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.. not available for a specific reference period
... not applicable
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0^s value rounded to 0 (zero) where there is a meaningful distinction between true zero and the value that was rounded
^{p} preliminary
^{r} revised
x suppressed to meet the confidentiality requirements of the *Statistics Act*
^{E} use with caution
F too unreliable to be published
* significantly different from reference category ($p < 0.05$)

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Deaths from congenital anomalies in Canada, 1974 to 2012

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Highlights

- In 2012, congenital anomalies were the leading cause of infant death in Canada.
- From 1974 to 2012, deaths from congenital anomalies in Canada decreased from 53 to 28 deaths per 1,000,000 people. This difference can mostly be explained by changes in the death rates for circulatory system anomalies and spina bifida.
- Overall, the average age at death from a congenital anomaly has increased by 19 years since 1974, when the average age at death was 8 years old.
- The average age at death from Down syndrome increased by 32 years between 1974 and 2012.

Introduction

Congenital anomalies, commonly referred to as "birth defects", describe conditions that develop in the fetus at conception or during pregnancy.[1,2] In Canada, approximately 1 in 25 babies is born with a congenital anomaly annually. While congenital anomalies can be present at the time of conception (e.g., in the case of Down syndrome), they more commonly develop by the end of the seventh week of pregnancy (e.g., in the case of spina bifida), or between the eighth and sixteenth weeks of pregnancy.[2] There are several hundred separate congenital anomalies, and for the majority the cause is unknown.[1]

Risk factors for congenital anomalies include: genetics, maternal age, lifestyle factors (e.g., smoking, drinking or using drugs during pregnancy), medication use during pregnancy, maternal chronic diseases (e.g., diabetes, obesity and thyroid disease), and environmental factors (e.g., pollution).[1,2] Although the vast majority (more than 93%) of infants born with a congenital anomaly survive, in Canada, congenital anomalies are one of the leading causes of infant death (a death before the age of 1).[1,2,3]

In this article, information on deaths from five congenital anomalies is presented: circulatory system anomalies, spina bifida, and three chromosomal anomalies—Down syndrome, Edwards' syndrome and Patau's syndrome. Death from a congenital anomaly refers to all deaths where the underlying cause of death was coded as a congenital anomaly since 1974 according to the International Classification of Diseases (ICD), 8th, 9th, and 10th revisions. Trends in the average age at death and death rates from 1974 to 2012 are presented using data from the Canadian Vital Statistics – Death Database.

Definitions of congenital anomalies examined in this article

Congenital anomalies describe developmental or physical disorders that occur during the development of the fetus. They can be identified during pregnancy, after birth, or later in life. There are hundreds of different congenital anomalies. The anomalies examined in this article include:

Circulatory system anomalies are a group of congenital anomalies that mainly affect the structure of the heart and how the heart functions. They are commonly referred to as "heart defects".

Spina bifida affects the central nervous system.[2] It occurs when a portion of the neural tube fails to develop or to close properly causing developmental problems in the bones of the spine and spinal cord. The condition results in medical complications throughout an individual's life and lowers life expectancy.[4]

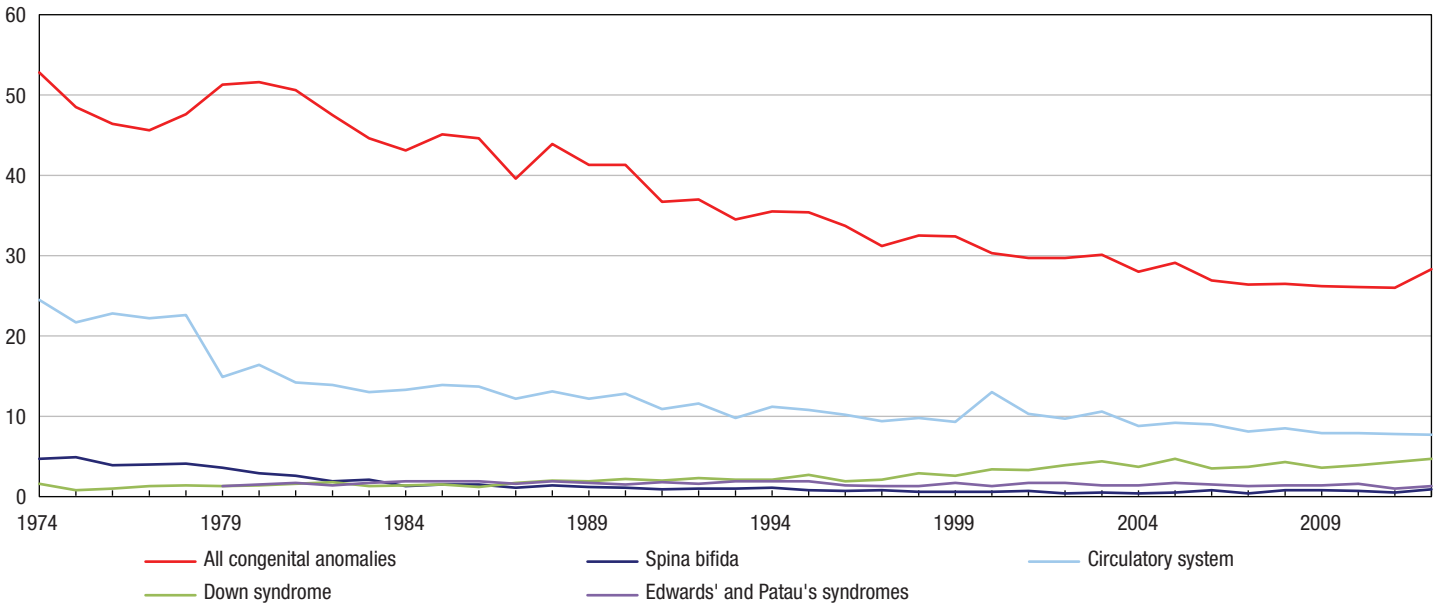
Chromosomal congenital anomalies are caused by problems that occur in the structures (chromosomes) that carry genetic material. This paper explores three chromosomal anomalies:

Down syndrome, also known as trisomy 21, is caused by the presence of extra genetic material on chromosome 21. It causes lifelong physical and intellectual delays in development that can range from mild to severe. Health concerns include a higher risk of heart problems, recurrent ear infections, sleep problems, thyroid disease, leukaemia, and early onset Alzheimer's disease.[1]

Edwards' syndrome, also known as trisomy 18, is caused by the presence of extra genetic material on chromosome 18. Infants with this syndrome have multiple problems that affect the heart, kidneys, and intestines. The majority of affected infants die within the first month after birth and only 10% survive until 1 year of age.[1]

Patau's syndrome, also known as trisomy 13, is caused by the presence of extra genetic material on chromosome 13. It is associated with intellectual disability and physical disabilities such as heart, brain and spine problems. Like infants born with Edwards' syndrome, many infants born with Patau's syndrome die within the first month of life, and only 5% to 10% survive until 1 year of age.[1]

Chart 1
Age-standardized mortality rate, by congenital anomaly, Canada, 1974 to 2012[1]



1. Data for Edwards' and Patau's syndromes are not available before 1979.
Note: See the "Data sources, methods and definitions" box for definitions of the variables in this chart.
Source: Statistics Canada, Canadian Vital Statistics – Death Database.

Deaths from congenital anomalies have been decreasing since 1974

In Canada, the age-standardized death rate from all congenital anomalies combined, has been decreasing since 1974, from 53 to 28 deaths per 1,000,000 people in 2012 (Chart 1).

Looking deeper at specific anomalies, the death rates from circulatory system anomalies and spina bifida have decreased markedly since 1974.

Decreases in deaths from circulatory system anomalies were likely a result of medical advancements. Studies have found that surgery during pregnancy, or shortly after birth, can improve the survival of infants with circulatory system anomalies that affect the heart.[2, 5, 6, 7]

Decreases in the death rate from spina bifida were also likely a result of early surgical intervention[6] as well as changes in prenatal care, including folic acid supplementation. It is recommended that women of childbearing age take an additional 0.4 milligrams (mg) of folic acid per day beyond the 0.4 mg recommended for the rest of the population. Furthermore, in 1998, Canada approved the mandatory folic acid fortification of white flour and other selected grains. Since folate deficiency is the most well-established risk factor for spina bifida, both the use of folic acid supplements and the folic acid fortification of some foods have reduced the number of babies born with spina bifida.[8, 9, 10]

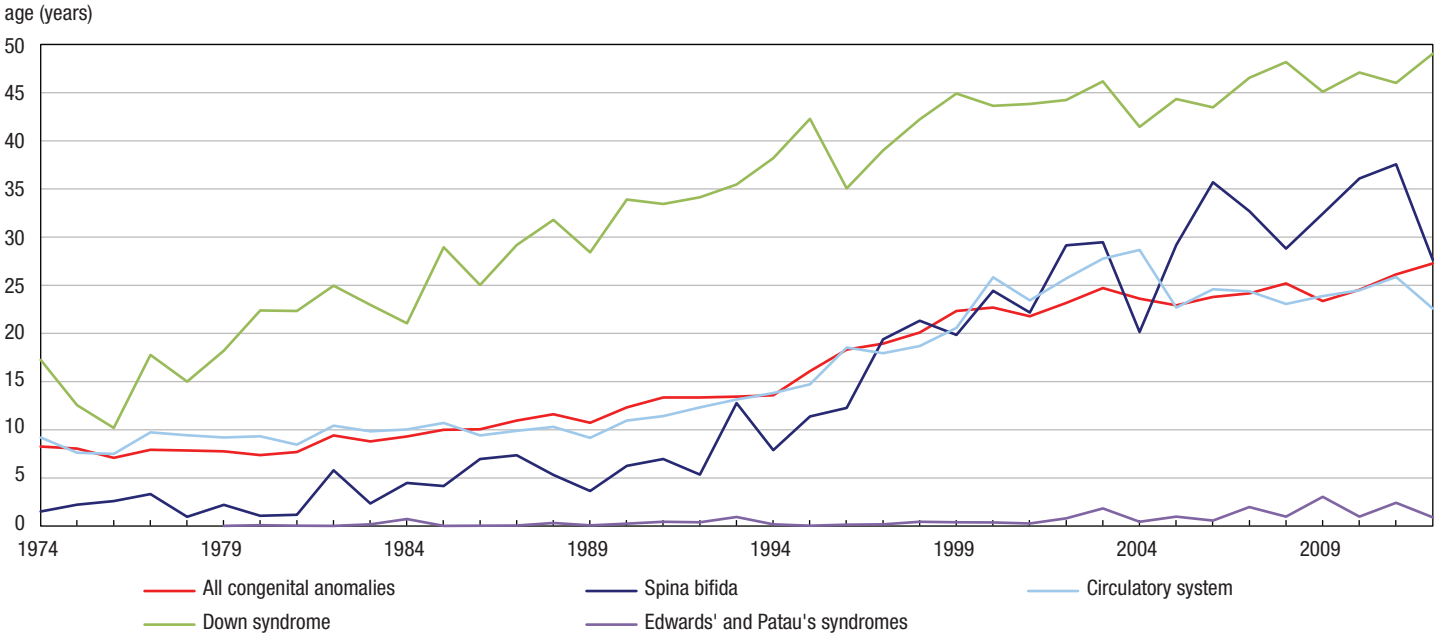
From 1974 to 2012, the death rate from the chromosomal anomalies studied in this article did not decrease. While the death rate from Edwards' and Patau's syndromes[11] remained relatively stable over this period, the death rate from Down syndrome increased from 2 deaths per 1,000,000 people in 1974 to 5 deaths per 1,000,000 people in 2012 (Chart 1).

The average age at death from a congenital anomaly has increased in recent decades

Most pregnancies and births affected by congenital anomalies do not result in the death of a fetus or an infant.[1] In Canada, those who are dying from a congenital anomaly are dying at older ages than in previous decades (Chart 2).

In Canada, the average age at death from all congenital anomalies increased by approximately 19 years between 1974 and 2012, from 8 to 27 years of age (Chart 2). This finding can partly be explained by changes in the infant death rate from these anomalies. In 1974, approximately 70% of deaths from all congenital anomalies occurred before the age of 1—this dropped to 42% by 2012 (data not shown).

Chart 2
Average age at death, by congenital anomaly, Canada, 1974 to 2012[1]



1. Data for Edwards' and Patau's syndromes are not available before 1979.
Note: See the "Data sources, methods and definitions" box for definitions of the variables in this chart.
Source: Statistics Canada, Canadian Vital Statistics – Death Database.

When specific anomalies were examined, the greatest increase in average age at death was for those who died from Down syndrome. For this syndrome, the average age at death increased by 32 years between 1974 and 2012. This was followed by spina bifida, where the average age at death increased by 26 years, from 2 years to 28 years of age over the same time period (Chart 2).

The average age at death from a circulatory system anomaly was 23 years in 2012 compared with 9 years in 1974.

In contrast, in the case of Edwards' and Patau's syndromes, where most infants do not survive past the first year of life, the average age at death has increased very little from 1974 to 2012.

The proportion of infant deaths caused by congenital anomalies has increased

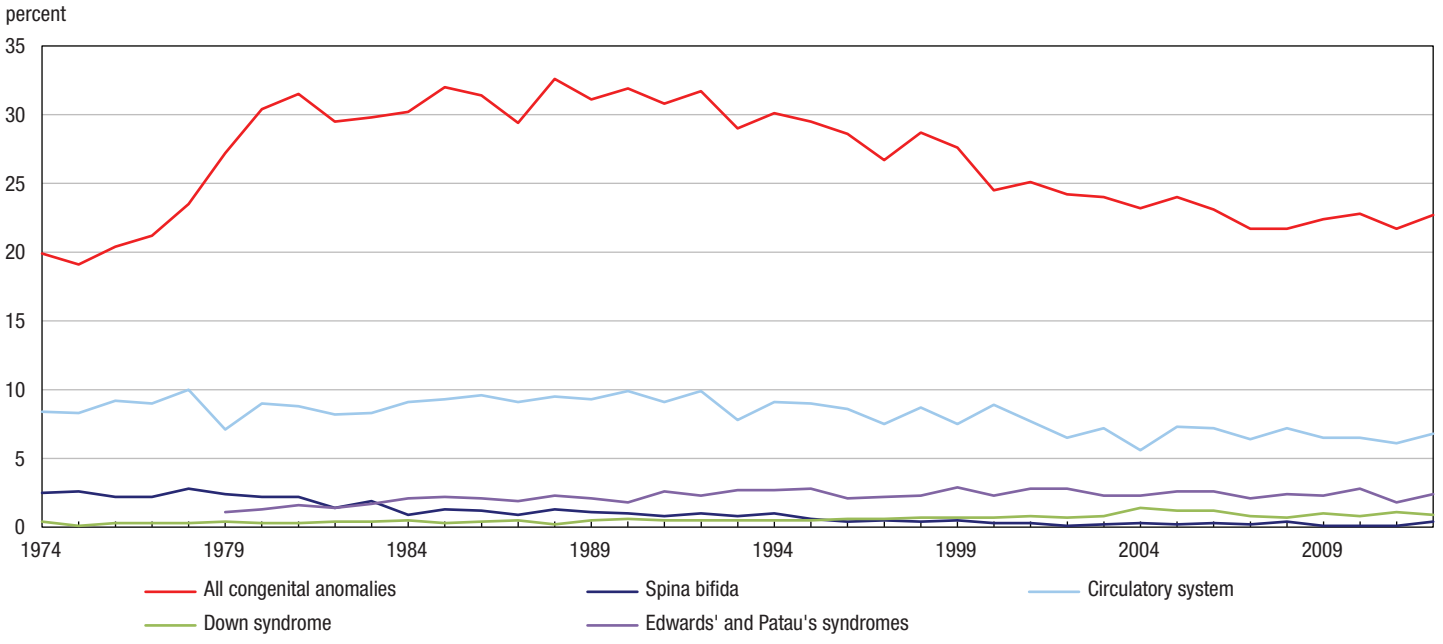
The proportion of all infant deaths, caused by congenital anomalies increased sharply between 1974 and 1981 but has been decreasing gradually since the early-1990s (Chart 3).

The increase in the proportion of infant deaths from congenital anomalies seen between 1974 and 1981, can be partly explained by decreases in other causes of infant death during this time period. The introduction of car seats for children led to fewer deaths from car accidents, and increased vaccine use led to fewer deaths from infectious diseases, just to name a few examples.[12]

That is, while preventable causes of infant death have been declining (e.g., accidents, death from infectious diseases), many congenital anomalies are not preventable so the proportion of infant deaths from congenital anomalies appears to stay the same or decrease little over the time period studied.[13] This does not mean there are more deaths from congenital anomalies, it means that relative to other causes, congenital anomalies have become one of the main causes of infant death. In fact, in 2012, congenital anomalies were the leading cause of infant death in Canada, followed by infant immaturity (i.e., short gestation and low birth weight), and complications related to pregnancy.[14]

Focusing on specific congenital anomalies, the percentage of infant deaths caused by circulatory system anomalies and spina bifida decreased between 1974 and 2012, while the percentage of infant deaths caused by Down syndrome increased. There was no change in the percentage of infant deaths caused by Edwards' and Patau's syndromes between 1974 and 2012. Together these two syndromes (Edwards' and Patau's) accounted for less than 3% of all infant deaths during this time period.

Chart 3
Percentage of all infant deaths[1] caused by congenital anomalies, Canada, 1974 to 2012[2]



1. Infant death is a death that occurs before the age of 1.
2. Data for Edwards' and Patau's syndromes are not available before 1979.
Note: See the "Data sources, methods and definitions" box for definitions of the variables in this chart.
Source: Statistics Canada, Canadian Vital Statistics – Death Database.

The infant death rate from congenital anomalies has declined

Overall, infant deaths declined in Canada from 15.0 deaths per 1,000 births in 1974 to 4.8 deaths per 1,000 births in 2012 (data not shown). Similarly, the infant death rate from all congenital anomalies has declined from 3.0 deaths per 1,000 births in 1974 to 1.1 deaths per 1,000 births in 2012 (Chart 4).

The decline in infant deaths from all congenital anomalies has been influenced by various factors, including: increased access to, and use of, prenatal screening; improved prenatal care; termination of pregnancies affected by major anomalies; mandatory folic acid fortification of certain foods; and lifestyle changes, such as smoking cessation during pregnancy and increased use of prenatal vitamins.[1,2]

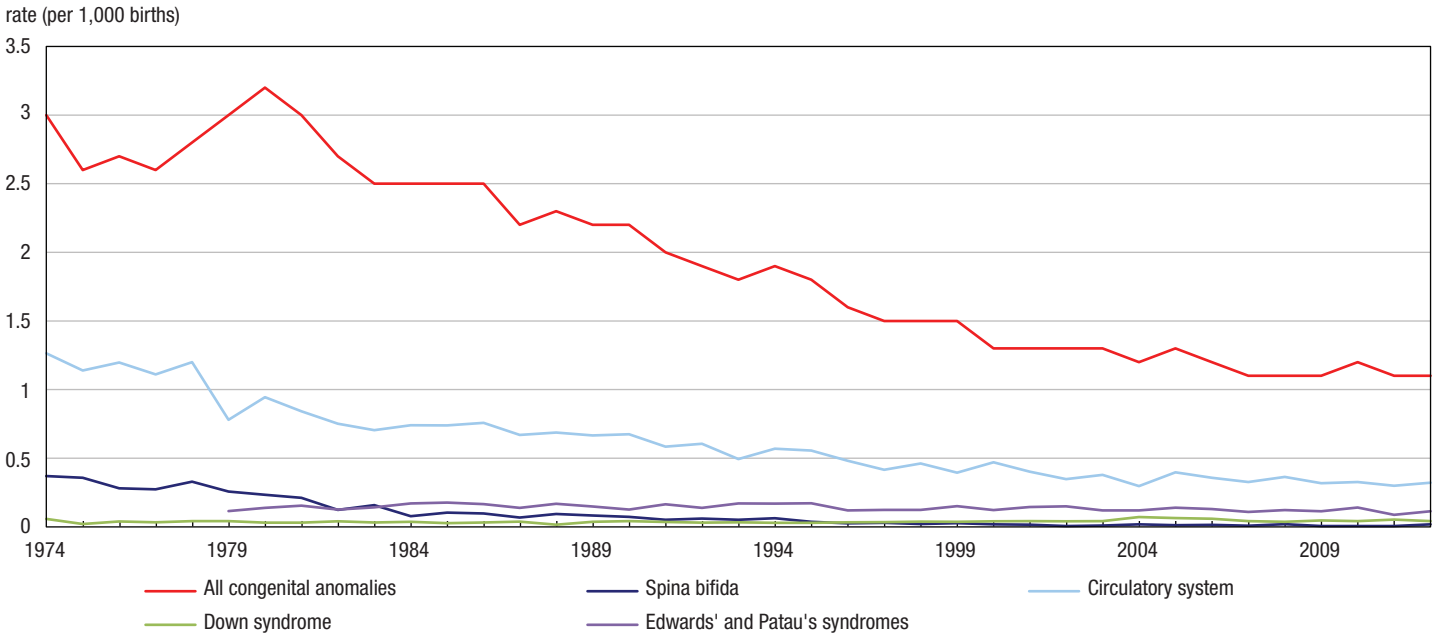
The greatest declines in the infant death rate were seen for circulatory system anomalies and spina bifida. For instance, infant deaths caused by circulatory system anomalies declined from a rate of 1.3 deaths per 1,000 births in 1974 to 0.3 deaths per 1,000 births in 2012. This decline was likely the result of increased early surgical repair.[2] For spina bifida, surgical intervention as well as the use of prenatal vitamins containing folic acid and the mandatory folic acid fortification in food products has resulted in fewer cases and, therefore, reduced the number of infant deaths from this congenital anomaly.[5,9]

Infant deaths caused by chromosomal anomalies such as Down syndrome, and Edwards' and Patau's syndromes have remained relatively stable since 1974 (Chart 4). Chromosomal anomalies are less responsive to surgical treatment.[1]

Conclusion

Deaths caused by all congenital anomalies have decreased in Canada since 1974. Deaths from circulatory system anomalies and spina bifida have decreased the most during this time period. Prenatal screening, surgical intervention, as well as changes to prenatal care (such as folic acid supplementation) and folic acid fortification of certain foods have contributed to this decline. Deaths from chromosomal anomalies, which represent a small percentage of all infant deaths, have not changed over the studied time period probably because prevention efforts have had less of an

Chart 4
Infant death[1] rate, by congenital anomaly, Canada, 1974 to 2012[2]



1. Infant death is a death that occurs before the age of 1.
2. Data for Edwards' and Patau's syndromes are not available before 1979.

Note: See the "Data sources, methods and definitions" box for definitions of the variables in this chart.
Source: Statistics Canada, Canadian Vital Statistics – Death Database and Birth Database.

impact on these anomalies. With the exception of Edwards' and Patau's syndromes, the average age at death from congenital anomaly is higher than it was in 1974.

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Data sources, methods and definitions

Data sources

The Canadian Vital Statistics – Death Database includes demographic and cause of death information for all deaths recorded on all provincial and territorial vital statistics registries in Canada. Prior to 2010, data were collected on Canadian residents who died in some American states; these deaths were excluded from this analysis. Starting with the 2010 reference year, data on Canadian residents who died in American states are no longer collected.

The Canadian Vital Statistics – Birth Database collects demographic information on all live births annually in Canada from all provincial and territorial vital statistics registries. Prior to 2012, some data were also collected on live births to Canadian women residing in some American states. These births were excluded from this analysis.

Methods

Age-standardized mortality rates can be used to control for population differences in age-structure when comparisons are made at different points in time and/or for different populations. In this study, the populations for all years assessed (1974 to 2012) were mathematically adjusted to have the same age structure as the 2011 Census population. In this way, all the years of data were given the same age distribution to present a clearer picture of the trend in deaths from congenital anomalies when rates are compared over time.

Classification of infant deaths caused by congenital anomalies

The **International Classification of Diseases (ICD)** is a coding system developed by the World Health Organization with the goal of permitting the comparison of different causes of death between and within different countries at different points in time.[15]

The following **ICD** diagnosis codes were used to classify deaths from congenital anomalies:[16]

Congenital Anomaly	ICD Code
All congenital anomalies	ICD-10 Q00-Q99; ICD-9 740-759; ICD-8 740-759
Spina bifida	ICD-10 Q05; ICD-9 741; ICD-8 741
Circulatory system disorders	ICD-10 Q20-28, Q290; ICD-9 746-747; ICD-8 746-747
Down syndrome	ICD-10 Q90; ICD-9 758.; ICD-8 759.3
Edwards' and Patau's syndromes	ICD-10 Q91; ICD-9 758.1-758.2; **Note:** No ICD-8 codes available.

The study period covers ICD-8, ICD-9 and ICD-10. New versions of the ICD are developed periodically to keep the classification system current. Revisions to the classification system may result in certain causes of death being placed in different categories (i.e., coded differently). Differences in coding could result in differences in death counts over time.[16, 17, 18]

Definitions

Underlying cause of death is defined as the disease or injury that initiated the series of events that led to death.[15]

Infant death is a death that occurs before the age of 1.

Infant death rate is the number of infant deaths per 1,000 live births.

Prenatal screening are tests that can be done during pregnancy to assess the risk that a fetus has a congenital anomaly.

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