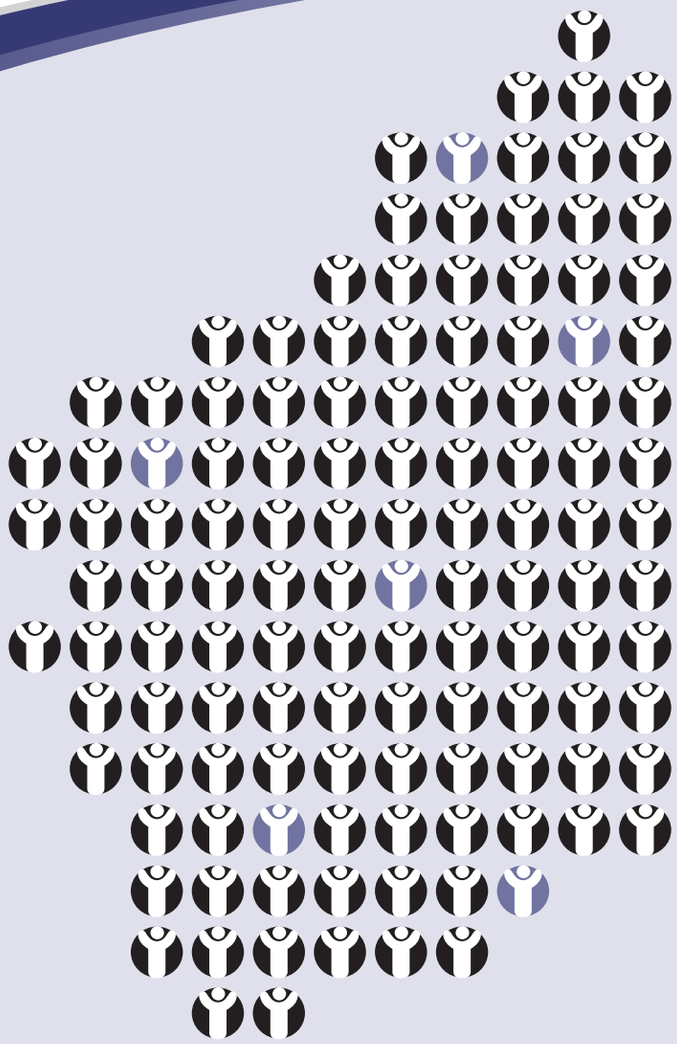
Government of **Western Australia**
Department of **Health**
Women and Newborn Health Service

Western Australian Register of Developmental Anomalies

1980-2014

November 2015



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REPORT OF THE WESTERN AUSTRALIAN REGISTER OF DEVELOPMENTAL ANOMALIES

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Our thanks to the specialist advisors to the Register and to the members of our Advisory Committees and Consumer Reference Group for their guidance and support.

FOREWORD

The Western Australian Register for Developmental Anomalies came into being in 2011 when the Western Australian Birth Defects Registry (WARDA-BD) and the Western Australian Cerebral Palsy Register combined. This year WARDA-BD celebrates 35th years of continuous statewide surveillance. WARDA is the oldest birth defects register in Australia and one of the leading birth defects registers in the world. Its 35 year history of surveillance, research and prevention provides an opportune moment for reflection on its accomplishments over the past 35 years. Who could have predicted that an idea for a birth defects register in Western Australia that began in the heart and mind of Professor Fiona Stanley 35 years ago would grow into an internationally recognized register impacting the lives of women, men and children in Western Australia, Australia and the world? But I get ahead of myself. Fiona Stanley and her band of merry epidemiologists: Drs. Jane Seward and Carol Bower officially began ascertaining cases of birth defects among 20,000 births among 1.27 million residents in Western Australia on January 1, 1980 as the Western Australia Congenital Malformation Register. Today, the register monitors approximately 33,000 births annually from a population of about 2.5 million residents. Since the publication of its first annual report in 1981, the register has published 34 such reports providing population counts, prevalence estimates and trends for birth defects monitored by the register. However, what is not included in the pages of this typed report with its neatly arranged rows and columns is the tremendous time and effort and the process that is required to conduct state-wide surveillance, ultimately resulting in an annual report. So let's take a brief "behind the scenes" look at the inner workings of WARDA. The register ascertains cases of birth defects (stillbirths, terminations or live births) to state residents using active methods. Prior to 2011, WARDA ascertained cases from statutory reporting sources (i.e., the Midwives Notification System, Hospital Morbidity data, and birth and death records) and voluntary notifications from healthcare professionals. On January 1, 2011 it became a statutory register which requires mandatory reporting by health practitioners. Trained register staff ascertain new cases and confirm notification cases by reviewing medical records from approximately 60 hospitals and medical facilities in Western Australia. Staff also ascertain and confirm cases by reviewing records from antenatal ultrasonography clinics, cytogenetic laboratories, specialty clinics, community health clinics, the state newborn screening laboratory and newborn hearing and vision screening programs.

All cases must have a documented confirmatory postnatal diagnosis, which may involve diagnostic tests (e.g. cardiac catherisation, echocardiogram, etc.) or surgical intervention for each defect identified in order for the defect(s) to be included in the Register. Once recorded, each case is reviewed by the Head of WARDA prior to entry into the database. All data entered into the Register database are reviewed periodically and annually by staff for accuracy and completeness. Case information is updated on an ongoing basis as new information becomes available or errors are found. Information not obtained through medical record abstraction is collected through routine annual linkage to the Western Australia Midwives' Notification System, Hospital Morbidity data, and state birth and death records. But then what? Well, as I stated before, after the data is collected each year, WARDA consolidates the data and prepares an annual report.

So that is how the data is collected but how has it been used? Well, for some reason, you have picked up this 1980-2015 report of the WARDA - most likely because of the information it contains. You might use this information to do research, to provide services, to provide information to other parents or family members concerned about a particular birth defect, or perhaps it just landed on your desk. Regardless of the reason, the information contained in this report was collected with the intention that it would be *used*. WARDA data is not collected to just sit in a drawer or on a computer server somewhere; it is and has been used extensively within the state, nationally and internationally for clinical, epidemiologic and public health studies of birth defects. Since its inception, the register has collaborated with students and faculty at universities, researchers at international organizations, institutes and agencies to conduct research. As of this year, WARDA data was used in more than 325 peer-reviewed publications plus reports and numerous state, national and international presentations at scientific conferences.

Okay, I concede that as impressive as that may sound you would be right to remain a little skeptical. After all, you might say, "publications are nice, but how has WARDA *directly* impacted the lives of Australians?" I agree; that is a very reasonable question. Well I am pleased to report that results from the annual report and scientific studies conducted using WARDA data are disseminated to the general public through lay summaries of scientific studies on the WARDA website and are routinely included in community organization newsletters and publications. But what is even more exciting is that WARDA data was the basis for implementation of new public health practices and policies in Australia. Findings from peer-reviewed publications using WARDA data and other published studies were instrumental in the passing of legislation mandating fortification of grain products with folic acid in Australia in 2009. Its data have also been used for other public policy triumphs including more recent efforts with fetal alcohol spectrum disorder (FASD). So has WARDA *directly* impacted the lives of Australians? The answer is - Yes!
Now I would be remiss if I did not highlight another important achievement of the register. Partnering with the community has been an important component to WARDA's successful 35-year history. The Consumer Reference Group was formally established in 2009 as an advisory group to assist in determining if the Register would require statutory notification; as a result of their incredible support, the register has statutory notification of birth defects and cerebral palsy in Western Australia. The Consumer Reference Group has also been actively engaged in the development of educational resources about birth defects and cerebral palsy and advising researchers on their proposed research projects.

Yes, WARDA has had 35-years of successful statewide surveillance *and* research in Western Australia. They have conducted studies, published papers, worked with community organizations and impacted public health policy. But what might not be apparent by rehearsing their list of accomplishments of 35-years, reading a scientific publication or reading this annual report is that the numbers contained on the pages of this report or the data contained in the database are not *just* numbers. To the staff of WARDA who toil each day to collect the information from medical records and other reports, each number corresponds to a child with a birth defect. Each "number" represented in each table of this report on the 35-year database has a family and a story. The staff know that and hope that as you read and use the information contained in this report that coincides with the 35th year of statewide surveillance in Western Australia, that you too will remember that behind every

number in every table on every page of this report and every other report produced from the register, there are names and there are stories – not just numbers. WARDA now hands the data they worked so hard to collect over 35-years to you. What will you do with the stories of these children? Go and tell the story of the children who are represented in this annual report.

Through the efforts of the WARDA staff and its collaboration with numerous researchers, clinicians, parents, community groups, public health workers and others over the past 35 years and now *you*, the ending of the story of some of the children represented in this report has changed and can be changed. And thanks to hard work and the public health impact of WARDA, the beginning of the story for children born during the past 35 years was changed so that they were *never* included in a WARDA annual report and hopefully the same will be true for many more unborn children in the future.

Happy 35th Birthday WARDA-BD!

Wendy N. Nembhard, MPH, PhD
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Associate Professor Nembhard collaborated with the Western Australia Register of Developmental Anomalies and the Telethon Kids Institute as a 2014-2015 Fulbright Senior Scholar funded through the Australian-American Fulbright Commission. During her time in Australia she worked on several research projects using WARDA data and also conducted a formal evaluation of the register.

SUMMARY

- The Western Australian Register of Developmental Anomalies (WARDA) (*Health (Western Australian Register of Developmental Anomalies) Regulations 2010*) monitors information based on the statutory notification of developmental anomalies in Western Australia.

- For the purposes of the Register, a developmental anomaly is defined as: cerebral palsy; or a structural or functional abnormality that is present at conception or occurs before the end of pregnancy and is diagnosed by six years of age.

- This Report provides information separately on birth defects (notified to the Register for births and terminations occurring between 1 January 1980 and 31 December 2014) and cerebral palsy in children born 1980-2009.

- For **birth defects**, the numerator data comprise anomalies occurring in live births and stillbirths in WA and in pregnancies terminated because of fetal abnormalities. Birth defects diagnosed prenatally and in children up to six years of age are included. The denominator data are all births in WA.
  - A total of 1789 cases of birth defects were notified relating to births and terminations of pregnancy in 2014, a proportion of 5.4%. This is expected to increase as birth defects continue to be diagnosed up to the age of six years in children born in 2014 (Table 1).
  - Birth defects were generally more common in male infants and multiple pregnancies and were reported slightly less frequently in Aboriginal compared with non-Aboriginal infants (Table 2).
  - Birth defects were also generally less frequently reported for rural compared with metropolitan regions (Table 3). Over the period 2002-2014, neural tube defects were more common in the Midwest (1.8 per 1000), Goldfields (1.6 per 1000) and Wheatbelt (1.5 per 1000), compared with 1.4 per 1000 or less elsewhere (Table 4).
  - In 2014, musculo-skeletal (26.6 per 1000 births) was by far the most common category of birth defects and higher than in previous years, the major contributing anomaly being developmental dysplasia of the hip (20.0 per 1000 (Table 5).
  - In 2014, neural tube defects (births plus terminations of pregnancy) occurred in 1.2 per 1000 births (Table 5 and Figure 1). This is likely due to mandatory fortification of flour with folic acid - continued monitoring is required.
  - Chromosomal defects generally have been increasing since 1980. The total rate for Down syndrome (births plus terminations) and the rate for terminations alone have steadily increased over time. Down syndrome in liveborn infants has been below 1 per 1000 since 2011 (Figure 2).
  - Birth defects are a major cause of death. For 2014 births, a birth defect was present in 12.4% of stillbirths and 28.9% of neonatal deaths (Table 6). Terminations of pregnancy for fetal anomaly occurred at a rate of 6.5 per 1000 births in 2014.
  - The major sources of notification are hospitals, private practitioners and investigative and treatment centres (Table 7).

- For **cerebral palsy (CP)**, the numerator data include all individuals identified as having cerebral palsy born in WA from 1956 onwards. Denominators are all live births in WA or, in some analyses, neonatal survivors (Table 8).
  - There are currently 4059 cases of CP on the Register from birth-year 1956 onwards. Of these, 461 (11.4%) are due to postneonatal causes (occurring after the first month of life) and 743 (18.3%) were not born in WA.
  - CP rates remain consistently higher in males than in females (Figure 4).
  - There has been little change in CP rates by severity over time. A concerning increase in severe CP in the 1990-94 year group has not persisted in subsequent years to 2009. For all WA-born cases born 1980-2009 combined, minimal and mild CP accounted for almost half (12.7% and 35% respectively), with moderate and severe CP representing about a quarter each (25.5% and 26.8% respectively.
  - Different types of CP can occur singly or in combinations, and proportions are reported by the predominant type. Spastic CP is the most commonly occurring type, accounting for more than 80% of all CP 1980-2009, though in widely varying distributions and severities. The remainder of cases are predominantly non-spastic: ataxic (7.1%), dyskinetic (8.7%) or hypotonic (1.5%).
  - CP is accompanied by intellectual disability (IQ less than 70) in approximately 40% of cases born 1980-2009, and this has not changed over time.

- Research using Register data is reported: This includes studies of oesophageal atresia, cleft lip and palate, birth defects in offspring of women with polycystic ovary syndrome and Down syndrome.

INTRODUCTION

As WARDA is a statutory register (*Health (Western Australian Register of Developmental Anomalies) Regulations 2010*), it is mandatory for developmental anomalies to be reported. The medical practitioner making the diagnosis or caring for the patient diagnosed and/or the chief executive officer of a hospital in which the diagnosis of a developmental anomaly is made are responsible for making the notification. This is required within six months of the diagnosis. Under the regulations, there are provisions to impose a fine for non-compliance with the regulations.

A developmental anomaly is defined in the Regulations as:

a. cerebral palsy; or
b. a structural or functional anomaly, which is present at conception or occurs before the end of pregnancy and is diagnosed during pregnancy, or after stillbirth or termination of pregnancy, or after live birth, but before 6 years of age (referred to as "birth defects" in this report)

WARDA has a commitment to obtain high quality, complete, and population-based information on birth defects and cerebral palsy for WA, and to use this information:

a. to monitor the number of cases of developmental anomaly in Western Australia (WA);
b. to plan, monitor and evaluate services for the prevention and alleviation of developmental anomalies and the care of persons with a developmental anomaly in WA;
c. to compile and publish general or statistical information relating to developmental anomalies; and
d. to carry out research into the causes of developmental anomalies and the effectiveness of prevention, screening and treatment services.

This report provides routine statistics on notifications of:

- **birth defects** received by 31 August 2015 for births occurring between 1 January 1980 and 31 December 2014.
- **cerebral palsy** as recorded at the age of five years, for births 1956-2009.

Data on children not born in WA but resident in the State are not included in this report. They are, however, recorded on the Register for such purposes as evaluation of treatment and planning of facilities for children with developmental anomalies in WA.

BIRTH DEFECTS DATA

Routine statistics

The numerator data in this report comprise birth defects occurring in live births and stillbirths in WA and in pregnancies terminated because of fetal malformation. Birth defects diagnosed in children up to six years of age are included. The denominator data in this Report are derived from information provided by the Department of Health and include only live births and stillbirths of 20 weeks' gestation or more. Amongst children born in 2008, who are now all over six years of age, 6.0% had a birth defect (Table 1). As children born from 2009 onwards are not yet six, the percentage with birth defects in more recent years of birth is expected to increase over time.

Table 1
Birth Defects in Western Australia, 1980 - 2014

Year	Total Births in WA	Cases of WA birth defects notified	Births with defects %
1980	20825	978	4.7
1981	22240	1038	4.7
1982	22400	1055	4.7
1983	23082	1161	5.0
1984	22989	1178	5.1
1985	23402	1158	4.9
1986	23961	1212	5.1
1987	24242	1246	5.1
1988	25191	1299	5.2
1989	25582	1393	5.4
1990	26039	1466	5.6
1991	25058	1462	5.8
1992	25358	1569	6.2
1993	25370	1568	6.2
1994	25450	1610	6.3
1995	25448	1634	6.4
1996	25586	1728	6.8
1997	25257	1761	7.0
1998	25668	1661	6.5
1999	25743	1731	6.7
2000	25229	1728	6.8
2001	24932	1665	6.7
2002	24782	1580	6.4
2003	24681	1486	6.0
2004	25530	1573	6.2
2005	26984	1726	6.4
2006	28665	1857	6.5
2007	30077	1794	6.0
2008	30674	1835	6.0
2009	31219	1922	6.2
2010	31265	1839	5.9
2011	32191	1936	6.0
2012	33862	1983	5.9
2013	34403	2051	6.0
2014	33073	1789	5.4

Demographic information

Race, sex and plurality (Table 2)

Birth defects are generally more common in multiple births and male infants. There is generally a lower prevalence of birth defects reported in Aboriginal children. This may be due to relative under-ascertainment.

Table 2

Birth Defects in Western Australia births by Aboriginality, Sex and Plurality, 1980 - 2014

(Percentages are for total Western Australian births in each category)

Year of Birth	Aboriginality		Sex		Plurality	
	Non-Aboriginal Aboriginal		Male Female Indeterminate Unknown		Single Multiple	
	No	%	No	%	No	%
1980-89	11148	(5.0)	6921	(5.8)	11389	(5.0)
	570	(4.6)	4751	(4.2)	329	(5.9)
			8			
			38			
1990-99	15373	(6.4)	9264	(7.1)	15615	(6.3)
	817	(5.4)	6851	(5.5)	575	(8.0)
			5			
			70			
2000-04	7569	(6.6)	4602	(7.2)	7728	(6.4)
	463	(5.7)	3370	(5.5)	304	(7.4)
			4			
			56			
2005-09	8616	(6.2)	5213	(6.9)	8779	(6.1)
	518	(5.9)	3842	(5.3)	355	(8.1)
			2			
			77			
2010	1751	(5.9)	1063	(6.7)	1770	(5.8)
	88	(5.2)	752	(4.5)	69	(8.2)
			0			
			24			
2011	1860	(6.1)	1040	(6.3)	1845	(5.9)
	76	(4.4)	878	(5.6)	91	(10.1)
			0			
			18			
2012	1912	(5.9)	1091	(6.3)	1912	(5.8)
	71	(4.3)	870	(5.3)	71	(7.6)
			0			
			22			
2013	1968	(6.0)	1017	(5.8)	1961	(5.9)
	83	(4.7)	999	(6.0)	90	(9.5)
			0			
			35			
2014	1729	(5.5)	831	(4.9)	1703	(5.3)
	60	(3.4)	915	(5.7)	86	(9.1)
			1			
			42			

Area of residence (Table 3)

Table 3 shows that the proportion of births with a birth defect. Proportions tend to be higher in the two metropolitan regions than in the rural regions. This may be due to under-ascertainment from rural regions rather than a real difference.

Table 3

Numbers and Proportions of Cases of Birth Defects by Year of Birth and Health Region, 1980 - 2014

No=Number, Prop=Proportion per 100

Health Region (WA Dept of Health)	1980-89	90-99	00-04	05-09	2010	2011	2012	2013	2014
	No Prop	No Prop	No Prop	No Prop	No Prop	No Prop	No Prop	No Prop	No Prop
North Metro	4573 5.3	6689 6.9	3348 6.8	3865 6.6	762 6.1	897 7.0	853 6.3	966 7.1	827 6.4
South Metro	3897 5.4	5567 6.6	2894 6.7	3219 6.1	659 5.7	702 5.7	772 5.9	743 5.5	646 5.0
Kimberley	212 3.9	338 5.5	193 5.7	192 5.5	29 4.2	28 4.3	31 4.5	32 4.5	30 4.6
Pilbara Gascoyne	467 3.7	474 4.6	227 5.4	229 5.5	58 6.8	44 5.1	40 4.5	33 3.8	36 3.8
Midwest Murchison	417 4.4	502 5.1	218 5.8	229 5.0	54 5.6	34 3.6	35 3.9	43 4.8	35 3.9
Wheatbelt	662 4.7	665 5.7	274 5.7	284 5.8	63 6.7	43 4.7	64 6.6	41 4.5	71 7.5
Goldfields SE Coastal	408 4.2	574 5.1	274 5.7	240 4.8	46 4.8	34 3.8	44 4.5	51 5.4	42 4.4
Great Southern	371 4.6	401 5.3	162 4.7	174 4.7	44 6.0	34 4.4	34 4.7	25 3.3	20 2.9
Southwest	694 4.5	941 5.8	418 5.0	641 6.4	107 5.1	101 4.8	88 4.0	102 4.6	72 3.3

Individual birth defects by Health Region 2001-2014 (Table 4)

The proportions per 1000 births for these major birth defects are generally similar across regions. However, neural tube defects are more common in the Midwest (1.8 per 1000), Wheatbelt (1.5 per 1000) and the Goldfields (1.6 per 1000), compared with other regions.

Table 4

Numbers and Proportions of Cases of Some Major Birth Defects by Health Region, 2002 – 2014 (No=Number, Prop=Proportion per 1000)

Diagnostic Category (British Paediatric Association Code)	North Metro No	North Metro Prop	South Metro No	South Metro Prop	Kimberley No	Kimberley Prop	Pilbara No	Pilbara Prop	Mid West No	Mid West Prop	Wheatbelt No	Wheatbelt Prop	Goldfields No	Goldfields Prop	Great Southern No	Great Southern Prop	South West No	South West Prop
Neural Tube Defects (74000 - 74209)	170	1.2	169	1.4	10	1.2	12	1.2	19	1.8	17	1.5	19	1.6	12	1.4	33	1.4
Congenital Hydrocephalus (excludes those with NTD) (74230 - 74239)	102	0.7	90	0.7	6	0.7	9	0.9	6	0.6	11	1.0	6	0.5	10	1.2	18	0.8
Cleft Palate only (74900 - 74909)	165	1.2	130	1.0	9	1.1	4	0.4	13	1.3	11	1.0	14	1.2	7	0.8	27	1.2
Cleft Lip with or without Cleft Palate (74910 - 74929)	142	1.0	151	1.2	19	2.3	10	1.0	16	1.5	19	1.7	8	0.7	12	1.4	24	1.0
Oesophageal Atresia/Stenosis (with or without fistula) (75030 - 75038)	52	0.4	49	0.4	2	0.2	5	0.5	5	0.5	2	0.2	6	0.5	0	0.0	14	0.6
Anorectal Stenosis/Atresia (75121 - 75125)	81	0.6	71	0.6	2	0.2	5	0.5	3	0.3	2	0.2	8	0.7	4	0.5	16	0.7
Hypospadias (75260, 75263 - 75269)	491	3.6	468	3.8	20	2.4	25	2.5	26	2.5	37	3.2	35	3.0	17	2.0	78	3.4
Renal Agenesis (75300 - 75301)	65	0.5	75	0.6	2	0.2	5	0.5	6	0.6	8	0.7	7	0.6	12	1.4	14	0.6
Total Limb Reduction Defects (75520 - 75529)	115	0.8	93	0.8	6	0.7	7	0.7	8	0.8	9	0.8	8	0.7	4	0.5	18	0.8
Diaphragmatic Hernia (75661)	56	0.4	46	0.4	4	0.5	2	0.2	3	0.3	1	0.1	2	0.2	3	0.3	10	0.4
Exomphalos (75670)	71	0.5	59	0.5	3	0.4	2	0.2	6	0.6	6	0.5	3	0.3	4	0.5	7	0.3
Gastroschisis (75671)	40	0.3	61	0.5	2	0.2	2	0.2	3	0.3	3	0.3	6	0.5	3	0.3	8	0.3
Transposition of Great Vessels (74510 - 74519)	64	0.5	54	0.4	7	0.9	6	0.6	6	0.6	7	0.6	7	0.6	5	0.6	5	0.2
Hypoplastic Left Heart Syndrome (74670)	28	0.2	29	0.2	2	0.2	2	0.2	4	0.4	1	0.1	1	0.1	0	0.0	4	0.2
Down Syndrome (75800 - 75809)	426	3.1	355	2.9	21	2.6	26	2.6	27	2.6	29	2.5	16	1.4	30	3.5	68	3.0
Anotia/Microtia (74400-74401)	31	0.2	18	0.1	2	0.2	2	0.2	1	0.1	2	0.2	2	0.2	2	0.2	3	0.1

Diagnostic information

The definition of a birth defect used by the Register is: *a structural or functional anomaly, which is present at conception or occurs before the end of pregnancy and is diagnosed during pregnancy, or after stillbirth or termination of pregnancy, or after live birth, but before 6 years of age.* This includes structural (eg spina bifida), chromosomal (eg Down syndrome) and metabolic (eg phenylketonuria) defects. Each individual defect (up to a maximum of 10 defects per case) is coded according to the 5-digit British Paediatric Association ICD-9 system. Syndrome diagnoses are coded along with the major individual defects seen in that infant (eg Down syndrome, VSD and duodenal atresia occurring in one child are all coded).

Most minor malformations are excluded unless they are disfiguring or require treatment. A list of exclusions is provided on the WARDA website, as well as lists (numeric and alphabetic) of the standard codes and text used by WARDA for birth defects and their major/minor classification.

Table 5 shows the number and proportion per 1000 total births of the main categories of defects, as well as the more common or important defects individually, by year of birth. Since about a quarter of the cases registered have more than one defect, the total number of defects exceeds the total number of cases. Not all individual birth defects are reported in Table 5, but information on any birth defect is available on request.

Figures 1 and 2 show livebirths, terminations and total cases with neural tube defects and Down syndrome respectively.

Some trends of note:

- There has been a fall in **neural tube defects** in total since 1995, from around 2 per 1000 to 1.2 per 1000 in 2014, and in **anencephaly** and **spina bifida** when considered separately. This is believed to be due to increased maternal intake of periconceptional folate, as folic acid supplements and food fortification (voluntary fortification for some foods from 1996 and, since September 2009, mandatory fortification of wheat flour for bread-making). Most cases of neural tube defects are detected prenatally and the pregnancy terminated (Figure 1), highlighting the importance of including terminations when monitoring trends in neural tube defects.
- There has been a marked increase in **developmental dysplasia of the hip** from 2011, up to 20.0 per 1000 in 2014, compared with <10 per 1000 in previous years. Possible reasons for the increase are being sought.
- The apparent fall in prevalence of **undescended testes** is partly due to the fact that this birth defect is mainly registered at the time surgery is undertaken, usually around 1-2 years of age. However, there has been a lower rate since 2005.
- The gradual rise in **chromosomal defects** since 1980, a result of increasing maternal age and possibly also the increased use of first trimester screening, appears to have peaked in 2010 at 6.7 per 1000 births for all chromosomal defects and 3.2 per 1000 for **Down syndrome**. The rate of Down syndrome in liveborn infants has been below 1 per 1000 since 2011 (Figure 2).

Table 5

Numbers and Proportions of Cases of Birth Defects by Year of Birth and Diagnosis, 1980 - 2014

(Proportions are per 1000 births and are only calculated if number of cases is greater than 13)
No=Number, Prop=Proportion

Diagnostic Category (British Paediatric Association Code)	80-89 No Prop	90-99 No Prop	00-04 No Prop	05-09 No Prop	2010 No Prop	2011 No Prop	2012 No Prop	2013 No Prop	2014 No Prop
NERVOUS SYSTEM DEFECTS (74000 - 74299)	**998 4.3**	**1198 4.7**	**657 5.2**	**769 5.2**	**126 4.0**	**150 4.7**	**153 4.5**	**148 4.3**	**138 4.2**
Neural Tube Defects (74000 - 74209)	446 1.9	455 1.8	182 1.5	202 1.4	36 1.2	46 1.4	37 1.1	40 1.2	41 1.2
Anencephalus (74000 - 74029)	202 0.9	201 0.8	79 0.6	85 0.6	12	16 0.5	16 0.5	18 0.5	22 0.7
Spina Bifida (74100 - 74199)	203 0.9	210 0.8	88 0.7	90 0.6	20 0.6	21 0.7	17 0.5	14 0.4	17 0.5
Encephalocoele (74200 - 74209)	41 0.2	44 0.2	15 0.1	27 0.2	4	9	4	8	2
Microcephaly (74210)	129 0.6	150 0.6	78 0.6	71 0.5	14 0.4	7	16 0.5	12	10
Congenital Hydrocephalus (excludes those with NTD) (74230 - 74239)	162 0.7	230 0.9	87 0.7	121 0.8	20 0.6	30 0.9	23 0.7	17 0.5	21 0.6
Congenital Deafness (74287)	160 0.7	214 0.8	172 1.4	198 1.3	33 1.1	34 1.1	44 1.3	39 1.1	31 0.9
CONGENITAL ANOMALIES OF EYE (74300 - 74399)	260 1.1	347 1.4	169 1.4	137 0.9	25 0.8	12	30 0.9	19 0.6	9
Anophthalmia (74300 - 74309)	13	14 0.1	6	5	0	0	2	1	0
Microphthalmia (74310 - 74319)	43 0.2	56 0.2	22 0.2	15 0.1	5	0	8	3	2
Congenital Cataract and Lens Anomalies (743300 - 74339)	57 0.2	101 0.4	35 0.3	29 0.2	6	3	5	2	2
CONGENITAL ANOMALIES OF EAR, FACE AND NECK (74400 - 74499)	568 2.4	982 3.9	581 4.6	654 4.4	95 3.0	74 2.3	87 2.6	79 2.3	52 1.6
Anotia, Microtia (74400 - 74401, 74421)	46 0.2	74 0.3	30 0.2	32 0.2	3	5	3	5	9
Branchial Remnants (74440 - 74448)	122 0.5	155 0.6	68 0.5	88 0.6	15 0.5	6	11	4	3

Table 5 (continued)

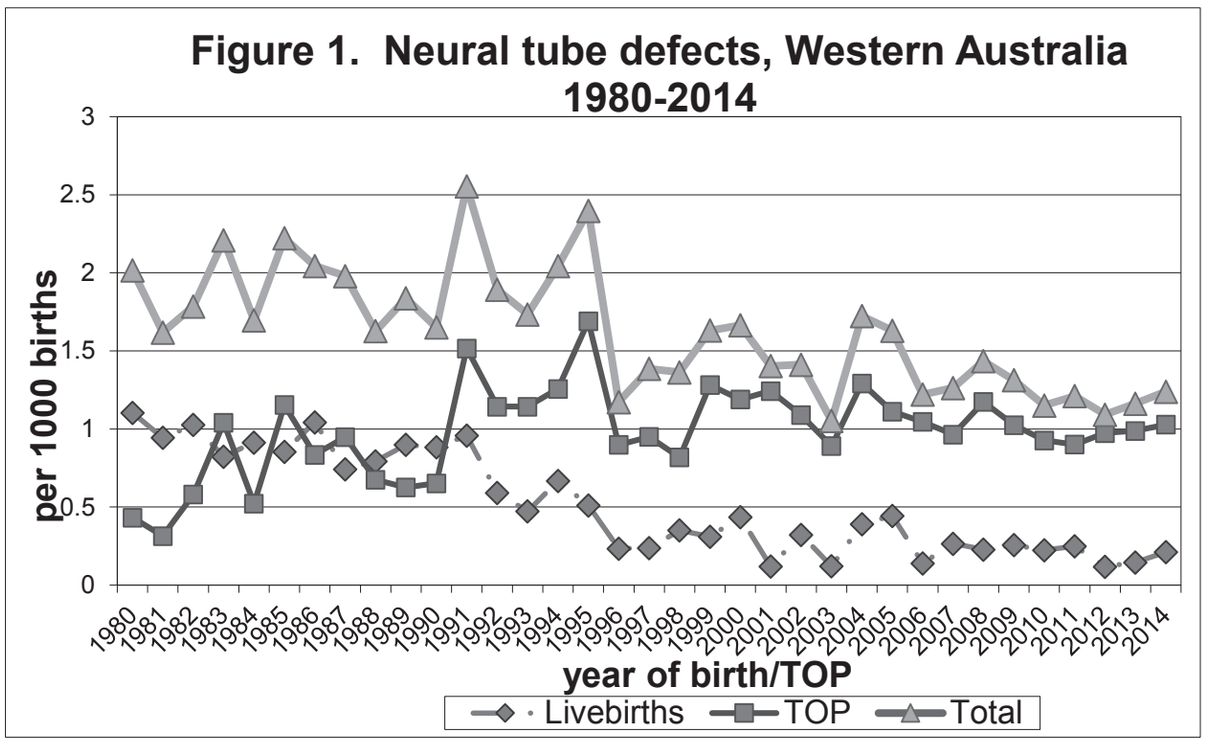
Diagnostic Category (British Paediatric Association Code)	80-89 No Prop	90-99 No Prop	00-04 No Prop	05-09 No Prop	2010 No Prop	2011 No Prop	2012 No Prop	2013 No Prop	2014 No Prop
CARDIOVASCULAR DEFECTS (74500 - 74799)	1897 8.1	3093 12.1	1559 12.5	1677 11.4	335 10.7	333 10.3	298 8.8	321 9.3	314 9.5
Transposition of Great Vessels (74510 - 74519)	97 0.4	114 0.4	64 0.5	67 0.5	7	14 0.4	16 0.5	16 0.5	11
Tetralogy of Fallot (74520)	71 0.3	98 0.4	37 0.3	38 0.3	13	10	8	11	17 0.5
Ventricular Septal Defect (74540 - 74549)	958 4.1	1727 6.8	865 6.9	881 6.0	180 5.8	179 5.6	166 4.9	179 5.2	174 5.3
Atrial Septal Defect (74551 - 74559)	303 1.3	488 1.9	222 1.8	269 1.8	55 1.8	48 1.5	52 1.5	54 1.6	50 1.5
Hypoplastic Left Heart Syndrome (74670)	43 0.2	49 0.2	20 0.2	32 0.2	8	5	6	9	10
Patent Ductus Arteriosus (74700)	288 1.2	445 1.7	192 1.5	273 1.8	54 1.7	53 1.6	50 1.5	37 1.1	29 0.9
Coarctation of Aorta (74710 - 74719)	118 0.5	140 0.5	81 0.6	77 0.5	14 0.4	10	5	17 0.5	14 0.4
RESPIRATORY SYSTEM DEFECTS (74800 - 74899)	202 0.9	243 1.0	144 1.2	158 1.1	32 1.0	37 1.1	30 0.9	38 1.1	19 0.6
Choanal Atresia (74800 - 74809)	35 0.1	32 0.1	19 0.2	12	2	1	2	5	0
GASTRO-INTESTINAL DEFECTS (74900 - 75199)	1465 6.3	1669 6.5	786 6.3	911 6.2	171 5.5	173 5.4	181 5.3	178 5.2	124 3.7
Cleft Palate only (74900 - 74909)	196 0.8	280 1.1	157 1.3	174 1.2	31 1.0	26 0.8	29 0.9	36 1.0	31 0.9
Cleft Lip only (74910 - 74919)	116 0.5	115 0.5	76 0.6	81 0.5	12	10	19 0.6	9	6
Cleft Lip and Palate (74920 - 74929)	194 0.8	171 0.7	91 0.7	92 0.6	14 0.4	20 0.6	14 0.4	25 0.7	19 0.6
Tracheo-Oesophageal Fistula, Oesophageal Atresia/Stenosis (75030 - 75038)	75 0.3	74 0.3	50 0.4	70 0.5	6	14 0.4	7	8	8
Pyloric Stenosis (75051 - 75058)	470 2.0	461 1.8	145 1.2	196 1.3	42 1.3	38 1.2	39 1.2	33 1.0	6
Stenosis/Atresia Small Intestine (75110 - 75119)	61 0.3	69 0.3	39 0.3	36 0.2	12	8	9	12	6
Stenosis/Atresia Anus (75123 - 75125)	118 0.5	158 0.6	86 0.7	78 0.5	21 0.7	10	17 0.5	19 0.6	18 0.5
Hirschprung's Disease (75130 - 75133)	38 0.2	54 0.2	17 0.1	37 0.3	11	6	9	7	8

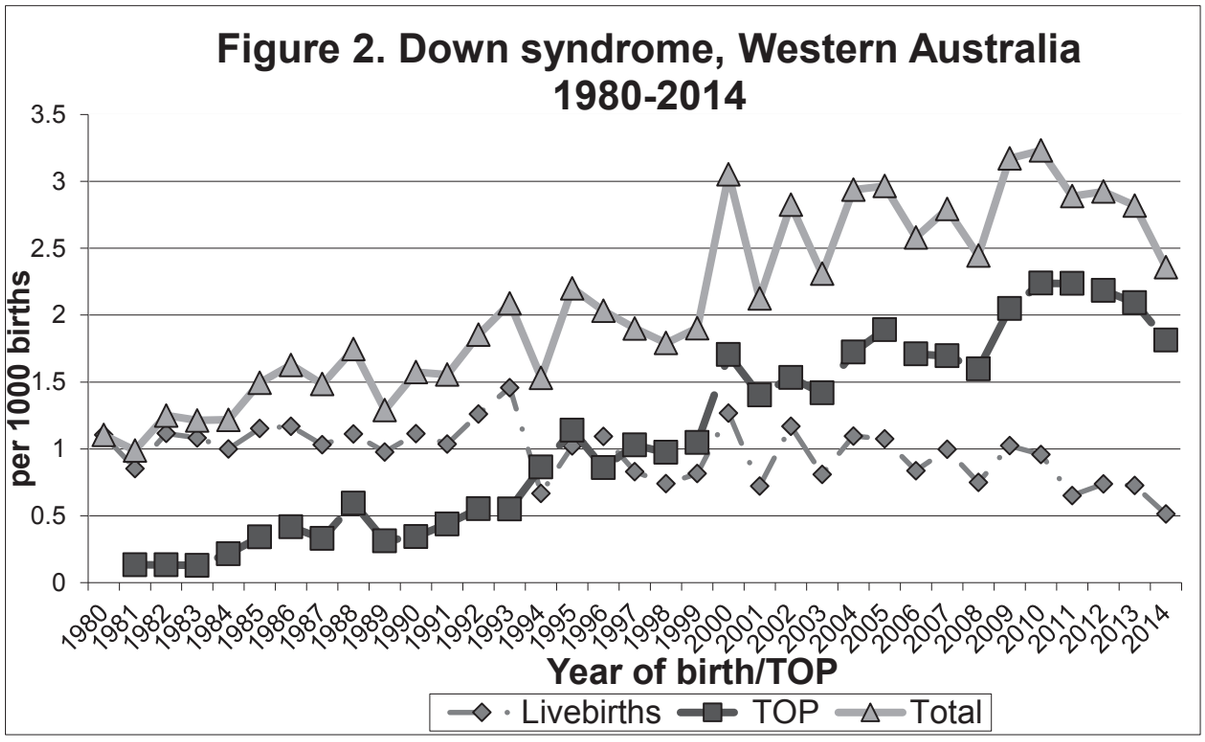
Table 5 (continued)

Diagnostic Category (British Paediatric Association Code)	80-89 No	80-89 Prop	90-99 No	90-99 Prop	00-04 No	00-04 Prop	05-09 No	05-09 Prop	2010 No	2010 Prop	2011 No	2011 Prop	2012 No	2012 Prop	2013 No	2013 Prop	2014 No	2014 Prop
URO-GENITAL DEFECTS (75200 - 75399)	3283	14.0	4834	19.0	2288	18.3	2623	17.8	551	17.6	536	16.7	538	15.9	435	12.6	324	9.8
Undescended Testis (treated) (75250 - 75254, 75257)	1546	6.6	1596	6.3	656	5.2	646	4.4	140	4.5	138	4.3	115	3.4	61	1.8	52	1.6
Hypospadias (75260, 75263 - 75269)	666	2.8	883	3.5	464	3.7	510	3.5	111	3.6	97	3.0	107	3.2	107	3.1	58	1.8
Renal Agenesis or Dysgenesis (75300 - 75306)	111	0.5	171	0.7	120	1.0	125	0.8	30	1.0	32	1.0	29	0.9	27	0.8	22	0.7
Cystic Kidney Disease (75310 - 75319)	83	0.4	190	0.7	128	1.0	126	0.9	26	0.8	38	1.2	29	0.9	36	1.0	37	1.1
Obstructive Defects Renal Pelvis (75320 - 75329)	156	0.7	496	1.9	297	2.4	662	4.5	157	5.0	140	4.3	147	4.3	111	3.2	104	3.1
Vesico-Ureteric Reflux (75344)	488	2.1	1156	4.5	440	3.5	335	2.3	49	1.6	50	1.6	66	1.9	50	1.5	25	0.8
Other Anomalies of Ureter (75340 - 75343, 75345 - 75349)	137	0.6	341	1.3	136	1.1	202	1.4	44	1.4	51	1.6	45	1.3	51	1.5	50	1.5
MUSCULO-SKELETAL DEFECTS (75400 - 75699)	3325	14.2	4142	16.2	2003	16.0	2233	15.1	491	15.7	607	18.9	678	20.0	856	24.9	881	26.6
Developmental Dysplasia of Hip (75430 - 75434, 75439)	1502	6.4	1705	6.7	716	5.7	816	5.5	213	6.8	330	10.3	411	12.1	597	17.4	660	20.0
Talipes (75450, 75454-75456,75473)	512	2.2	520	2.0	302	2.4	305	2.1	55	1.8	49	1.5	42	1.2	71	2.1	68	2.1
Polydactyly (75500 - 75509)	238	1.0	293	1.1	143	1.1	174	1.2	44	1.4	44	1.4	38	1.1	37	1.1	15	0.5
Syndactyly (75510 - 75519)	155	0.7	151	0.6	72	0.6	104	0.7	20	0.6	12		16	0.5	11		10	
Reduction Deformities Upper and/or Lower Limbs (75520 - 75549)	177	0.8	286	1.1	155	1.2	164	1.1	24	0.8	27	0.8	27	0.8	26	0.8	30	0.9
Craniosynostosis (75600, 75601)	105	0.4	161	0.6	60	0.5	86	0.6	14	0.4	14	0.4	16	0.5	8		5	
Diaphragmatic Hernia (75661)	71	0.3	102	0.4	43	0.3	48	0.3	8		20	0.6	7		17	0.5	12	
Exomphalos (75670)	58	0.2	88	0.3	57	0.5	60	0.4	12		18	0.6	6		25	0.7	15	0.5
Gastroschisis (75671)	37	0.2	84	0.3	38	0.3	62	0.4	10		13		13		8		17	0.5
CONGENITAL ANOMALIES OF INTEGUMENT (75700 - 75799)	715	3.1	1229	4.8	665	5.3	548	3.7	77	2.5	81	2.5	83	2.5	85	2.5	29	0.9
Birth Marks, Naevus (75738)	408	1.7	706	2.8	426	3.4	333	2.3	41	1.3	40	1.2	52	1.5	49	1.4	9	

Table 5 (continued)

Diagnostic Category (British Paediatric Association Code)	80-89 No	80-89 Prop	90-99 No	90-99 Prop	00-04 No	00-04 Prop	05-09 No	05-09 Prop	2010 No	2010 Prop	2011 No	2011 Prop	2012 No	2012 Prop	2013 No	2013 Prop	2014 No	2014 Prop
CHROMOSOME DEFECTS (75800 - 75899)	555	2.4	1013	4.0	708	5.7	903	6.1	208	6.7	207	6.4	202	6.0	199	5.8	166	5.0
Down Syndrome (75800 - 75809)	316	1.4	470	1.8	332	2.7	410	2.8	101	3.2	93	2.9	99	2.9	97	2.8	78	2.4
Trisomy 13 (75810 - 75819)	22	0.1	40	0.2	40	0.3	48	0.3	6		9		8		8		5	
Trisomy 18 (75820 - 75829)	40	0.2	110	0.4	91	0.7	121	0.8	27	0.9	25	0.8	27	0.8	32	0.9	27	0.8
Turner Syndrome (75860 - 75861, 75869)	29	0.1	78	0.3	63	0.5	60	0.4	13		23	0.7	13		10		13	
OTHER																		
Congenital Hypothyroidism (24390 - 24399)	49	0.2	94	0.4	65	0.5	69	0.5	7		22	0.7	25	0.7	16	0.5	11	
Adrenogenital Syndome (25520 - 25529)	15	0.1	25	0.1	15	0.1	14	0.1	2		0		3		0		3	
Disorders of Amino Acid Transport and Metabolism (27000 - 27099)	38	0.2	62	0.2	26	0.2	51	0.3	2		9		11		2		6	
Phenylketonuria (27010)	13		14	0.1	10		13		1		2		2		0		4	
Disorders of Carbohydrate Transport and Metabolism (27100 - 27199)	20	0.1	20	0.1	8		4		5		4		3		3		1	
Cystic Fibrosis (27700)	76	0.3	72	0.3	54	0.4	66	0.4	12		13		11		15	0.4	14	0.4
G6PD Deficiency (28220)	44	0.2	66	0.3	30	0.2	20	0.1	4		5		4		1		4	
Thalassemias (28240 - 28249)	5		12		3		5		0		1		0		3		2	
Haemophilia (28600 - 28620)	19	0.1	23	0.1	23	0.2	13		4		0		0		0		0	
Muscular Dystrophies and Myopathies (35900 - 35999)	70	0.3	52	0.2	14	0.1	35	0.2	3		2		4		3		1	
Fetal Alcohol Spectrum Disorder (75995-75998)	36	0.2	51	0.2	74	0.6	72	0.5	6		6		5		2		0	
Congenital Rubella Syndrome (77100)	24	0.1	9		2		0		0		0		0		0		0	





Deaths

Table 6 shows the number (and percentage) of stillbirths, neonatal and post-neonatal deaths known to have a birth defect. Terminations of pregnancy are those which occurred following prenatal diagnosis of a fetal abnormality. Between 8% and 15% of stillbirths have a reported birth defect, as do 27% - 47% of neonatal deaths and 13% - 36% of post-neonatal deaths.

Terminations of pregnancy for fetal abnormality have increased from 1.6 per 1000 births in 1980-1989 to 7.5 per 1000 in 2013 and 6.5 per 1000 in 2014.

Table 6
Deaths with Birth Defects 1980 - 2014

Year of Birth	Stillbirths (% is of all stillbirths)		Neonatal Deaths (% is of all neonatal deaths)		Postneonatal Deaths (% is of all postneonatal deaths)		Terminations of Pregnancy for Fetal Anomaly
	No	%	No	%	No	%	No
1980-89	254	13.7	509	39.4	213	27.5	403
1990-99	273	15.2	331	39.4	173	32.2	1109
2000-04	96	10.7	90	30.0	52	31.9	820
2005-09	106	10.0	100	30.4	62	31.5	1012
2010	18	8.3	23	34.3	7	13.2	229
2011	26	9.6	19	30.6	8	30.8	239
2012	19	8.0	13	27.7	11	26.8	225
2013	19	9.0	28	47.5	9	36.0	254
2014	29	12.4	11	28.9	13	*	222

* Complete data on all postneonatal deaths

Notifications

Table 7 documents the number of notifications of birth defects received from different sources by year of birth of the child. Most sources provide very consistent levels of notification.

Table 7

Numbers of Notifications by Source and Year of Birth of Cases Notified, 1980 - 2014

Notifiers	1980-89	90-99	00-04	05-09	2010	2011	2012	2013	2014
HOSPITAL MORBIDITY	1421	3167	2161	2595	462	413	389	320	0
PAEDIATRIC HOSPITALS EXCL SPECIAL DEPTS	5503	5114	1478	1353	218	193	190	224	204
PAEDIATRIC HOSPITALS SPECIAL DEPARTMENTS	2140	3780	1547	1572	367	364	437	536	636
OBSTETRIC HOSPITALS EXCL SPECIAL DEPTS	1771	2058	853	739	161	200	164	138	154
OBSTETRIC HOSPITALS SPECIAL DEPARTMENTS	327	1003	671	840	211	226	207	264	257
OTHER HOSPITALS	422	102	90	61	13	10	16	5	7
CYTOGENETIC SERVICES	402	926	655	880	160	176	184	172	154
PATHOLOGY SERVICES	814	1215	738	862	198	203	163	180	165
GENETICS SERVICES	1946	3170	1695	1779	321	357	373	289	190
PRIVATE PRACTITIONERS	4977	6776	3250	3063	668	739	754	837	799
CHILD & COMMUNITY HEALTH NURSES & DOCTORS	1199	672	86	39	5	1	5	0	4
RURAL PAEDIATRIC SERVICE	285	635	186	129	20	13	11	4	2
OTHER	617	121	391	490	14	3	35	29	26
REGISTER CHECK	769	451	133	236	38	63	36	29	28
TOTAL	25923	32207	15288	16107	3147	337	3394	3267	2626

- Hospital morbidity data for 2014 not available in time for reporting

CEREBRAL PALSY DATA

Unlike other developmental anomalies that can be well described at the time they are recognised, the physical expression of cerebral palsy tends to change over time. Signs and symptoms can sometimes resolve altogether, or a syndrome in its early stages can be mistaken for cerebral palsy. For these reasons, information for all cerebral palsy cases is updated at the age of five years in order to confirm and report data at a meaningful and consistent age. There is therefore always a five-year delay in reporting cerebral palsy data.

Routine statistics

The numerator data include all individuals identified as having cerebral palsy born in WA from 1956 onwards. Cases born outside WA are included on the Register in order to estimate numbers of people requiring services but are excluded from data reported here. Cases due to causes occurring after the first month of life, such as head injury, stroke or meningitis, are also included on the Register but usually analysed separately.

Overall rates of cerebral palsy (CP) have shown little variation over time (Figure 3; Table 8). Increases from the early 1970s accompanied the introduction of neonatal intensive care, which resulted in greater survival of preterm infants who later developed CP. Continued improvements in neonatal intensive care may be responsible for reduction in rates seen from the late 1990s and sustained into the 2000s. While the lower rate in 2005-09 is likely to be related to under-ascertainment of cases due to the unavailability of previously included sources of data, particularly the Ability Centre (previously known as the Centre for Cerebral Palsy), it could be at least partly due to falling rates of CP in preterm births, as has been observed in other Australian States (ACPR Group. Report of the Australian Cerebral Palsy Register, Birth Years 1993-2006, February 2013). Restored access to the Hospital Morbidity Data System in 2015 and imminent linkage to the Midwives Notification System will provide some of the missing data for an updated report on CP to birth year 2009.

Figure 3. Rates per 1000 live births (3-year moving averages) for total CP, CP excluding postneonatal CP (PNN), and CP excluding both postneonatal and minimal CP, Western Australia, 1956-2009

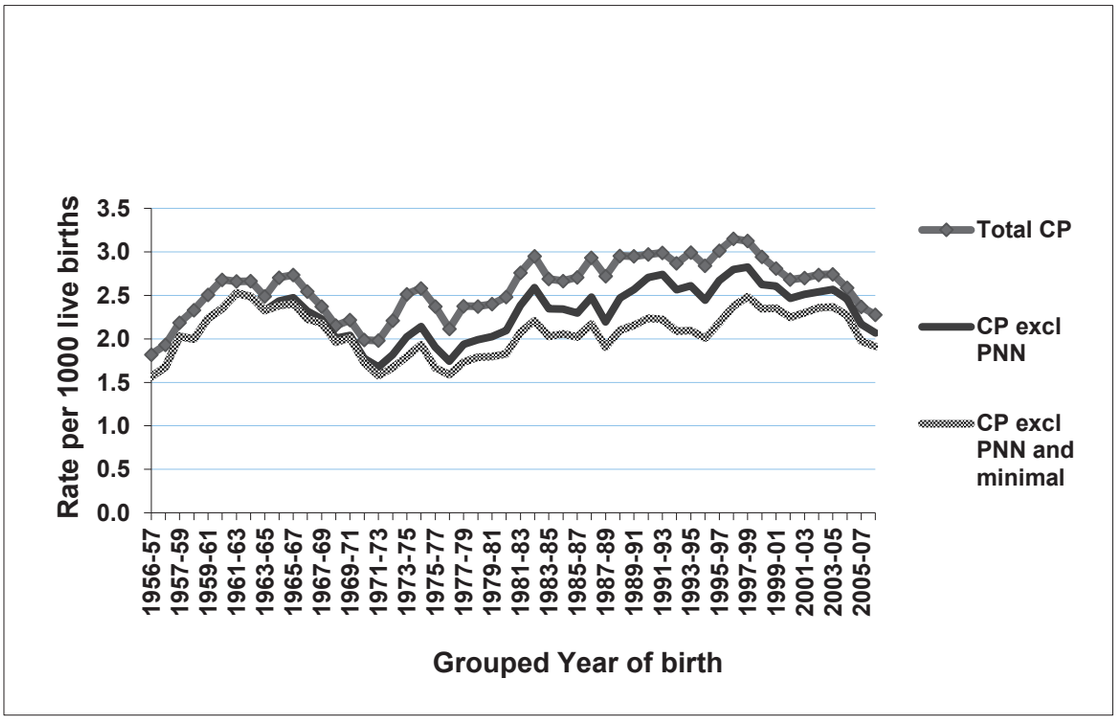


Table 8
Cerebral palsy (CP) birth prevalence rates per 1000 live births (LB) for total CP, CP excluding postneonatally acquired (PNN), and CP excluding both postneonatally acquired and minimal severity, Western Australia, 1956-2009 in Western Australia

Year of birth	Total CP	Rate/1000 LB	*95% Confidence Interval*	CP excl PNN	Rate/1000 LB	*95% Confidence Interval*	CP excl PNN and minimal	Rates excl PNN and minimal
1956-59	125	1.85	*1.52 - 2.17*	108	1.60	*1.29 - 1.90*	108	1.60
1960-64	222	2.61	*2.27 - 2.95*	202	2.38	*2.05 - 2.70*	202	2.38
1965-69	231	2.52	*2.20 - 2.85*	211	2.31	*1.99 -2.62*	206	2.25
1970-74	226	2.08	*1.81 - 2.35*	200	1.84	*1.58 - 2.09*	192	1.77
1975-79	247	2.40	*2.10 - 2.70*	202	1.96	*1.69 - 2.23*	183	1.78
1980-84	297	2.69	*2.38 - 2.99*	252	2.28	*2.0 - 2.56*	220	1.99
1985-89	334	2.76	*2.46 - 3.05*	285	2.35	*2.08 - 2.62*	248	2.05
1990-94	374	2.96	*2.66 - 3.26*	332	2.63	*2.35 - 2.91*	274	2.17
1995-99	378	2.98	*2.68 - 3.28*	329	2.59	*2.31 - 2.87*	277	2.18
2000-04	340	2.74	*2.45 - 3.03*	314	2.53	*2.25 - 2.81*	281	2.26
2005-09	362	2.47	*2.22 - 2.72*	335	2.29	*2.04 - 2.53*	312	2.13

REGISTER ACTIVITIES

1. Provision of data

The Register is a comprehensive source of information on birth defects and cerebral palsy in WA for use in all relevant areas of health service provision, policy development, research and evaluation. Provision of data from the Register may take two forms: (1) unnamed tabulated information similar to that contained in this report; and (2) identified or de-identified unit data for specific research projects. Requests for the latter must be discussed with the data custodian for the Register in the first instance and, it should be noted that access to CP data requires the prior approval of the CP Advisory sub-committee. Requests must then be submitted to the Department of Health WA Human Research Ethics Committee for approval.

2. Consumer Reference Group (CRG)

The CRG is now formally a sub-committee of the WARDA Advisory Board. Our thanks to Dr Rachel Skoss for chairing the CRG for several years. Rachel resigned this role earlier this year and the CRG is now in the process of membership renewal.

3. Information on developmental anomalies

Over the past year, 49 (32 birth defects; 17 CP) requests for information or data on developmental anomalies have been received. Three of these requests were from the state Department of Health, 38 were from health professionals and institutions in WA, Australia or overseas and 8 were from the general public, the media or students. Half the requests required a considerable amount of computing, analysis and discussion, and responses to most of the remainder involved provision and/or interpretation of published data.

4. Website

The WARDA website has been improved over the last year to make information more accessible and relevant to the public, health professionals and prospective researchers. In collaboration with the Consumer Reference Group we have created sections to include Family Stories and Plain Language Research Summaries and have included links to other relevant sites.The website can be accessed on:
http://www.kemh.health.wa.gov.au/services/register_developmental_anomalies.htm

We also provide a section on the website of the codes used for birth defects and a complete list of publications arising from the Register.

5. International Clearinghouse for Birth Defect Surveillance and Research

Data based on 2012 births were provided to the International Clearinghouse for inclusion in the 2015 Annual Report of the Clearinghouse. WARDA has collaborated with the Clearinghouse on research projects and training programs for birth defects surveillance.

6. World Birth Defects Day

The inaugural World Birth Defects Day, co-sponsored by several international agencies (including the March of Dimes, the US Centers for Disease Control and Prevention, Eurocat and the International Clearinghouse for Birth Defects Surveillance and Research), was held on 3 March 2015.
WARDA promoted World Birth Defects Day by raising awareness of the Register at King Edward Memorial Hospital.
We set up a table and pinup board in the Staff Dining Room, with information on display including Annual Reports, WARDA Brochures, WARDA information sheets, and posters with information on World Birth Defects Day, Cerebral Palsy, Fetal Alcohol Spectrum

Disorders, neural tube defects, Down syndrome and the Register in general. WARDA staff promoted the information to customers in the dining room, explaining who we are and what we do. We also had a very popular WARDA Quiz with prizes.
The results were excellent – all the staff members we approached were interested and feedback from the day will be used to run a bigger and broader event next year.

7. The Australian Cerebral Palsy Register (ACPR)
This national collaboration was spearheaded by the WA cerebral palsy team in 2002 when only three State registers were in existence, covering 45% of the Australian live born population. This progressed to 100% coverage by 2007 at which time the administrative centre of the ACPR moved from WA to the CP Alliance NSW where it continues to thrive with CP data collections now well established in all States and Territories. State representatives meet annually to discuss data and research matters. The first ACPR Report, covering birth years 1993-2003, was published in 2009 and the second to birth year 2006, in 2013. In 2014, the ACPR Group commenced work on a Supplement of the journal *Developmental Medicine and Child Neurology* to showcase CP research conducted throughout Australia. The second 'World Survey of CP Registers' will published as part of this Supplement and will create a resource to foster international collaborations by providing a means of readily ascertaining characteristics and availability of data collected by CP registers around the world.

8. Register-based Research

8.1. Long term outcomes for WA children born with orofacial clefts.
Using data from the WARDA, records of children with orofacial clefts were linked to WA Health Department data and school test data from the Education Department. This provided the opportunity to compare hospital admissions, survival up to 20 years old, and school test (WALNA and NAPLAN) results between children born 1980 to 2010 in WA with and without orofacial clefts. The data linkage was funded by The Developmental Pathways in WA Children Project and performed by Data Linkage Branch in the WA Health Department.

Children born with clefts had higher rates of hospital admission and spent longer time in hospital than children without clefts. This difference was greatest in infancy and childhood, and continued up to adulthood. Even if admissions related directly to cleft care were excluded, children with clefts still had higher admission rates, especially for respiratory, middle ear, and digestive (mainly dental) system conditions.

Babies born with a cleft and no other anomaly had the same survival rates in infancy, and up to 20 years old, as children without clefts. Babies with clefts and co-existing anomalies had lower survival up to one year old, but after one year, babies with cleft lip, or cleft lip and palate and co-existing anomalies had the same survival as children without clefts. Babies with cleft palate and co-existing anomalies had slightly lower survival between one and five years, but after that, their survival was the same as the general population too.

Papers based on these two studies have been submitted for publication.

The study comparing the proportions of children with and without clefts that reach the minimum standard in the WALNA and NAPLAN tests is continuing. A paper will be submitted late in 2015.

8.2 Implications of polycystic ovary syndrome (PCOS) for pregnancy and for the health of offspring.

In a project led by Professors Hart and Doherty from the School of Women's and Infants' Health and the Women and Infants Research Foundation, using record linkage, the records of women with a diagnosis of PCOS were linked to birth and birth defects records and compared with a randomly selected aged matched comparison group. 69.7% of women with a PCOS diagnosis and 62.9% of comparison women had one or more births recorded. The offspring of women with PCOS were more likely to have a birth defect (6.3% compared with 4.9%; OR 1.20 (95% CI 1.03-1.40)) and OR excluded unity for cardiovascular and urogenital defect categories separately. Possible reasons for this increased risk include maternal diabetes, obesity and/or medication use. A paper on this study has been published (Doherty D et al. Obstet Gynecol 2015;125:1397-1406).

8.3 Oesophageal atresia and tracheo-oesophageal fistula in Western Australia: Prevalence and trends

This study was undertaken because higher prevalence of oesophageal atresia with or without tracheo-oesophageal fistula (OA±TOF) was identified in in Western Australia, compared with data from other registers internationally. Based on the WARDA records of all infants born in WA, 1980-2009 with OA ±TOF, we found that OA±TOF and TOF alone affected 3.00 and 0.42 per 10 000 births, respectively. There was an increase of 2.0% per annum in the prevalence of OA±TOF with associated anomalies (64% of cases). Isolated TOF rates were stable. In 2000-2009, there was a 30% fall in OA±TOF live births with 61 (58%) cases diagnosed in first week of life, 10 (9%) prenatally and 34 (32%) at post-mortem only. Almost all cases of TOF alone (94%) were reported in liveborn infants. We concluded that the observed higher prevalence of OA±TOF in WA and the increase over time are attributable to increases with associated anomalies. Consistent reporting, availability of prenatal diagnosis and ascertainment of cases following termination of pregnancy or post-mortem examinations can significantly affect prevalence. A paper has been published based on this work.

8.4 Trends in Down syndrome.

Aggregated data from WARDA were used to assess the effects of prenatal screening and diagnostic testing on trends in birth prevalence and terminations for Down syndrome in Western Australia, from 1980-2013. Between 1980 and 2013, the rate of Down syndrome pregnancies increased, corresponding to a greater proportion of babies born to older women. Following the introduction of screening in 1994, the rate of liveborn infants with Down syndrome fell, while the rate of terminations of pregnancy for Down syndrome remained stable. In the absence of termination, the Down syndrome live-birth rate was estimated to have risen from 1.1 to 2.17/1000 between 1980 and 2013. A paper reporting these results has been published.

8.5 Monogenic Causes of Birth Defects in Rare Diseases

WARDA continues to collaborate on identifying the cause of monogenic rare diseases manifesting birth defects. In partnership with the SeqNextGen project lead by Professor Jennifer Blackwell and the Rare and Undiagnosed Diseases Diagnostic Service at Genetic Services of Western Australia, King Edward Memorial Hospital. Mutations in genes associated with rare monogenic disorders have been identified in approximately 25% of assessed cases. Many of these cases have been associated with birth defects. This work is informing the development of models of care and it has been presented locally and internationally. A paper will be submitted imminently.

PUBLICATIONS

Publications relating to WARDA in 2014 and 2015 are listed below. For a full listing of all publications since 1979, please go to our website:
http://www.kemh.health.wa.gov.au/services/register_developmental_anomalies.htm

2014

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2015

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This document can be made available in alternative formats on request for a person with a disability.

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