

## 9. Congenital malformations

Congenital malformations are anatomical defects, chromosomal abnormalities or other genetic diseases that are present at birth. Major congenital malformations are defined as those malformations which are either lethal or significantly affect the child's function and/or appearance. Minor congenital malformations are defined as those which do not have functional or cosmetic importance, but sometimes may signify an underlying genetic disorder (AIHW NPSU: Hurst et al. 2001).

The causes of most human congenital malformations cannot be determined. One review suggests that the cause is unknown for approximately 60% of malformations (Kalter & Warkany 1983a, 1983b). The causes that are known can be classified into certain groups. Congenital malformations that are caused by a single major mutant gene are thought to account for approximately 7.5% of all congenital malformations. Chromosomal abnormalities are estimated to be the cause of 6% of all malformations. Congenital malformations can also be caused by an interaction between genetic and non-genetic factors, as seen with Down syndrome, which becomes more common as maternal age increases. It has been suggested that such interactions account for 20% of malformations (Kalter & Warkany 1983a). As well as genetic factors, there are specific environmental factors which have been found to cause congenital malformations. These may include maternal infections such as rubella, maternal illness such as diabetes, environmental substances such as mercury, teratogenic agents taken by the mother (drugs or chemicals which can cause congenital malformations), and nutritional deficiencies such as a lack of folate. Maternal illness and teratogenic drugs are thought to account for approximately 5% of all congenital malformations.

Knowing that some congenital malformations result from environmental factors means that it is possible to decrease occurrence through primary prevention. For example, thalidomide, a teratogenic drug previously used to treat morning sickness, is no longer given to women, and doctors are more careful when prescribing drugs to pregnant women. Immunisation against rubella in adolescence is a preventive measure to ensure women do not contract rubella while pregnant. Increasing the consumption of folate prior to conception can decrease the likelihood of neural tube defects occurring in the baby.

Prenatal tests can be done to screen for and to diagnose congenital malformations and abnormalities, such as amniocentesis for chromosomal abnormalities, and ultrasound for anencephalus, hydrocephalus, spina bifida and other malformations (Williams et al. 1989; Garne & Berman 1999).

### Notifications

Infants and foetuses with major congenital malformations are reported to the AIHW National Perinatal Statistics Unit (NPSU) by the State and Territory birth defects registers, and these data are collated into a national data collection. These data describe major congenital malformations diagnosed in liveborn infants in the first 28 days, in stillbirths of at least 20 weeks gestation or 400 grams birthweight, and in induced abortions related to congenital malformations.

The indicator for congenital malformations is the total number of births diagnosed with a major congenital malformation in a given year as a rate per 10,000 total births.

In 1997, 4,489 infants, or 175 infants per 10,000 total births (both live and still births), had a major congenital malformation notified to the NPSU (Table 9.1). Of these infants, 77.6% had malformations affecting a single body system, 6.7% had malformations affecting more than one system and 15.6% had an identifiable chromosomal defect.

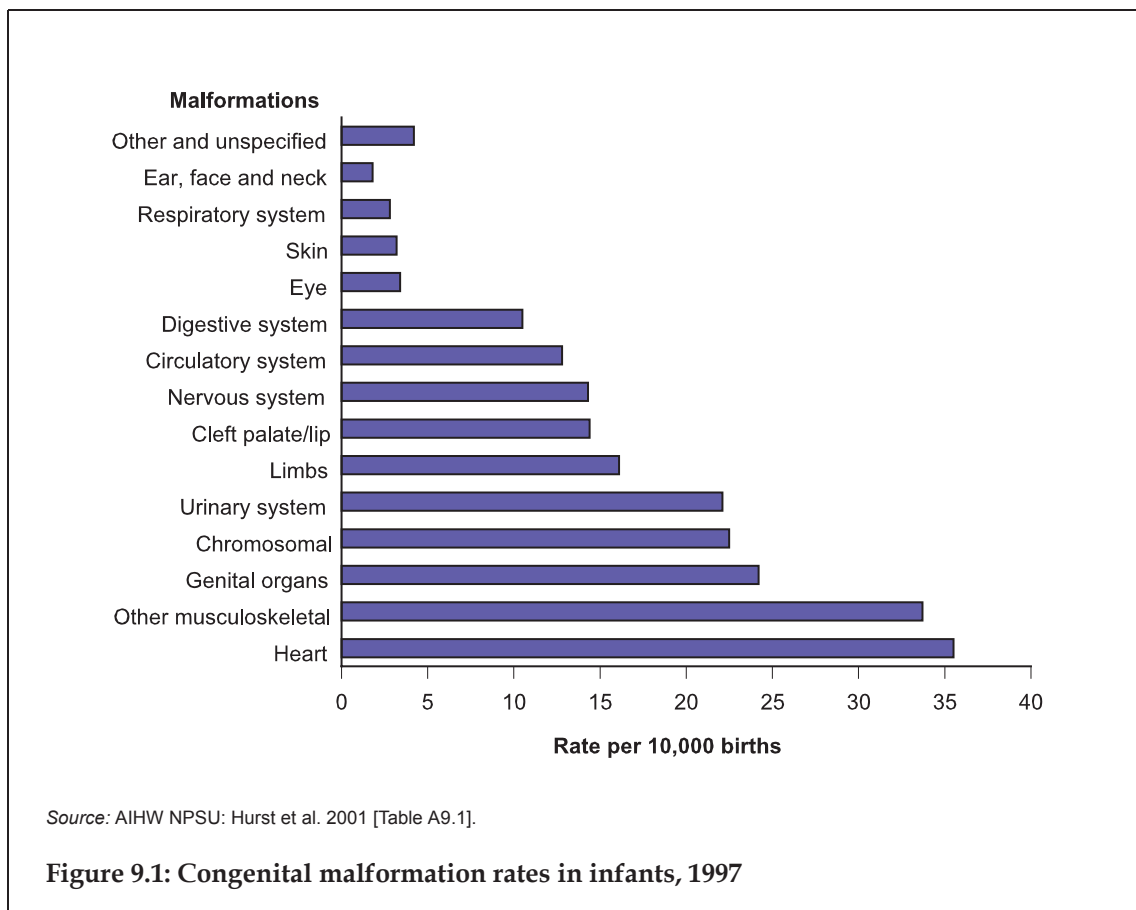
**Table 9.1: Notifications of births with major congenital malformations, 1990–97**

	1990	1991	1992	1993	1994	1995	1996	1997
Notifications	4,751	4,589	4,500	4,426	4,297	4,561	4,522	4,489
Rate per 10,000 births	179.8	177.4	169.4	169.9	164.4	175.4	175.9	175.2

Source: AIHW NPSU: Hurst et al. 1999, 2001; AIHW NPSU: Lancaster et al. 1997; AIHW NPSU: Lancaster & Pedisich 1995; AIHW NPSU 1993.

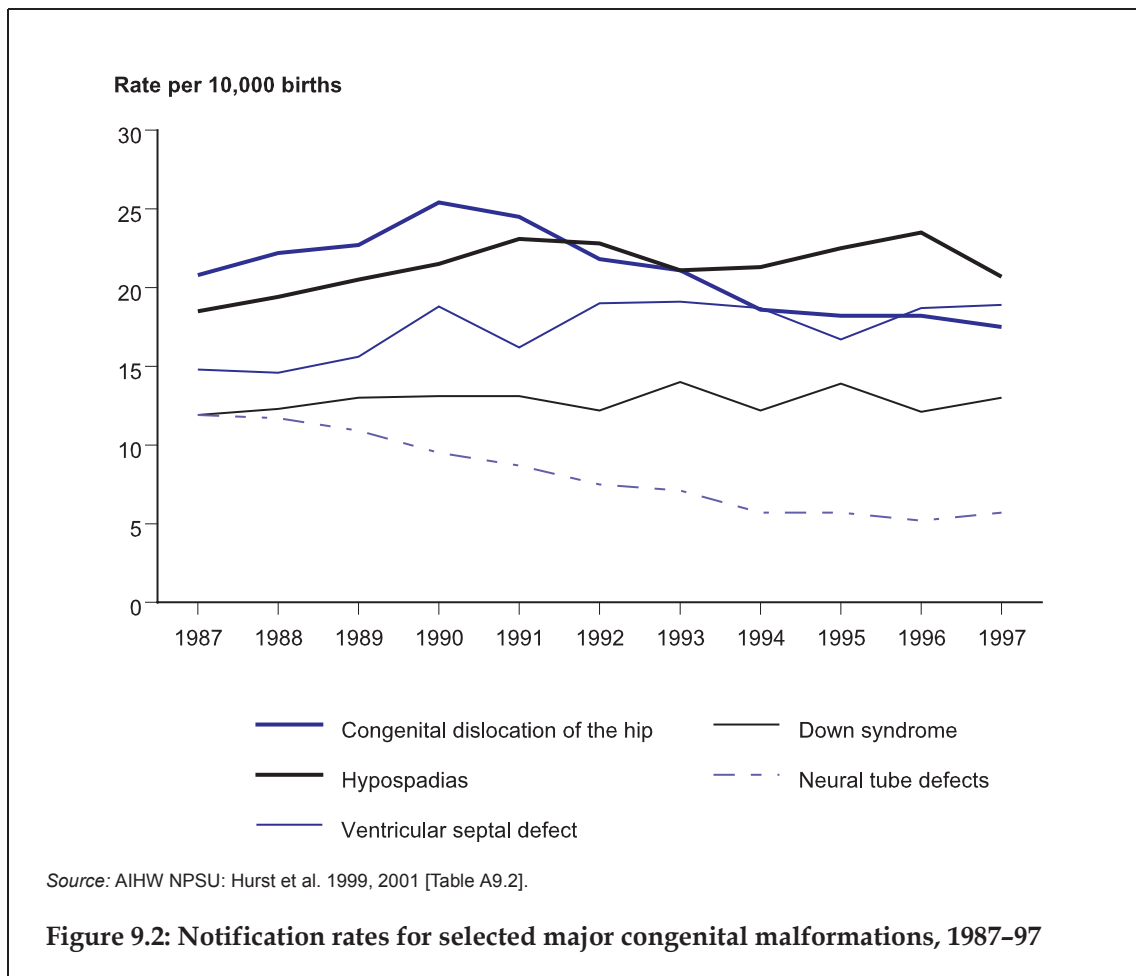
- Between 1990 and 1994, the rate of major congenital malformations declined from 179.8 to 164.4 per 10,000 births. Between 1995 and 1997, the rate remained fairly constant at around 175 per 10,000 births.

The major anatomical systems affected by congenital malformations are shown in Figure 9.1.



- In 1997, malformations of the heart were the most common congenital malformations (35.5 per 10,000 births, accounting for 20% of all congenital malformations). These were followed by malformations of the musculoskeletal system (33.7, or 19%), malformations of the genital organs (24.2, or 14%), chromosomal malformations (22.5, or 13%), and malformations of the urinary system (22.1, or 13%).

Major congenital malformations



- Hypospadias, a malformation of the male genital organs, was the most common congenital malformation in 1997 (531 births). It is characterised by an opening of the urethra on the underside of the penis. The notification rate for this condition in 1997 was 20.7 per 10,000 total births, compared with 18.5 in 1987.
- Ventricular septal defect is a congenital malformation of the heart characterised by an abnormal connection between the ventricles of the heart (sometimes known as ‘hole in the heart’), and is usually characterised by a heart murmur. The hole sometimes spontaneously closes in early childhood (AIHW: Hurst et al. 1999). In 1997, 484 infants were born with this malformation. The notification rate for this condition has also increased since 1987 (18.9 compared with 14.8).
- Congenital dislocation of the hip affected 449 infants in 1997. In this condition, the head of the femur is either displaced, or is able to be displaced, from the pelvis. The notification rate for this condition was 17.5 per 10,000 total births in 1997, compared with 20.8 in 1987.
- Down syndrome was the most common chromosomal abnormality in 1997, affecting 332 infants. This syndrome is characterised by a specific pattern of malformations, including muscular weakness, flat face, slanted eyes, small ears, intellectual disability, and variable occurrence of other minor and major congenital

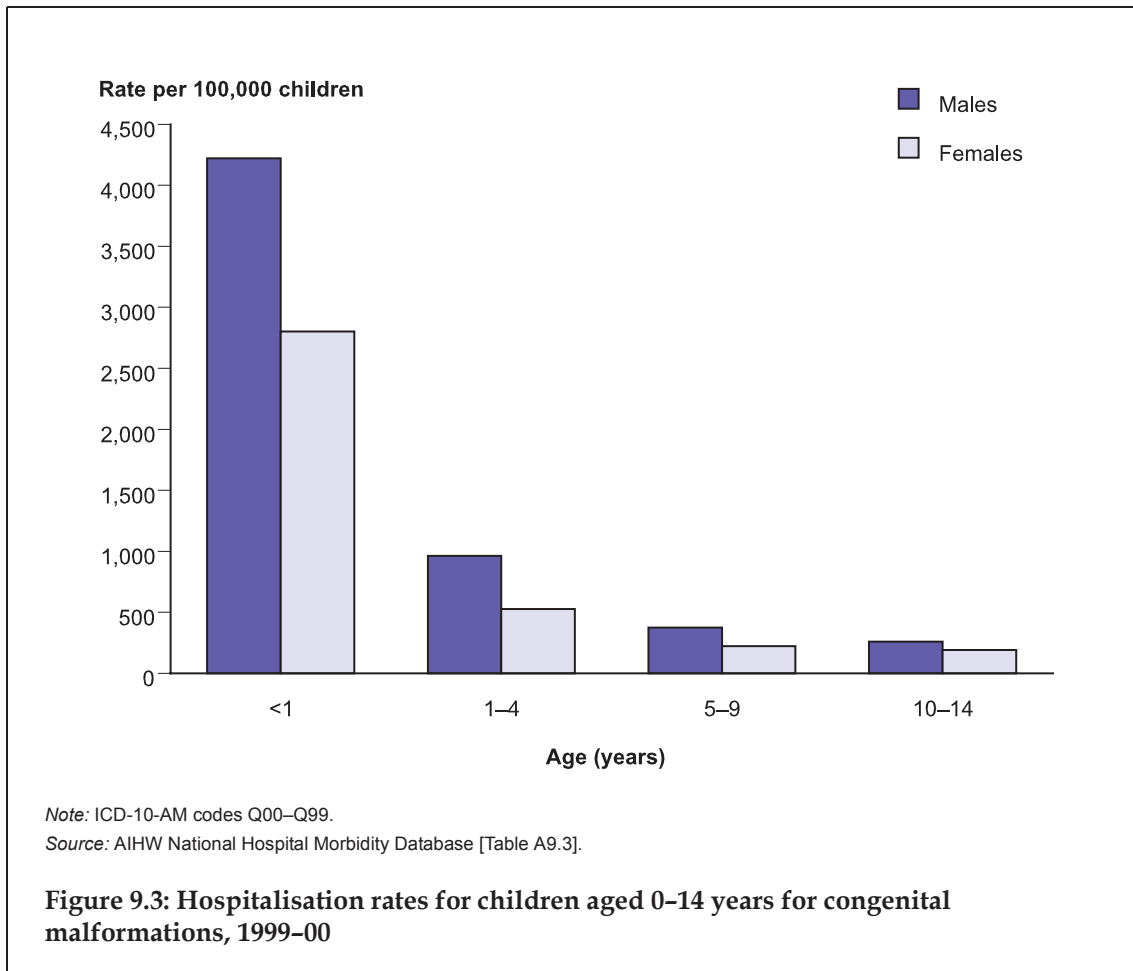
malformations. The notification rate for Down syndrome was 13.0 per 10,000 births in 1997.

- Neural tube defects (spina bifida, anencephalus and encephalocele) affected 146 infants in 1997. Such defects have decreased by about half since 1987 (5.7 compared with 11.9 per 10,000 births), with the greatest reductions seen in spina bifida and encephalocele. The occurrence of neural tube defects is thought to be reduced by giving folic acid to pregnant women and even before pregnancy (Kennedy 1998).

In 1996, 527 terminations of pregnancy for foetal malformations were notified to the NPSU. Ascertainment of terminations is incomplete and varies by State and Territory. The most common reasons for termination of pregnancy were Down syndrome, other chromosomal abnormalities, neural tube defects (including spina bifida, anencephalus and encephalocele) and cardiac defects.

The duration of pregnancy was not known for 68.1% of notified terminations. Among pregnancies for which the gestational age was known, chromosomal malformations, particularly Down syndrome, were the main reasons for the majority of terminations occurring before 16 weeks gestation. Terminations due to anencephalus, spina bifida and other malformations were more likely to occur just before 20 weeks, when most pregnant women are screened by ultrasound examination (AIHW NPSU: Hurst et al. 1999).

## Hospitalisations



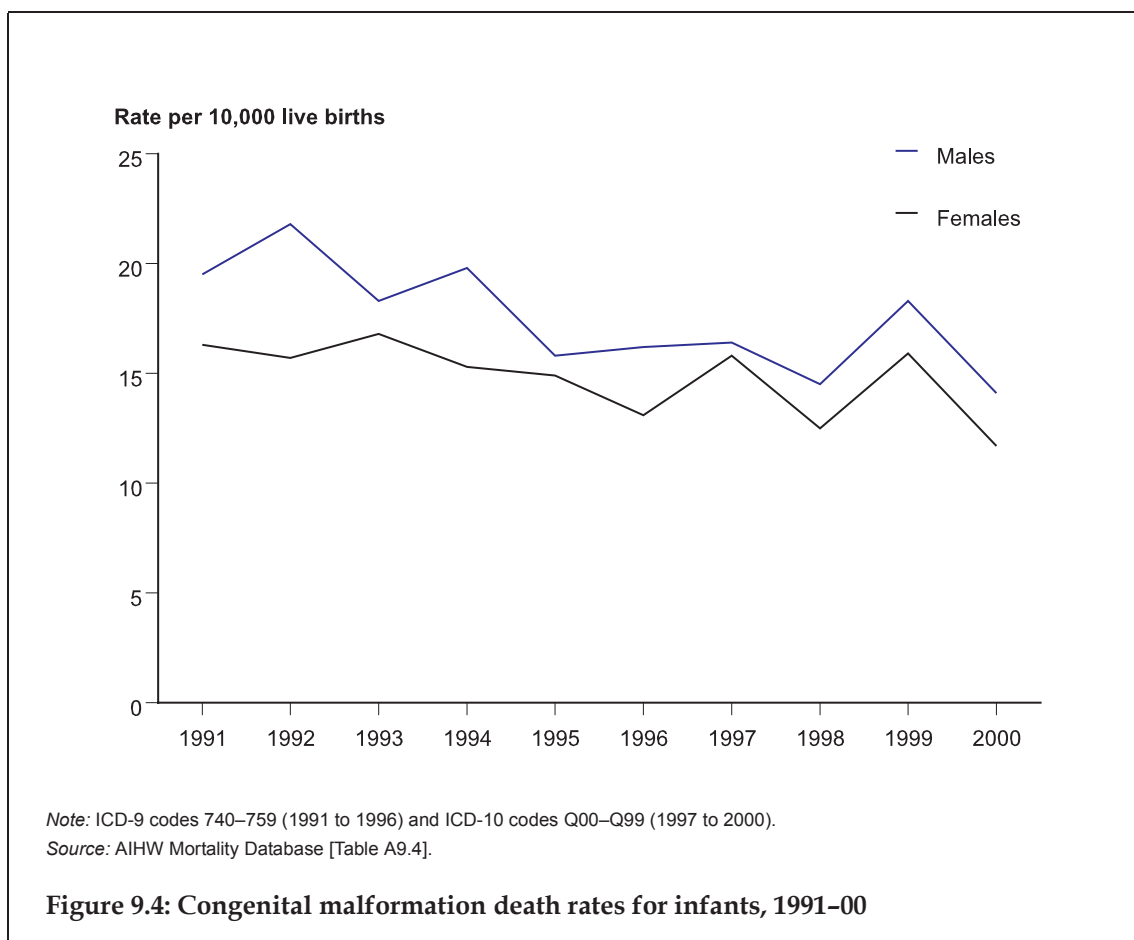
- In 1999–00, there were 23,489 hospitalisations of children aged 0–14 years for congenital malformations. Of these, 8,842 or 38% were infants.
- Children under the age of 1 year had the highest hospitalisation rate among children for congenital malformations (4,220.8 per 100,000 boys and 2,802.4 per 100,000 girls).
- For all age groups, boys had higher hospitalisation rates than girls. This difference was greatest among children aged 1–4 years, where boys were hospitalised at a rate 1.8 times that of girls. The difference was least among children aged 10–14 years.

The impact of congenital malformations can also be examined in terms of the length of time children spent in hospital. In 1999–00, the average length of stay was 2.7 days, with 81,136 hospital bed days with a principal diagnosis of congenital malformations. Congenital malformations were also responsible for an additional 22,976 bed days where they were not the main reason for hospital stay but where they had to be managed during hospitalisations for other conditions.

## Deaths

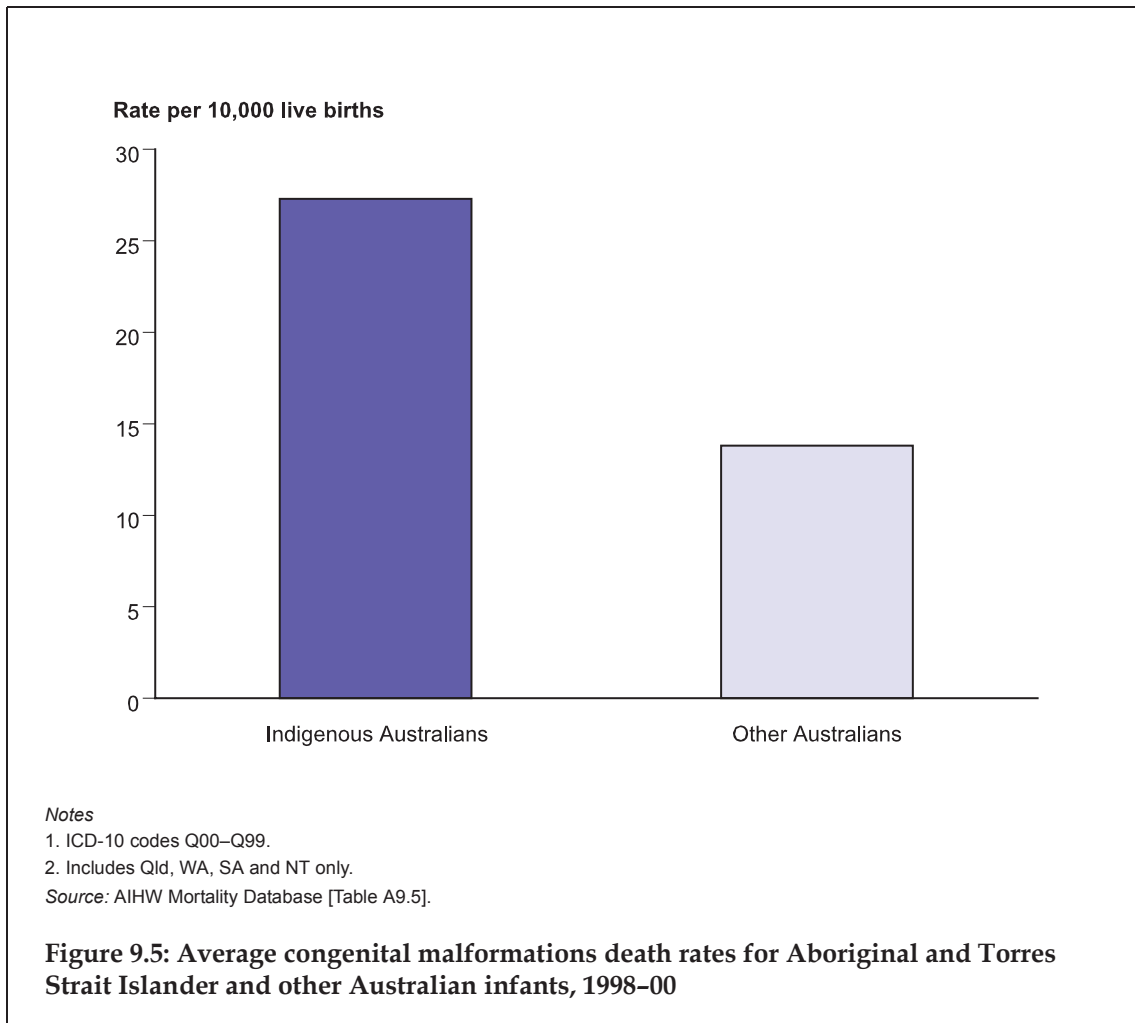
Congenital malformations are a major cause of childhood mortality in Australia. In 2000, they were responsible for 25% of all deaths in children aged under 1 year and for 8% of all deaths in children aged 1–14 years. Congenital malformations of the circulatory system and of the nervous system were the two largest contributors, accounting for 8% and 4% respectively of all infant deaths from congenital malformations. This was followed by malformations of the respiratory system, which accounted for 2% of infant deaths from congenital malformations (ABS 2001b).

The indicator for congenital malformation deaths is the number of deaths of infants from congenital malformations in a given year as a rate per 10,000 live births. In 2000, there were 323 deaths of infants from congenital malformations.



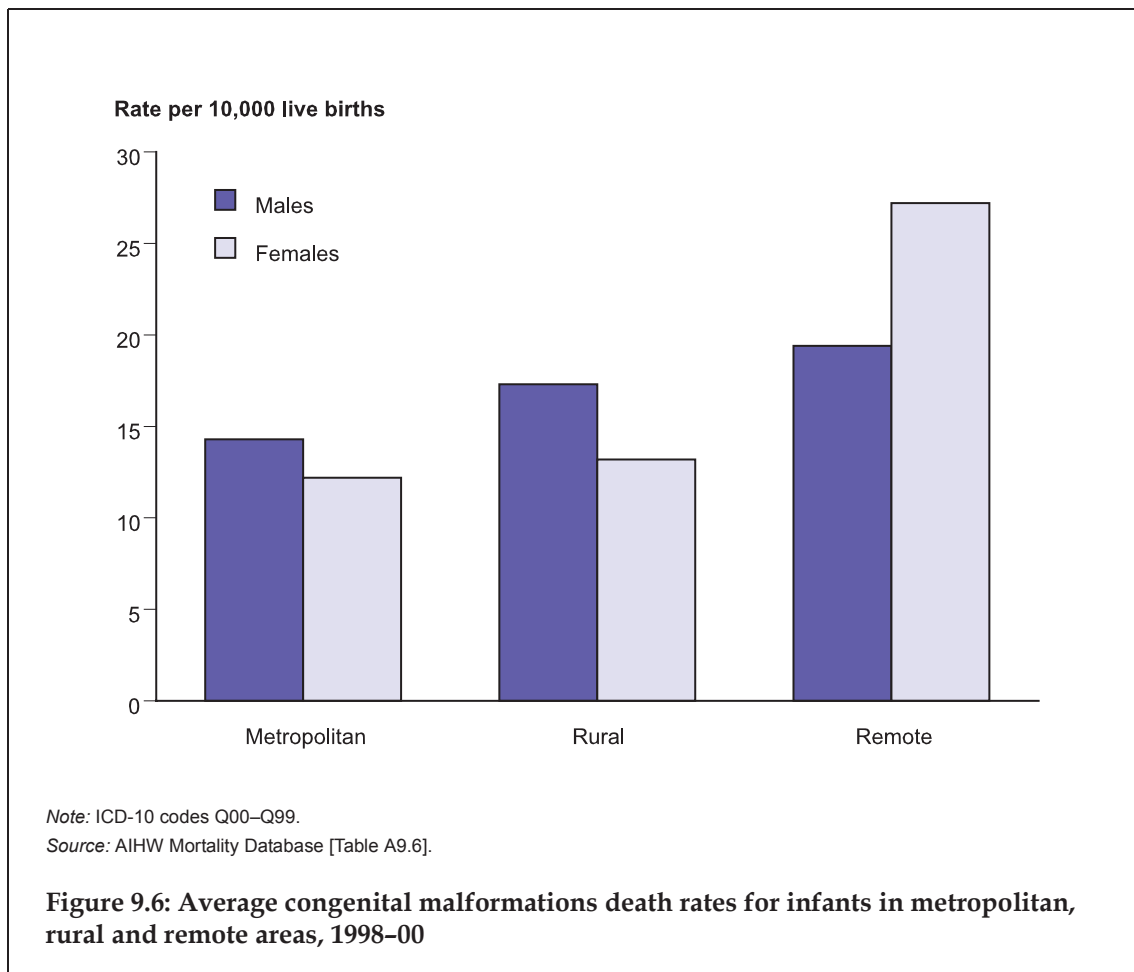
- There was a general decline in the death rate of infants from congenital malformations over the period 1991–00, from 18.0 to 12.9 per 10,000 live births. However, there was a rise in 1997 for females, and in 1999 for all infants.
- Male infants had a higher death rate from all congenital malformations than did females. In 2000, the death rate of male infants was 1.2 times that of female infants.

Aboriginal and Torres Strait Islander children



- Between 1998 and 2000 in Queensland, Western Australia, South Australia and the Northern Territory, there were 55 deaths of Aboriginal and Torres Strait Islander infants from congenital malformations.
- Aboriginal and Torres Strait Islander infants died at a rate twice as high as that for other Australian infants (27.3 compared with 13.8 per 10,000 live births).

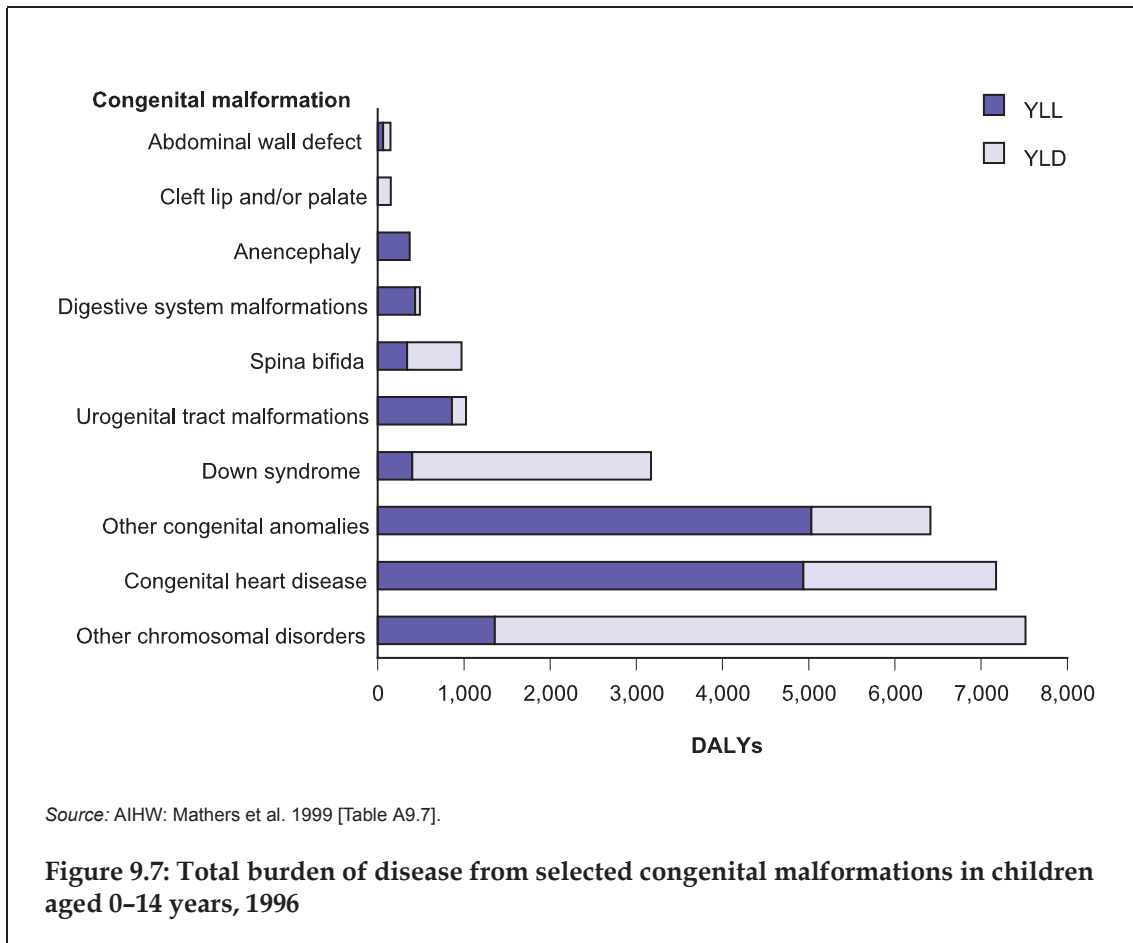
### Children in metropolitan, rural and remote areas



- Between 1998 and 2000, there were 709 infants in metropolitan areas, 283 in rural areas and 68 in remote areas who died from congenital malformations.
- Average death rates were higher in remote areas than in rural or metropolitan areas (23.2 compared with 15.3 and 13.3 per 10,000 live births, respectively).
- In metropolitan and rural areas, death rates were higher among male infants than female infants. However, the pattern was reversed in remote areas, where female infants died at a rate of 27.2 per 10,000 live births, and male infants died a rate of 19.4.

### Burden of disease attributable to congenital malformations

In 1996, congenital malformations were estimated to account for 12.9% of the total disease burden in children aged 0–14 years (27,422 DALYs). The total burden of disease was higher in boys (56% of the burden in children) than in girls (44%). The congenital malformations disability and mortality burdens were relatively even (13,635 YLD, 13,787 YLL). The total burden of disease from selected congenital malformations is shown in Figure 9.7.



- 'Other chromosomal disorders' were responsible for the greatest total burden of disease in 1996 among children aged 0-14 years, followed by congenital heart disease, 'other congenital anomalies' and Down syndrome.
- Most of the disease burden in congenital heart disease, urogenital tract malformations, digestive system malformation and anencephaly was from premature mortality, while in spina bifida, Down syndrome and other chromosomal disorders the main burden was from disability.

