



Victorian Birth Defects Bulletin

No. 5 June 2008

Welcome to the fifth edition of the *Victorian Birth Defects Bulletin*. Its contents are based upon the most recent available data in the Victorian Birth Defects Register (VBDR) – 2005 and 2006

In August, 2008, we will be releasing our biennial report, **Birth Defects in Victoria 2005-2006**. It will be available on our website:

www.health.vic.gov.au/perinatal.

We are producing a limited number of printed copies, so if **you would like us to mail Birth Defects in Victoria 2005-2006** to you please contact us via email :

perinatal.data@dhs.vic.gov.au

OR

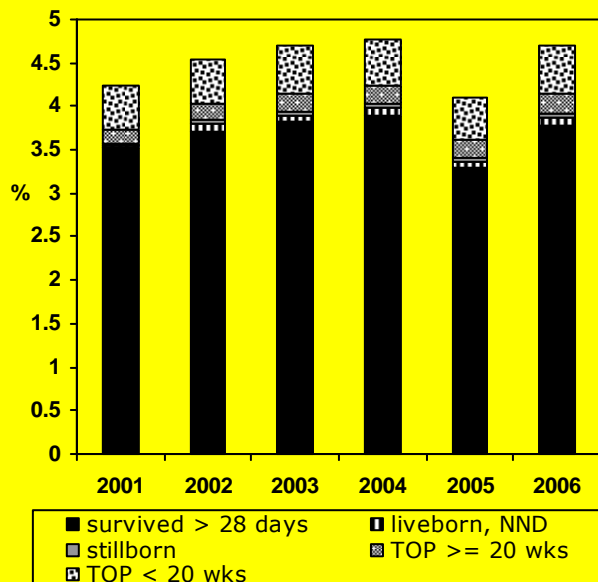
by phone:

1300 858 505.

As usual, any feedback or requests for further information are most welcome.

Jane Halliday and Merylyn Riley

Prevalence of birth defects by outcome, 2001-2006



Overall prevalence of birth defects, 2005 - 2006

- In 2005-2006 there were 5,067 babies born at, or after, 20 weeks gestation with a birth defect. There were another 669 identified as terminations of pregnancy before 20 weeks gestation for a birth defect. This gives an **overall birth defect prevalence rate of 42/1,000 or 4.2%**.

NB: There were also an additional 716 notifications of conditions that are collected by the VBDR but not reported in our routine publications (eg. undescended testes ≥ 37 weeks, vesicoureteric reflux). If these conditions are included the overall birth defect prevalence rate for 2005-2006 is 47/1,000 or 4.7%.

- The **livebirth birth defect prevalence rate** in 2005–2006 was 35/1,000 livebirths or 3.5%.
- 12% of all pregnancies with birth defects were terminated before 20 weeks gestation, and another 5% of birth defects were terminated at 20 weeks or later.
- Of babies with a birth defect born at 20 weeks or later, excluding terminations, 62 (1.1%) were stillborn and 95 (1.6%) were neonatal deaths. This gives a **perinatal mortality rate of babies with birth defects of 33/1,000**. If terminations of pregnancy at 20 weeks or later are included, the perinatal mortality rate increases to 87/1,000 birth defects.

Provision of data

The VBDR encourages the release of data to all health professionals; foremost consideration is that the release of data will not endanger confidentiality of information. All requests must be made **in writing** via email, post or fax. A "Request for Access to Perinatal Data" form may be accessed on the perinatal website:

www.health.vic.gov.au/perinatal

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Trends in prevalence of selected birth defects

Of the twenty-eight major defects regularly presented in our report, Births Defects in Victoria, significant linear trends between **1995 and 2006** were found for seven defects:

- All neural tube defects decreasing
- Spina bifida decreasing
- Hypoplastic left heart syndrome increasing
- Obstructive defects of renal pelvis increasing
- Trisomy 21 (Down syndrome) increasing
- Trisomy 13 (Patau syndrome) increasing
- Trisomy 18 (Edward syndrome) increasing

A significant increasing trend was also found for oesophageal atresia and/or stenosis for the period 2001 – 2006.

- The decreases in neural tube defects and spina bifida specifically were most likely related to the voluntary fortification of food products with folate, and increased public awareness of the role of periconceptual folate in reducing neural tube defects.
- The increase in hypoplastic left heart syndrome (HLHS) is thought to be related to non-Victorian residents coming to Victoria (tertiary hospitals) for birth and subsequent treatment, following prenatal diagnosis of HLHS.
- The steady increase in prevalence of obstructive defects of the renal pelvis (ODRP)

probably reflects increasing ascertainment through prenatal ultrasound diagnosis.

- The increase in prevalence of the three trisomies (21, 13, 18) is most likely related to the increase in maternal age and improved early antenatal ascertainment of cases that may once have been missed due to early fetal loss.
- It is uncertain why there has been an increase in the prevalence of oesophageal atresia and/or stenosis over the last five years.

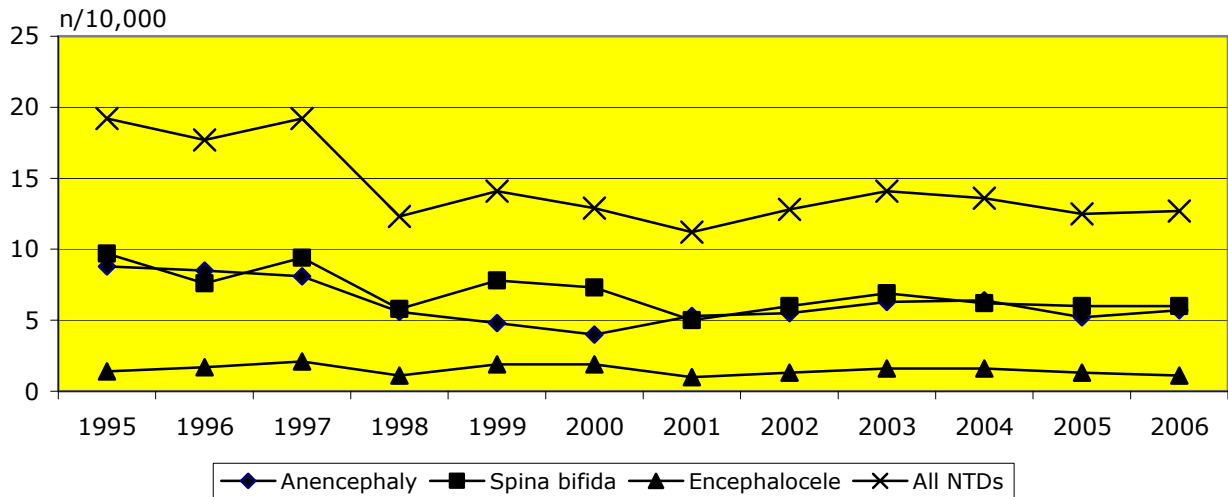
Ten most common birth defects, 2005-2006

Defect	N/10,000
Hypospadias*	74*
Obstructive defects of the renal pelvis	40
Ventricular septal defect	32.2
Trisomy 21	29.5
Develop. dysplasia of hip	27.5
Trisomy 18	8.4
Hydrocephalus	8.1
Cleft palate	8.0
Cystic kidney disease	6.8
Renal agenesis/dysgenesis	6.6

*Male babies only used as denominator.

These were also the ten most common birth defects in 2003–2004.

Trends in prevalence of neural tube defects, 2005-2006



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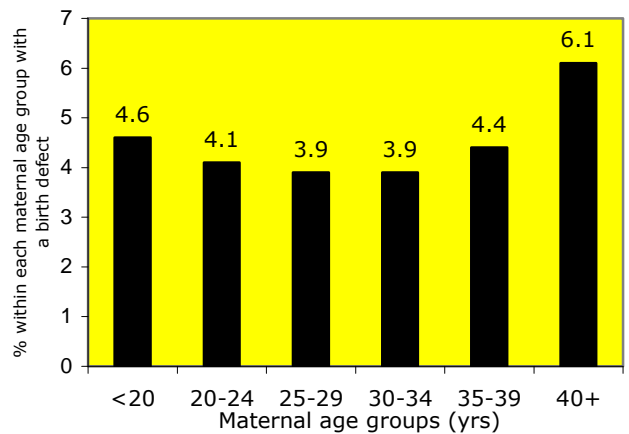
Factors which can influence the prevalence of birth defects

Maternal age

The frequency of birth defects is highest amongst mothers aged 35 years and over. In 2005 - 2006, 26.8% of all mothers who had a pregnancy affected by a birth defect were aged 35 years and over. Once chromosomal defects are excluded there is no overall increased risk of having a pregnancy affected by a birth defect compared to women less than 35 years of age.

Ventricular septal defect, cleft palate, cystic kidney disease, developmental dysplasia of the hip, trisomy 21, trisomy 18 and trisomy 13 are all more likely to occur with older mothers (using pooled data from 2001-2006). All neural tube defects (combined), obstructive defects of the renal pelvis and gastroschisis are associated with younger maternal age.

Maternal age and birth defects, 2005 - 2006

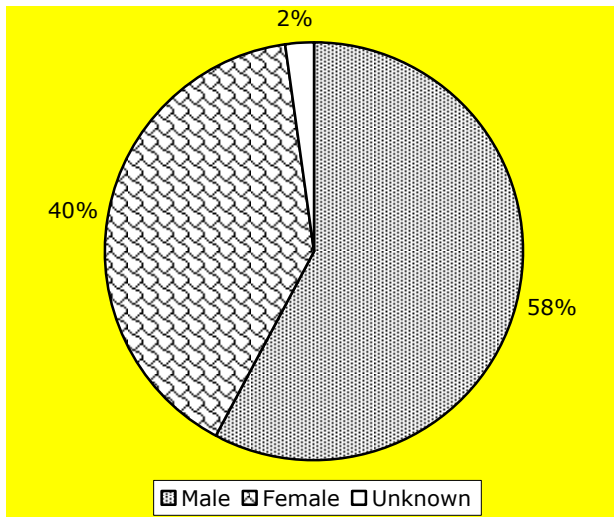


Sex

Male babies are more likely to have a birth defect than female babies; 4.7% of males compared with 3.7% of female babies had a birth defect in 2005 - 2006. Higher prevalence for males was seen for some cardiac conditions (such as transposition of the great arteries and hypoplastic left heart syndrome) cleft lip and palate, anorectal atresia and/or stenosis, renal agenesis/dysgenesis, obstructive defects of the renal pelvis, cystic kidney disease, diaphragmatic

hernia and Trisomy 21. Developmental dysplasia of the hip was three times more prevalent in females compared to males.

Birth defects cases by sex of infant, 2005-2006



• **Plurality**

Multiple births (twin, triplets, and quads) are also slightly more likely to be affected by a birth defect than singleton births; 4.6% of multiple births had a birth defect compared to 4.3% of singleton births in 2005 – 2006. The only significant increased prevalence rates

were for hydrocephalus and ventricular septal defect in multiples.

An update on electronic birth defects notification forms

To date we have received 56 notifications via the electronic form, since January 2008. Thankyou to all of you who have used this system to notify us of babies/children with birth defects.

Remember, it can be found on our website: www.health.vic.gov.au/perinatal under the "What's new" section on the home page.

If you need a new Birth Defect Notification Booklet or any more Victorian Birth Defect Register Pamphlets please don't hesitate to contact us via email or phone.

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An audit on developmental dysplasia of the hip (DDH) in Victoria, 2007 – an update (Merylyn Riley, VBDR; Jane Halliday, VBDR; Leo Donnan, RCH; Michelle Vu, RCH)

As many readers of this Bulletin will be aware, in 2007 we decided to conduct a statewide audit to follow-up all cases of hip abnormalities reported to the VBDR within a six month period.

For children born between January 1, 2007 to June 30, 2007, there were 236 cases of "hip anomalies for follow-up/investigation" reported on the Perinatal Morbidity Statistics Form and another 31 cases notified through other sources. Over several months we have been going through the process of obtaining the name of the treating doctor, and contacting them for outcome information. We are now at the point of analysing the data. Preliminary analysis shows that of these original 236 cases reported on the Perinatal Form, 37 cases were confirmed as DDH and 9 cases which were reported as DDH did not have this condition.

Recent publications using BDR data or relevant to the BDR

1. Collins VR, Muggli EE, Riley M, Palma S, Halliday JL. Is Down Syndrome a disappearing birth defect? *J Pediatrics* 152:20-4 (2008)
2. Nagle C, Gunn J, Bell R, Lewis S, Meiser B, Metcalfe S, Ukoumunne O and Halliday J. Use of a decision aid for prenatal testing of fetal abnormalities to improve women's informed decision making: a cluster randomised controlled trial *BJOG* 115:339-347 (2008)
3. Herlihy A, Halliday J. Is paternal age playing a role in the changing prevalence of Klinefelter syndrome? *Eur J Hum Genetics* May 21 (2008) [Epub ahead of print].

Birth defect research

The VBDR is involved in a wide range of collaborative projects with State, National and International organisations. Two recently completed projects being prepared for publication include:

1) a study of the use of genetic counselling after a baby is diagnosed with a birth defect. This record linkage project between the VBDR and records held by Genetic Health Services Victoria examined trends in utilisation over a 15 year period and categorised birth defects into high, moderate and low need for genetic counselling. Overall, 20% have genetic counselling, but approximately 50% in the high need category do not access counselling.

2) a study of birth defects after assisted reproduction, comparing outcomes for singleton babies of women having IVF, ICSI, or GIFT, with women who are subfertile or have 'normal fertility'. This record-linkage study combines data from all ART providers in Victoria, is funded by the BUPA Foundation in the UK. The study has examined the influence on outcome of freezing embryos before transfer, the number of multiple pregnancies early in pregnancy, aetiology of infertility and the number of stimulated cycles.

For more information please contact the Victorian Birth Defects Register, Victorian Perinatal Data Collection Unit on 1300 858 505 or by email on perinatal.data@dhs.vic.gov.au

Related Reports (available on our website)

Riley M & Halliday J, *Birth Defects in Victoria 2005-2006*, Victorian Perinatal Data Collection Unit, Victorian Government Department of Human Services, Melbourne, 2008

Davey M-A, Taylor O, Oats, J, and Riley M, *Births in Victoria 2005-2006*, Victorian Perinatal Data Collection Unit, Victorian Government Department of Human Services, Melbourne, 2008 (August)

The Consultative Council on Obstetric and Paediatric Mortality and Morbidity. *Annual Report for the year 2006, incorporating the 45th survey of perinatal deaths in Victoria*. Melbourne, 2008

Muggli E & Halliday J (2008) *Report on prenatal diagnostic testing in Victoria, 2007*, The Murdoch Childrens Research Institute and the Victorian Birth Defects Register (soon)