



## NSW BIRTH DEFECTS REGISTER 1994 REPORT

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**A**bout 2,000 infants are born with birth defects each year in NSW. The fourth annual report of the NSW Birth Defects Register presents information reported on birth defects which were detected during pregnancy or at birth, or up to one year of age for the years 1990-93 and during pregnancy or at birth for 1994.

The reported number of liveborn and stillborn infants with birth defects rose from 1,659 in 1990 and 1,632 in 1991 to 2,142 in both 1992 and 1993. In 1994, 1,127 infants were reported as having a birth defect, but this figure includes only those infants whose malformation was detected during pregnancy or at birth. The improved reporting in 1992 followed the introduction of a notification system for individual health care providers in that year, and improved reporting from cytogenetic laboratories and paediatric referral hospitals.

Among liveborn and stillborn infants, malformations of the cardiovascular system were most commonly reported (Table 1). More than half of these comprised atrial and ventricular septal defects and heart valve defects. Defects of the musculoskeletal system were the second most commonly reported group. About one-quarter of these were congenital dislocation of the hips. The third most commonly reported group was defects of the genitourinary system, one-third of which were hypospadias. Compared with previous years, there was an increase in the number of ventricular and atrial septal defects and heart valve defects reported in 1993 and 1994. This is probably due to improved diagnosis and reporting of less severe defects detected some time after birth.

In 1994, 136 terminations of pregnancy were reported after diagnosis of a malformation, compared with 140 for 1993 and 254 for 1990-92. More than half the reported terminations were associated with chromosomal defects, most commonly Down syndrome, and almost one-third with neural tube defects (Table 2).

From 1993 to 1994 there was a slight increase in the reported number of terminations of pregnancy and a slight decrease in the number of stillbirths associated with neural tube defects, which may indicate an early trend towards increasing prenatal diagnosis of these conditions.

Birth defects were slightly more common among male than female infants. The rate of birth defects was lowest in the 25-29 year maternal age group and increased with increasing maternal age, with almost one in 20 infants born to mothers over 35 years of age reported as having a malformation (Figure 1).

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## Birth Defects Register 1994 Report

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**TABLE 1**

**BIRTH DEFECTS AMONG STILLBIRTHS AND LIVEBIRTHS BY DIAGNOSTIC CATEGORY, 1990-94**

Diagnostic category	No. defects	Rate per 1,000 births	Diagnostic category	No. defects	Rate per 1,000 births
<b>DEFECTS OF NERVOUS SYSTEM</b>	914	2.1	Other gastrointestinal defects	444	1.0
Anencephaly	75	0.2	<b>DEFECTS OF GENITOURINARY SYSTEM</b>	2,930	6.7
Spina bifida	217	0.5	Defects of female genitals	86	0.2
Encephalocele	41	0.1	Undescended testis	321	0.7
Microcephaly	116	0.3	Hypospadias	999	2.3
Congenital hydrocephalus	212	0.5	Epispadias	17	0.0
Other nervous system defects	253	0.6	Chordee	230	0.5
<b>DEFECTS OF EYE</b>	323	0.7	Indeterminate sex/ambiguous genitalia	53	0.1
Anophthalmos/microphthalmos	66	0.2	Renal agenesis/dysgenesis	146	0.3
Buphthalmos/congenital glaucoma	29	0.1	Obstructive defects of renal pelvis and ureter	469	1.1
Congenital cataract	69	0.2	Other genitourinary system defects	609	1.4
Other eye defects	159	0.4	<b>DEFECTS OF MUSCULOSKELETAL SYSTEM</b>	3,721	8.6
<b>DEFECTS OF EAR, FACE AND NECK</b>	216	0.5	Congenital dislocation of the hips	873	2.0
Absence/stricture of auditory canal	36	0.1	Talipes equinovarus	185	0.4
Absence of auricle	9	0.0	Polydactyly	418	1.0
Defects of face and neck	37	0.1	Syndactyly	1,187	0.4
Other ear defects	134	0.3	Reduction deformities of limbs	355	0.8
<b>DEFECTS OF CARDIOVASCULAR SYSTEM</b>	4,440	10.2	Craniosynostosis	394	0.9
Transposition of great vessels	169	0.4	Diaphragmatic hernia	116	0.3
Tetralogy of Fallot	127	0.3	Exomphalos	67	0.2
Ventricular septal defect	933	2.1	Gastroschisis	66	0.2
Atrial septal defect	817	1.9	Other musculoskeletal defects	1,060	2.4
Heart valve defects	646	1.5	<b>DEFECTS OF THE INTEGUMENTARY SYSTEM</b>	195	0.4
Patent ductus arteriosus >37 weeks	598	1.4	CYSTIC HYGROMA	39	0.1
Coarctation of aorta	153	0.4	<b>CHROMOSOMAL DEFECTS</b>	815	1.9
Other defects of aorta	94	0.2	Trisomy 21	495	1.1
Defects of pulmonary artery	129	0.3	Trisomy 13	23	0.1
Other cardiovascular defects	774	1.8	Trisomy 18	92	0.2
<b>DEFECTS OF RESPIRATORY SYSTEM</b>	270	0.6	Turner syndrome	35	0.1
Defects of nose	64	0.1	Other chromosomal defects	171	0.4
Defects of larynx, trachea and bronchus	83	0.2	<b>SITUS INVERSUS</b>	19	0.0
Defects of lung	120	0.3	<b>CONGENITAL MALFORMATION SYNDROMES</b>	175	0.4
Other respiratory defects	3	0.0	CONGENITAL RUBELLA SYNDROME	3	0.0
<b>DEFECTS OF GASTROINTESTINAL SYSTEM</b>	1,591	3.7	CONGENITAL CYTOMEGALOVIRUS INFECTION	7	0.0
Cleft palate only	348	0.8	CONGENITAL TOXOPLASMOSIS	3	0.0
Cleft lip only	161	0.4	NON-IMMUNE HYDROPS FOETALIS	64	0.1
Cleft palate and cleft lip	248	0.6	<b>OTHER AND UNSPECIFIED BIRTH DEFECTS</b>	178	0.4
Oesophageal atresia only	18	0.0	<b>TOTAL</b>	15,905	36.6
Oesophageal atresia with TOF	85	0.2			
Tracheo-oesophageal fistula (TOF) only	38	0.1			
Atresia/stenosis of small intestine	116	0.3			
Atresia/stenosis of anus	133	0.3			

Source: NSW Birth Defects Register, Epidemiology and Surveillance Branch, NSW Health Department.

Note: For 1990-93, cases reported during pregnancy and up to one year of age are included. For 1994, cases reported during pregnancy or at birth are reported.

**TABLE 2**

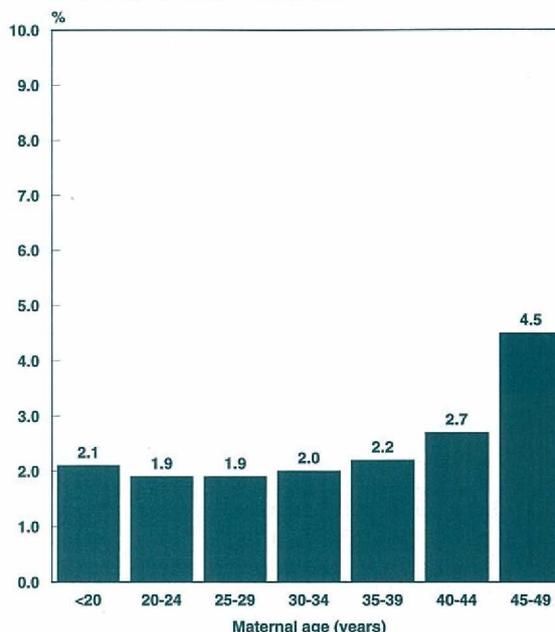
**BIRTH DEFECTS AMONG TERMINATIONS OF PREGNANCY, 1990-94**

Diagnostic category	No. terminations
DEFECTS OF NERVOUS SYSTEM	160
Neural tube defects	111
Other nervous system defects	39
DEFECTS OF EYE	1
DEFECTS OF EAR, FACE AND NECK	1
DEFECTS OF CARDIOVASCULAR SYSTEM	20
DEFECTS OF RESPIRATORY SYSTEM	6
DEFECTS OF GASTROINTESTINAL SYSTEM	22
DEFECTS OF GENITOURINARY SYSTEM	53
DEFECTS OF MUSCULOSKELETAL SYSTEM	120
CYSTIC HYGROMA	58
CHROMOSOMAL DEFECTS	353
Trisomy 21	144
Trisomy 13	15
Trisomy 18	53
Turner Syndrome	30
Other chromosomal defects	111
CONGENITAL MALFORMATION SYNDROMES	4
NON-IMMUNE HYDROPS FOETALIS	15
OTHER AND UNSPECIFIED BIRTH DEFECTS	16
<b>TOTAL</b>	<b>829</b>

Source: NSW Birth Defects Register, Epidemiology and Surveillance Branch, NSW Health Department.

**FIGURE 1**

**LIVEBORN AND STILLBORN INFANTS WITH BIRTH DEFECTS BY MATERNAL AGE, 1990-94**



Source: NSW Birth Defects Register, Epidemiology and Surveillance Branch, NSW Health Department.

Note: For 1990-93, cases reported during pregnancy and up to one year of age are included. For 1994, cases reported during pregnancy or at birth are reported.

## THE NSW BIRTH DEFECTS REGISTER

The NSW Birth Defects Register (BDR) is a population-based surveillance system established to monitor congenital malformations detected during pregnancy or at birth, or diagnosed in infants up to one year of age. The BDR was set up in 1990 and is based in the Epidemiology and Surveillance Branch of the NSW Health Department.

Activities of the BDR include publication of an annual report on the occurrence of birth defects; provision of information to Area Health Services to assist in service planning, monitoring of child health and investigation of specific issues; provision of information in response to specific requests from the public, health professionals, and other government departments; provision of data to the AIHW National Perinatal Statistics Unit (NPSU) for monitoring of birth defects at a national level; and special studies.

The BDR is supported by an advisory committee, comprising a panel of clinical experts representing the following specialities: genetics, dysmorphology, neonatology, obstetrics and gynaecology, bioethics and

epidemiology; and a community representative from the Association of Genetic Support of Australasia.

Notifications of birth defects are received from individual health care providers, the NSW Midwives Data Collection, paediatric referral hospitals and cytogenetic laboratories. Congenital malformations detected at birth are required to be notified to the NSW Midwives Data Collection under the NSW Public Health Act 1991. All other notifications are voluntary.

Annual reports and notification kits may be obtained from:

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