

Victorian Birth Defects Bulletin

No.4 December 2007



Welcome to the fourth edition of the *Victorian Birth Defects Bulletin*. In this edition we look at some current initiatives of the Victorian Birth Defects Register, significant trends in birth defects from 1994 to 2005, some issues related to common chromosomal anomalies and current research.

Our next detailed report, ***Birth Defects in Victoria 2005-2006***, will be produced and ready for distribution by mid-2008. We welcome any feedback or suggestions on topics you would find relevant in future issues of the Bulletin.

Jane Halliday and Merylyn Riley
Victorian Birth Defects Register (VBDR)

Victorian Birth Defects Subcommittee

The Victorian Birth Defects Register works under the oversight of the Consultative Council on Obstetric and Paediatric Mortality and Morbidity (CCOPMM). Under the jurisdiction of CCOPMM there are several expert committees that provide insight into, and review, perinatal, paediatric and maternal deaths.

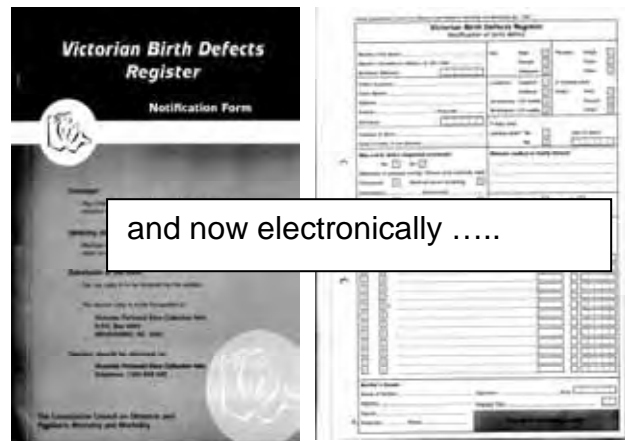
Another committee of CCOPMM is the Victorian Birth Defects Subcommittee, which meets three times a year to provide advice on issues related to collection and use of, and communication and operational matters about, data on birth defects in Victoria. Professor Agnes Bankier, Director of Genetic Health Services Victoria, has ably chaired this committee for the past three years, and we wish to formally acknowledge our appreciation to Agnes as she relinquishes this role. We also wish to welcome the new incoming chair, Dr Donna Henderson, from the Association for Children with a Disability.

The current members of this subcommittee are listed on the website:

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

Electronic birth defects notification form

In our June 2007 edition of the Bulletin we indicated that the VBDR now has an on-line mechanism for notification of birth defects to the Register, along with maintaining our notification booklets for those who prefer to use hardcopy.



To date, we have received very few notifications on-line. We would like to encourage people to notify us electronically of any babies/children with birth defects who may be encountered in the course of their work. We would also like any feedback on this notification process as it is a new and untested initiative of the VBDR.

To access this electronic form, go to the Perinatal Home Page

www.health.vic.gov.au/perinatal

and follow these steps:

1. Go to the box on the left-hand side titled "Perinatal Home".
2. Select "Forms".
3. Select "Complete VBDR notification on-line".
4. Complete the form.
5. Print off the form if you want to keep a copy.
6. Send.

The information will be encrypted and sent directly to a server at the Department of Human Services. Access to the data is restricted to only authorised staff at the Victorian Perinatal Data Collection Unit.

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Trends in prevalence of selected birth defects

One of the primary functions of the VBDR is to monitor trends in birth defects (prevalence and survival data). This is a complex area and may be affected by the time period over which the trend is reported.

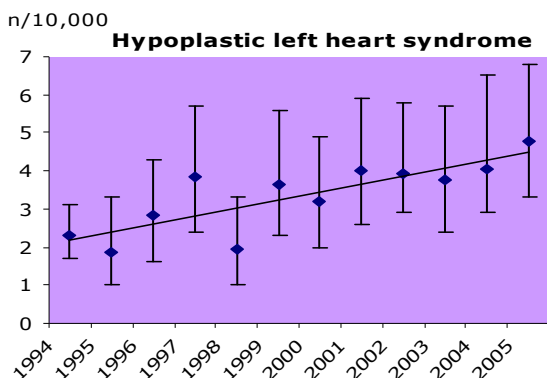
For example, of the twenty-eight major defects regularly presented in our report, Births Defects in Victoria, significant linear trends between **1994-2005** were found for seven defects:

- Spina bifida decreasing
- Hypoplastic left heart synd. increasing
- Anorectal atresia/stenosis decreasing
- Obstructive defects of renal pelvis increasing
- Trisomy 21 (Down Synd.) increasing
- Trisomy 13 (Patau Synd.) increasing
- Trisomy 18 (Edward Synd.) increasing

However, if the time period of the trend is changed to **1999-2005**, then only four of the twenty-eight major defects had significant linear trends:

- Anencephaly increasing
- Hypoplastic L heart synd. increasing
- Gastroschisis decreasing
- Trisomy 18 increasing

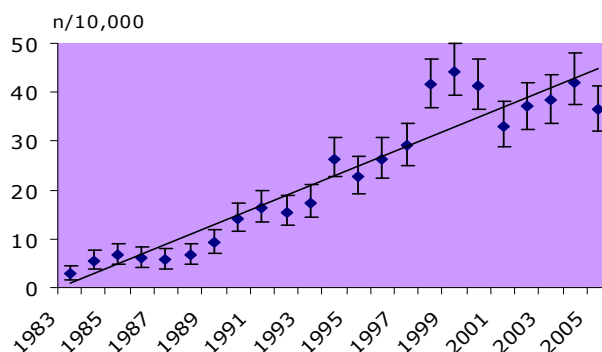
There were only two birth defects that had a consistently increasing significant linear trend over both time periods: Trisomy 18 (see p.3) and hypoplastic left heart syndrome (see below). This latter increase is mainly due to an increase in the number of interstate cases that are antenatally diagnosed and then the mother electively delivers in Melbourne for ready access to surgical treatment.



Trends in obstructive defects of the renal pelvis (ODRP)

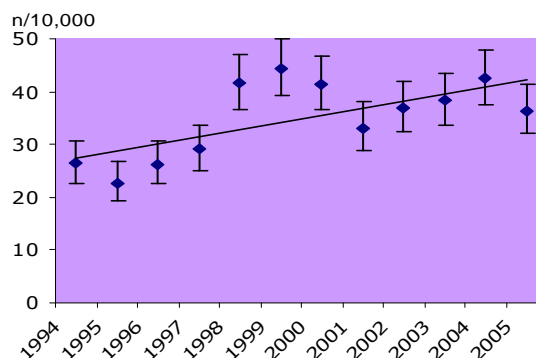
The choice of time periods for monitoring trends can influence prevalence rates. This can be seen with the congenital anomaly, obstructive defects of the renal pelvis. From 1983-2005 there has been a significant increase in this condition mainly due to improved ascertainment through an increased use of antenatal ultrasound.

ODRP, 1983-2005



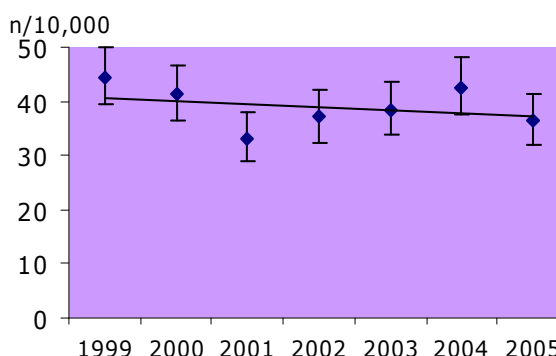
The significant increase in the prevalence of ODRP is also observed for the period 1994-2005.

ODRP, 1994-2005



However if you limit the time period of analysis to 1999-2005 then the significant increase in ODRP disappears.

ODRP, 1999-2005



Chromosomal anomalies in Victoria in 2005

Introduction:

There are many different types of chromosome anomalies possible, involving any of the 23 pairs of chromosomes in each human cell, and they are numbered according to their size. Only a minority of the 23 chromosomes are commonly present in a trisomic form i.e. where there are three whole copies of the chromosome instead of the usual two. The majority of trisomies, especially trisomy 16, lead to spontaneous miscarriage. Reported to the VBDR most frequently are trisomies of chromosomes 13, 18 and 21, and less frequently are some trisomies of the X and Y or sex chromosomes.

Other chromosomal anomalies involve rearrangement or loss of chromosomal material, and include a wide variety of perinatal outcomes and associated morbidities.

Prevalence:

In 2005, 392 birth defect cases reported to the VBDR had a chromosomal anomaly. This represents 15% of all birth defects. The overall birth defect prevalence rate for all chromosomal anomalies reported to the VBDR in 2005 was 59.6/10,000 pregnancies.

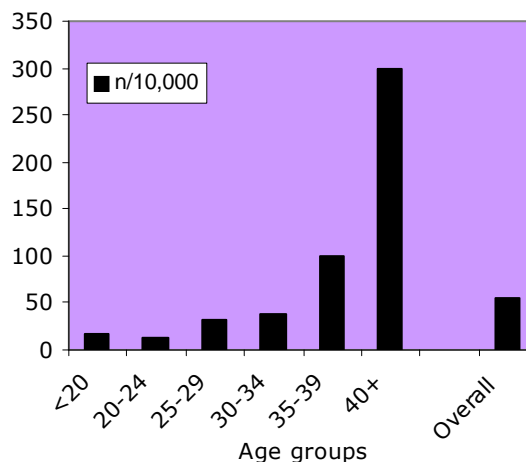
Birth Outcome

In 2005, most (67.9%) pregnancies affected by chromosomal anomalies that were reported to the VBDR were terminated either prior to 20 weeks gestation (58.7%) or very soon after 20 weeks gestation (9.2%). There were an additional 3.8% of pregnancies affected by a chromosomal anomaly that resulted in a perinatal death, with 28.3% of births affected by a chromosomal anomaly surviving beyond 28 days.

Maternal Age:

It is well documented that the risk of a baby having a chromosomal anomaly increases with maternal age. Our 2005 data revealed that 0.13% of pregnancies in women aged between 20-24 years were affected by a chromosomal anomaly compared to 3.0% in women aged 40+ years.

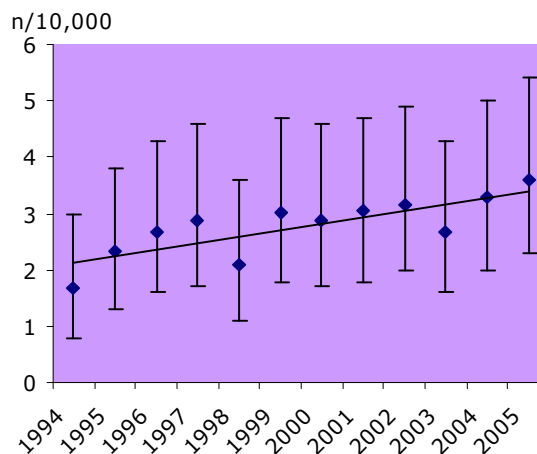
Maternal age effect for all chromosomal anomalies, 2005



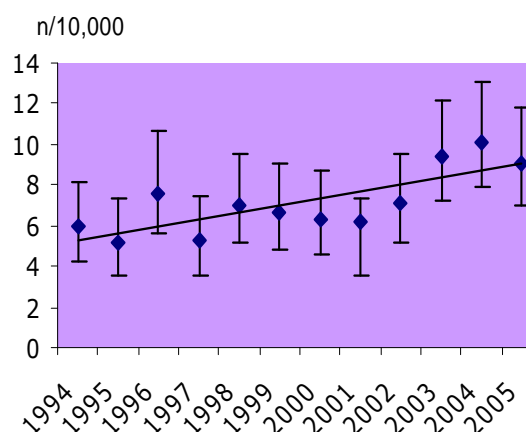
Trends in Trisomy 13 and 18

The significant increase in prevalence for Trisomy 13 and Trisomy 18 from 1994-2005 is primarily related to more widespread early prenatal screening, identifying cases that would previously have miscarried.

Trends in prevalence, Trisomy 13, 1994-2005



Trends in prevalence, Trisomy 18, 1994-2005



An audit on developmental dysplasia of the hip in Victoria, 2007

In our June Bulletin we included an insert regarding this project. We decided to conduct a statewide audit to follow-up all cases of hip abnormality reported to the VBDR within a six month period in order to:

1. improve reporting and classification of hip anomalies,
2. determine the prevalence of DDH/CDH in Victoria over a six month period.

To date, we have sent letters to 45 hospitals requesting details of treating clinicians for over 220 babies born between 1/1/2007 and 30/6/2007 who have been reported to the Perinatal Unit with some form of hip anomaly or hip requiring follow-up. The next phase will be to contact these clinicians for further information regarding the hip anomaly.

Publications

Is Down Syndrome a disappearing birth defect? The epidemiology of Down Syndrome in Victoria, Australia from 1986 to 2004

(Collins V., MCRI; Muggli E., MCRI; Riley M., VBDR, DHS; Palma S., VBDR, DHS; Halliday J., MCRI & VBDR, DHS)

Objective: We have utilised two high quality data collections on prenatal diagnosis and birth defects to study the epidemiology of Down Syndrome in a geographically defined population, specifically to examine the net effect of increasing maternal age and uptake of prenatal testing from 1986 to 2004.

J of Paediatrics (2007)

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

Birth defect research

The VBDR is involved in a wide range of collaborative research projects with State, National and International organisations. Some recent projects include:

Comprehensive perinatal follow-up and evaluation of prenatal screening and diagnosis in Victoria during 2002-2004

(Jaques A., MCRI; Collins V., MCRI; Sheffield L., GHSV; Francis I., VCGS; Bonaquisto L., VCGS; Halliday J., MCRI & VBDR, DHS)

All prenatal screening and diagnostic testing records were linked to the birth and birth defect records at the VPDCU. Fifty-two percent of women had screening and/or diagnostic testing, with those not tested more likely to be younger, public patients, and from rural Victoria. Preliminary evaluation of first and second trimester screening tests showed detection rates for Down syndrome between 78-92%, with false positive rates of 4.6-7.7%.

Updating predictors of having a baby with neural tube defects

(du Plessis L., RCH, Hunt R., RCH; Fletcher, A., Genetics, DHS; Riley M., VBDR, DHS; Jane Halliday, VBDR, DHS)

Aim: This study aims to describe changes in the epidemiology of births affected by neural tube defects (NTDS), following introduction of voluntary fortification and a folate awareness campaign. This study is currently being written up for publication.

Related Reports/Publications (available on our website)

Muggli E & Halliday J (2007) *Report on prenatal diagnostic testing in Victoria, 2006*, The Murdoch Childrens Research Institute and the Victorian Birth Defects Register, DHS

Muggli E., MCRI; Collins, V., MCRI; Marraffa, C., RCH (2007) *First information needs of families with a baby with Down Syndrome*

The report was called "Going down a different road", and was based upon a qualitative interview study that investigated first support and information needs of families with a baby with Down syndrome to assist clinicians with practice points derived directly from parent feed-back.