



**Summary of ten most frequently reported birth defects in Victoria  
2003-2004**

The Victorian Birth Defects Register has collected information on birth defects in all reported pregnancies occurring in Victoria since January 1st, 1982. The maintenance of this register is an ongoing function of the Victorian Perinatal Data Collection Unit at The Department of Human Services. One of its many functions is to provide information to those concerned about having a baby with a birth defect. This publication, a summary of their latest report: 'Birth Defects in Victoria 2003-2004' is designed to make the information easily accessible to the general public.

When facing the possibility of having a child with a birth defect, parents are often overwhelmed with concerns and questions. To help provide answers to some of their questions now and in the future the Victorian Birth Defects Register uses the information it collects to conduct research that helps find factors that may lead to a birth defect. We hope this publication will show you what we found in the 2003-2004 period. At the end of this report we have included a list of websites that you may find useful. A glossary of terms frequently used when talking about birth defects has also been attached to aid your understanding.

For the full report please visit the **Victorian Birth Defects Register Website:**

<http://www.health.vic.gov.au/perinatal/vbdr/index.htm>

#### **What is a birth defect?**

A birth defect is any abnormality that may be detected during pregnancy, at birth or in early childhood. This includes structural, functional, genetic, chromosomal and biochemical changes. Birth defects can range from minor to very severe conditions and can affect pregnancies and babies in a variety of ways.

#### **How often are babies born with a birth defect?**

In Victoria approximately 1 in every 25 babies (4%) is affected by a birth defect each year. This represents approximately 2,700 babies a year in Victoria that have a birth defect. In the case of a serious birth defect that can be detected during pregnancy, parents can elect to discontinue the pregnancy. Therefore not all reported cases of birth defect represent live births.

#### **What are the more common birth defects?**

The ten most common birth defects are discussed over the next few pages of this publication in the order of frequency in the Victoria population.

#### **What factors influence the prevalence of birth defects?**

As well as sex of the child and the number of fetuses (twins, triplets, quads), the Birth Defects Register also collects maternal information such as the mother's country of birth, region of residence and age. Some of these factors have been shown to influence the chance of having a baby with a birth defect.

##### Maternal age

The frequency of birth defects is highest among mothers aged 35 years and over. These women have almost a one and a half times increased risk of having a pregnancy affected by a birth defect compared to younger women. By the time a woman is 40 years and over her chance is almost twice as high (7.4%). Hydrocephalus, ventricular septal defect, cleft palate, cleft lip and palate, congenital dislocated hip, trisomy 21 and trisomy 18 are all more likely to occur with older mothers. Younger maternal age was significantly associated with all neural tube defects, and obstructive defects of the renal pelvis.

##### Sex

Female babies are more likely to have a birth defect than male babies; 4.7% of males compared with 3.7% of female babies had a birth defect in 2003-2004.

##### Plurality

Multiple births (twin, triplets, and quads) are also slightly more likely to be affected by a birth defect than singleton births; 5% of multiple births had a birth defect compared to 4.3% of singleton births that had a birth defect.

## 1. Hypospadias

Hypospadias is a birth defect of the penis that involves an abnormally placed urethral opening. The urethra is a tube that connects the bladder to the outside of the body. In hypospadias the urethra does not extend to the tip of the penis like it normally should; instead, the opening of the urethra is located somewhere along the underside of the penis. Hypospadias is rarely severe and our recent data shows a 99% survival rate. Hypospadias is usually treated with surgery, which extends the urethra to the top of the penis. This occurs when the child is between 6 and 18 months old. In the vast majority of babies with Hypospadias, no other birth defects were present.

## 2. Obstructive defects of the renal pelvis

An obstructive defect of the renal pelvis is caused by a blockage in the ureters and results in an inability to properly drain urine from the kidneys into the bladder. Without treatment an obstructive defect of the renal pelvis can lead to persistent urinary tract infections and kidney failure. The blockage is usually corrected with open surgery. Possible complications of surgery include infection and trauma to structures of the urinary system, however an obstructive defect of the renal pelvis is not a severe birth defect and has a high survival rate (94% in the 2003-2004 period). This birth defect can be detected during a pregnancy ultrasound. A subsequent ultrasound of the baby is usually needed, once it is born, to confirm the diagnosis. From 1993 to 2004 the number of recorded cases of obstructive defects of the renal pelvis has increase is due to a greater ability to detect the defect during pregnancy. Male babies have a two and half times increased risk of having an obstructive defect of the renal pelvis compared to females. Affected babies rarely have other birth defects and association with lethal birth defects is especially uncommon.

## 3. Ventricular septal defect

A ventricular septal defect is a hole in the wall (septum) that divides the lower right and left chambers of the heart. This causes the blood to circulate improperly making the heart work harder than normal. Small holes may heal by themselves or not need repair at all. Alternatively, surgery can be used to close the hole. This defect is rarely

severe and has a high survival rate (88% in 2003-2004). Ventricular septal defects are often associated with other birth defects as shown in the figure below. There is a significantly higher frequency of the defect in babies born to women aged 40 years and over when compared to younger women.

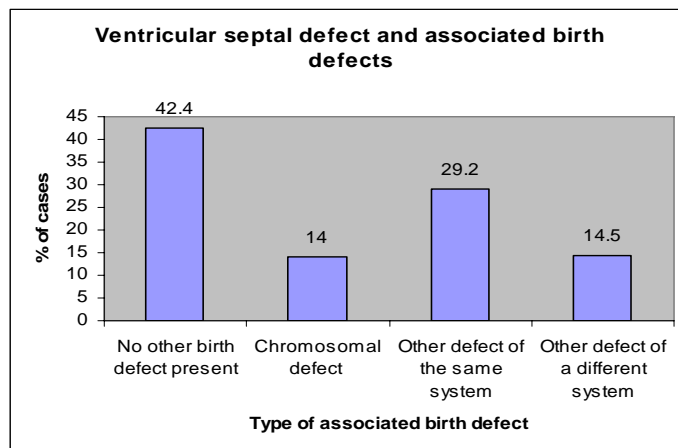


Figure 1: Ventricular septal defect and associated birth defects.

## 4. Congenital dislocated hip

A congenital dislocated hip, also known as developmental dysplasia of the hip is when the ball of the thigh bone doesn't fit snugly into the hip bone socket. Dislocation of both hips is not uncommon. Congenital dislocated hip has a 100% survival rate and around 95% of babies can be successfully treated. Most babies affected by a congenital dislocated hip do not have another birth defect. Female babies are three times more likely to have a congenital dislocated hip when compared to male babies.

## 5. Trisomy 21 or Down syndrome

Trisomy 21 or Down syndrome is quite a well known birth defect. The presence of an extra copy of chromosome 21 affects mental and physical characteristics. The number of identified cases of Down syndrome is increasing each year in Victoria mostly due to the use of first trimester screening. The early screening identifies some fetuses that may have spontaneously miscarried and would normally not have entered the statistics. In addition there are more older women having babies and advanced maternal age is association with an increased frequency of Down syndrome as shown in the graph below. Due to the severity of this birth defect a large

proportion (72%) of pregnancies affected by Down syndrome are discontinued. However the number of babies that are live born with Down syndrome has not declined.

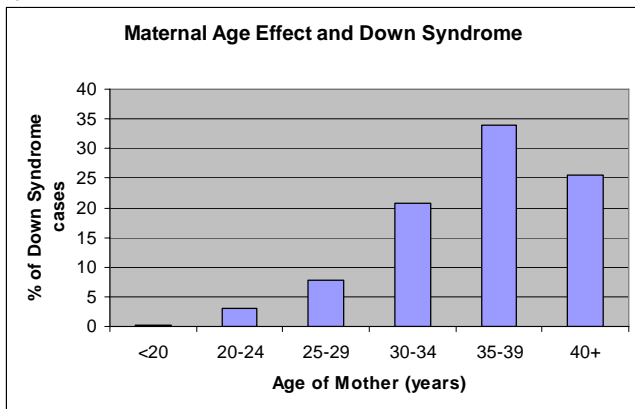


Figure 2: Maternal Age Effect and Down syndrome.

### 6. Hydrocephalus

Hydrocephalus is an abnormal buildup of the brain's regular water-like fluid, cerebrospinal fluid (CSF.) Hydrocephalus cannot be cured, just treated; untreated it can result in brain damage or death. Treatment depends on the underlying cause of the fluid build up; often surgery is used to place a small synthetic tube (a shunt) into the ventricle to drain the excess fluid. Other medical conditions are often associated with hydrocephalus creating more serious problems. Hydrocephalus can be a severe birth defect and some parents (42%) elect to discontinue a pregnancy when an ultrasound detects this condition. The graph below shows the proportion of pregnancy outcomes for the 2003-2004 period.

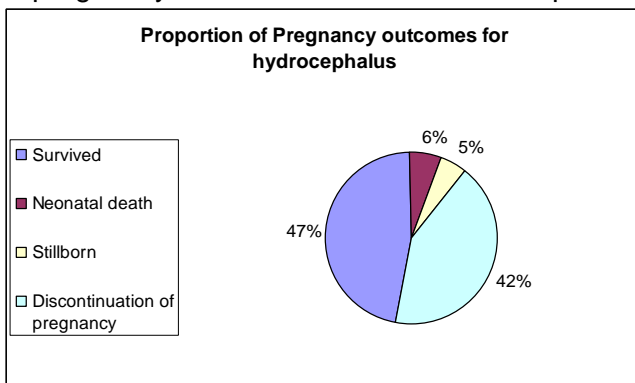


Figure 3: Proportion of pregnancy outcomes for hydrocephalus

### 7. Cleft Palate

A cleft is a birth defect caused by the insufficient

fusion of the mouth parts during development. During the first two months of pregnancy a baby's tissue grows inwards from the sides of the upper jaw and joins in the middle to form the roof of the mouth (palate). Failure to fuse results in a cleft or split in the palate that can affect one or both sides of the palate and lip. Cleft palate is more of a cosmetic, rather than a functional defect although feeding problems are common. The cleft can be surgically corrected at around three months of age, with subsequent surgery on the palate between six months and one year. Pregnancies affected by a cleft palate have a high survival rate (87%). The remainder of cases that are discontinued or result in neonatal death are most likely due to an association with another birth defect, which can occur in a quarter of all cleft palate cases. Cleft palate is almost one and a half times more likely to occur in females than in males.

### 8. Trisomy 18 or Edward Syndrome

Trisomy 18 is a severe birth defect; affected babies rarely survive. Multiple abnormalities of the heart, diaphragm, lungs, kidneys, ureters and palate are caused by an additional copy of chromosome 18. Trisomy 18 has increased in overall prevalence from 1993-2004. This is mostly due to the number of recognized affected pregnancies and subsequent terminations. In the 2003 -2004 period there were 123 identified cases of Trisomy 18; the outcome of these pregnancies is shown in the figure below. As with other trisomies, like Down syndrome, the overall prevalence of Trisomy 18 increases with increasing maternal age

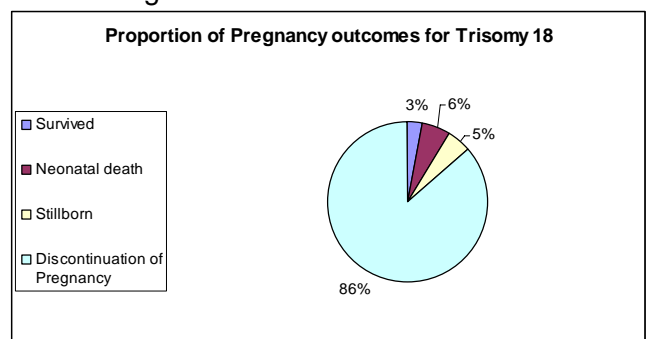


Figure 4: Proportion of Pregnancy outcomes for trisomy 18

### 9. Renal Agenesis/dysgenesis

A birth defect of the urinary system, renal agenesis means that one or both kidneys are missing. A baby can manage with one functioning kidney, since the remaining organ enlarges to cope with the extra workload but if both kidneys are missing, the baby

will not survive. Dysgenesis refers to a malformation of the kidneys. Some of the reduction in neonatal death and stillbirth since 1993 may be due to the more severe cases being identified *in utero* and being represented amongst the increased proportion of terminations (~31%). Just over half of pregnancies affected with renal agenesis/dysgenesis go on to a successful birth. Renal agenesis/dysgenesis is often associated with other birth defects. Males are almost twice as likely to be affected as females.

### 10. Cleft lip and palate

A cleft lip and palate birth defect is a cleft or split in the upper lip as well as in the palate. The lip is formed when two tabs of skin unite from the sides of the face at around 4-6 weeks of the baby's development. A failure to join up properly results in an open space or cleft in the upper lip and palate. Cleft lip and palate is considered a separate birth defect to cleft palate, and like cleft palate, it can be corrected with surgery. Cleft lip and palate occur with another defect in 33.7% of cases. Discontinuation of pregnancy occurs and babies do not survive when a serious birth defect is also present. In a third of cleft lip and palate cases another birth defect was also present.

### Summary of the ten most common birth defects.

With a few exceptions, the majority of birth defects that appear amongst the ten most common are not severe or life threatening to the child. Research often focuses on severe but rare birth defects that in turn become widely known to the general public. In the next section we will explain more about neural tube defects that were just outside the top ten most common birth defects but are widely known.

### Neural Tube Defects

Anencephaly and spina bifida form a group known as neural tube defects. The neural tube, affected in both of these defects, is the embryonic structure that develops into the brain and spinal cord. The prevalence of neural tube defects has decreased over time most likely due to an increased awareness by the public of their prevention by taking folic acid.

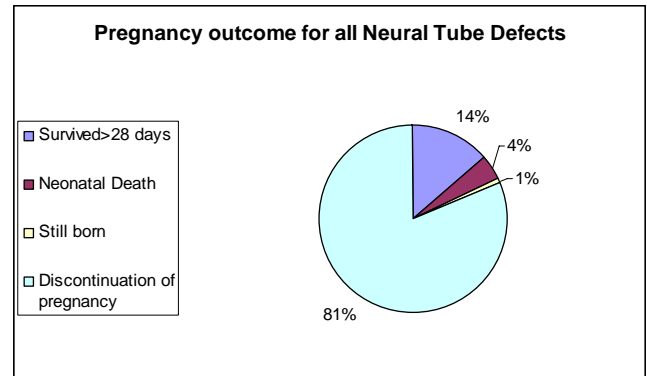


Figure 5: Pregnancy outcomes for all neural tube defects.

### Spina Bifida

Spina bifida is one of the most common severe birth defects. It involves a malformation of the spinal column and affects the backbone and sometimes, the spinal cord. A large number (73%) of parents choose to discontinue a pregnancy affected by spina bifida. Of those that do continue with the pregnancy the survival rate is high. A quarter of spina bifida cases are affected by another birth defect of a different body system and in two thirds of cases Spina bifida is present as the only birth defect.

### Anencephaly

Anencephaly, like spina bifida, is a birth defect of the neural tube and of brain development. When the neural tube fails to close at the head, the brain and the skull bones do not develop normally. Anencephaly is a severe birth defect that is incompatible with life. Most parents (94%) elect to discontinue a pregnancy affected by anencephaly.

## **USEFUL WEBSITES:**

### **Better Health Channel:**

<http://www.betterhealth.vic.gov.au>

The Better Health Channel has fact sheets on most birth defects and disorders. The birth defects page lists all the common birth defects in Victoria from 1983-1998 in order of prevalence with a brief description of each and statistics on how often they occur. The site also contains links to support groups and other related sites.

### **Brain Foundation of Australia:**

[http://www.brainaustralia.org.au/AZ\\_of\\_Brain\\_Disorders/hydrocephalus](http://www.brainaustralia.org.au/AZ_of_Brain_Disorders/hydrocephalus)

The brain foundation website has an extensive description of hydrocephalus and links to support groups.

### **Disability Online**

<http://www.disability.vic.gov.au>

Disability online provides information for people with a disability, their families and carers.

### **Genetic Health Services Victoria**

<http://www.genetichealthvic.net.au>

This website contains information on health services available for those with or concerned about a genetic condition

### **Genetic Support Network Victoria**

<http://www.gsnv.org.au/>

The Genetic Support Network's site contains links and contact information to the major support groups for people affected by a genetic disorder and their families.

### **Hearts of hope**

[www.hearts-of-hope.com/](http://www.hearts-of-hope.com/)

A support group for families living with complex congenital heart conditions.

### **Royal Children's Hospital Website**

[www.rch.org.au](http://www.rch.org.au)

The RCH site has a 'for parents' section which contains fact sheets for a range of disorders including some of the birth defects discussed in this publication.

### **The Cleft palate and lip Society, Victoria.**

<http://www.cleftpalsvic.com/>

### **Down syndrome association of Victoria**

<http://www.dsav.asn.au/>

### **Australian Spina Bifida and Hydrocephalus association**

[www.asbha.org.au/index.htm](http://www.asbha.org.au/index.htm)

This website provides fact sheets on different areas relating to Spina bifida and hydrocephalus.

## **GLOSSARY**

**Cerebrospinal fluid (CSF):** The brain's normal water-like fluid

**Chromosomal birth defect:** A birth defect caused by an alteration in the number or structure of chromosomes

**Chromosome:** Genetic structure of a cell that carries the DNA

**Congenital:** A medical condition that is present at birth

**First Trimester:** The period from conception to 12 weeks of pregnancy

**Gastroschisis:** An abnormality (defect or hole) in the abdominal wall that allows the abdominal contents to protrude outside the body

***In Utero:*** In the uterus, during pregnancy.

**Isolated defects:** Cases of a birth defect present with only a single condition

**Neonatal:** Neonatal refers to the period immediately after birth, up to 28 days

**Neural Tube:** Embryonic structure that develops into the brain and spinal cord.

**Palate:** The roof of the mouth

**Perinatal:** Perinatal refers to the period from 20 weeks pregnancy (or five months before birth) to one month after birth.

**Perinatal death:** Perinatal death is a stillbirth or neonatal death

**Post natal:** Post natal refers to anytime after birth

**Renal pelvis:** The area at the centre of the kidney where urine is collected

**Spinal Column:** A column of vertebrae that houses the spinal cord and its spinal canal.

**Septum:** A wall that divides the left and right sides of the heart

**Severe birth defect:** Birth defects that cause death, hospitalization, mental retardation, interfere with physical performance or; require significant or repeated surgery.

**Trisomies:** A classification of birth defects characterised by the presence of an additional chromosome giving three instead of the usual two copies. Children affected by trisomy usually have a range of birth defects, including delayed development and intellectual disabilities.

**Ureters:** The ducts that carry urine from the kidneys to the urinary bladder

**Urethra:** A tube which connects the urinary bladder to the outside of the body.

**Urinary system:** Body system that produces stores and eliminates urine. It includes kidney, ureters, urinary bladder, urethra and surrounding muscles.