	22.8	20.4
758.0	Trisomy 21 (Down)	4,107	361	312	4,780	12.2	13.9	12.1	12.3
758.1	Trisomy 13 (Patau)	312	25	25	362	0.9	1.0	1.0	0.9
758.2	Trisomy 18 (Edwards)	703	84	56	843	2.1	3.2	2.2	2.2
758.3	Autosomal deletion	280	29	46	355	0.8	1.1	1.8	0.9
758.5	Other autosomal	614	76	76	766	1.8	2.9	3.0	2.0
758.6	Turner syndrome	319	35	28	382	0.9	1.3	1.1	1.0
758.7	Klinefelter syndrome	113	13	17	143	0.3	0.5	0.7	0.4
758.8	Other sex chromosomes	236	35	25	296	0.7	1.3	1.0	0.8
758.9	Unspecified	31	1	2	34	0.1	0.0	0.1	0.1
759	Other & unspecified	1,361	103	113	1,577	4.0	4.0	4.4	4.1
759.0	Spleen	114	5	9	128	0.3	0.2	0.4	0.3
759.1	Adrenal gland	59	2	1	62	0.2	0.1	0.0	0.2
759.2	Other endocrine glands	130	14	14	158	0.4	0.5	0.5	0.4
759.3	Situs inversus	114	8	17	139	0.3	0.3	0.7	0.4
759.4	Conjoined twins	30	-	1	31	0.1	-	0.0	0.1
759.6	Hamartoses NEC	17	3	2	22	0.1	0.1	0.1	0.1
759.7	Multiple, so described	81	11	5	97	0.2	0.4	0.2	0.2
759.8	Other specified	879	63	68	1,010	2.6	2.4	2.6	2.6
759.9	Unspecified	16	1	1	18	0.0	0.0	0.0	0.0
760.2	Congenital rubella	23	1	2	26	0.1	0.0	0.1	0.1
760.70	Fetal hydantoin synd.	11	-	-	11	0.0	-	-	0.0
760.76	Fetal alcohol synd.	23	1	-	24	0.1	0.0	-	0.1

Note: Data for 1981 exclude Vic, WA, ACT, NT and certain hospital in NSW; data for 1982-1985 exclude NT.

Table 2.6: Selected congenital malformations, by State or Territory of birth, 1991-1996

Codes	Malformations	NSW	Vic	Qld	WA	SA	Tas	ACT	NT	Australia
						Number				
	Total									
	1995	1,227	1,530	945	229	392	51	143	44	4,561
	1996	1,151	1,501	977	265	352	33	139	40	4,458
	1991-96	7,565	8,746	5,486	1,704	2,235	407	557	252	26,952
740	Anencephalus									
	1995	5	14	8	5	3	-	1	-	36
	1996	13	14	9	4	1	-	-	1	42
	1991-96	73	82	66	42	9	2	5	9	288
741	Spina bifida									
	1995	23	29	21	9	6	2	1	1	92
	1996	20	26	19	1	4	2	3	2	77
	1991-96	200	166	128	64	31	13	9	9	620
742.0	Encephalocele									
	1995	9	6	2	1	1	-	-	-	19
	1996	5	5	2	1	1	-	-	-	14
	1991-96	40	45	13	13	9	1	2	1	124
742.1	Microcephalus									
	1995	17	20	8	2	1	-	-	-	48
	1996	14	6	11	4	1	-	2	-	38
	1991-96	81	69	43	22	12	1	3	5	236
742.3	Hydrocephalus									
	1995	24	38	21	7	4	2	2	2	100
	1996	23	33	17	5	-	3	2	1	84
	1991-96	160	199	101	44	25	16	15	8	568
745.1	Transposition of great vessels									
	1995	20	34	16	10	7	1	2	-	90
	1996	23	28	22	9	8	1	3	-	94
	1991-96	150	191	105	47	53	10	5	5	566
745.2	Tetralogy of Fallot									
	1995	17	19	9	-	7	-	4	1	57
	1996	16	17	7	2	5	3	2	3	55
	1991-96	108	125	45	23	25	3	10	10	349
745.4	Ventricular septal defect									
	1995	92	166	99	17	47	2	8	3	434
	1996	98	145	132	38	46	3	16	2	480
	1991-96	699	949	588	204	273	28	42	32	2,815
745.5	Ostium secundum atrial septal defect									
	1995	30	44	11	15	16	1	1	2	120
	1996	32	72	35	12	15	-	1	2	169
	1991-96	243	337	114	85	82	2	7	9	879

Table 2.6: Selected congenital malformations, by State or Territory of birth, 1991-1996 (cont.)

Codes	Malformations	NSW	Vic	Qld	WA	SA	Tas	ACT	NT	Australia
						Number				
746.00	Pulmonary valve atresia									
	1995	6	5	8	-	3	-	-	-	22
	1996	8	8	6	3	4	-	-	-	29
	1991-96	51	48	36	23	25	4	1	-	188
746.01	Pulmonary valve stenosis									
	1995	9	29	15	-	6	-	1	-	60
	1996	16	32	8	8	2	1	-	-	67
	1991-96	91	185	61	26	30	8	3	4	408
746.7	Hypoplastic left heart									
	1995	5	11	6	4	2	2	3	3	36
	1996	15	15	12	9	5	1	1	1	59
	1991-96	81	91	58	40	35	6	5	10	326
747.0	Patent ductus arteriosus									
	1995	49	75	13	9	6	1	1	1	155
	1996	61	94	19	19	8	1	9	1	212
	1991-96	429	449	102	101	59	7	18	4	1,169
747.1	Coarctation of aorta									
	1995	12	34	11	3	7	-	2	-	69
	1996	11	20	13	7	6	-	1	-	58
	1991-96	100	164	62	36	33	3	3	2	403
748.0	Choanal atresia									
	1995	2	23	3	3	-	1	1	-	33
	1996	8	8	2	2	1	-	-	-	21
	1991-96	45	79	19	13	10	3	2	1	172
749.0	Cleft palate									
	1995	73	59	38	5	13	4	2	3	197
	1996	56	47	33	22	15	-	4	1	178
	1991-96	357	266	165	82	75	20	16	13	994
749.1	Cleft lip									
	1995	36	19	18	10	13	1	2	-	99
	1996	37	13	12	6	6	2	2	-	78
	1991-96	171	145	93	33	44	10	10	1	507
749.2	Cleft palate + lip									
	1995	55	28	21	15	13	2	1	-	135
	1996	36	30	31	10	13	2	1	3	126
	1991-96	286	209	165	75	68	25	8	13	849
750.3	TOF, oesophageal atresia/stenosis									
	1995	12	23	18	9	9	1	1	-	73
	1996	17	24	7	4	9	2	1	-	64
	1991-96	130	139	85	34	59	13	3	2	465

Table 2.6: Selected congenital malformations, by State or Territory of birth, 1991-1996 (cont.)

Codes	Malformations	NSW	Vic	Qld	WA	SA	Tas	ACT	NT	Australia
						Number				
751.1	Small intestine atresia/stenosis									
	1995	15	7	15	4	7	-	-	-	48
	1996	12	20	6	4	2	1	1	-	46
	1991-96	91	87	69	28	35	5	3	1	319
751.2	Large intestine, rectum, anal canal atresia/stenosis									
	1995	28	31	10	6	5	2	-	-	82
	1996	24	26	8	8	12	-	2	2	82
	1991-96	146	162	79	48	49	3	2	7	496
751.3	Hirschsprung disease, etc									
	1995	6	17	5	-	2	-	1	1	32
	1996	7	9	9	2	1	-	1	-	29
	1991-96	44	89	30	19	12	1	4	1	200
752.60,	Hypospadias									
752.63-5	1995	177	192	112	33	48	3	16	3	584
	1996	146	215	132	41	53	2	10	5	604
	1991-96	1,021	1,149	632	259	296	45	72	14	3,488
752.7	Indeterminate sex, etc									
	1995	14	20	10	1	2	-	-	-	47
	1996	14	10	6	7	4	1	-	-	42
	1991-96	63	95	41	31	13	4	-	4	251
753.0	Renal agenesis/dysgenesis									
	1995	11	40	21	5	10	2	3	3	95
	1996	26	42	18	6	10	1	1	2	106
	1991-96	124	182	119	34	56	7	7	8	537
753.1	Cystic kidney disease									
	1995	12	31	20	8	8	3	1	1	84
	1996	20	36	20	5	5	2	3	1	92
	1991-96	120	158	96	42	49	10	6	14	495
753.2	Obstructive defects renal pelvis/ureter									
	1995	64	114	52	8	35	-	14	3	290
	1996	73	137	53	9	31	-	11	2	316
	1991-96	356	628	228	53	139	8	45	17	1,474
754.30	Dislocation of hip									
	1995	71	151	182	11	43	-	10	4	472
	1996	74	169	142	19	42	-	20	3	469
	1991-96	524	996	1,112	113	339	20	43	35	3,182

Table 2.6: Selected congenital malformations, by State or Territory of birth, 1991-1996 (cont.)

Codes	Malformations	NSW	Vic	Qld	WA	SA	Tas	ACT	NT	Australia
						Number				
755.0	Polydactyly									
	1995	81	59	25	10	18	1	6	4	204
	1996	66	53	31	8	15	1	7	1	182
	1991-96	437	334	182	55	92	10	22	5	1,137
755.1	Syndactyly									
	1995	25	23	15	-	12	1	1	-	77
	1996	8	34	12	3	7	-	1	1	66
	1991-96	118	134	86	19	54	5	5	1	422
755.2-4	Limb reduction									
	1995	35	38	25	7	14	4	2	2	127
	1996	31	36	16	13	11	-	2	1	110
	1991-96	247	179	142	50	71	12	8	10	719
756.00	Craniosynostosis									
	1995	9	20	4	-	1	-	-	-	34
	1996	5	16	3	1	1	-	-	1	27
	1991-96	88	92	18	5	16	-	-	1	220
756.4	Chondrodystrophy									
	1995	5	7	6	-	-	-	-	1	19
	1996	10	8	4	1	1	2	-	-	26
	1991-96	50	34	25	14	10	3	4	2	142
756.5	Osteodystrophies									
	1995	1	4	1	2	-	-	1	-	9
	1996	4	9	-	1	-	-	1	-	15
	1991-96	22	49	13	6	6	3	3	-	102
756.61	Diaphragmatic hernia									
	1995	17	32	13	6	2	3	1	-	74
	1996	22	23	14	11	5	1	-	-	76
	1991-96	121	139	95	43	28	14	5	-	445
756.70	Exomphalos									
	1995	14	11	10	2	4	-	1	-	42
	1996	18	10	10	1	1	-	1	1	42
	1991-96	86	69	48	27	24	5	4	2	265
756.71	Gastroschisis									
	1995	15	11	15	4	4	-	3	-	52
	1996	11	16	12	9	6	1	-	-	55
	1991-96	78	63	69	38	22	6	6	1	283

Table 2.6: Selected congenital malformations, by State or Territory of birth, 1991-1996 (cont.)

Codes	Malformations	NSW	Vic	Qld	WA	SA	Tas	ACT	NT	Australia
						Number				
758.0	Trisomy 21 (Down)									
	1995	148	85	58	29	25	7	6	3	361
	1996	132	55	75	22	16	2	9	1	312
	1991-96	757	494	379	160	121	44	44	16	2,015
758.1	Trisomy 13 (Patau)									
	1995	14	4	2	3	2	-	-	-	25
	1996	9	10	1	1	3	-	1	-	25
	1991-96	50	40	23	15	12	7	6	1	154
758.2	Trisomy 18 (Edwards)									
	1995	46	16	8	3	7	1	1	2	84
	1996	19	21	5	8	2	-	1	-	56
	1991-96	164	103	58	29	22	6	6	7	395
758.6	Turner syndrome									
	1995	16	10	5	3	1	-	-	-	35
	1996	14	4	6	1	3	-	-	-	28
	1991-96	85	48	29	14	11	2	1	1	191
758.3-5,	Other chromosomal									
758.7-9	1995	71	39	17	12	10	3	2	-	154
	1996	59	60	26	12	6	1	1	1	166
	1991-96	329	275	113	61	60	20	13	3	874

Table 2.7: Selected congenital malformation rates, by State or Territory of birth, 1991-1996

Codes	Malformations	NSW	Vic	Qld	WA	SA	Tas	ACT	NT	Australia
						Rate per 10,000 births				
	Total									
	1995	140.4	240.1	194.7	90.0	199.8	75.2	291.9	120.4	175.4
	1996	133.2	238.4	203.6	103.6	184.2	48.7	290.3	115.6	173.4
	1991-96	143.9	225.5	193.6	112.0	188.8	98.7	195.4	118.2	172.9
740	Anencephalus									
	1995	0.6	2.2	1.6	2.0	1.5	-	2.0	-	1.4
	1996	1.5	2.2	1.9	1.6	0.5	-	-	2.9	1.6
	1991-96	1.4	2.1	2.3	2.8	0.8	0.5	1.8	4.2	1.8
741	Spina bifida									
	1995	2.6	4.6	4.3	3.5	3.1	2.9	2.0	2.7	3.5
	1996	2.3	4.1	4.0	0.4	2.1	2.9	6.3	5.8	3.0
	1991-96	3.8	4.3	4.5	4.2	2.6	3.2	3.2	4.2	4.0
742.0	Encephalocele									
	1995	1.0	0.9	0.4	0.4	0.5	-	-	-	0.7
	1996	0.6	0.8	0.4	0.4	0.5	-	-	-	0.5
	1991-96	0.8	1.2	0.5	0.9	0.8	0.2	0.7	0.5	0.8
742.1	Microcephalus									
	1995	1.9	3.1	1.6	0.8	0.5	-	-	-	1.8
	1996	1.6	1.0	2.3	1.6	0.5	-	4.2	-	1.5
	1991-96	1.5	1.8	1.5	1.4	1.0	0.2	1.1	2.3	1.5
742.3	Hydrocephalus									
	1995	2.7	6.0	4.3	2.8	2.0	2.9	4.1	5.5	3.8
	1996	2.7	5.2	3.5	2.0	-	4.4	4.2	2.9	3.3
	1991-96	3.0	5.1	3.6	2.9	2.1	3.9	5.3	3.8	3.6
745.1	Transposition of great vessels									
	1995	2.3	5.3	3.3	3.9	3.6	1.5	4.1	-	3.5
	1996	2.7	4.4	4.6	3.5	4.2	1.5	6.3	-	3.7
	1991-96	2.9	4.9	3.7	3.1	4.5	2.4	1.8	2.3	3.6
745.2	Tetralogy of Fallot									
	1995	1.9	3.0	1.9	-	3.6	-	8.2	2.7	2.2
	1996	1.9	2.7	1.5	0.8	2.6	4.4	4.2	8.7	2.1
	1991-96	2.1	3.2	1.6	1.5	2.1	0.7	3.5	4.7	2.2
745.4	Ventricular septal defect									
	1995	10.5	26.1	20.4	6.7	24.0	2.9	16.3	8.2	16.7
	1996	11.3	23.0	27.5	14.9	24.1	4.4	33.4	5.8	18.7
	1991-96	13.3	24.5	20.7	13.4	23.1	6.8	14.7	15.0	18.1
745.5	Ostium secundum atrial septal defect									
	1995	3.4	6.9	2.3	5.9	8.2	1.5	2.0	5.5	4.6
	1996	3.7	11.4	7.3	4.7	7.8	-	2.1	5.8	6.6
	1991-96	4.6	8.7	4.0	5.6	6.9	0.5	2.5	4.2	5.6

Table 2.7: Selected congenital malformation rates, by State or Territory of birth, 1991-1996 (cont.)

Codes	Malformations	NSW	Vic	Qld	WA	SA	Tas	ACT	NT	Australia
						Rate per 10,000 births				
746.00	Pulmonary valve atresia									
	1995	0.7	0.8	1.6	-	1.5	-	-	-	0.8
	1996	0.9	1.3	1.3	1.2	2.1	-	-	-	1.1
	1991-96	1.0	1.2	1.3	1.5	2.1	1.0	0.4	-	1.2
746.01	Pulmonary valve stenosis									
	1995	1.0	4.6	3.1	-	3.1	-	2.0	-	2.3
	1996	1.9	5.1	1.7	3.1	1.0	1.5	-	-	2.6
	1991-96	1.7	4.8	2.2	1.7	2.5	1.9	1.1	1.9	2.6
746.7	Hypoplastic left heart									
	1995	0.6	1.7	1.2	1.6	1.0	2.9	6.1	8.2	1.4
	1996	1.7	2.4	2.5	3.5	2.6	1.5	2.1	2.9	2.3
	1991-96	1.5	2.3	2.0	2.6	3.0	1.5	1.8	4.7	2.1
747.0	Patent ductus arteriosus									
	1995	5.6	11.8	2.7	3.5	3.1	1.5	2.0	2.7	6.0
	1996	7.1	14.9	4.0	7.4	4.2	1.5	18.8	2.9	8.2
	1991-96	8.2	11.6	3.6	6.6	5.0	1.7	6.3	1.9	7.5
747.1	Coarctation of aorta									
	1995	1.4	5.3	2.3	1.2	3.6	-	4.1	-	2.7
	1996	1.3	3.2	2.7	2.7	3.1	-	2.1	-	2.3
	1991-96	1.9	4.2	2.2	2.4	2.8	0.7	1.1	0.9	2.6
748.0	Choanal atresia									
	1995	0.2	3.6	0.6	1.2	-	1.5	2.0	-	1.3
	1996	0.9	1.3	0.4	0.8	0.5	-	-	-	0.8
	1991-96	0.9	2.0	0.7	0.9	0.8	0.7	0.7	0.5	1.1
749.0	Cleft palate									
	1995	8.4	9.3	7.8	2.0	6.6	5.9	4.1	8.2	7.6
	1996	6.5	7.5	6.9	8.6	7.8	-	8.4	2.9	6.9
	1991-96	6.8	6.9	5.8	5.4	6.3	4.9	5.6	6.1	6.4
749.1	Cleft lip									
	1995	4.1	3.0	3.7	3.9	6.6	1.5	4.1	-	3.8
	1996	4.3	2.1	2.5	2.3	3.1	2.9	4.2	-	3.0
	1991-96	3.3	3.7	3.3	2.2	3.7	2.4	3.5	0.5	3.3
749.2	Cleft palate + lip									
	1995	6.3	4.4	4.3	5.9	6.6	2.9	2.0	-	5.2
	1996	4.2	4.8	6.5	3.9	6.8	2.9	2.1	8.7	4.9
	1991-96	5.4	5.4	5.8	4.9	5.7	6.1	2.8	6.1	5.4
750.3	TOF, oesophageal atresia/stenosis									
	1995	1.4	3.6	3.7	3.5	4.6	1.5	2.0	-	2.8
	1996	2.0	3.8	1.5	1.6	4.7	2.9	2.1	-	2.5
	1991-96	2.5	3.6	3.0	2.2	5.0	3.2	1.1	0.9	3.0

Table 2.7: Selected congenital malformation rates, by State or Territory of birth, 1991-1996 (cont.)

Codes	Malformations	NSW	Vic	Qld	WA	SA	Tas	ACT	NT	Australia
						Rate per 10,000 births				
751.1	Small intestine atresia/stenosis									
	1995	1.7	1.1	3.1	1.6	3.6	-	-	-	1.8
	1996	1.4	3.2	1.3	1.6	1.0	1.5	2.1	-	1.8
	1991-96	1.7	2.2	2.4	1.8	3.0	1.2	1.1	0.5	2.0
751.2	Large intestine, rectum, anal canal atresia/stenosis									
	1995	3.2	4.9	2.1	2.4	2.5	2.9	-	-	3.2
	1996	2.8	4.1	1.7	3.1	6.3	-	4.2	5.8	3.2
	1991-96	2.8	4.2	2.8	3.2	4.1	0.7	0.7	3.3	3.2
751.3	Hirschsprung disease, etc									
	1995	0.7	2.7	1.0	-	1.0	-	2.0	2.7	1.2
	1996	0.8	1.4	1.9	0.8	0.5	-	2.1	-	1.1
	1991-96	0.8	2.3	1.1	1.2	1.0	0.2	1.4	0.5	1.3
752.60,	Hypospadias									
752.63-5	1995	20.3	30.1	23.1	13.0	24.5	4.4	32.7	8.2	22.5
	1996	16.9	34.2	27.5	16.0	27.7	2.9	20.9	14.4	23.5
	1991-96	19.4	29.6	22.3	17.0	25.0	10.9	25.3	6.6	22.4
752.7	Indeterminate sex, etc									
	1995	1.6	3.1	2.1	0.4	1.0	-	-	-	1.8
	1996	1.6	1.6	1.3	2.7	2.1	1.5	-	-	1.6
	1991-96	1.2	2.4	1.4	2.0	1.1	1.0	-	1.9	1.6
753.0	Renal agenesis/dysgenesis									
	1995	1.3	6.3	4.3	2.0	5.1	2.9	6.1	8.2	3.7
	1996	3.0	6.7	3.8	2.3	5.2	1.5	2.1	5.8	4.1
	1991-96	2.4	4.7	4.2	2.2	4.7	1.7	2.5	3.8	3.4
753.1	Cystic kidney disease									
	1995	1.4	4.9	4.1	3.1	4.1	4.4	2.0	2.7	3.2
	1996	2.3	5.7	4.2	2.0	2.6	2.9	6.3	2.9	3.6
	1991-96	2.3	4.1	3.4	2.8	4.1	2.4	2.1	6.6	3.2
753.2	Obstructive defects renal pelvis/ureter									
	1995	7.3	17.9	10.7	3.1	17.8	-	28.6	8.2	11.2
	1996	8.4	21.8	11.0	3.5	16.2	-	23.0	5.8	12.3
	1991-96	6.8	16.2	8.0	3.5	11.7	1.9	15.8	8.0	9.5
754.30	Dislocation of hip									
	1995	8.1	23.7	37.5	4.3	21.9	-	20.4	10.9	18.2
	1996	8.6	26.8	29.6	7.4	22.0	-	41.8	8.7	18.2
	1991-96	10.0	25.7	39.2	7.4	28.6	4.9	15.1	16.4	20.4

Table 2.7: Selected congenital malformation rates, by State or Territory of birth, 1991-1996 (cont.)

Codes	Malformations	NSW	Vic	Qld	WA	SA	Tas	ACT	NT	Australia
						Rate per 10,000 births				
755.0	Polydactyly									
	1995	9.3	9.3	5.2	3.9	9.2	1.5	12.2	10.9	7.8
	1996	7.6	8.4	6.5	3.1	7.8	1.5	14.6	2.9	7.1
	1991-96	8.3	8.6	6.4	3.6	7.8	2.4	7.7	2.3	7.3
755.1	Syndactyly									
	1995	2.9	3.6	3.1	-	6.1	1.5	2.0	-	3.0
	1996	0.9	5.4	2.5	1.2	3.7	-	2.1	2.9	2.6
	1991-96	2.2	3.5	3.0	1.2	4.6	1.2	1.8	0.5	2.7
755.2-4	Limb reduction									
	1995	4.0	6.0	5.2	2.8	7.1	5.9	4.1	5.5	4.9
	1996	3.6	5.7	3.3	5.1	5.8	-	4.2	2.9	4.3
	1991-96	4.7	4.6	5.0	3.3	6.0	2.9	2.8	4.7	4.6
756.00	Craniosynostosis									
	1995	1.0	3.1	0.8	-	0.5	-	-	-	1.3
	1996	0.6	2.5	0.6	0.4	0.5	-	-	2.9	1.1
	1991-96	1.7	2.4	0.6	0.3	1.4	-	-	0.5	1.4
756.4	Chondrodystrophy									
	1995	0.6	1.1	1.2	-	-	-	-	2.7	0.7
	1996	1.2	1.3	0.8	0.4	0.5	2.9	-	-	1.0
	1991-96	1.0	0.9	0.9	0.9	0.8	0.7	1.4	0.9	0.9
756.5	Osteodystrophies									
	1995	0.1	0.6	0.2	0.8	-	-	2.0	-	0.3
	1996	0.5	1.4	-	0.4	-	-	2.1	-	0.6
	1991-96	0.4	1.3	0.5	0.4	0.5	0.7	1.1	-	0.7
756.61	Diaphragmatic hernia									
	1995	1.9	5.0	2.7	2.4	1.0	4.4	2.0	-	2.8
	1996	2.5	3.7	2.9	4.3	2.6	1.5	-	-	3.0
	1991-96	2.3	3.6	3.4	2.8	2.4	3.4	1.8	-	2.9
756.70	Exomphalos									
	1995	1.6	1.7	2.1	0.8	2.0	-	2.0	-	1.6
	1996	2.1	1.6	2.1	0.4	0.5	-	2.1	2.9	1.6
	1991-96	1.6	1.8	1.7	1.8	2.0	1.2	1.4	0.9	1.7
756.71	Gastroschisis									
	1995	1.7	1.7	3.1	1.6	2.0	-	6.1	-	2.0
	1996	1.3	2.5	2.5	3.5	3.1	1.5	-	-	2.1
	1991-96	1.5	1.6	2.4	2.5	1.9	1.5	2.1	0.5	1.8

Table 2.7: Selected congenital malformation rates, by State or Territory of birth, 1991-1996 (cont.)

Codes	Malformations	NSW	Vic	Qld	WA	SA	Tas	ACT	NT	Australia
						Rate per 10,000 births				
758.0	Trisomy 21 (Down)									
	1995	16.9	13.3	12.0	11.4	12.7	10.3	12.2	8.2	13.9
	1996	15.3	8.7	15.6	8.6	8.4	2.9	18.8	2.9	12.1
	1991-96	14.4	12.7	13.4	10.5	10.2	10.7	15.4	7.5	12.9
758.1	Trisomy 13 (Patau)									
	1995	1.6	0.6	0.4	1.2	1.0	-	-	-	1.0
	1996	1.0	1.6	0.2	0.4	1.6	-	2.1	-	1.0
	1991-96	1.0	1.0	0.8	1.0	1.0	1.7	2.1	0.5	1.0
758.2	Trisomy 18 (Edwards)									
	1995	5.3	2.5	1.6	1.2	3.6	1.5	2.0	5.5	3.2
	1996	2.2	3.3	1.0	3.1	1.0	-	2.1	-	2.2
	1991-96	3.1	2.7	2.0	1.9	1.9	1.5	2.1	3.3	2.5
758.6	Turner syndrome									
	1995	1.8	1.6	1.0	1.2	0.5	-	-	-	1.3
	1996	1.6	0.6	1.3	0.4	1.6	-	-	-	1.1
	1991-96	1.6	1.2	1.0	0.9	0.9	0.5	0.4	0.5	1.2
758.3-5,	Other chromosomal									
758.7-9	1995	8.1	6.1	3.5	4.7	5.1	4.4	4.1	-	5.9
	1996	6.8	9.5	5.4	4.7	3.1	1.5	2.1	2.9	6.5
	1991-96	6.3	7.1	4.0	4.0	5.1	4.9	4.6	1.4	5.6

Table 2.8: Proportion of notified births with missing information, 1993-1996

Characteristic	Births								
	NSW	Vic	Qld	WA	SA	Tas	ACT	NT	Australia
					Per cent				
Maternal age	7.4	1.0	2.4	3.4	0.5	12.9	1.1	3.0	3.3
Maternal country of birth	0.4	0.5	0.6	24.8	1.5	4.6	0.2	2.4	2.1
Aboriginality	15.9	2.5	3.3	48.6	4.0	19.4	5.8	3.6	9.5
Plurality	2.9	0.7	0.9	1.0	-	5.1	-	0.6	1.3
Infant's sex	1.1	0.1	0.4	0.1	0.3	0.9	0.2	1.2	0.5
Birthweight	13.2	1.7	2.5	5.8	3.2	14.3	7.6	4.2	5.7
Gestational age	10.4	2.5	2.0	47.6	0.9	12.9	6.5	6.6	7.4

Table 2.9: Terminations of pregnancy for fetal malformations, Australia, 1993-1996

Congenital malformation	Induced abortions* (less than 20 weeks)				Induced births (20-27 weeks)				All terminations (up to 27 weeks)			
	1993	1994	1995	1996	1993	1994	1995	1996	1993	1994	1995	1996
						Number						
Anencephalus	57	85	79	70	23	9	2	-	80	94	81	70
Spina bifida	51	71	55	65	17	7	8	1	68	78	63	66
Encephalocele	9	15	10	5	3	2	2	1	12	17	12	6
Hydrocephalus	30	32	32	25	14	18	6	-	44	50	38	25
Cardiac defects	39	55	60	61	32	40	13	8	71	95	73	69
Renal abnormalities	15	19	16	10	7	10	7	2	22	29	23	12
Limb reduction defects	11	16	17	15	8	5	1	1	19	21	18	16
Diaphragmatic hernia	6	8	8	8	4	3	2	-	10	11	10	8
Exomphalos	24	23	21	17	5	2	4	2	29	25	25	19
Trisomy 21	118	130	106	116	20	25	17	4	138	155	123	120
Trisomy 13	22	16	26	12	6	7	2	2	28	23	28	14
Trisomy 18	34	47	46	45	9	10	8	-	43	57	54	45
Other autosomal	43	48	30	33	8	12	7	-	51	60	37	33
Gonadal dysgenesis	18	26	24	28	2	8	1	1	20	34	25	29
Sex chromosomal	16	22	16	18	2	2	3	1	18	24	19	19
Other	140	189	172	168	50	59	29	16	190	248	201	184
Single	135	214	174	183	62	54	23	6	197	268	197	189
Multiple	51	66	57	47	14	10	8	4	65	76	65	51
Syndrome	269	308	261	276	55	70	42	11	324	378	303	287
All fetuses	455	588	492	506	131	134	73	21	586	722	565	527
						Ratio per 10,000 births						
Anencephalus	2.2	3.3	3.0	2.7	0.9	0.3	0.1	-	3.1	3.6	3.1	2.7
Spina bifida	2.0	2.7	2.1	2.5	0.7	0.3	0.3	0.0	2.6	3.0	2.4	2.6
Encephalocele	0.3	0.6	0.4	0.2	0.1	0.1	0.1	0.0	0.5	0.7	0.5	0.2
Hydrocephalus	1.2	1.2	1.2	1.0	0.5	0.7	0.2	-	1.7	1.9	1.5	1.0
Cardiac defects	1.5	2.1	2.3	2.4	1.2	1.5	0.5	0.3	2.7	3.6	2.8	2.7
Renal abnormalities	0.6	0.7	0.6	0.4	0.3	0.4	0.3	0.1	0.8	1.1	0.9	0.5
Limb reduction defects	0.4	0.6	0.7	0.6	0.3	0.2	0.0	0.0	0.7	0.8	0.7	0.6
Diaphragmatic hernia	0.2	0.3	0.3	0.3	0.2	0.1	0.1	-	0.4	0.4	0.4	0.3
Exomphalos	0.9	0.9	0.8	0.7	0.2	0.1	0.2	0.1	1.1	1.0	1.0	0.7
Trisomy 21	4.5	5.0	4.1	4.5	0.8	1.0	0.7	0.2	5.3	5.9	4.7	4.7
Trisomy 13	0.8	0.6	1.0	0.5	0.2	0.3	0.1	0.1	1.1	0.9	1.1	0.5
Trisomy 18	1.3	1.8	1.8	1.8	0.3	0.4	0.3	-	1.7	2.2	2.1	1.8
Other autosomal	1.7	1.8	1.2	1.3	0.3	0.5	0.3	-	2.0	2.3	1.4	1.3
Gonadal dysgenesis	0.7	1.0	0.9	1.1	0.1	0.3	0.0	0.0	0.8	1.3	1.0	1.1
Sex chromosomal	0.6	0.8	0.6	0.7	0.1	0.1	0.1	0.0	0.7	0.9	0.7	0.7
Other	5.4	7.2	6.6	6.5	1.9	2.3	1.1	0.6	7.3	9.5	7.7	7.2
Single	5.2	8.2	6.7	7.1	2.4	2.1	0.9	0.2	7.6	10.3	7.6	7.4
Multiple	2.0	2.5	2.2	1.8	0.5	0.4	0.3	0.2	2.5	2.9	2.5	2.0
Syndrome	10.3	11.8	10.0	10.7	2.1	2.7	1.6	0.4	12.4	14.5	11.7	11.2
All fetuses	17.5	22.5	18.9	19.7	5.0	5.1	2.8	0.8	22.5	27.6	21.7	20.5

* *Includes terminations at unstated gestational ages*

Table 2.10: Terminations of pregnancy for fetal malformations by gestational age, Australia, 1993-1996

Years	Gestational age (weeks)										
	<10	10	11	12	13	14	15	16	17	18	19
1993	1	3	6	12	19	27	16	29	38	104	119
1994	4	5	6	16	15	14	22	31	47	102	102
1995	3	4	-	6	4	6	8	13	25	69	57
1996	1	-	-	4	7	3	5	14	18	42	53
All years	9	12	12	38	45	50	51	87	128	317	331

	20	21	22	23	24	25	26	27	Not stated	All terminations
1993	42	27	27	8	10	10	4	3	81	586
1994	48	34	20	16	6	2	5	3	224	722
1995	22	20	9	3	8	9	2	-	297	565
1996	7	5	7	1	-	1	-	-	359	527
All years	119	86	63	28	24	22	11	6	961	2,400

Table 2.11: Terminations of pregnancy for selected malformations by gestational age, Australia, 1993-1996

Malformation	Gestational age (weeks)										
	<10	10	11	12	13	14	15	16	17	18	19
Anencephalus	1	-	-	3	3	10	9	15	33	53	34
Spina bifida	1	-	-	2	-	1	1	5	21	62	45
Trisomy 21	5	4	8	15	18	15	13	24	20	63	70
Other malformations	2	8	5	21	26	28	32	54	76	181	215
All terminations	9	12	12	38	45	50	51	87	128	317	331

	20	21	22	23	24	25	26	27	Not stated	All terminations
Anencephalus	8	8	6	2	3	2	2	3	130	325
Spina bifida	12	7	5	1	1	5	2	-	104	275
Trisomy 21	26	17	12	5	5	1	-	-	215	536
Other malformations	93	65	44	23	18	17	9	3	614	1,534
All terminations	119	86	63	28	24	22	11	6	961	2,400

Table 2.12: Terminations of pregnancy for fetal malformations, States and Territories, 1993-1996

State / Territory	Induced abortions* (less than 20 weeks)				Induced births (20-27 weeks)				All terminations (up to 27 weeks)			
	1993	1994	1995	1996	1993	1994	1995	1996	1993	1994	1995	1996
	Number											
New South Wales	133	154	126	92	6	11	7	2	139	165	133	94
Victoria	149	231	229	241	72	60	5	-	221	291	234	241
Queensland	19	16	24	28	7	4	9	2	26	20	33	30
Western Australia	70	91	43	77	14	10	-	-	84	101	43	77
South Australia	59	58	61	61	25	36	44	15	84	94	105	76
Tasmania	12	20	5	7	1	2	4	1	13	22	9	8
Aust. Capital Territory	9	14	4	-	5	8	-	-	14	22	4	-
Northern Territory	4	4	-	-	1	3	4	1	5	7	4	1
Australia	455	588	492	506	131	134	73	21	586	722	565	527

** Includes terminations of unstated gestational ages*

Table 2.13: Ratios of terminations of pregnancy for fetal malformations, States and Territories, 1996

State / Territory	Terminations of pregnancy			Total births	Ratio of TOPs per 10,000 births		
	<20	20+	All TOPs		<20	20+	All TOPs
	Number				Ratio		
New South Wales	92	2	94	86,429	10.6	0.2	10.9
Victoria	241	-	241	62,951	38.3	-	38.3
Queensland	28	2	30	47,987	5.8	0.4	6.3
Western Australia	77	-	77	25,584	30.1	-	30.1
South Australia	61	15	76	19,111	31.9	7.8	39.8
Tasmania	7	1	8	6,781	10.3	1.5	11.8
Australian Capital Territory	-	-	-	4,788	-	-	-
Northern Territory	-	1	1	3,461	-	2.9	2.9
Australia	506	21	527	257,092	19.7	0.8	20.5

TOPs: Terminations of pregnancy

Table 2.14: Terminations of pregnancy for fetal malformations by States, Australia, 1993-1996

State	Gestational age (weeks)										
	<10	10	11	12	13	14	15	16	17	18	19
New South Wales	7	8	6	10	10	15	22	39	56	121	103
Victoria	-	2	5	14	19	15	12	17	15	37	40
Queensland	2	-	-	-	-	-	4	6	8	19	24
Western Australia	-	1	-	2	3	6	5	14	20	33	71
South Australia	-	1	1	11	11	10	4	8	23	75	74
Other	-	-	-	1	2	4	4	3	6	32	19
Australia	9	12	12	38	45	50	51	87	128	317	331

State	20	21	22	23	24	25	26	27	Not stated	All terminations
New South Wales	14	4	2	3	2	1	-	-	108	531
Victoria	42	34	27	18	7	4	4	1	674	987
Queensland	6	5	3	-	3	3	1	1	24	109
Western Australia	8	1	6	-	2	4	1	2	126	305
South Australia	40	33	21	7	5	8	4	2	21	359
Other	9	9	4	-	5	2	1	-	8	109
Australia	119	86	63	28	24	22	11	6	961	2,400

Table 2.15: Proportion of notified terminations of pregnancy with missing information, 1993-1996

Characteristic	Terminations of pregnancy (<20 weeks)								
	NSW	Vic	Qld	WA	SA	Tas	ACT	NT	Australia
	Per cent								
Maternal age	8.1	2.0	23.0	3.6	0.8	-	-	-	4.4
Maternal country of birth	0.6	55.3	29.9	27.4	25.5	20.5	-	-	31.7
Aboriginality	42.8	99.3	97.7	100.0	100.0	100.0	92.6	37.5	85.1
Plurality	5.3	1.5	4.6	0.7	-	22.7	-	75.0	3.0
Infant's sex	5.5	19.3	-	4.3	18.4	52.3	-	87.5	13.6
Birthweight	59.8	69.9	72.4	77.6	81.2	81.8	88.9	100.0	70.5
Gestational age	21.4	79.3	27.6	44.8	8.8	11.4	11.1	0.0	47.1

Table 2.16: Congenital malformations by maternal age, Australia, 1993-1996

Maternal age (years)	Type of malformation				
	Isolated	Multiple	Syndrome		All types
			Chromosomal	Non-chromosomal	
			Number		
Births (20+ weeks)					
Less than 20	695	81	55	39	870
20 - 24	2,552	212	204	100	3,068
25 - 29	4,320	361	407	159	5,247
30 - 34	3,885	306	622	158	4,971
35 - 39	1,573	139	529	56	2,297
40 years and over	258	23	251	22	554
Not stated	193	32	350	15	590
All ages	13,476	1,154	2,418	549	17,597
Terminations of pregnancy (<20 weeks)					
Less than 20	35	15	10	1	61
20 - 24	146	39	65	19	269
25 - 29	262	81	163	30	536
30 - 34	245	72	233	27	577
35 - 39	103	33	426	11	573
40 years and over	20	5	261	1	287
Not stated	40	12	38	7	97
All ages	851	257	1,196	96	2,400
			Rate per 10,000 births		
Births (20+ weeks)					
Less than 20	127.2	14.8	10.1	7.1	159.3
20 - 24	129.2	10.7	10.3	5.1	155.4
25 - 29	125.9	10.5	11.9	4.6	153.0
30 - 34	126.1	9.9	20.2	5.1	161.4
35 - 39	134.9	11.9	45.4	4.8	197.0
40 years and over	136.7	12.2	133.0	11.7	293.5
All ages	129.7	11.1	23.3	5.3	169.4
Terminations of pregnancy (<20 weeks)					
			Ratio per 10,000 births		
Less than 20	6.4	2.7	1.8	0.2	11.2
20 - 24	7.4	2.0	3.3	1.0	13.6
25 - 29	7.6	2.4	4.8	0.9	15.6
30 - 34	8.0	2.3	7.6	0.9	18.7
35 - 39	8.8	2.8	36.5	0.9	49.1
40 years and over	10.6	2.6	138.3	0.5	152.0
All ages	8.2	2.5	11.5	0.9	23.1

Table 2.17: Congenital malformations by plurality, Australia, 1993-1996

Plurality	Type of malformation				
	Isolated	Multiple	Syndrome		All types
			Chromosomal	Non-chromosomal	
			Number		
Births (20+ weeks)					
Singleton	12,902	1,068	2,210	515	16,695
Twins	475	72	64	27	638
Others	20	3	1	1	25
Not stated	79	11	143	6	239
All pluralities	13,476	1,154	2,418	549	17,597
Terminations of pregnancy (<20 weeks)					
Singleton	810	248	1,146	92	2,296
Twins	17	5	12	2	36
Others	-	-	3	-	3
Not stated	24	4	35	2	65
All pluralities	851	257	1,196	96	2,400
			Rate per 10,000 births		
Births (20+ weeks)					
Singleton	127.7	10.6	21.9	5.1	165.3
Twins	171.4	26.0	23.1	9.7	230.3
Others	170.2	25.5	8.5	8.5	212.8
All pluralities	129.7	11.1	23.3	5.3	169.4
Terminations of pregnancy (<20 weeks)					
			Ratio per 10,000 births		
Singleton	8.0	2.5	11.3	0.9	22.7
Twins	6.1	1.8	4.3	0.7	13.0
Others	-	-	25.5	-	25.5
All pluralities	8.2	2.5	11.5	0.9	23.1

Table 2.18: Live births, stillbirths and total births, States and Territories, 1981-1996

Year		NSW	Vic	Qld	WA	SA	Tas	ACT	NT	Australia
1981	Live births	47,362	-	38,834	-	19,351	7,188	-	-	112,735
	Stillbirths	353	-	253	-	129	44	-	-	779
	Total births	47,715*	-	39,087	-	19,480	7,232	-	-	113,514
1982	Live births	83,489	59,983	40,540	22,236	19,294	7,002	4,479	-	237,023
	Stillbirths	600	490	225	146	121	48	31	-	1,661
	Total births	84,089	60,473	40,765	22,382	19,415	7,050	4,510	-	238,684
1983	Live births	82,739	60,123	42,000	23,046	19,901	7,028	4,622	-	239,459
	Stillbirths	526	439	262	150	115	49	30	-	1,571
	Total births	83,265	60,562	42,262	23,196	20,016	7,077	4,652	-	241,030
1984	Live births	81,792	59,763	40,356	21,601	20,149	7,098	4,590	-	235,349
	Stillbirths	545	422	245	142	131	45	24	-	1,554
	Total births	82,337	60,185	40,601	21,743	20,280	7,143	4,614	-	236,903
1985	Live births	82,780	61,726	40,275	23,066	19,889	7,213	4,619	-	239,568
	Stillbirths	477	396	251	142	143	52	30	-	1,491
	Total births	83,257	62,122	40,526	23,208	20,032	7,265	4,649	-	241,059
1986	Live births	84,009	60,387	40,166	24,175	19,826	6,911	4,627	3,307	243,408
	Stillbirths	530	403	252	146	125	66	28	35	1,585
	Total births	84,539	60,790	40,418	24,321	19,951	6,977	4,655	3,342	244,993
1987	Live births	85,650	61,642	39,100	23,271	19,345	6,752	4,680	3,519	243,959
	Stillbirths	497	363	231	142	92	29	32	46	1,432
	Total births	86,147	62,005	39,331	23,413	19,437	6,781	4,712	3,565	245,391
1988	Live births	84,268	62,347	40,240	25,123	19,231	6,745	4,817	3,422	246,193
	Stillbirths	523	360	235	120	111	55	36	33	1,473
	Total births	84,791	62,707	40,475	25,243	19,342	6,800	4,853	3,455	247,666
1989	Live births	85,464	64,185	41,714	25,019	19,703	6,788	4,614	3,366	250,853
	Stillbirths	465	411	220	112	132	34	32	45	1,451
	Total births	85,929	64,596	41,934	25,131	19,835	6,822	4,646	3,411	252,304
1990	Live births	90,260	67,158	44,533	25,322	19,981	7,001	4,859	3,534	262,648
	Stillbirths	574	404	245	132	119	45	39	32	1,590
	Total births	90,834	67,562	44,778	25,454	20,100	7,046	4,898	3,566	264,238
1991	Live births	86,220	64,660	44,460	24,815	19,622	6,902	4,490	3,459	254,628
	Stillbirths	692	529	327	192	127	55	41	43	2,006
	Total births	86,912	65,189	44,787	25,007	19,749	6,957	4,531	3,502	256,634
1992	Live births	88,401	65,853	46,307	25,159	20,004	6,975	4,678	3,582	260,959
	Stillbirths	572	447	305	165	148	51	33	46	1,767
	Total births	88,973	66,300	46,612	25,324	20,152	7,026	4,711	3,628	262,726
1993	Live births	87,362	64,323	47,156	25,160	19,844	6,809	4,754	3,505	258,913
	Stillbirths	536	414	292	176	123	47	37	40	1,665
	Total births	87,898	64,737	47,448	25,336	19,967	6,856	4,791	3,545	260,578
1994	Live births	87,488	64,448	47,716	25,237	19,673	6,790	4,747	3,490	259,589
	Stillbirths	496	484	330	188	128	46	36	37	1,745
	Total births	87,984	64,932	48,046	25,425	19,801	6,836	4,783	3,527	261,334
1995	Live births	86,870	63,245	48,172	25,257	19,472	6,734	4,853	3,618	258,221
	Stillbirths	521	470	363	191	148	47	46	37	1,823
	Total births	87,391	63,715	48,535	25,448	19,620	6,781	4,899	3,655	260,044
1996	Live births	85,884	62,484	47,625	25,383	18,979	6,734	4,751	3,434	255,274
	Stillbirths	545	467	362	201	132	47	37	27	1,818
	Total births	86,429	62,951	47,987	25,584	19,111	6,781	4,788	3,461	257,092
1981-	Live births	1,330,038	942,327	689,194	363,870	314,264	110,670	70,180	38,236	3,858,779
1996	Stillbirths	8,452	6,499	4,398	2,345	2,024	760	512	421	25,411
	Total births	1,338,490	948,826	693,592	366,215	316,288	111,430	70,692	38,657	3,884,190

** Data for NSW were incomplete as some hospitals did not report births in 1981*

Note: Sources of data: 1981-1990 from ABS; 1991-1996 from AIHW National Perinatal Statistics Unit

3 Selected major congenital malformations

The twenty-five major congenital malformations or chromosomal abnormalities presented in this chapter are chosen because they satisfy one or more of the following:

- Lethal,
- surgical intervention necessary,
- require intensive care,
- increased length of stay in hospital after birth,
- future physical or mental disabilities, possibly requiring special assistance,
- significant consequences for surviving children and their families,
- or, are relatively common.

3.1 Anencephalus

- Anencephalus is a congenital malformation characterised by total or partial absence of the cranial vault, the covering skin and the brain.

- The International Classification of Diseases codes for anencephalus are 740.0-740.2.

- The national rate of anencephalus in births showed a marked decline from 3.8 per 10,000 births in 1987, to 1.4 per 10,000 in 1995. The rate increased slightly to 1.6 per 10,000 births in 1996, with an overall annual decline of approximately 0.26 per 10,000 births (Table 3.1, Figure 3.1). During the same period, the number of induced abortions performed before 20 weeks' gestation for anencephalus increased, but notification of these abortions was incomplete.

- In the years 1987-1996, induced abortions were reported in 46.6% of all recorded notifications of anencephalus, rising from 21.6% in 1987 to 62.2% in 1996.

- Among 613 infants with anencephalus and known outcome, 69.0% were stillborn; neonatal deaths were reported in all but 15 liveborn infants. As anencephalus is always a lethal malformation, occasional failure to report the death of a liveborn infant is the most likely explanation for these 15 instances.

- Associated major malformations were reported in 13.2% of the births with anencephalus and only 2 infants had a chromosomal abnormality.

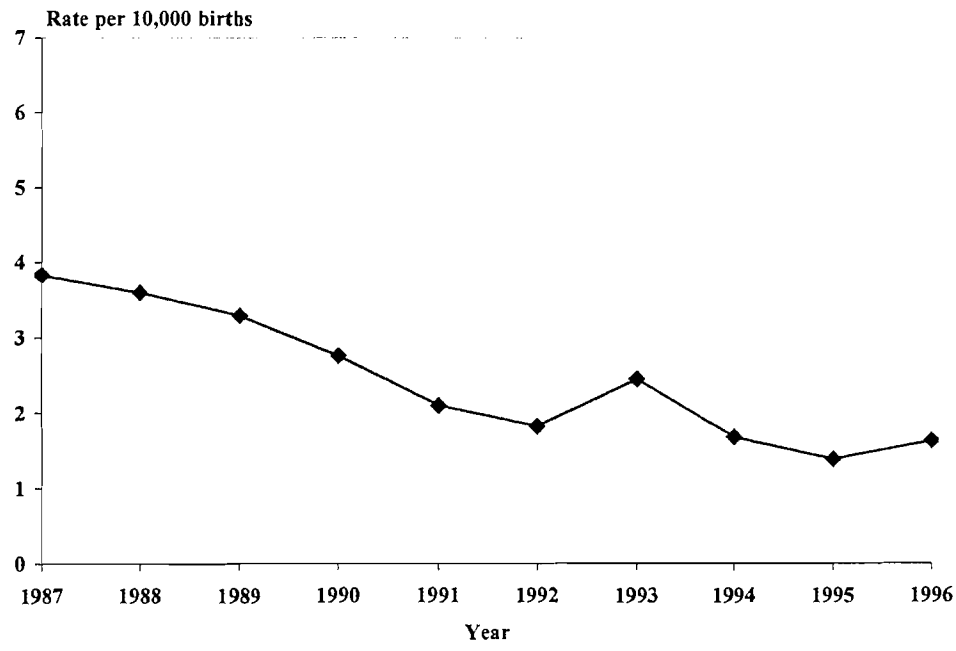
- Following reports in the early 1990s that periconceptional use of folic acid can reduce the occurrence of neural tube defects (anencephalus, spina bifida and encephalocele), educational programs aimed at improving the intake of folic acid by women in the reproductive age group have been developed in many countries, including Australia. Evaluation of the impact of these programs requires complete ascertainment of all neural tube defects among births and terminations of pregnancy. Studies are in progress to determine the influence of folic acid supplementation on trends in neural tube defects in Australia.

Table 3.1: Anencephalus by outcome and type of malformation, Australia, 1987-1996

Outcome	1987	1988	1989	1990	1991	1992	1993	1994	1995	1996	1987-96
						Number					
Live births	29	26	28	25	15	10	18	18	10	11	190
Stillbirths	63	63	51	47	37	38	43	26	26	29	423
Total births*	94	89	83	73	54	48	64	44	36	42	627
Induced abortions	26	30	39	53	52	62	56	84	78	69	549
Neonatal deaths	29	25	28	25	15	8	15	14	7	9	175
						Rate per 10,000 births					
Total births	3.8	3.6	3.3	2.8	2.1	1.8	2.5	1.7	1.4	1.6	2.4
						Number					
Isolated	85	72	71	68	45	41	56	38	31	35	542
Associated	9	17	12	5	9	7	7	5	5	7	83
Chromosomal	-	-	-	-	-	-	1	1	-	-	2
						Rate per 10,000 births					
Isolated	3.5	2.9	2.8	2.6	1.8	1.6	2.1	1.5	1.2	1.4	2.1
Associated	0.4	0.7	0.5	0.2	0.4	0.3	0.3	0.2	0.2	0.3	0.3
Chromosomal	-	-	-	-	-	-	0.0	0.0	-	-	0.0

** Total includes 'not stated'*

Figure 3.1: Anencephalus, Australia, 1987-1996



3.2 Spina bifida

- Spina bifida is a congenital malformation characterised by herniation or exposure of the spinal cord and/or meninges through an incompletely closed spine. It is not counted as a separate malformation when present with anencephalus; this combination of malformation is often described as craniorachischisis.

- The International Classification of Diseases codes for spina bifida are 741.0-741.9.

- The national rate of spina bifida in births showed a gradual decline from 7.1 per 10,000 births in 1987 to 3.0 per 10,000 births in 1994, increasing to 3.5 in 1995, and then declining again in 1996. The national rate has been decreasing annually by approximately 0.45 per 10,000 births (Table 3.2, Figure 3.2). During the same period, the number of induced abortions performed before 20 weeks' gestation for spina bifida increased, but notification of these abortions was incomplete.

- In the years 1987-1996, induced abortions were reported in 23.8% of all recorded notifications of spina bifida, increasing by a factor of six from 7.4% in 1987 to its highest in 1994 of 48.0%, then declining slightly to 46.1% in 1996.

- Among 1,279 infants with spina bifida and known outcome, 22.0% were stillborn and 19.8% of liveborn infants died in the neonatal period.

- Associated major malformations were reported in 15.3% of the births with spina bifida and another 2.9% had a chromosomal abnormality.

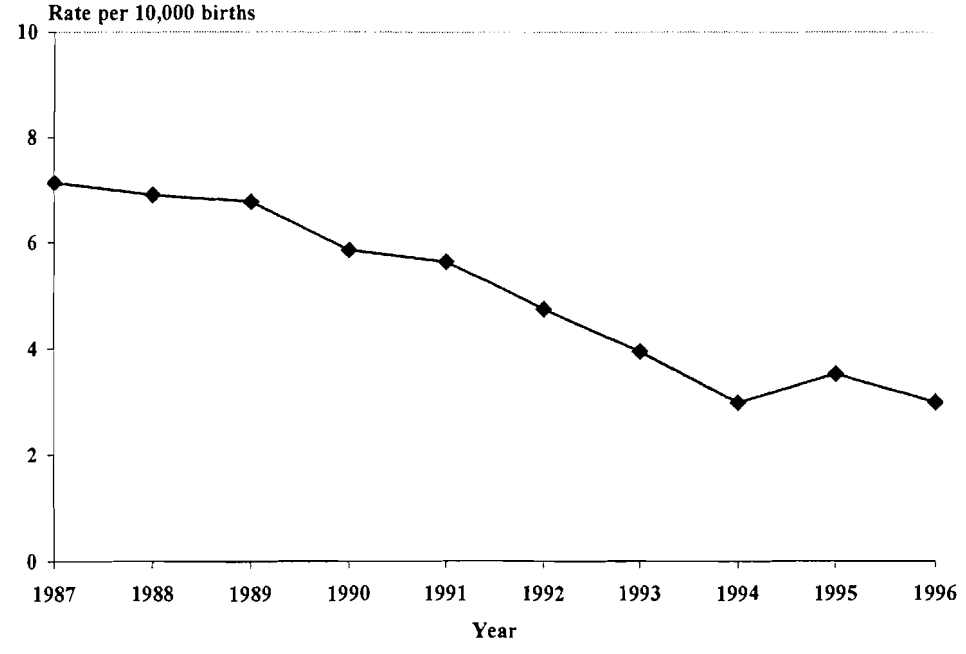
- See comment in Section 3.1 regarding periconceptional folic acid and neural tube defects.

Table 3.2: Spina bifida by outcome and type of malformation, Australia, 1987-1996

Outcome	1987	1988	1989	1990	1991	1992	1993	1994	1995	1996	1987-96
						Number					
Live births	136	141	139	129	110	89	72	61	67	54	998
Stillbirths	38	29	31	26	33	33	29	16	25	21	281
Total births*	175	171	171	155	145	125	103	78	92	77	1,292
Induced abortions	14	21	26	29	26	45	51	71	55	66	404
Neonatal deaths	32	34	35	29	20	15	7	8	7	11	198
						Rate per 10,000 births					
Total births	7.1	6.9	6.8	5.9	5.7	4.8	4.0	3.0	3.5	3.0	5.0
						Number					
Isolated	151	145	136	122	118	102	80	66	70	66	1,056
Associated	20	22	26	28	23	20	17	12	20	10	198
Chromosomal	4	4	9	5	4	3	6	-	2	1	38
						Rate per 10,000 births					
Isolated	6.2	5.9	5.4	4.6	4.6	3.9	3.1	2.5	2.7	2.6	4.1
Associated	0.8	0.9	1.0	1.1	0.9	0.8	0.7	0.5	0.8	0.4	0.8
Chromosomal	0.2	0.2	0.4	0.2	0.2	0.1	0.2	-	0.1	0.0	0.1

** Total includes 'not stated'*

Figure 3.2: Spina bifida, Australia, 1987-1996



3.3 Encephalocele

- Encephalocele is a congenital malformation characterised by herniation of the brain and/or meninges through a defect in the skull. It is not counted as a separate malformation when present with spina bifida. Anencephalus, spina bifida and encephalocele are collectively known as neural tube defects.

- The International Classification of Diseases code for encephalocele is 742.0.

- The national rate of encephalocele in births showed a slow downward trend from a high of 1.2 per 10,000 births in 1988 to a low of 0.5 per 10,000 births in 1996 (Table 3.3, Figure 3.3). During the same period, the number of induced abortions performed before 20 weeks' gestation for encephalocele increased.

- In the years 1987-1996, induced abortions were reported in 23.3% of all recorded notifications of encephalocele, ranging from 3.2% in 1988 to 36.6% in 1994.

- Among 218 infants with encephalocele and known outcome, 28.4% were stillborn; 35.3% of liveborn infants died in the neonatal period.

- Associated major malformations were reported in 35.5% of the births with encephalocele and 5 (2.3%) infants had a chromosomal abnormality.

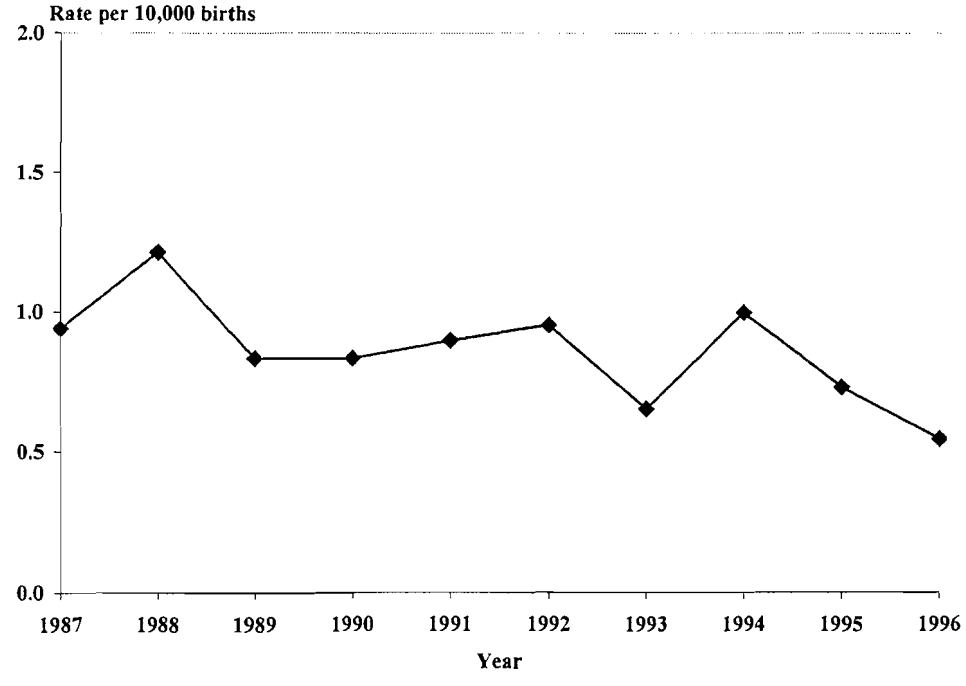
- See comment in Section 3.1 regarding periconceptional folic acid and neural tube defects.

Table 3.3: Encephalocele by outcome and type of malformation, Australia, 1987-1996

Outcome	1987	1988	1989	1990	1991	1992	1993	1994	1995	1996	1987-96
						Number					
Live births	18	21	17	18	15	19	9	16	14	9	156
Stillbirths	4	9	4	4	8	6	8	10	5	4	62
Total births*	23	30	21	22	23	25	17	26	19	14	220
Induced abortions	3	1	3	7	8	6	9	15	10	5	67
Neonatal deaths	9	10	8	6	4	5	4	2	5	2	55
						Rate per 10,000 births					
Total births	0.9	1.2	0.8	0.8	0.9	1.0	0.7	1.0	0.7	0.5	0.9
						Number					
Isolated	16	23	11	13	15	15	6	19	7	12	137
Associated	7	7	9	9	7	10	10	6	11	2	78
Chromosomal	-	-	1	-	1	-	1	1	1	-	5
						Rate per 10,000 births					
Isolated	0.7	0.9	0.4	0.5	0.6	0.6	0.2	0.7	0.3	0.5	0.5
Associated	0.3	0.3	0.4	0.3	0.3	0.4	0.4	0.2	0.4	0.1	0.3
Chromosomal	-	-	0.0	-	0.0	-	0.0	0.0	0.0	-	0.0

** Total includes 'not stated'*

Figure 3.3: Encephalocele, Australia, 1987-1996



3.4 Microcephalus

- Microcephalus is a congenital malformation characterised by a reduced brain size and head circumference. The head circumference is more than three standard deviations below the mean measurement of infants of the same gestational age.

- The International Classification of Diseases code for microcephalus is 742.1.

- There was no clear trend in the national rate of microcephalus in births during 1987 to 1996, with an overall rate of 1.6 per 10,000 births (Table 3.4, Figure 3.4).

- In the years 1987-1996, induced abortions were reported in 1.7% of all recorded notifications of microcephalus.

- Among 404 infants with microcephalus and known outcome, 6.9% were stillborn; 21.5% of liveborn infants died in the neonatal period, decreasing from 30.8% in 1988 to 8.1% in 1996.

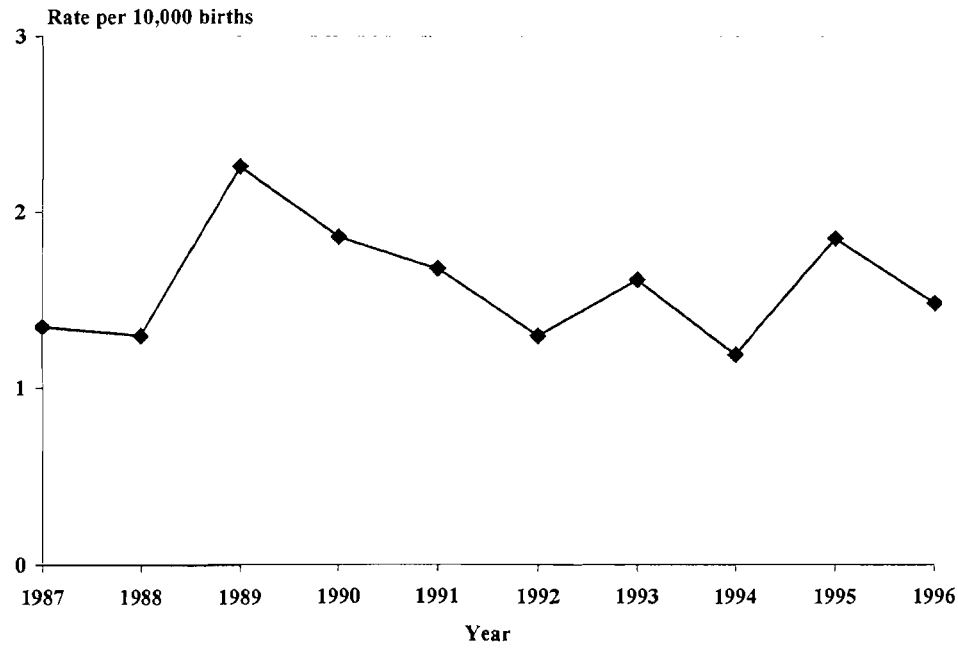
- Associated major malformations were reported in 32.9% of the births with microcephalus and another 20.1% had a chromosomal abnormality.

Table 3.4: Microcephalus by outcome and type of malformation, Australia, 1987-1996

Outcome	1987	1988	1989	1990	1991	1992	1993	1994	1995	1996	1987-96
						Number					
Live births	30	26	55	45	39	32	38	29	45	37	376
Stillbirths	3	6	2	4	4	2	1	2	3	1	28
Total births*	33	32	57	49	43	34	42	31	48	38	407
Induced abortions	-	-	1	-	2	-	-	3	1	-	7
Neonatal deaths	8	8	14	11	9	7	7	6	8	3	81
						Rate per 10,000 births					
Total births	1.3	1.3	2.3	1.9	1.7	1.3	1.6	1.2	1.8	1.5	1.6
						Number					
Isolated	13	14	27	27	16	19	15	14	24	22	191
Associated	9	10	18	10	18	11	20	14	13	11	134
Chromosomal	11	8	12	12	9	4	7	3	11	5	82
						Rate per 10,000 births					
Isolated	0.5	0.6	1.1	1.0	0.6	0.7	0.6	0.5	0.9	0.9	0.7
Associated	0.4	0.4	0.7	0.4	0.7	0.4	0.8	0.5	0.5	0.4	0.5
Chromosomal	0.4	0.3	0.5	0.5	0.4	0.2	0.3	0.1	0.4	0.2	0.3

** Total includes 'not stated'*

Figure 3.4: Microcephalus, Australia, 1987-1996



3.5 Hydrocephalus

- Hydrocephalus is a congenital malformation characterised by dilation of the ventricular system, not associated with primary brain atrophy, with or without enlargement of the head, and diagnosed before birth or during the first week of life. It is not counted as a separate malformation when present with encephalocele or open spina bifida.

- The International Classification of Diseases code for hydrocephalus is 742.3.

- The national rate of hydrocephalus in births showed no clear trend during the period 1987 to 1996, ranging from a high of 4.7 per 10,000 births in 1990 to a low of 3.3 per 10,000 births in 1996 (Table 3.5, Figure 3.5). Relatively more induced abortions performed before 20 weeks' gestation for hydrocephalus were reported in 1993 to 1996 than in earlier years.

- In the years 1987-1996, induced abortions were reported in 15.8% of all notifications of hydrocephalus, ranging from less than 10% in the 1980s to over 20% in the mid 1990s.

- Among 961 infants with hydrocephalus and known outcome, 37.1% were stillborn; 26.7% of liveborn infants died in the neonatal period.

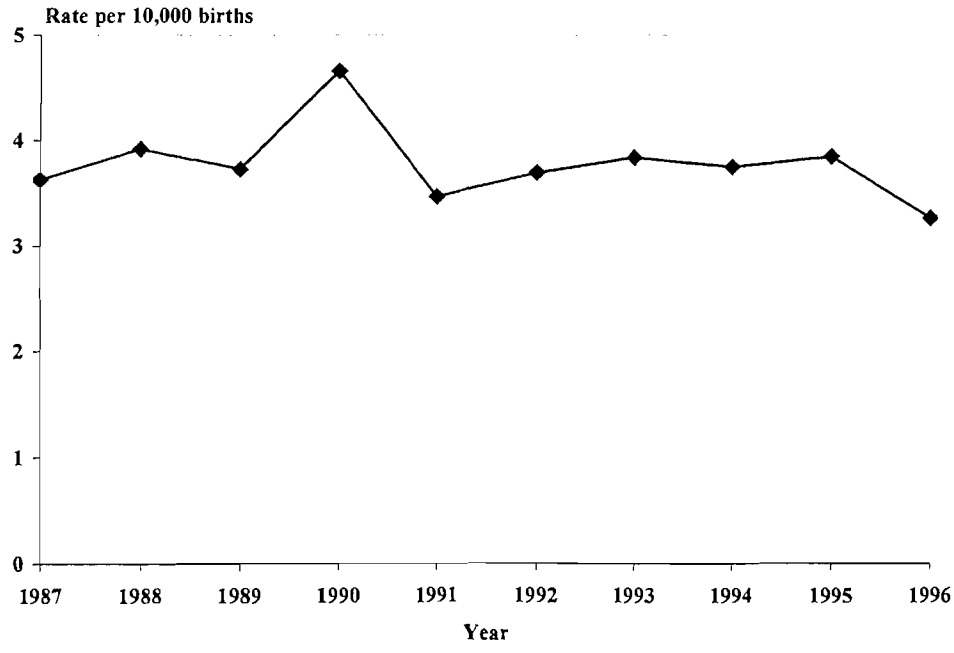
- Associated major malformations were reported in 28.5% of the births with hydrocephalus and another 8.7% had a chromosomal abnormality.

Table 3.5: Hydrocephalus by outcome and type of malformation, Australia, 1987-1996

Outcome	1987	1988	1989	1990	1991	1992	1993	1994	1995	1996	1987-96
						Number					
Live births	55	63	63	82	56	55	65	64	53	48	604
Stillbirths	34	33	30	40	33	40	33	34	44	36	357
Total births*	89	97	94	123	89	97	100	98	100	84	971
Induced abortions	6	7	8	8	17	17	30	32	32	25	182
Neonatal deaths	24	30	12	15	14	15	16	10	14	11	161
						Rate per 10,000 births					
Total births	3.6	3.9	3.7	4.7	3.5	3.7	3.8	3.7	3.8	3.3	3.8
						Number					
Isolated	63	56	58	88	55	55	62	64	61	48	610
Associated	22	32	32	26	31	31	26	24	26	27	277
Chromosomal	4	9	4	9	3	11	12	10	13	9	84
						Rate per 10,000 births					
Isolated	2.6	2.3	2.3	3.3	2.1	2.1	2.4	2.4	2.3	1.9	2.4
Associated	0.9	1.3	1.3	1.0	1.2	1.2	1.0	0.9	1.0	1.1	1.1
Chromosomal	0.2	0.4	0.2	0.3	0.1	0.4	0.5	0.4	0.5	0.4	0.3

* *Total includes 'not stated'*

Figure 3.5: Hydrocephalus, Australia, 1987-1996



3.6 Transposition of great vessels

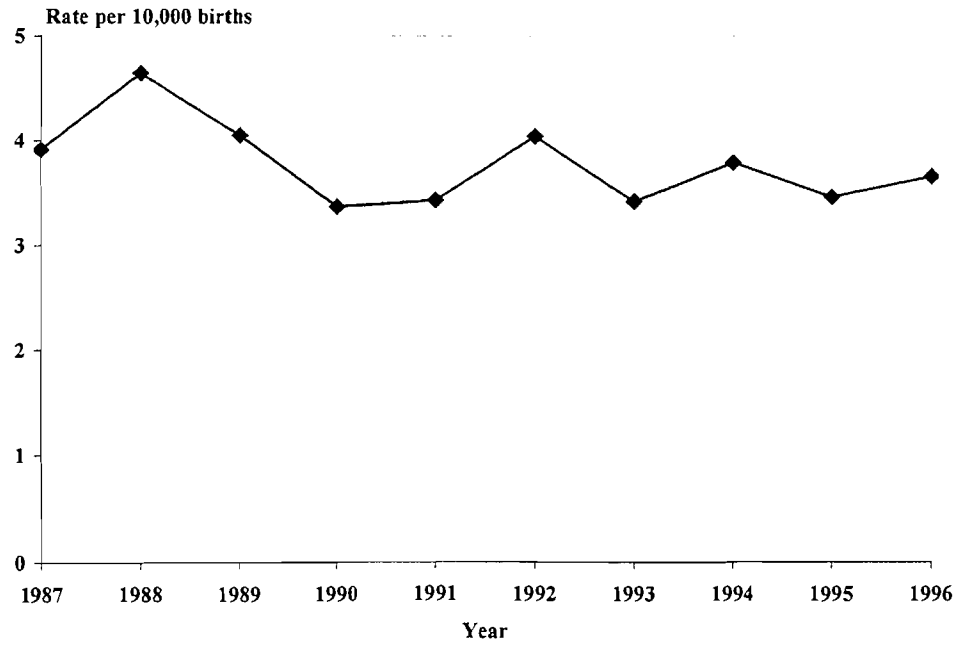
- Transposition of the great vessels is a congenital heart defect usually characterised by the aorta arising from the right ventricle and the pulmonary artery from the left ventricle.
- The International Classification of Diseases code for transposition of the great vessels is 745.1.
- The national rate of transposition of the great vessels showed a slight decline between 1988 and 1996; the overall rate for this period was 3.8 per 10,000 births (Table 3.6, Figure 3.6). Increasing numbers of induced abortions were reported in 1995 and 1996.
- Among 967 infants with transposition of the great vessels and known outcome, 5.0% were stillborn; 16.6% of liveborn infants died in the neonatal period.
- Associated major malformations were reported in 27.2% of the births with transposition of the great vessels and another 4.9% had a chromosomal abnormality.

Table 3.6: Transposition of the great vessels by outcome and type of malformation, Australia, 1987-1996

Outcome	1987	1988	1989	1990	1991	1992	1993	1994	1995	1996	1987-96
						Number					
Live births	92	114	98	83	82	100	87	96	79	88	919
Stillbirths	4	1	4	6	6	6	2	3	11	5	48
Total births*	96	115	102	89	88	106	89	99	90	94	968
Induced abortions	-	2	-	-	1	1	1	2	5	10	22
Neonatal deaths	20	31	23	12	19	12	14	6	10	6	153
						Rate per 10,000 births					
Total births	3.9	4.6	4.0	3.4	3.4	4.0	3.4	3.8	3.5	3.7	3.8
						Number					
Isolated	67	73	65	58	48	84	60	74	66	63	658
Associated	25	37	34	25	33	19	24	24	17	25	263
Chromosomal	4	5	3	6	7	3	5	1	7	6	47
						Rate per 10,000 births					
Isolated	2.7	2.9	2.6	2.2	1.9	3.2	2.3	2.8	2.5	2.5	2.6
Associated	1.0	1.5	1.3	0.9	1.3	0.7	0.9	0.9	0.7	1.0	1.0
Chromosomal	0.2	0.2	0.1	0.2	0.3	0.1	0.2	0.0	0.3	0.2	0.2

** Total includes 'not stated'*

Figure 3.6: Transposition of the great vessels, Australia, 1987-1996



3.7 Ventricular septal defect

- Ventricular septal defect is an abnormal communication between the ventricles of the heart, usually characterised by a heart murmur and sometimes by spontaneous closure in early childhood.

- The International Classification of Diseases code for ventricular septal defect is 745.4.

- The national rate of ventricular septal defect in births showed a gradual increase from a low of 14.6 per 10,000 births in 1988 to a high of 19.1 per 10,000 births in 1993 (Table 3.7, Figure 3.7). In the 1990s, the number of induced abortions of fetuses with a ventricular septal defect increased. Other malformations or chromosomal abnormalities were often present.

- Among 4,410 infants with ventricular septal defect and known outcome, 6.2% were stillborn; 8.5% of liveborn infants died in the neonatal period.

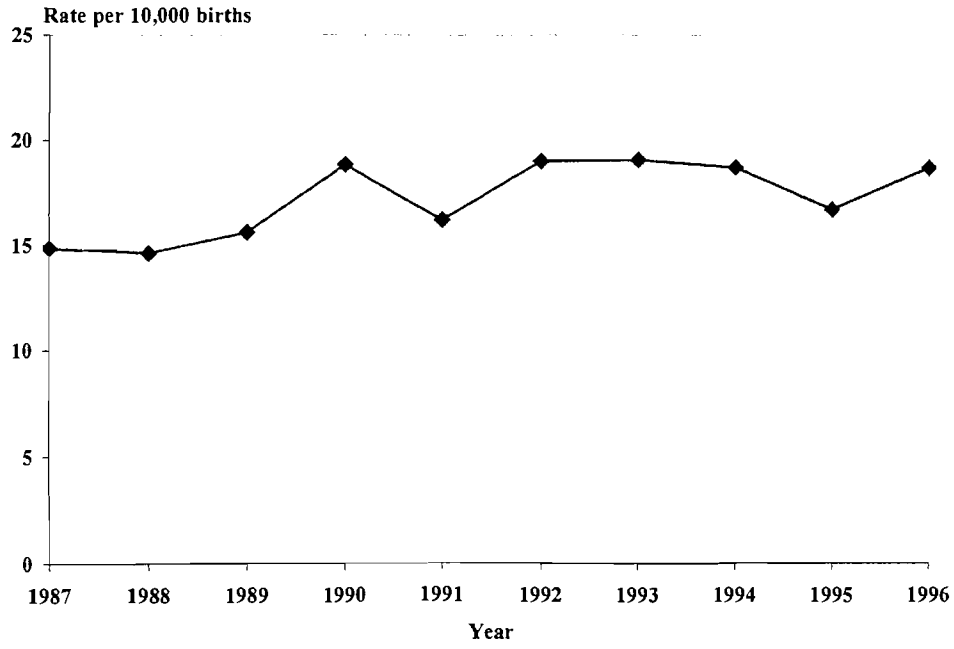
- Associated major malformations were reported in 32.7% of the births with ventricular septal defect and another 13.5% had a chromosomal abnormality.

Table 3.7: Ventricular septal defect by outcome and type of malformation, Australia, 1987-1996

Outcome	1987	1988	1989	1990	1991	1992	1993	1994	1995	1996	1987-96
						Number					
Live births	340	339	370	465	389	463	465	459	405	440	4,135
Stillbirths	24	23	24	27	24	34	27	29	28	35	275
Total births*	364	362	394	498	416	499	497	489	434	480	4,433
Induced abortions	2	5	7	6	9	7	13	19	20	25	113
Neonatal deaths	51	48	43	39	37	28	29	27	28	23	353
						Rate per 10,000 births					
Total births	14.8	14.6	15.6	18.8	16.2	19.0	19.1	18.7	16.7	18.7	17.3
						Number					
Isolated	185	167	200	263	228	289	279	273	242	258	2,384
Associated	138	150	136	166	139	161	156	143	127	135	1,451
Chromosomal	41	45	58	69	49	49	62	73	65	87	598
						Rate per 10,000 births					
Isolated	7.5	6.7	7.9	10.0	8.9	11.0	10.7	10.4	9.3	10.0	9.3
Associated	5.6	6.1	5.4	6.3	5.4	6.1	6.0	5.5	4.9	5.3	5.7
Chromosomal	1.7	1.8	2.3	2.6	1.9	1.9	2.4	2.8	2.5	3.4	2.3

** Total includes 'not stated'*

Figure 3.7: Ventricular septal defect, Australia, 1987-1996



3.8 Hypoplastic left heart

- Hypoplastic left heart is a congenital malformation resulting from an obstructive valvular and vascular lesion of the left side of the heart with varying degrees of hypoplasia of the left ventricle.

- The International Classification of Diseases code for hypoplastic left heart is 746.7.

- The national rate of hypoplastic left heart in births showed a decline from 2.8 per 10,000 births in 1991 to 1.4 per 10,000 births in 1995, then increased again in 1996 to 2.3 per 10,000 births (Table 3.8, Figure 3.8).

- There were more induced abortions for hypoplastic left heart reported in the most recent years than in the earlier period.

- Among 543 infants with hypoplastic left heart and known outcome, 11.4% were stillborn; 68.6% of liveborn infants died in the neonatal period. As this is a lethal condition, there may be incomplete reporting of neonatal deaths, or the initial diagnosis reported on perinatal forms may sometimes be incorrect.

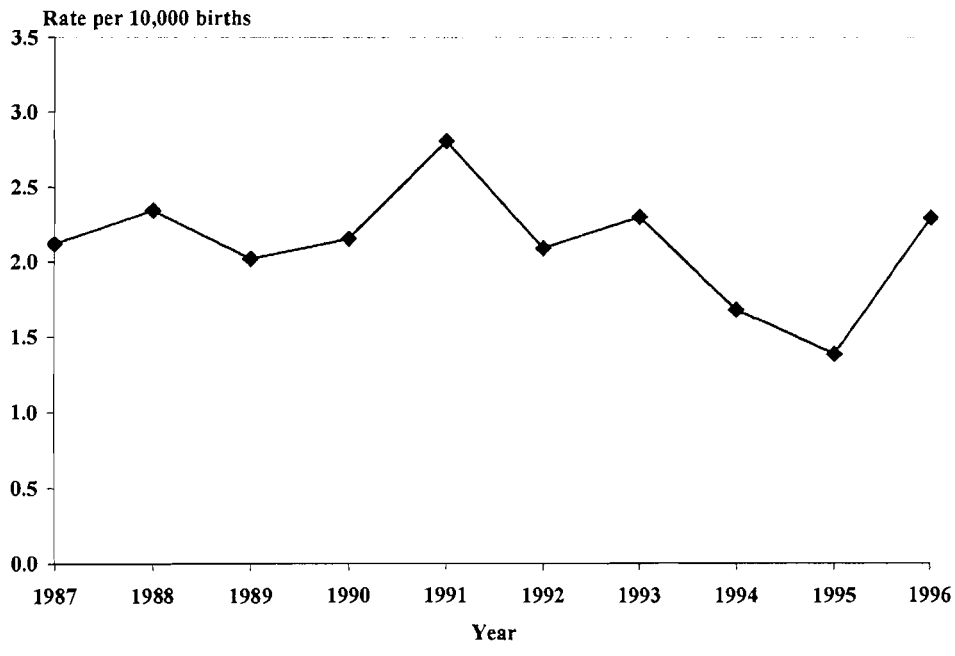
- Associated major malformations were reported in 17.1% of the births with hypoplastic left heart and another 6.4% had a chromosomal abnormality.

Table 3.8: Hypoplastic left heart by outcome and type of malformation, Australia, 1987-1996

Outcome	1987	1988	1989	1990	1991	1992	1993	1994	1995	1996	1987-96
						Number					
Live births	50	56	48	52	67	46	52	33	26	51	481
Stillbirths	2	2	3	5	4	9	8	11	10	8	62
Total births*	52	58	51	57	72	55	60	44	36	59	544
Induced abortions	-	-	1	3	6	2	3	6	4	6	31
Neonatal deaths	44	45	39	43	37	26	32	18	17	29	330
						Rate per 10,000 births					
Total births	2.1	2.3	2.0	2.2	2.8	2.1	2.3	1.7	1.4	2.3	2.1
						Number					
Isolated	43	49	39	43	53	42	40	36	29	42	416
Associated	8	8	6	10	12	8	18	6	4	13	93
Chromosomal	1	1	6	4	7	5	2	2	3	4	35
						Rate per 10,000 births					
Isolated	1.8	2.0	1.5	1.6	2.1	1.6	1.5	1.4	1.1	1.6	1.6
Associated	0.3	0.3	0.2	0.4	0.5	0.3	0.7	0.2	0.2	0.5	0.4
Chromosomal	0.0	0.0	0.2	0.2	0.3	0.2	0.1	0.1	0.1	0.2	0.1

* *Total includes 'not stated'*

Figure 3.8: Hypoplastic left heart, Australia, 1987-1996



3.9 Coarctation of aorta

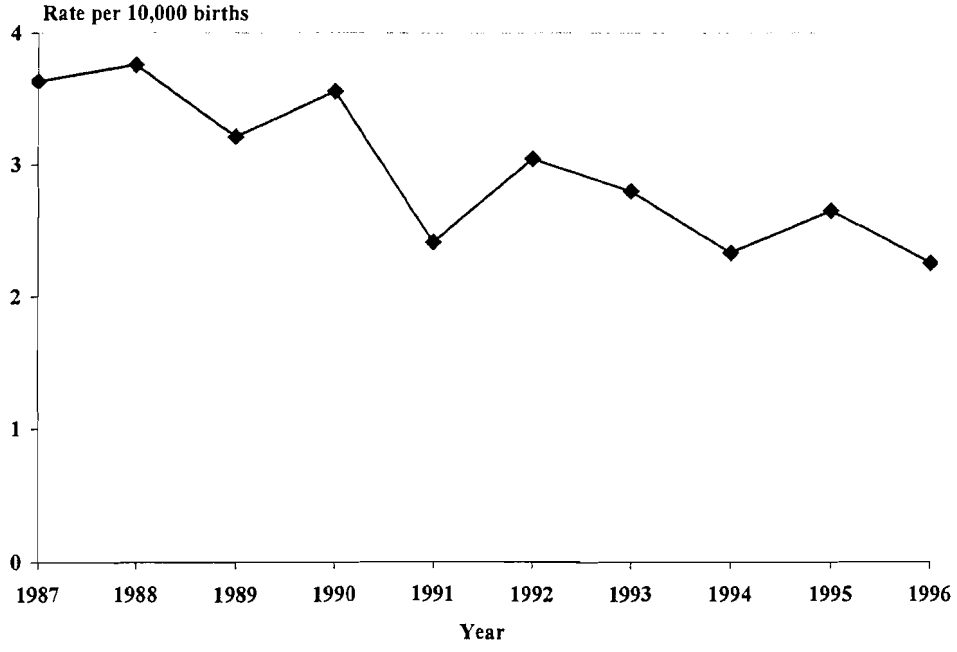
- Coarctation of the aorta is a congenital malformation resulting from narrowing of the aorta, either proximal or distal to the ductus arteriosus.
- The International Classification of Diseases code for coarctation of the aorta is 747.1.
- The national rate of coarctation of the aorta in births showed a decline since the late 1980s from 3.8 per 10,000 births in 1988 to 2.3 per 10,000 births in 1996 (Table 3.9, Figure 3.9).
- In the mid 1990s, there were more induced abortions of fetuses with coarctation of the aorta than in the earlier period.
- Among 757 infants with coarctation of the aorta and known outcome, 4.5% were stillborn; 15.4% of liveborn infants died in the neonatal period.
- Associated major malformations were reported in 36.4% of the births with coarctation of the aorta and another 8.6% had a chromosomal abnormality.

Table 3.9: Coarctation of the aorta by outcome and type of malformation, Australia, 1987-1996

Outcome	1987	1988	1989	1990	1991	1992	1993	1994	1995	1996	1987-96
						Number					
Live births	87	92	78	91	57	76	66	57	63	56	723
Stillbirths	2	1	3	2	4	4	6	4	6	2	34
Total births*	89	93	81	94	62	80	73	61	69	58	760
Induced abortions	-	-	1	-	1	2	-	3	3	3	13
Neonatal deaths	20	21	11	14	7	7	10	7	6	8	111
						Rate per 10,000 births					
Total births	3.6	3.8	3.2	3.6	2.4	3.0	2.8	2.3	2.7	2.3	3.0
						Number					
Isolated	53	49	46	51	32	45	34	31	39	38	418
Associated	31	41	29	34	27	31	29	21	24	10	277
Chromosomal	5	3	6	9	3	4	10	9	6	10	65
						Rate per 10,000 births					
Isolated	2.2	2.0	1.8	1.9	1.2	1.7	1.3	1.2	1.5	1.5	1.6
Associated	1.3	1.7	1.1	1.3	1.1	1.2	1.1	0.8	0.9	0.4	1.1
Chromosomal	0.2	0.1	0.2	0.3	0.1	0.2	0.4	0.3	0.2	0.4	0.3

* *Total includes 'not stated'*

Figure 3.9: Coarctation of the aorta, Australia, 1987-1996



3.10 Cleft palate

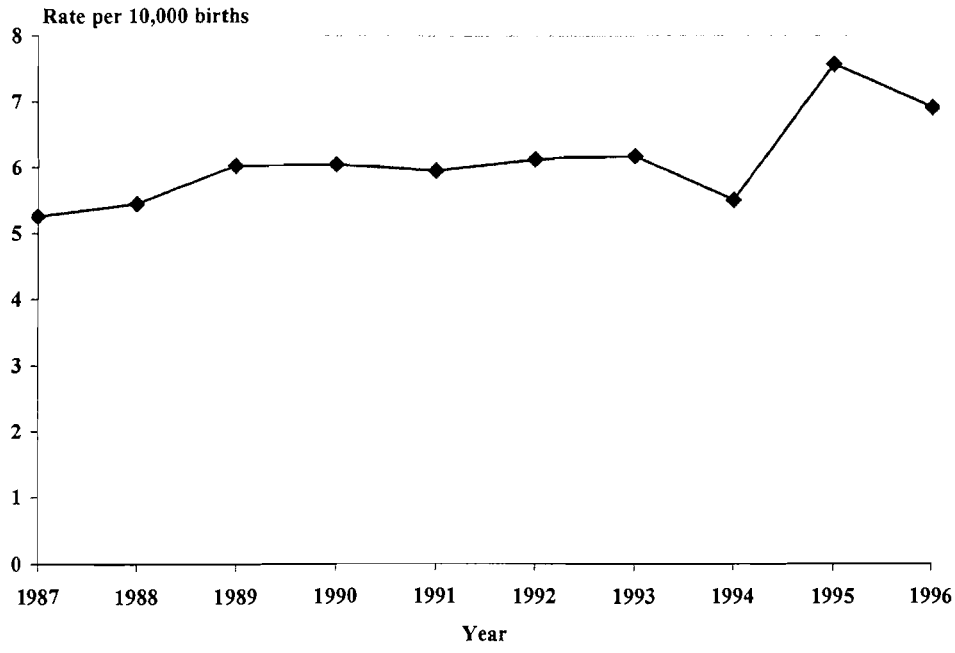
- Cleft palate is a congenital malformation characterised by a fissure defect of the hard and/or soft palate behind the foramen incisivum without cleft lip.
- The International Classification of Diseases code for cleft palate is 749.0.
- The national rate of cleft palate in births showed very little change between 1987 and 1994, then increased to 7.6 per 10,000 births in 1995, with an overall rate of 6.1 per 10,000 births in the years 1987 to 1996 (Table 3.10, Figure 3.10).
- Among 1,563 infants with cleft palate and known outcome, 4.7% were stillborn; 6.7% of liveborn infants died in the neonatal period.
- Associated major malformations were reported in 18.4% of the births with cleft palate and another 6.9% had a chromosomal abnormality.

Table 3.10: Cleft palate by outcome and type of malformation, Australia, 1987-1996

Outcome	1987	1988	1989	1990	1991	1992	1993	1994	1995	1996	1987-96
						Number					
Live births	118	128	143	154	147	153	152	131	192	171	1,489
Stillbirths	11	7	9	4	4	7	8	12	5	7	74
Total births*	129	135	152	160	153	161	161	144	197	178	1,570
Induced abortions	-	-	2	3	6	2	3	11	4	6	37
Neonatal deaths	16	13	16	9	7	8	6	12	6	7	100
						Rate per 10,000 births					
Total births	5.3	5.5	6.0	6.1	6.0	6.1	6.2	5.5	7.6	6.9	6.1
						Number					
Isolated	89	96	97	130	114	125	125	97	160	139	1,172
Associated	29	30	44	25	29	23	28	27	28	26	289
Chromosomal	11	9	11	5	10	13	8	20	9	13	109
						Rate per 10,000 births					
Isolated	3.6	3.9	3.8	4.9	4.4	4.8	4.8	3.7	6.2	5.4	4.6
Associated	1.2	1.2	1.7	0.9	1.1	0.9	1.1	1.0	1.1	1.0	1.1
Chromosomal	0.4	0.4	0.4	0.2	0.4	0.5	0.3	0.8	0.3	0.5	0.4

* *Total includes 'not stated'*

Figure 3.10: Cleft palate, Australia, 1987-1996



3.11 Cleft lip

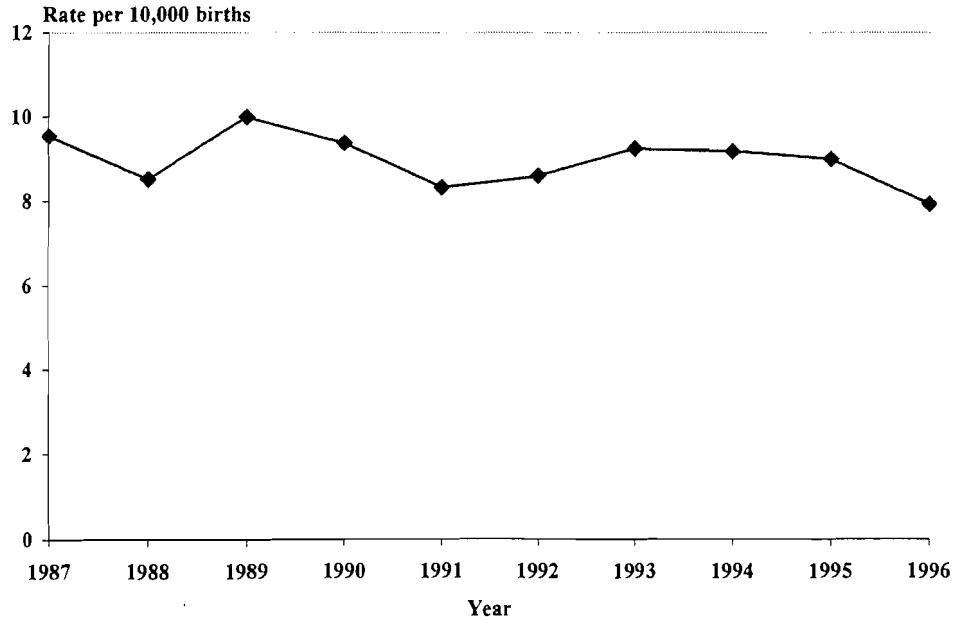
- Cleft lip is a congenital malformation characterised by clefting of the upper lip, with or without clefting of the alveolar ridge and palate.
- The International Classification of Diseases codes are 749.1 for isolated cleft lip and 749.2 for cleft lip with cleft palate.
- The national rate of cleft lip in births showed very little change between 1987 and 1996, with an overall rate of 9.0 per 10,000 births in this period (Table 3.11, Figure 3.11).
- Among 2,296 infants with cleft lip and known outcome, 6.2% were stillborn; 5.7% of liveborn infants died in the neonatal period.
- Associated major malformations were reported in 11.7% of the births with cleft lip and another 6.3% had a chromosomal abnormality.

Table 3.11: Cleft lip with or without cleft palate by outcome and type of malformation, Australia, 1987-1996

Outcome	1987	1988	1989	1990	1991	1992	1993	1994	1995	1996	1987-96
						Number					
Live births	225	194	239	231	200	204	221	226	221	193	2,154
Stillbirths	9	17	12	15	13	22	19	13	12	10	142
Total births*	234	211	252	248	214	226	241	240	234	204	2,304
Induced abortions	-	2	5	8	6	6	8	13	16	16	80
Neonatal deaths	20	9	26	7	17	13	9	9	5	7	122
						Rate per 10,000 births					
Total births	9.5	8.5	10.0	9.4	8.3	8.6	9.2	9.2	9.0	7.9	9.0
						Number					
Isolated	187	176	196	215	169	186	197	196	199	169	1,890
Associated	32	18	34	23	30	27	23	34	24	25	270
Chromosomal	15	17	22	10	15	13	21	10	11	10	144
						Rate per 10,000 births					
Isolated	7.6	7.1	7.8	8.1	6.6	7.1	7.6	7.5	7.7	6.6	7.4
Associated	1.3	0.7	1.3	0.9	1.2	1.0	0.9	1.3	0.9	1.0	1.1
Chromosomal	0.6	0.7	0.9	0.4	0.6	0.5	0.8	0.4	0.4	0.4	0.6

* *Total includes 'not stated'*

Figure 3.11: Cleft lip with or without cleft palate, Australia, 1987-1996



3.12 Oesophageal atresia or stenosis

- Oesophageal atresia or stenosis is a congenital malformation characterised by occlusion or narrowing of the oesophagus, with or without tracheo-oesophageal fistula.

- The International Classification of Diseases code for oesophageal atresia or stenosis is 750.3.

- The national rate of oesophageal atresia or stenosis in births showed a slight decline between 1987 and 1996, with an overall rate of 3.0 per 10,000 births during this period (Table 3.12, Figure 3.12), ranging from 3.8 per 10,000 births in 1993 to 2.5 per 10,000 births in 1994 and 1996.

- Among 776 infants with oesophageal atresia or stenosis and known outcome, 9.7% were stillborn; 14.1% of liveborn infants died in the neonatal period.

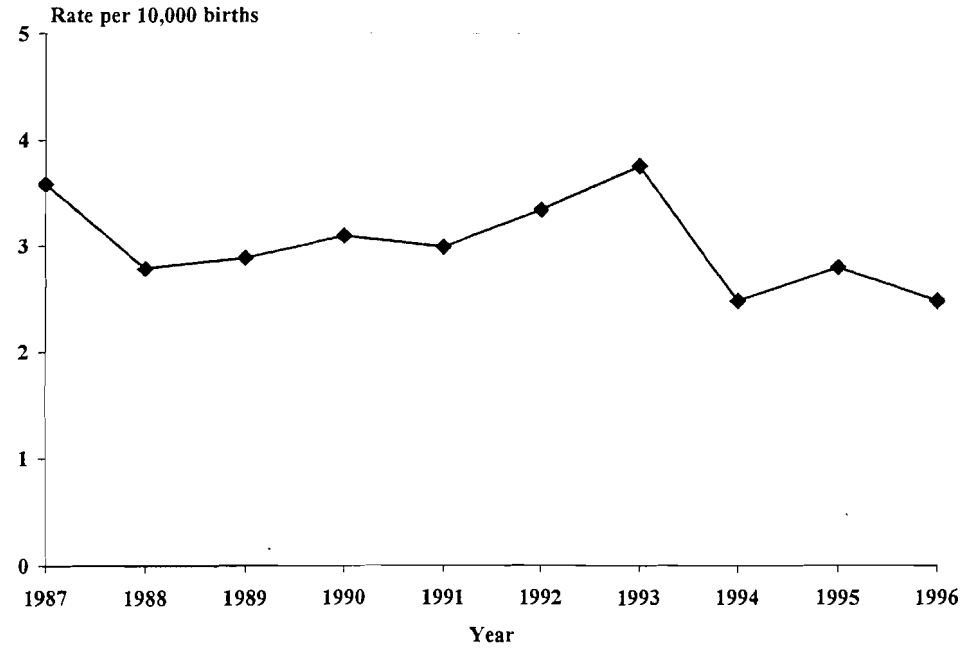
- Associated major malformations were reported in 42.3% of the births with oesophageal atresia or stenosis and another 9.3% had a chromosomal abnormality.

Table 3.12: Oesophageal atresia or stenosis by outcome and type of malformation, Australia, 1987-1996

Outcome	1987	1988	1989	1990	1991	1992	1993	1994	1995	1996	1987-96
						Number					
Live births	79	62	65	73	71	79	89	58	62	63	701
Stillbirths	9	7	8	9	6	9	9	7	10	1	75
Total births*	88	69	73	82	77	88	98	65	73	64	777
Induced abortions	-	-	-	-	1	-	2	1	1	3	8
Neonatal deaths	14	15	6	16	10	6	15	2	9	6	99
						Rate per 10,000 births					
Total births	3.6	2.8	2.9	3.1	3.0	3.3	3.8	2.5	2.8	2.5	3.0
						Number					
Isolated	45	38	34	43	44	45	37	27	34	29	376
Associated	32	26	29	31	27	38	53	33	31	29	329
Chromosomal	11	5	10	8	6	5	8	5	8	6	72
						Rate per 10,000 births					
Isolated	1.8	1.5	1.3	1.6	1.7	1.7	1.4	1.0	1.3	1.1	1.5
Associated	1.3	1.0	1.1	1.2	1.1	1.4	2.0	1.3	1.2	1.1	1.3
Chromosomal	0.4	0.2	0.4	0.3	0.2	0.2	0.3	0.2	0.3	0.2	0.3

* *Total includes 'not stated'*

Figure 3.12: Oesophageal atresia or stenosis, Australia, 1987-1996



3.13 Small intestinal atresia or stenosis

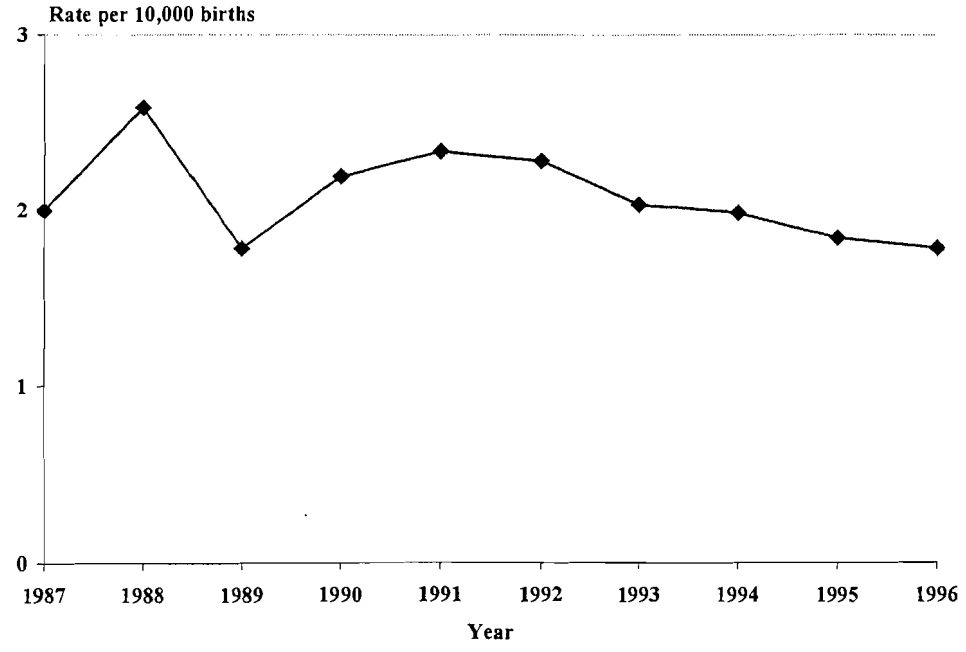
- Small intestinal atresia or stenosis is a congenital malformation characterised by occlusion or narrowing of the duodenum, jejunum or ileum.
- The International Classification of Diseases code for small intestinal atresia or stenosis is 751.1.
- The national rate of small intestinal atresia or stenosis in births showed a slight decline between 1987 and 1996, with an overall rate of 2.1 per 10,000 births during this period (Table 3.13, Figure 3.13).
- Among 532 infants with small intestinal atresia or stenosis and known outcome, 5.5% were stillborn; 8.9% of liveborn infants died in the neonatal period.
- Associated major malformations were reported in 23.9% of the births with small intestinal atresia or stenosis and another 20.4% had a chromosomal abnormality.

Table 3.13: Small intestinal atresia or stenosis by outcome and type of malformation, Australia, 1987-1996

Outcome	1987	1988	1989	1990	1991	1992	1993	1994	1995	1996	1987-96
						Number					
Live births	46	62	42	54	58	57	50	44	47	43	503
Stillbirths	3	2	3	4	2	2	3	7	1	2	29
Total births*	49	64	45	58	60	60	53	52	48	46	535
Induced abortions	-	-	-	-	-	-	-	1	-	1	2
Neonatal deaths	8	8	8	4	8	1	3	2	1	2	45
						Rate per 10,000 births					
Total births	2.0	2.6	1.8	2.2	2.3	2.3	2.0	2.0	1.8	1.8	2.1
						Number					
Isolated	22	36	20	36	31	40	33	30	29	21	298
Associated	14	13	12	15	19	12	9	9	14	11	128
Chromosomal	13	15	13	7	10	8	11	13	5	14	109
						Rate per 10,000 births					
Isolated	0.9	1.5	0.8	1.4	1.2	1.5	1.3	1.1	1.1	0.8	1.2
Associated	0.6	0.5	0.5	0.6	0.7	0.5	0.3	0.3	0.5	0.4	0.5
Chromosomal	0.5	0.6	0.5	0.3	0.4	0.3	0.4	0.5	0.2	0.5	0.4

** Total includes 'not stated'*

Figure 3.13: Small intestinal atresia or stenosis, Australia, 1987-1996



3.14 Anorectal atresia or stenosis

- Anorectal atresia or stenosis is a congenital malformation characterised by absence of the anus or of the communication between rectum and anus, or narrowing of the canal, with or without fistula to neighbouring organs. Clinically, these malformations are often termed imperforate anus.

- The International Classification of Diseases code for anorectal atresia or stenosis is 751.2. The British Paediatric Association Classification codes for atresia or stenosis of the rectum or anus are 751.21-751.24.

- The national rate of anorectal atresia or stenosis in births has varied little since 1988, with an overall rate of 3.2 per 10,000 births during 1987 to 1996 (Table 3.14, Figure 3.14).

- There were relatively more induced abortions of fetuses with anorectal atresia or stenosis in the mid 1990s than in earlier years.

- Among 832 infants with anorectal atresia or stenosis and known outcome, 11.9% were stillborn; 16.1% of liveborn infants died in the neonatal period, decreasing over the years, with a high of 29.9% in 1988 to a low of 5.3% in 1992.

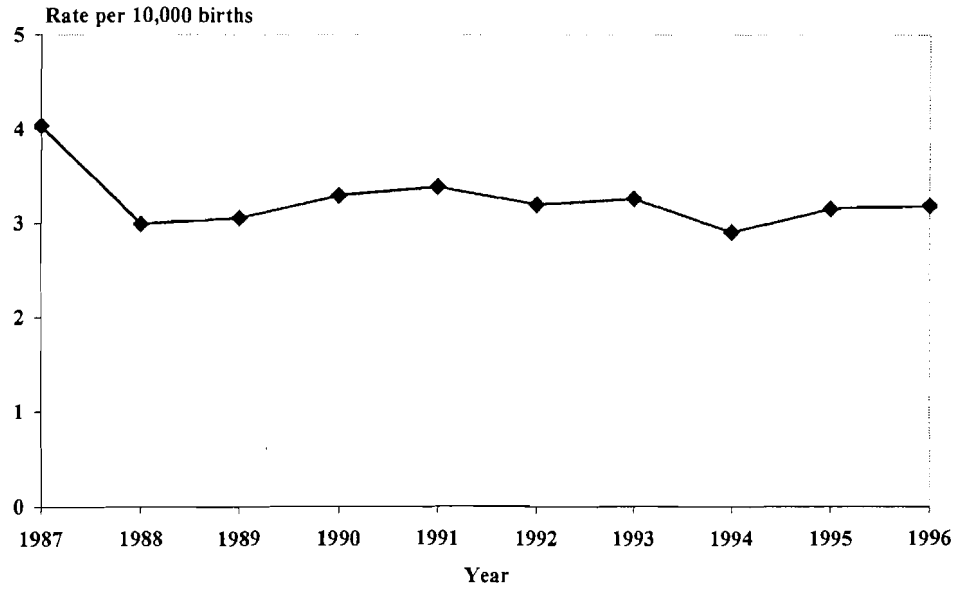
- Associated major malformations were reported in 53.4% of the births with anorectal atresia or stenosis and another 7.4% had a chromosomal abnormality.

Table 3.14: Anorectal atresia or stenosis by outcome and type of malformation, Australia, 1987-1996

Outcome	1987	1988	1989	1990	1991	1992	1993	1994	1995	1996	1987-96
						Number					
Live births	89	67	66	73	74	75	79	71	67	72	733
Stillbirths	10	7	11	14	13	9	6	5	14	10	99
Total births*	99	74	77	87	87	84	85	76	82	82	833
Induced abortions	-	2	1	5	5	1	8	12	9	8	51
Neonatal deaths	22	20	10	15	12	4	8	8	9	10	118
						Rate per 10,000 births					
Total births	4.0	3.0	3.1	3.3	3.4	3.2	3.3	2.9	3.2	3.2	3.2
						Number					
Isolated	35	30	26	34	32	39	39	31	30	30	326
Associated	56	41	44	49	50	37	41	38	47	42	445
Chromosomal	8	3	7	4	5	8	5	7	5	10	62
						Rate per 10,000 births					
Isolated	1.4	1.2	1.0	1.3	1.2	1.5	1.5	1.2	1.2	1.2	1.3
Associated	2.3	1.7	1.7	1.9	1.9	1.4	1.6	1.5	1.8	1.6	1.7
Chromosomal	0.3	0.1	0.3	0.2	0.2	0.3	0.2	0.3	0.2	0.4	0.2

** Total includes 'not stated'*

Figure 3.14: Anorectal atresia or stenosis, Australia, 1987-1996



3.15 Hypospadias

- Hypospadias is a congenital malformation characterised by opening of the urethra on the ventral side of the penis, irrespective of the degree of severity.

- The International Classification of Diseases code for hypospadias is 752.6. Although epispadias and congenital chordee are also included under this code in ICD, separate 5-digit codes in the British Paediatric Association Classification enable distinction between hypospadias and these other malformations.

- The national rate of hypospadias in births increased each year from 18.5 per 10,000 births in 1987 to 23.1 per 10,000 births in 1991 and then was relatively stable to 1996 (Table 3.15, Figure 3.15).

- Among 5,496 infants with hypospadias and known outcome, 0.8% were stillborn; 1.5% of liveborn infants died in the neonatal period.

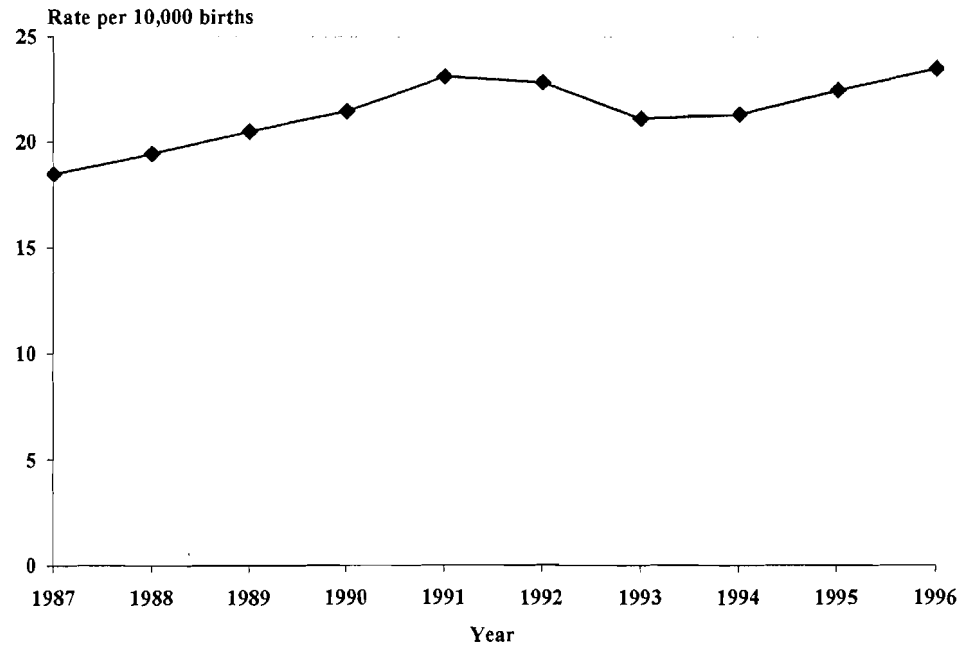
- Associated major malformations were reported in 6.4% of the births with hypospadias and another 1.4% had a chromosomal abnormality.

Table 3.15: Hypospadias by outcome and type of malformation, Australia, 1987-1996

Outcome	1987	1988	1989	1990	1991	1992	1993	1994	1995	1996	1987-96
						Number					
Live births	451	477	513	561	591	585	548	553	576	597	5,452
Stillbirths	2	4	4	6	1	13	2	2	6	4	44
Total births*	453	481	517	567	593	600	550	557	584	604	5,506
Induced abortions	-	-	1	-	-	1	4	2	-	2	10
Neonatal deaths	9	10	12	11	6	9	5	8	7	5	82
						Rate per 10,000 births					
Total births	18.5	19.4	20.5	21.5	23.1	22.8	21.1	21.3	22.5	23.5	21.4
						Number					
Isolated	402	451	470	523	559	571	508	504	532	556	5,076
Associated	45	23	35	38	27	26	33	44	43	40	354
Chromosomal	6	7	12	6	7	3	9	9	9	8	76
						Rate per 10,000 births					
Isolated	16.4	18.2	18.6	19.8	21.8	21.7	19.5	19.3	20.5	21.6	19.8
Associated	1.8	0.9	1.4	1.4	1.1	1.0	1.3	1.7	1.7	1.6	1.4
Chromosomal	0.2	0.3	0.5	0.2	0.3	0.1	0.3	0.3	0.3	0.3	0.3

* *Total includes 'not stated'*

Figure 3.15: Hypospadias, Australia, 1987-1996



3.16 Renal agenesis and dysgenesis

- Renal agenesis and dysgenesis are congenital malformations characterised by absence of kidneys or severely dysplastic kidneys and occurring bilaterally or unilaterally. Polycystic kidneys are not included in this group.

- The International Classification of Diseases code for renal agenesis and dysgenesis is 753.0.

- The national rate of renal agenesis and dysgenesis in births fluctuated during the decade 1987 to 1996, varying between the highest rate of 4.2 per 10,000 births in 1991 and the lowest rate of 2.6 per 10,000 births in 1992 (Table 3.16, Figure 3.16).

- There were relatively more induced abortions performed for renal agenesis and dysgenesis in the mid 1990s than in the earlier period.

- Among 894 infants with renal agenesis and dysgenesis and known outcome, 26.6% were stillborn; 39.2% of liveborn infants died in the neonatal period. Over half (57.8%) of all infants had bilateral agenesis and dysgenesis (Table 2.5), which is a lethal malformation.

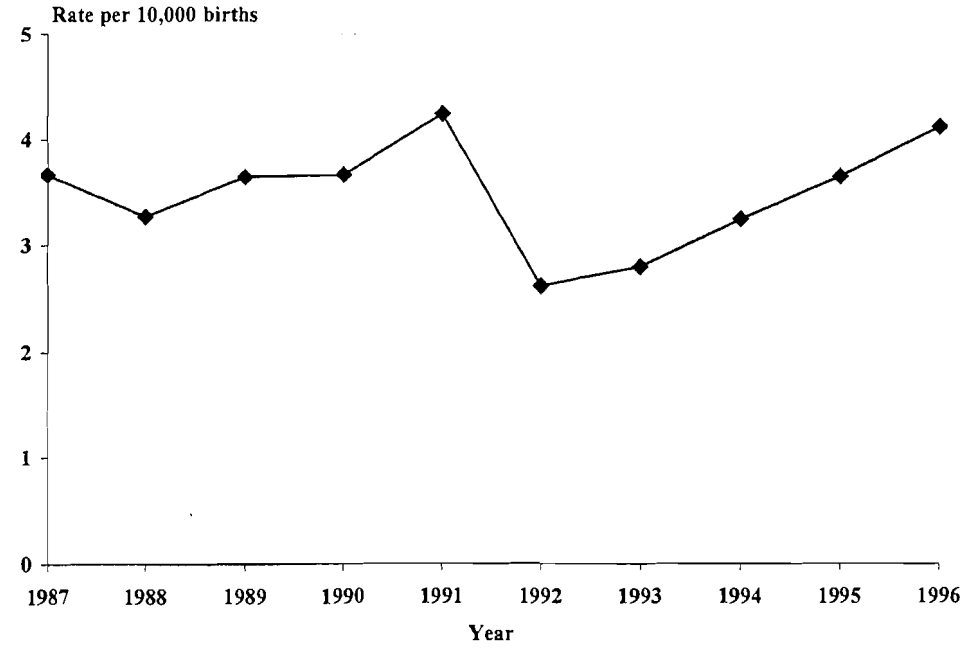
- Associated major malformations were reported in 41.2% of the births with renal agenesis and dysgenesis and another 5.6% had a chromosomal abnormality.

Table 3.16: Renal agenesis and dysgenesis by outcome and type of malformation, Australia, 1987-1996

Outcome	1987	1988	1989	1990	1991	1992	1993	1994	1995	1996	1987-96
						Number					
Live births	69	61	59	73	77	49	52	67	72	77	656
Stillbirths	20	19	33	24	32	20	20	18	23	29	238
Total births*	90	81	92	97	109	69	73	85	95	106	897
Induced abortions	-	8	8	4	3	6	15	16	16	10	86
Neonatal deaths	47	38	30	35	37	9	15	15	13	18	257
						Rate per 10,000 births					
Total births	3.7	3.3	3.6	3.7	4.2	2.6	2.8	3.3	3.7	4.1	3.5
						Number					
Isolated	45	35	42	48	60	41	36	39	60	71	477
Associated	40	42	44	43	44	27	33	37	30	30	370
Chromosomal	5	4	6	6	5	1	4	9	5	5	50
						Rate per 10,000 births					
Isolated	1.8	1.4	1.7	1.8	2.3	1.6	1.4	1.5	2.3	2.8	1.9
Associated	1.6	1.7	1.7	1.6	1.7	1.0	1.3	1.4	1.2	1.2	1.4
Chromosomal	0.2	0.2	0.2	0.2	0.2	0.0	0.2	0.3	0.2	0.2	0.2

** Total includes 'not stated'*

Figure 3.16: Renal agenesis and dysgenesis, Australia, 1987-1996



3.17 Cystic kidney disease

- Cystic kidney disease includes a heterogeneous group of malformations characterised by renal cysts of variable size and extent and described as polycystic or multicystic kidneys, occurring bilaterally or unilaterally.

- The International Classification of Diseases code for cystic kidney disease is 753.1. The British Paediatric Association Classification enables distinction between the various pathological types.

- The national rate of cystic kidney disease in births was higher in the 1990s than in the late 1980s, increasing from 2.0 per 10,000 births in 1987 to a peak of 3.6 per 10,000 births in 1996 (Table 3.17, Figure 3.17).

- There were considerably more induced abortions performed before 20 weeks' gestation for cystic kidney disease in 1994-1996 than in previous years.

- Among 739 infants with cystic kidney disease and known outcome, 12.4% were stillborn; 25.3% of liveborn infants died in the neonatal period.

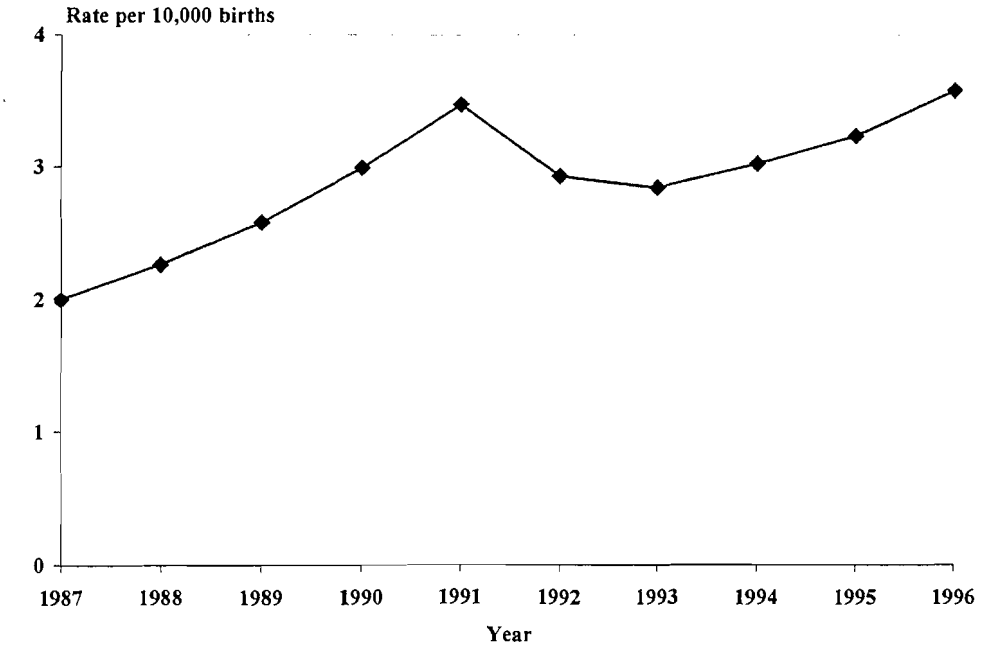
- Associated major malformations were reported in 31.6% of the births with cystic kidney disease and another 6.5% had a chromosomal abnormality.

Table 3.17: Cystic kidney disease by outcome and type of malformation, Australia, 1987-1996

Outcome	1987	1988	1989	1990	1991	1992	1993	1994	1995	1996	1987-96
						Number					
Live births	46	47	54	73	77	67	63	67	74	79	647
Stillbirths	3	9	11	5	12	9	8	12	10	13	92
Total births*	49	56	65	79	89	77	74	79	84	92	744
Induced abortions	4	3	2	4	3	7	6	15	19	10	73
Neonatal deaths	23	19	19	24	22	11	8	13	14	11	164
						Rate per 10,000 births					
Total births	2.0	2.3	2.6	3.0	3.5	2.9	2.8	3.0	3.2	3.6	2.9
						Number					
Isolated	20	29	37	48	57	57	53	42	58	60	461
Associated	24	21	20	26	29	18	14	31	24	28	235
Chromosomal	5	6	8	5	3	2	7	6	2	4	48
						Rate per 10,000 births					
Isolated	0.8	1.2	1.5	1.8	2.2	2.2	2.0	1.6	2.2	2.3	1.8
Associated	1.0	0.8	0.8	1.0	1.1	0.7	0.5	1.2	0.9	1.1	0.9
Chromosomal	0.2	0.2	0.3	0.2	0.1	0.1	0.3	0.2	0.1	0.2	0.2

* *Total includes 'not stated'*

Figure 3.17: Cystic kidney disease, Australia, 1987-1996



3.18 Obstructive defects of renal pelvis and ureter

- This group of malformations includes hydronephrosis and other obstructive defects of the renal pelvis and ureter that result in dilation of the renal collecting systems, occurring bilaterally or unilaterally.

- The International Classification of Diseases code for obstructive defects of renal pelvis and ureter is 753.2.

- The national reported rate of obstructive defects of renal pelvis and ureter in births more than doubled from 4.7 per 10,000 births in 1987 to 12.3 per 10,000 births in 1996 (Table 3.18, Figure 3.18). These malformations are increasingly detected by prenatal ultrasound screening.

- There were more reported induced abortions for these obstructive renal defects in the mid 1990s than in the earlier period.

- Among 2,021 infants with obstructive defects of renal pelvis and ureter and known outcome, 4.5% were stillborn; 6.0% of liveborn infants died in the neonatal period.

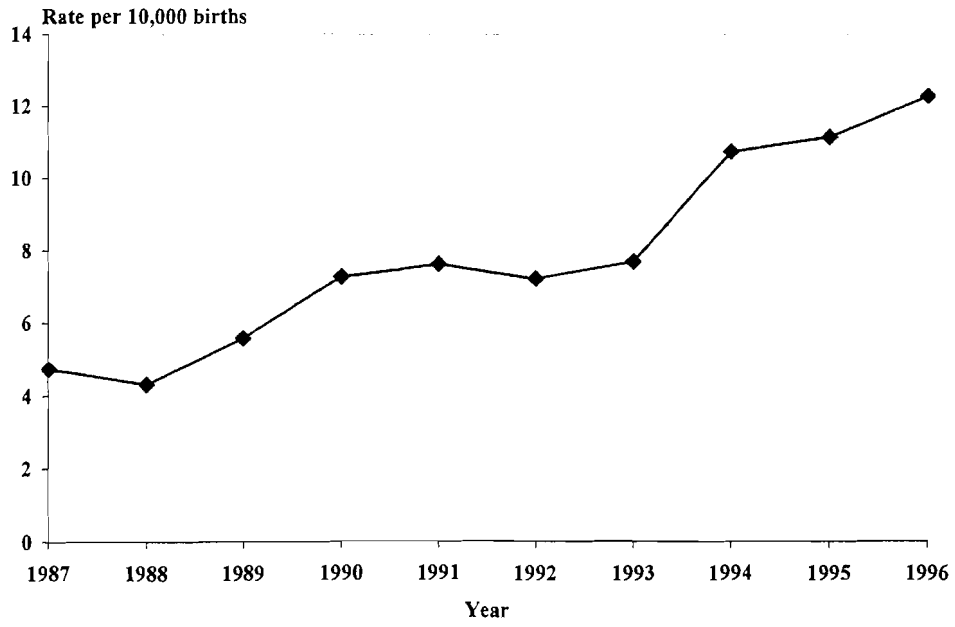
- Associated major malformations were reported in 20.2% of the births with obstructive defects of renal pelvis and ureter and another 4.6% had a chromosomal abnormality.

Table 3.18: Obstructive defects of the renal pelvis and ureter by outcome and type of malformation, Australia, 1987-1996

Outcome	1987	1988	1989	1990	1991	1992	1993	1994	1995	1996	1987-96
						Number					
Live births	107	100	135	184	186	181	186	267	280	304	1,930
Stillbirths	9	7	6	7	9	9	12	12	8	12	91
Total births*	116	107	141	193	196	190	201	281	290	316	2,031
Induced abortions	-	-	4	5	-	3	8	6	11	8	45
Neonatal deaths	16	15	10	9	10	11	12	11	12	10	116
						Rate per 10,000 births					
Total births	4.7	4.3	5.6	7.3	7.6	7.2	7.7	10.8	11.2	12.3	7.9
						Number					
Isolated	67	74	95	137	148	151	153	221	243	238	1,527
Associated	40	26	36	46	39	29	42	45	41	66	410
Chromosomal	9	7	10	10	9	10	6	15	6	12	94
						Rate per 10,000 births					
Isolated	2.7	3.0	3.8	5.2	5.8	5.7	5.9	8.5	9.3	9.3	5.9
Associated	1.6	1.0	1.4	1.7	1.5	1.1	1.6	1.7	1.6	2.6	1.6
Chromosomal	0.4	0.3	0.4	0.4	0.4	0.4	0.2	0.6	0.2	0.5	0.4

** Total includes 'not stated'*

Figure 3.18: Obstructive defects of the renal pelvis and ureter, Australia, 1987-1996



3.19 Congenital dislocation of the hip

- Congenital dislocation of the hip is a condition in which the femoral head is either displaced or displaceable from the acetabulum of the pelvis. Many newborn infants have clinical findings such as the so-called 'clicky hip' that may be confused with congenital dislocation of the hip; there may be considerable variation in the terminology used to describe these findings.

- The International Classification of Diseases code for congenital dislocation of the hip is 754.3.

- The national rate of congenital dislocation of the hip in births increased from 20.8 per 10,000 births in 1987 to 25.4 per 10,000 births in 1990, and then declined to 18.2 per 10,000 births in 1996 (Table 3.19, Figure 3.19).

- Among 5,470 infants with congenital dislocation of the hip and known outcome, 0.4% were stillborn; 0.9% of liveborn infants died in the neonatal period.

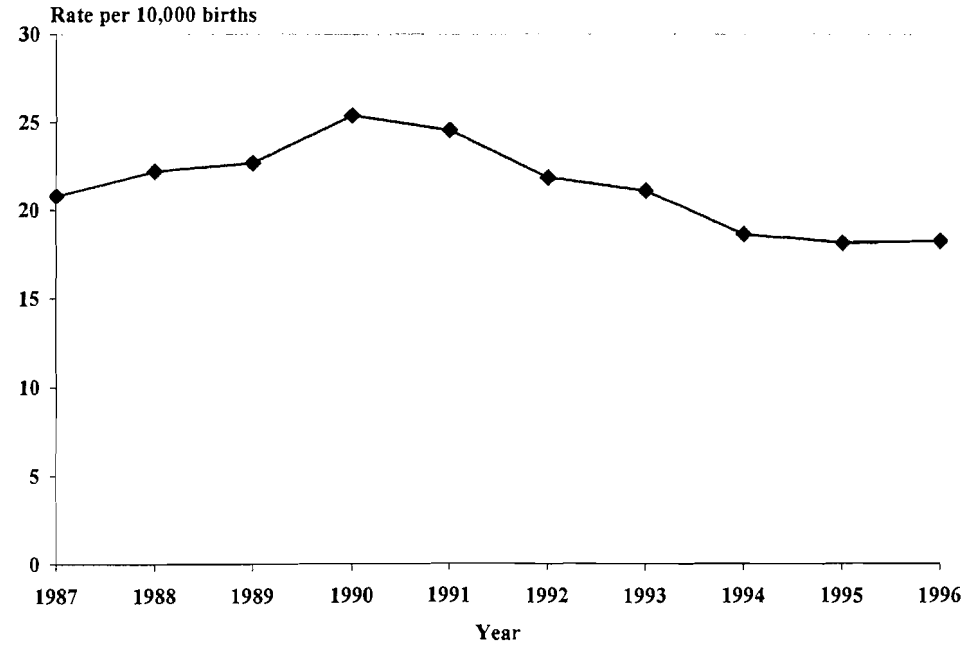
- Associated major malformations were reported in 5.5% of the births with congenital dislocation of the hip and another 0.7% had a chromosomal abnormality.

Table 3.19: Congenital dislocation of the hip by outcome and type of malformation, Australia, 1987-1996

Outcome	1987	1988	1989	1990	1991	1992	1993	1994	1995	1996	1987-96
						Number					
Live births	509	546	569	666	629	565	544	486	470	465	5,449
Stillbirths	1	3	1	3	1	6	3	-	1	2	21
Total births*	510	549	572	670	630	574	550	487	472	469	5,483
Induced abortions	-	-	1	-	-	-	-	-	2	1	4
Neonatal deaths	6	7	9	8	3	5	4	4	3	1	50
						Rate per 10,000 births					
Total births	20.8	22.2	22.7	25.4	24.5	21.8	21.1	18.6	18.2	18.2	21.4
						Number					
Isolated	477	511	533	616	602	545	512	468	453	426	5,143
Associated	27	34	36	47	27	26	30	18	17	37	299
Chromosomal	6	4	3	7	1	3	8	1	2	6	41
						Rate per 10,000 births					
Isolated	19.4	20.6	21.1	23.3	23.5	20.7	19.6	17.9	17.4	16.6	20.0
Associated	1.1	1.4	1.4	1.8	1.1	1.0	1.2	0.7	0.7	1.4	1.2
Chromosomal	0.2	0.2	0.1	0.3	0.0	0.1	0.3	0.0	0.1	0.2	0.2

* *Total includes 'not stated'*

Figure 3.19: Congenital dislocation of the hip, Australia, 1987-1996



3.20 Limb reduction defects

- Limb reduction defects are congenital malformations characterised by total or partial absence or severe hypoplasia of skeletal structures of the limbs. These are heterogeneous in type and are often analysed in the following groups: preaxial longitudinal, postaxial longitudinal, transverse, intercalary, multiple and unspecified.

- The International Classification of Diseases codes for limb reduction defects are 755.2-755.4. The NPSU coding of limb reduction defects was modified to include the groups specified above.

- From 1987 to 1996 the national rate of limb reduction defects in births varied between 5.2 per 10,000 births in 1989 and 3.9 per 10,000 births in 1994, with an overall rate of 4.6 per 10,000 births (Table 3.20, Figure 3.20).

- The reported number of induced abortions of fetuses with limb reduction defects (and often other malformations) increased during the 1990s.

- Among 1,182 infants with limb reduction defects and known outcome, 14.8% were stillborn; 10.2% of liveborn infants died in the neonatal period.

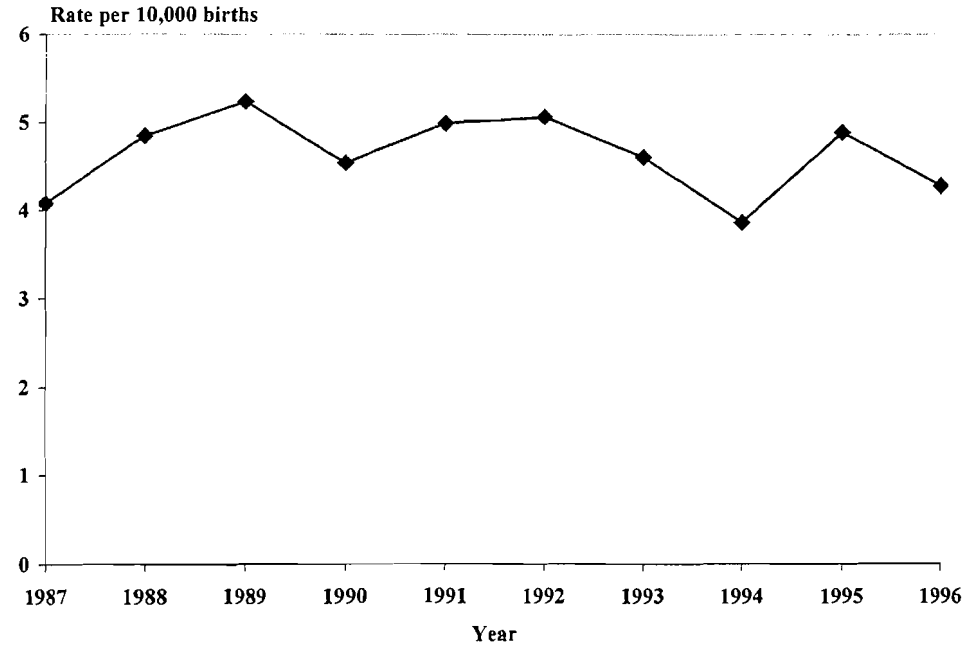
- Associated major malformations were reported in 30.7% of the births with limb reduction defects and another 6.5% had a chromosomal abnormality.

Table 3.20: Limb reduction defects by outcome and type of malformation, Australia, 1987-1996

Outcome	1987	1988	1989	1990	1991	1992	1993	1994	1995	1996	1987-96
						Number					
Live births	83	105	115	99	113	114	99	86	103	90	1,007
Stillbirths	17	14	17	21	13	18	21	13	24	17	175
Total births*	100	120	132	120	128	133	120	101	127	110	1,191
Induced abortions	-	6	7	5	5	8	10	16	17	15	89
Neonatal deaths	20	8	15	13	8	7	8	11	10	3	103
						Rate per 10,000 births					
Total births	4.1	4.8	5.2	4.5	5.0	5.1	4.6	3.9	4.9	4.3	4.6
						Number					
Isolated	55	89	80	68	87	85	78	57	82	66	747
Associated	36	27	39	44	33	41	35	39	37	35	366
Chromosomal	9	4	13	8	8	7	7	5	8	9	78
						Rate per 10,000 births					
Isolated	2.2	3.6	3.2	2.6	3.4	3.2	3.0	2.2	3.2	2.6	2.9
Associated	1.5	1.1	1.5	1.7	1.3	1.6	1.3	1.5	1.4	1.4	1.4
Chromosomal	0.4	0.2	0.5	0.3	0.3	0.3	0.3	0.2	0.3	0.4	0.3

** Total includes 'not stated'*

Figure 3.20: Limb reduction defects, Australia, 1987-1996



3.21 Diaphragmatic hernia

- Diaphragmatic hernia is a congenital malformation characterised by herniation into the thorax of abdominal contents through a defect of the diaphragm, but excluding eventration of the diaphragm.

- The International Classification of Diseases code for diaphragmatic hernia is 756.61.

- The national rate of diaphragmatic hernia in births varied between 2.1 (1988 and 1993) and 3.8 (1991) per 10,000 births, with an overall rate of 2.8 per 10,000 births during 1987 to 1996 (Table 3.21, Figure 3.21).

- In the years 1987-1996, induced abortions were reported in 6.7% of all recorded notifications of diaphragmatic hernia. The proportion of induced abortions has increased in this period from none in 1987 to about 10% in the mid-1990s.

- Among 723 infants with diaphragmatic hernia and known outcome, 11.3% were stillborn; 46.2% of liveborn infants died in the neonatal period.

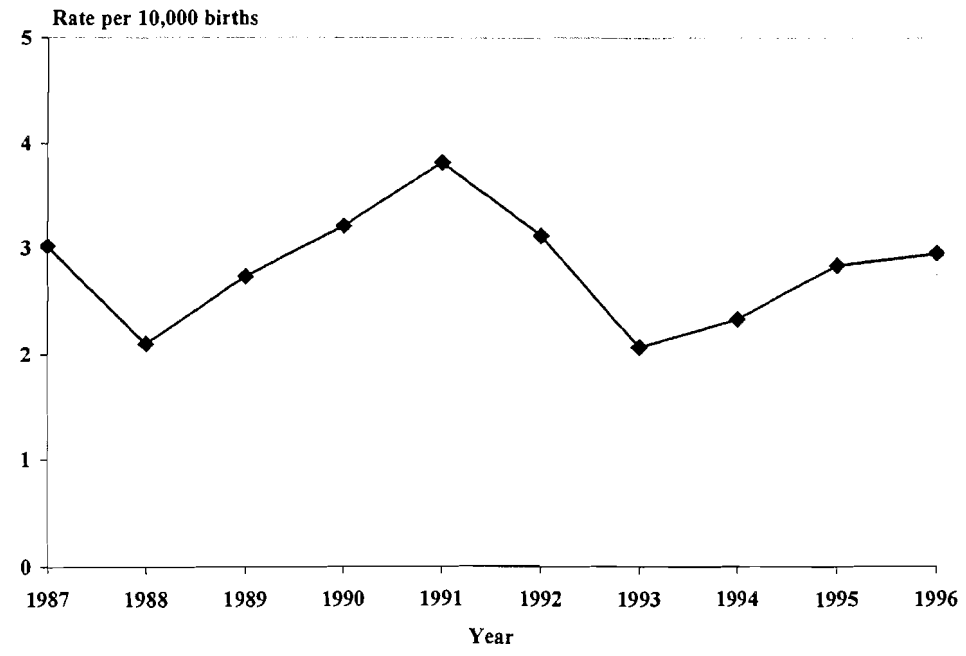
- Associated major malformations were reported in 24.7% of the births with diaphragmatic hernia and another 7.7% had a chromosomal abnormality.

Table 3.21: Diaphragmatic hernia by outcome and type of malformation, Australia, 1987-1996

Outcome	1987	1988	1989	1990	1991	1992	1993	1994	1995	1996	1987-96
						Number					
Live births	69	48	65	77	83	70	48	54	65	62	641
Stillbirths	5	4	4	8	15	12	6	6	8	14	82
Total births*	74	52	69	85	98	82	54	61	74	76	725
Induced abortions	-	2	1	5	6	8	6	8	8	8	52
Neonatal deaths	50	27	37	38	42	27	11	19	16	29	296
						Rate per 10,000 births					
Total births	3.0	2.1	2.7	3.2	3.8	3.1	2.1	2.3	2.8	3.0	2.8
						Number					
Isolated	43	28	51	63	74	65	37	41	45	43	490
Associated	28	17	16	19	16	11	13	15	20	24	179
Chromosomal	3	7	2	3	8	6	4	5	9	9	56
						Rate per 10,000 births					
Isolated	1.8	1.1	2.0	2.4	2.9	2.5	1.4	1.6	1.7	1.7	1.9
Associated	1.1	0.7	0.6	0.7	0.6	0.4	0.5	0.6	0.8	0.9	0.7
Chromosomal	0.1	0.3	0.1	0.1	0.3	0.2	0.2	0.2	0.3	0.4	0.2

** Total includes 'not stated'*

Figure 3.21: Diaphragmatic hernia, Australia, 1987-1996



3.22 Exomphalos

- Exomphalos is a congenital malformation characterised by herniation of the abdominal contents through the umbilical insertion and covered by a membrane which may or may not remain intact. Omphalocele is another term used to describe the same malformation.

- In the International Classification of Diseases, exomphalos is included under the code for anomalies of the abdominal wall (756.7). The British Paediatric Association code is 756.70, enabling distinction between exomphalos and other abdominal wall defects such as gastroschisis.

- The national rate of exomphalos in births has declined from a peak of 2.7 per 10,000 births in 1990 to 1.6 per 10,000 births in 1996 (Table 3.22, Figure 3.22).

- In the years 1987-1996, induced abortions were reported in 22.3% of all recorded notifications of exomphalos. The proportion of induced abortions reported has been steadily increasing, from 7.5% in 1987 and 1988 to 44.4% in 1994.

- Among 493 infants with exomphalos and known outcome, 32.0% were stillborn; 24.5% of liveborn infants died in the neonatal period.

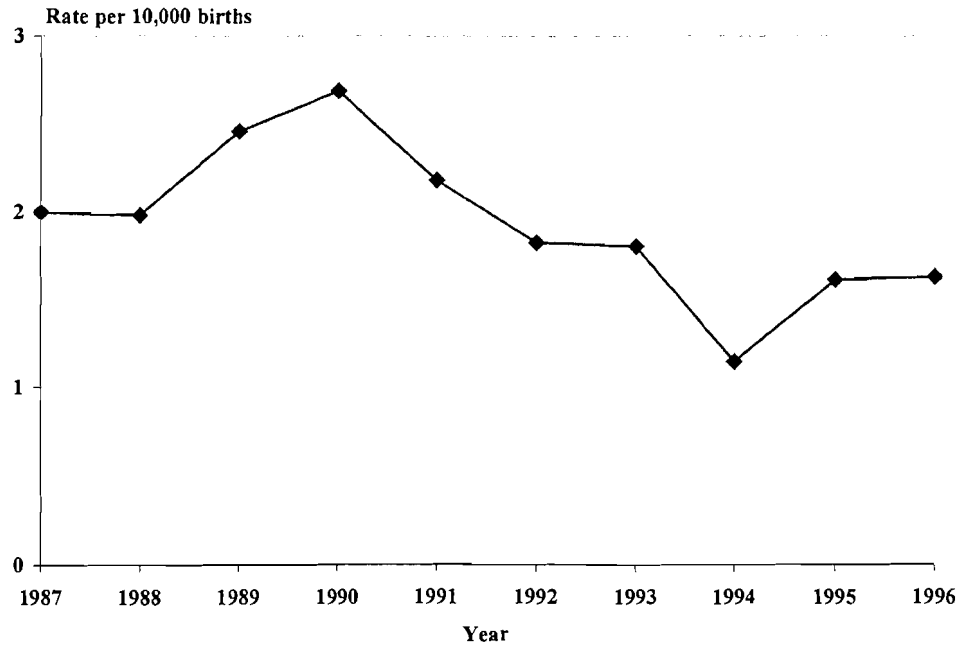
- Associated major malformations were reported in 37.3% of the births with exomphalos and another 17.1% had a chromosomal abnormality.

Table 3.22: Exomphalos by outcome and type of malformation, Australia, 1987-1996

Outcome	1987	1988	1989	1990	1991	1992	1993	1994	1995	1996	1987-96
						Number					
Live births	32	33	39	48	36	32	33	20	31	31	335
Stillbirths	17	16	23	22	18	16	14	10	11	11	158
Total births*	49	49	62	71	56	48	47	30	42	42	496
Induced abortions	4	4	13	10	12	13	23	24	21	18	142
Neonatal deaths	11	11	13	11	10	3	9	3	8	3	82
						Rate per 10,000 births					
Total births	2.0	2.0	2.5	2.7	2.2	1.8	1.8	1.1	1.6	1.6	1.9
						Number					
Isolated	18	22	28	36	25	22	21	12	18	24	226
Associated	25	19	22	27	24	15	11	13	15	14	185
Chromosomal	6	8	12	8	7	11	15	5	9	4	85
						Rate per 10,000 births					
Isolated	0.7	0.9	1.1	1.4	1.0	0.8	0.8	0.5	0.7	0.9	0.9
Associated	1.0	0.8	0.9	1.0	0.9	0.6	0.4	0.5	0.6	0.5	0.7
Chromosomal	0.2	0.3	0.5	0.3	0.3	0.4	0.6	0.2	0.3	0.2	0.3

* *Total includes 'not stated'*

Figure 3.22: Exomphalos, Australia, 1987-1996



3.23 Gastroschisis

- Gastroschisis is a congenital malformation (para-umbilical hernia) characterised by visceral herniation through an abdominal wall defect lateral to an intact umbilical cord.

- In the International Classification of Diseases, gastroschisis is included under the code for anomalies of the abdominal wall (756.7). The British Paediatric Association code is 756.71, enabling distinction between gastroschisis and other abdominal wall defects such as exomphalos.

- The national rate of gastroschisis in births increased during the period from 1987 to 1996 and varied between 0.9 per 10,000 births in 1987 and 1990, and 2.1 per 10,000 births in 1996 (Table 3.23, Figure 3.23). There were relatively few induced abortions for gastroschisis during this period.

- Among 408 infants with gastroschisis and known outcome, 11.0% were stillborn; 5.2% of liveborn infants died in the neonatal period.

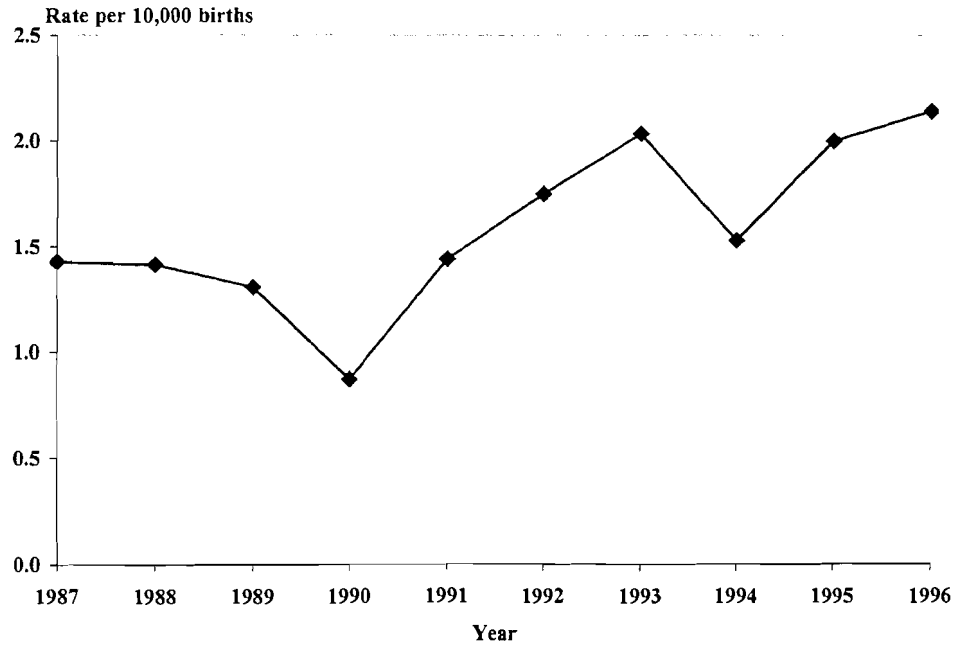
- Associated major malformations were reported in 14.9% of the births with gastroschisis and two infants had a chromosomal abnormality.

Table 3.23: Gastroschisis by outcome and type of malformation, Australia, 1987-1996

Outcome	1987	1988	1989	1990	1991	1992	1993	1994	1995	1996	1987-96
						Number					
Live births	30	32	27	21	34	41	47	38	46	47	363
Stillbirths	5	3	6	1	3	5	6	2	6	8	45
Total births*	35	35	33	23	37	46	53	40	52	55	409
Induced abortions	1	2	-	3	2	3	3	7	2	1	24
Neonatal deaths	6	5	1	-	1	3	1	-	-	2	19
						Rate per 10,000 births					
Total births	1.4	1.4	1.3	0.9	1.4	1.8	2.0	1.5	2.0	2.1	1.6
						Number					
Isolated	22	28	27	21	31	37	52	40	46	42	346
Associated	13	7	5	2	6	9	1	-	6	12	61
Chromosomal	-	-	1	-	-	-	-	-	-	1	2
						Rate per 10,000 births					
Isolated	0.9	1.1	1.1	0.8	1.2	1.4	2.0	1.5	1.8	1.6	1.3
Associated	0.5	0.3	0.2	0.1	0.2	0.3	0.0	-	0.2	0.5	0.2
Chromosomal	-	-	0.0	-	-	-	-	-	-	0.0	0.0

** Total includes 'not stated'*

Figure 3.23: Gastroschisis, Australia, 1987-1996



3.24 Trisomy 21 (Down syndrome)

- Trisomy 21 (Down syndrome) is characterised by a specific pattern of abnormalities including hypotonia, flat facies, slanted palpebral fissures, small ears, intellectual disability and variable occurrence of other minor and major congenital malformations. On chromosomal analysis, there is an additional chromosome 21 or part of its long arm.

- The International Classification of Diseases code for Down syndrome is 758.0. The 5-digit British Paediatric Association Classification enables separate codes for the different types of chromosomal abnormality (trisomy 21, translocation, mosaic).

- The national rate of Down syndrome in births was relatively constant at around 12.8 per 10,000 births during the period 1987 to 1996, ranging between a high of 14.0 per 10,000 births in 1993 and a low of 11.9 per 10,000 births in 1987 (Table 3.24, Figure 3.24).

- The reported number of induced abortions performed after prenatal diagnosis of trisomy 21 by amniocentesis or chorionic villus sampling increased substantially during this period, reaching the highest number of 130 in 1994. In the years 1987-1996, induced abortions were reported in 21.1% of all recorded notifications of Down syndrome, increasing from under 15% in 1987 to over 20% in the 1990s.

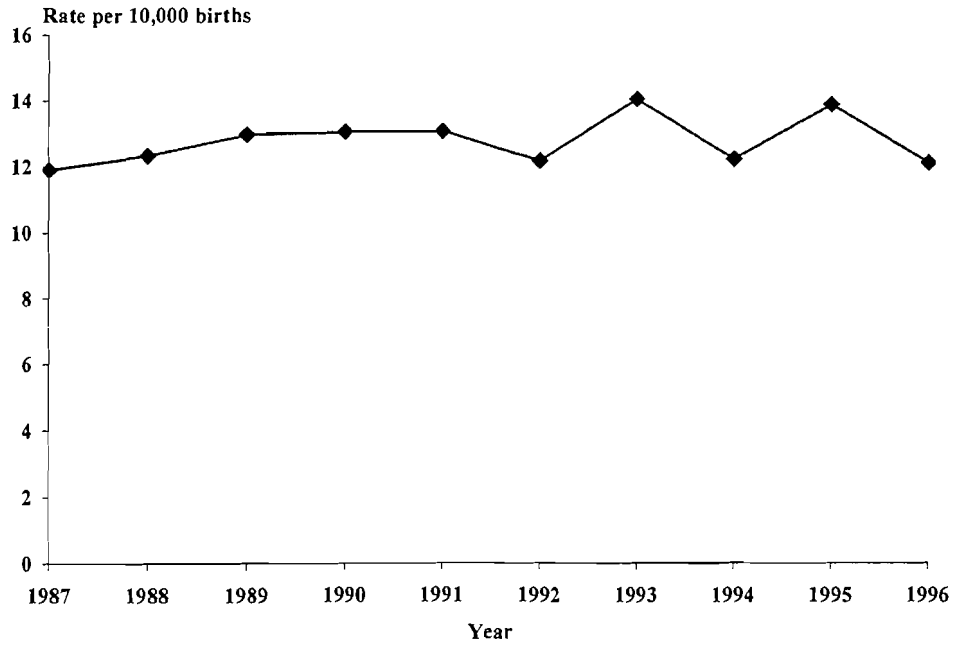
- Among 3,127 infants with trisomy 21 and known outcome, 9.2% were stillborn; 3.9% of liveborn infants died in the neonatal period.

Table 3.24: Trisomy 21 by outcome and type of malformation, Australia, 1987-1996

Outcome	1987	1988	1989	1990	1991	1992	1993	1994	1995	1996	1987-96
						Number					
Live births	265	282	306	312	305	277	291	261	284	255	2,838
Stillbirths	24	21	19	29	26	42	25	41	35	27	289
Total births*	292	305	327	345	336	320	366	320	361	312	3,284
Induced abortions	46	55	37	71	94	113	117	130	102	113	878
Neonatal deaths	16	8	24	8	12	5	7	11	11	9	111
						Rate per 10,000 births					
Total births	11.9	12.3	13.0	13.1	13.1	12.2	14.0	12.2	13.9	12.1	12.8
						Number					
Isolated	-	-	-	-	-	-	-	-	-	-	-
Associated	-	-	-	-	-	-	-	-	-	-	-
Chromosomal	292	305	327	345	336	320	366	320	361	312	3,284
						Rate per 10,000 births					
Isolated	-	-	-	-	-	-	-	-	-	-	-
Associated	-	-	-	-	-	-	-	-	-	-	-
Chromosomal	11.9	12.3	13.0	13.1	13.1	12.2	14.0	12.2	13.9	12.1	12.8

** Total includes 'not stated'*

Figure 3.24: Trisomy 21, Australia, 1987-1996



3.25 Trisomy 18 (Edwards syndrome)

- Trisomy 18 (Edwards syndrome) is characterised by a specific pattern of abnormalities including clenched hand and overlapping fingers, abnormal dermal ridge pattern of the fingertips, developmental disability and variable occurrence of other minor and major congenital malformations. On chromosomal analysis, there is an additional chromosome 18 or part of this chromosome.

- The International Classification of Diseases code for Edwards syndrome is 758.2. The 5-digit British Paediatric Association Classification enables separate codes for the different types of chromosomal abnormality (trisomy 18, translocation, mosaic).

- The national rate of trisomy 18 in births has slowly increased from 1.9 per 10,000 births in 1987 and 1989 to a peak of 3.2 per 10,000 births in 1995, with an overall rate of 2.4 per 10,000 births in the years 1987-1996 (Table 3.25, Figure 3.25).

- The reported number of induced abortions performed after prenatal diagnosis of trisomy 18 by amniocentesis or chorionic villus sampling increased substantially during this period, reaching the highest number of 47 in 1994. In the years 1987-1996, induced abortions were reported in 33.4% of all recorded notifications of Edwards syndrome. The proportion of induced abortions has increased recently, from 24.7% in 1987-1991 to 39.6% in 1992-1996.

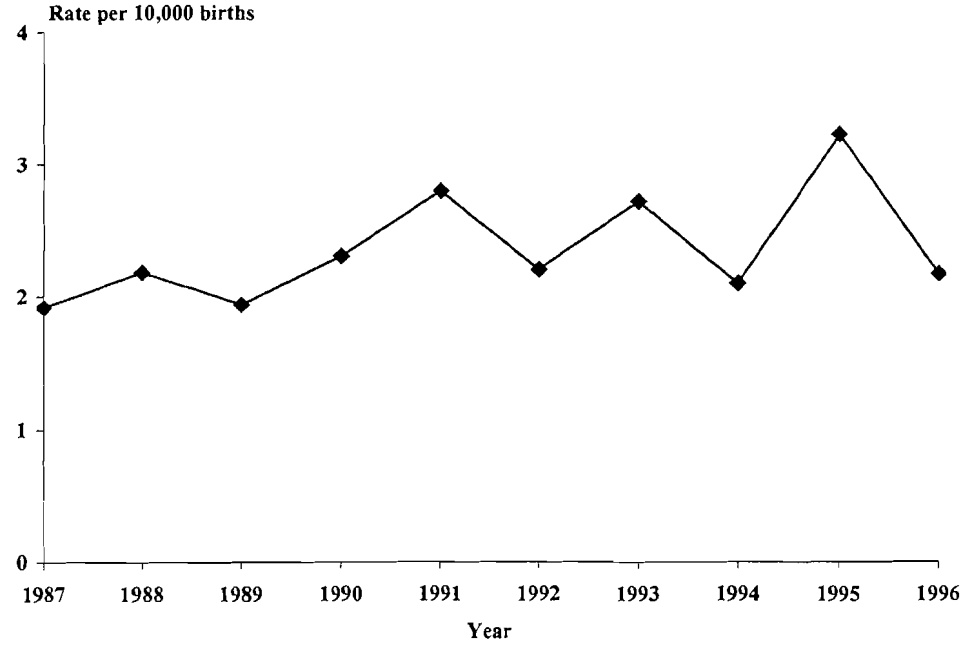
- Among 552 infants with trisomy 18 and known outcome, 38.4% were stillborn; 62.6% of liveborn infants died in the neonatal period.

Table 3.25: Trisomy 18 by outcome and type of malformation, Australia, 1987-1996

Outcome	1987	1988	1989	1990	1991	1992	1993	1994	1995	1996	1987-96
						Number					
Live births	35	41	30	37	49	37	30	24	32	25	340
Stillbirths	12	12	18	21	22	21	25	26	32	23	212
Total births*	47	54	49	61	72	58	71	55	84	56	607
Induced abortions	11	12	21	20	29	40	34	47	46	45	305
Neonatal deaths	28	26	22	22	32	17	15	13	21	17	213
						Rate per 10,000 births					
Total births	1.9	2.2	1.9	2.3	2.8	2.2	2.7	2.1	3.2	2.2	2.4
						Number					
Isolated	-	-	-	-	-	-	-	-	-	-	-
Associated	-	-	-	-	-	-	-	-	-	-	-
Chromosomal	47	54	49	61	72	58	71	55	84	56	607
						Rate per 10,000 births					
Isolated	-	-	-	-	-	-	-	-	-	-	-
Associated	-	-	-	-	-	-	-	-	-	-	-
Chromosomal	1.9	2.2	1.9	2.3	2.8	2.2	2.7	2.1	3.2	2.2	2.4

** Total includes 'not stated'*

Figure 3.25: Trisomy 18, Australia, 1987-1996



4 Perinatal, infant and childhood deaths due to congenital malformations

Congenital malformations are an important cause of perinatal and infant deaths in Australia. Although the causes of most malformations remain unknown, their occurrence at birth is increasingly being influenced by prenatal diagnosis and possible termination of pregnancy if lethal or severe malformations are detected. Many induced abortions of malformed fetuses occur at gestational ages below the usual limits that define perinatal deaths. Without effective notification of induced abortions, it is likely that the public health significance of congenital malformations and their contribution to fetal and early childhood death will be underestimated.

National data on perinatal deaths and infant deaths from the Australian Bureau of Statistics (ABS) were analysed for the period 1973 to 1996, and 1980 to 1996, respectively. This analysis determined the overall trends in death rates for specific types of malformations, the trends in the proportion of perinatal deaths due to malformations for various gestational age and birthweight groups, and the relative proportion of deaths due to malformations in each age group.

4.1 Data and methods

There are differing legal and statistical definitions in Australia for registering and reporting perinatal deaths. For legal purposes, all fetal and neonatal deaths of at least 20 weeks' gestation or at least 400g birthweight are registered. The Australian Bureau of Statistics (ABS) has published annual data on perinatal deaths based upon recommendations of the World Health Organization (WHO) for reporting national perinatal statistics. Fetal deaths are included if the birthweight is at least 500 grams or, when birthweight is not available, if the gestational age is at least 22 weeks, and there is no evidence of life after birth. Neonatal deaths are liveborn infants dying within 28 days of birth. Perinatal deaths include fetal deaths and neonatal deaths.

Fetal, neonatal and perinatal death numbers and rates in this section of the report are based on these ABS criteria. Denominators used in the calculation of death rates are annual births (live births for neonatal deaths and total births for fetal and perinatal death rates). Rates are expressed per 10,000 births in this report to maintain consistency with the malformation rates. Annual data are based on the year of registration.

National data on infant and childhood deaths from the ABS have also been analysed. An infant death is defined as the death of a liveborn child occurring within one year of birth. Childhood deaths are deaths in children aged between 1 and 14 years. Denominators used in the calculation of death rates are annual live births for infant deaths and resident population in the 1 to 14 year old age group for childhood deaths. Rates are expressed per 10,000 live births or 10,000 resident population.

Causes of death due to congenital malformations are classified according to the Ninth (1975) Revision of the World Health Organisation's International Classification of Diseases (ICD-9).

Trend lines have been fitted using a second order polynomial regression model to approximate a line of best fit (Kleinbaum et al. 1988).

4.2 Results

The overall perinatal death rate from congenital malformations declined from 35.9 per 10,000 births in 1973 to the lowest rate of 15.6 in 1996 (Figure 4.1). There was a marked fall in the death rate due to anencephalus, and lesser falls for the death rates due to spina bifida, hydrocephalus, and congenital heart defects. The perinatal death rate due to chromosomal abnormalities showed a slight increase in

the same period, from 2.2 per 10,000 births in 1973 to 2.7 per 10,000 in 1996, with higher rates in some intervening years (Figure 4.2).

The proportion of perinatal deaths due to malformations showed an initially increasing trend after 1973, rising from 16.5% in that year to a peak of 25.8% in 1983, then fluctuating until 1996, when the proportion had declined to 18.3% (Figure 4.1). A similar trend was evident for neonatal deaths, but the proportion of fetal deaths due to congenital malformations showed little change.

There was a gradual rise in the proportion of perinatal deaths that had relatively short gestational ages (Figure 4.3) or relatively low birthweights (Figure 4.4). These trends probably reflected an increasing number of terminations of pregnancy that followed prenatal diagnosis of fetal malformations.

Infant deaths from congenital malformations declined from 28.8 per 10,000 live births in 1980 to 14.8 per 10,000 in 1996 (Figure 4.5). This decline was more pronounced for malformations of the central nervous system than for congenital heart defects or other types of malformations. The proportion of infant deaths that were due to malformations also fluctuated during this period, varying from 26.8% in 1980 to 25.5% in 1996, the highest proportion (30.6%) occurring in 1983 and the lowest (24.6%) in 1989 (Figure 4.6).

The death rates of children aged 1-14 years also declined during the same period, from 3.6 per 10,000 children in 1980 to 2.2 per 10,000 in 1996 (Figure 4.5). The proportion of childhood deaths due to malformations rose slightly from 8.5% in 1980 to 9.7 % in 1996.

Figure 4.1: Proportion of fetal, neonatal and perinatal deaths due to congenital malformations, Australia, 1973-1996

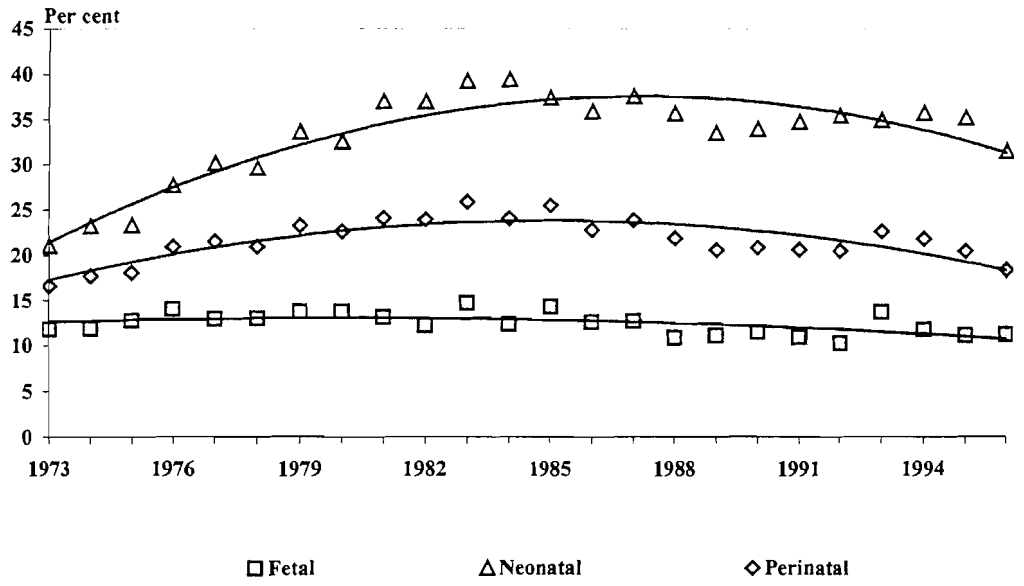


Figure 4.2: Trends in perinatal death rates for specific types of congenital malformations, Australia, 1973-1996

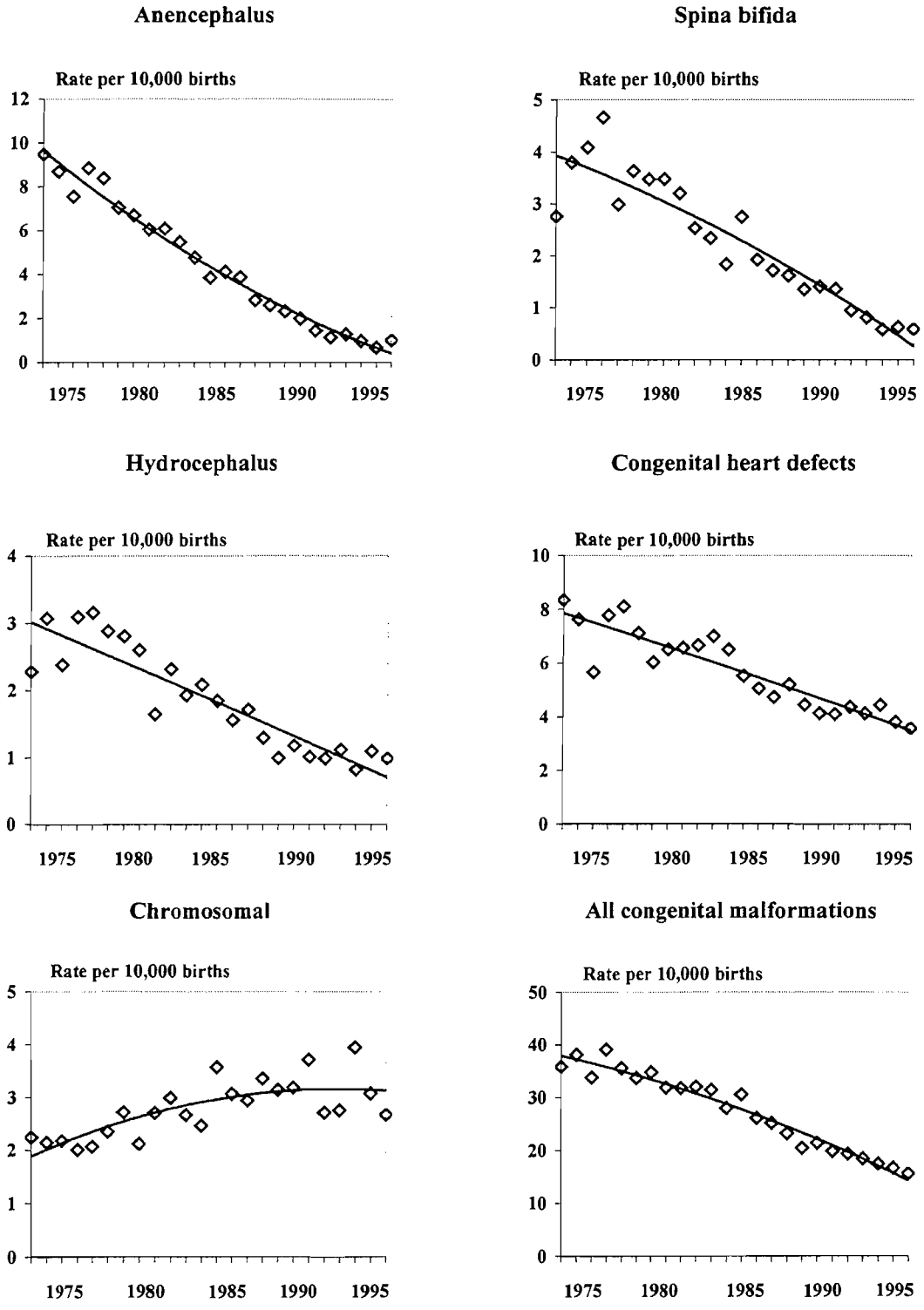


Figure 4.3: Proportion of perinatal deaths due to congenital malformations in selected gestational age groups, Australia, 1973-1996

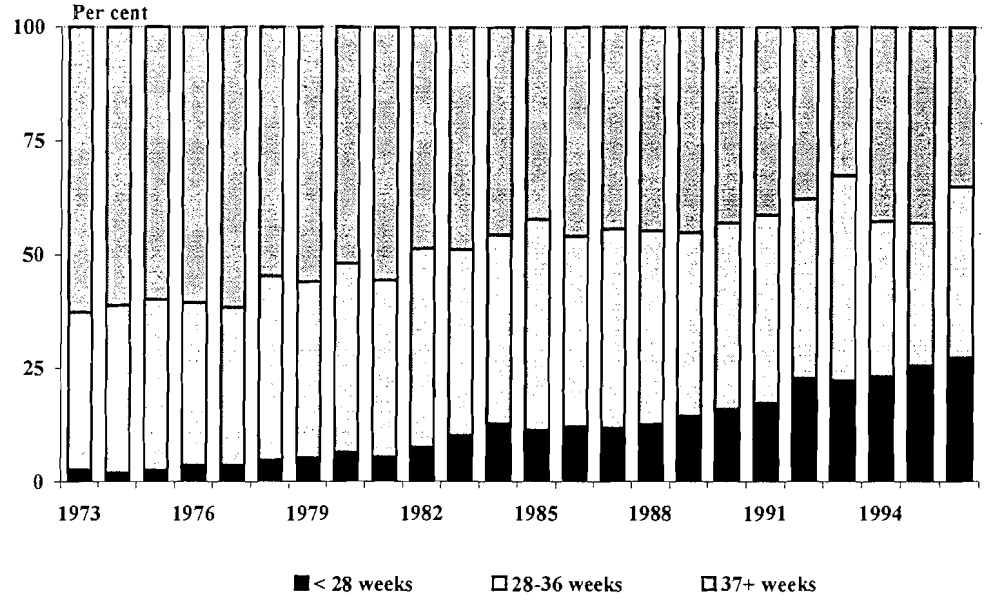


Figure 4.4: Proportion of perinatal deaths due to congenital malformations in selected birthweight groups, Australia, 1973-1996

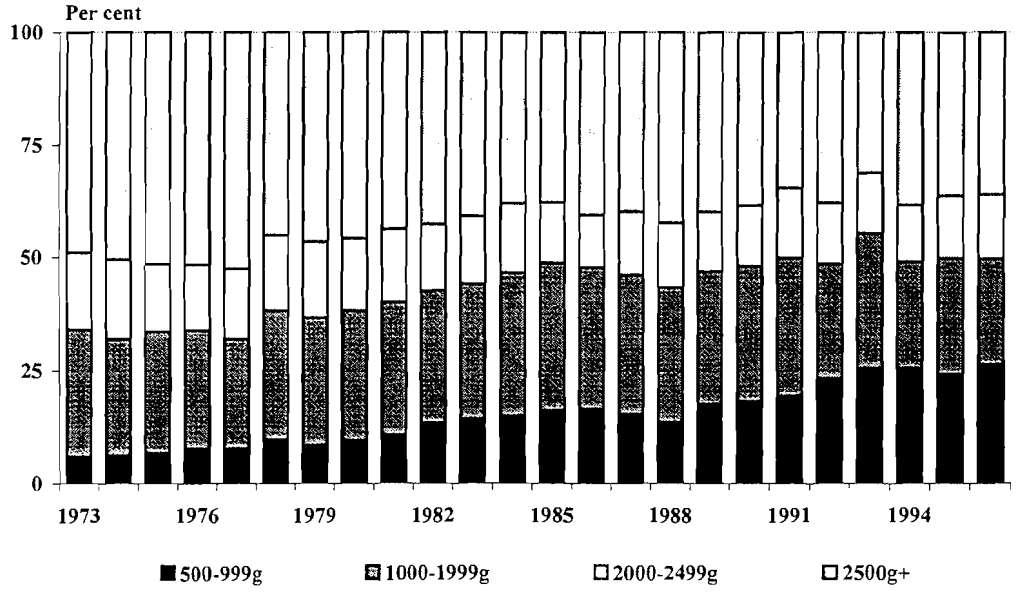


Figure 4.5: Trends in infant and childhood deaths for specific types of congenital malformations, Australia, 1980-1996

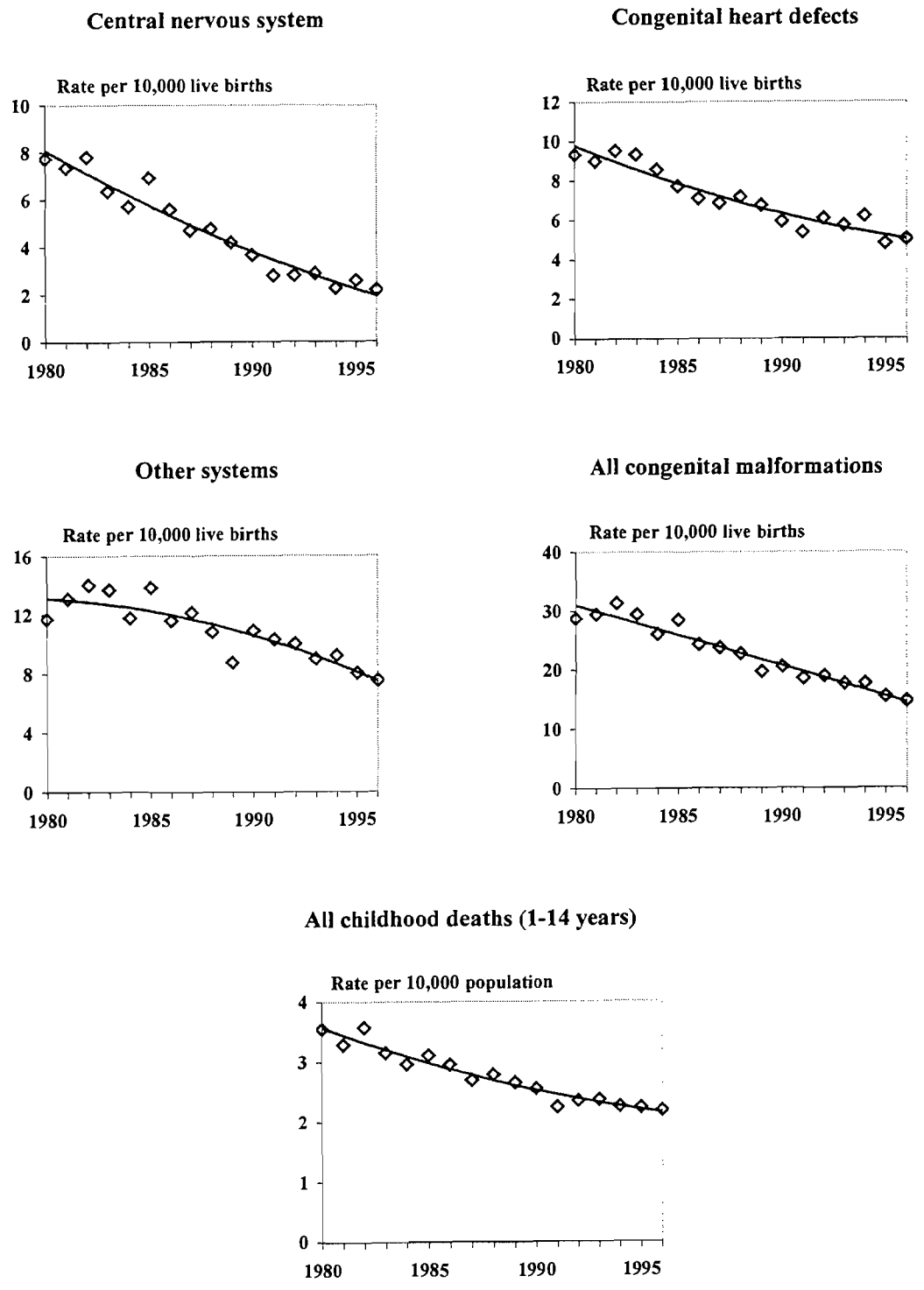
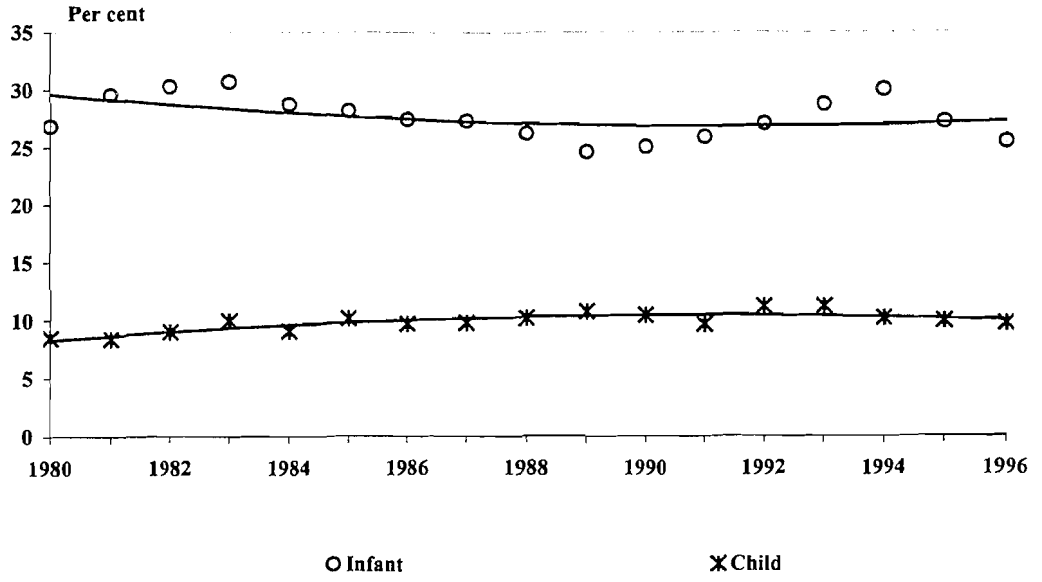


Figure 4.6: Proportion of infant and childhood deaths due to congenital malformations, Australia, 1980-1996



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Appendix 1 Minor congenital malformations

Unless an infant or fetus has other major congenital malformations, the following minor malformations and other conditions are not notified to the AIHW National Perinatal Statistics Unit.

Skin

- Skin cysts
- Noncavernous single small hemangioma (less than 10cm diameter)
- Benign skin neoplasms
- Nevus flammeus
- Birth mark
- Mongolian spots
- Cutis marmorata
- Café au lait spots
- Scalp defects, cutis aplasia
- Lanugo excessive or persistent
- Accessory nipple
- Pilonidal or sacral dimple

Skull

- Brachycephaly, dolichocephaly, plagiocephaly
- Craniotabes
- Large, small or absent fontanelles
- Macrocephaly
- Head asymmetry

Face

- Facial palsy
- Facial asymmetry
- Micrognathia
- Flat or wide nasal bridge, upturned nose, or other minor nose malformation
- Deviation of nasal septum

Eyes

- Esotropia, exotropia, strabismus
- Nystagmus
- Blue sclera
- Brushfield spots
- Epicanthal folds
- Eye slant (upward or downward)
- Narrow palpebral fissures
- Nasolacrimal duct obstruction/Dacryostenosis

Ears

- Ear tags
- Bat, cauliflower, elfin, lop, pointed, posteriorly rotated, or low-set ears
- Darwin's tubercle
- Preauricular sinus, cyst or pit
- Macrotia

Mouth, tongue and palate

- Tongue-tie
- Tongue cyst
- Ranula
- Cleft gum
- Macroglossia
- Microglossia
- Natal teeth
- Big, wide or small lips
- High-arched palate
- Bifid uvula

Neck

- Branchial cleft or sinus
- Redundant neck skin folds
- Webbing of neck
- Short neck

Cardiovascular system

- Patent ductus arteriosus or foramen ovale (gestational age <37 weeks or birthweight <2500g)
- Mild, trivial, or physiologic valvular regurgitation
- Cardiomegaly
- Dextroposition of heart
- Heart block
- Persistent fetal circulation
- Single umbilical artery

Gastrointestinal system

- Hepatomegaly
- Splenomegaly
- Meckel's diverticulum
- Anal tags
- Anal or rectal fissures
- Inguinal hernia in males
- Inguinal hernia in females (birthweight <2500g)
- Umbilical hernia (skin covered)

Urogenital system

- Imperforate hymen
- Prominent clitoris
- Fusion of vulva
- Vaginal or hymenal tags
- Cyst of vagina, vulva, canal of Nuck, or ovary
- Hydrocele
- Undescended testis (gestational age <37 weeks, birthweight <2500g)
- Small penis
- Chordee
- Patent urachus or urachal cyst
- Ectopic kidney

Limbs

- Skin tags on hands and feet
- Partial syndactyly of toes, webbing of toes
- Brachydactyly, unspecified
- Clinodactyly
- Camptodactyly
- Flexion deformities of digits
- Long fingers and toes
- Nail hypoplasia
- Enlarged or hypertrophic nails
- Widely spaced first and second toes
- Overlapping toes
- Tibial torsion or bowing
- Genu valgum, varum or recurvatum
- Dislocation of subluxation of knee
- Hallux valgus
- Hallux varus
- Talipes calcaneovalgus, equinovarus
- Cervical rib, other extra ribs
- Rocker-bottom feet
- Simian or Sydney lines, abnormal palmar creases
- Hip subluxation, clicky hips

Respiratory system

- Hypoplastic lungs (gestational age <37 weeks)
- Laryngeal stridor
- Laryngomalacia

Other conditions

- Balanced autosomal translocations
- Birth injuries
- Cephalhaematoma
- Cystic fibrosis
- Enzyme deficiencies
- Hydrops fetalis
- Meconium ileus
- Metabolic disorders
- Pyloric stenosis
- Sternomastoid tumour
- Torticollis
- Volvulus

Appendix 2 Data items in national monitoring system on congenital malformations

Demographic data:

State/Territory of birth
Reference number
State/Territory record number

Maternal data:

Local Government Area of residence
Date of birth/Age
Marital status
Previous pregnancies and outcome
Country of birth
Aboriginality
Accommodation status in hospital
Date of last menstrual period

Infant/fetus data:

Hospital of birth (or termination of pregnancy)
Sources of notification
Date of birth (or termination of pregnancy)
Sex
Plurality and birth order
Birthweight
Gestational age
Method of prenatal diagnosis (for aborted fetuses)
Outcome
Date of death (if applicable)
Autopsy performed
British Paediatric Association codes for congenital malformations
Malformation type (e.g. isolated, multiple, syndrome)
Sources of diagnosis
Comment (optional), including cytogenetic diagnosis
Follow-up information requested

Appendix 3 Definitions

Birthweight: The first weight of the baby (stillborn or liveborn) obtained after birth (usually measured to the nearest five grams and obtained within one hour of birth).

Congenital malformations: Structural or anatomical abnormalities that are present at birth, usually resulting from abnormal development in the first trimester of pregnancy.

Gestational age: The duration of pregnancy in completed weeks calculated from the date of a woman's last menstrual period and her infant's date of birth, or derived from clinical assessment during pregnancy or from examination of the infant after birth.

Induced abortion: Termination of pregnancy by medical or mechanical means before 20 weeks' gestation.

Infant death: Death of a liveborn child under 1 year of age. Infant death rates are expressed per 10,000 live births in this report.

Isolated malformation: Only one major congenital malformation is present in the infant or fetus.

Live birth: Live birth is the complete expulsion or extraction from its mother of a product of conception, irrespective of the duration of the pregnancy, which, after such separation, breathes or shows any other evidence of life, such as beating of the heart, pulsation of the umbilical cord, or definite movement of voluntary muscles, whether or not the umbilical cord has been cut or the placenta is attached; each product of such a birth is considered liveborn (WHO definition).

Low birthweight: Birthweight of less than 2500g.

Major congenital malformations: Congenital malformations that are either lethal or significantly affect the individual's function or appearance.

Maternal age: Mother's age at her child's birth.

Multiple malformations: More than one independent major congenital malformation is present in the infant or fetus.

Neonatal death: A death of a liveborn infant within 28 days of birth. Neonatal death rates are expressed per 10,000 live births in this report.

Perinatal death: Stillbirth (fetal death) or neonatal death. Perinatal death rates are expressed per 10,000 total births in this report.

Plurality: The number of births resulting from a pregnancy.

Preterm birth: Birth before 37 completed weeks of gestation.

Stillbirth (fetal death): Stillbirth is a fetal death prior to the complete expulsion or extraction from its mother of a product of conception of 20 or more completed weeks of gestation or of 400g or more birthweight; the death is indicated by the fact that after such separation the fetus does not breathe or show any other evidence of life, such as beating of the heart, pulsation of the umbilical cord, or definite movement of voluntary muscles. Fetal death rates are expressed per 10,000 total births in this report.