CONGENITAL ANOMALIES EVIDENT AT BIRTH

Introduction

Congenital anomalies (sometimes referred to as birth defects or congenital malformations), range in severity from minor conditions that are of no functional or cosmetic importance, to conditions that are incompatible with life. There are a large number of rare syndromes which are characterised by multiple congenital anomalies [1]. Congenital anomalies are one of the leading causes of fetal and infant deaths in New Zealand [2].

In New Zealand, the NZ Birth Defects Registry (NZBDR) has collected data on all babies with a diagnosed birth defect born or treated in a public hospital since 1977 [3]. The NZBDR contributes New Zealand data on 39 categories of birth defects to the International Clearinghouse for Birth Defects Surveillance Research (ICBDSR) based in Rome [4]. New Zealand data published in the latest (2010) ICBDSR report indicated an overall rate of congenital anomalies for 2004–2008 of around two anomalies per 100 births [4]. The Plunket National Child Health Study of 4286 children born in 1990–1 reported an overall prevalence of birth defects at six weeks of age of 4.3% [5].

Other developed countries have also reported similar figures [6,7,8]. The 2008 report from the Victorian Perinatal Data Collection Unit states that combined data from 2005–2006 showed that there was a birth defect in 4.2% of all births in Victoria [7]. EUROCAT (European Surveillance of Congenital Anomalies) collates data from 38 registries in 21 European countries. It collects information on major congenital anomalies that require surgical treatment, have serious effects on health or development, or have significant cosmetic impact. In 2010 the live birth prevalence of all EUROCAT anomalies was 176 per 10,000 (1.76%) [9].

The following section uses the National Minimum Dataset to review the number of congenital anomalies evident at birth, as well as the number of babies born with one or more congenital anomalies. Subsequent chapters consider cardiovascular anomalies, Down syndrome and neural tube defects in more detail. Note: In reviewing this data, it is important to remember that the analysis includes all congenital anomalies in the ICD-10-AM Q00–Q99 range (structural and chromosomal anomalies but not metabolic disorders), irrespective of whether they were minor (e.g. skin tags) or major (e.g. spina bifida). For this reason the overall prevalence estimates presented here may be higher than comparable overseas estimates (which may have included only major anomalies).

Data Source and Methods

Definition

1. *Number of congenital anomalies identified at birth (by anomaly type)*

2. *Number of babies with one or more congenital anomalies identified at birth*

Data Source

1. National Minimum Dataset

Numerator: Hospital Admissions with event type = birth and a congenital anomaly (Q00–Q99) listed in any of the first 15 diagnoses.

For this indicator, the unit of analysis is the number of congenital anomalies rather than the number of babies, with many babies having more than one anomaly.

For a list of the ICD-10-AM codes used to assign anomaly type see Appendix 8 (Congenital Anomaly Codes)

2. National Minimum Dataset

Numerator: Hospital admissions with event type = birth and a congenital anomaly (Q00–Q99) listed in any of the first 15 diagnoses.

For this indicator, the unit of analysis is the number of babies with one or more congenital anomalies.

Denominator: All hospital admissions with and event type = birth

Notes on Interpretation

Note 1: This analysis includes all admissions recorded in the National Minimum Dataset (NMDS) where the Event Type was listed as a Birth. In the NMDS only one birth event is allowed per NHI number, with babies born prior to hospital admission, or readmitted shortly after discharge, being listed as a routine inpatient event. Thus the analysis excludes babies born prior to hospital admission, babies born at home, or babies whose congenital anomaly was overlooked at the time of initial discharge, but who re-presented shortly thereafter.

Note 2: This analysis is likely to significantly undercount those conditions where the congenital or chromosomal anomaly usually only becomes evident at a later age, when the child fails to achieve their normal developmental milestones (e.g. many chromosomal or CNS anomalies), or where the condition may be difficult to detect on routine newborn examination.

Note 3: Because of the large number of ICD-10-AM diagnoses in the Q00–Q99 range, and the lack of additional supporting information, no attempt has been made to grade the severity of the congenital anomalies identified. The reader must thus bear in mind that in this analysis, minor anomalies such as skin tags, and anomalies which may (in some cases) be considered part of normal physiological development (e.g. isolated patent ductus arteriosus in preterm babies), have been counted equally alongside more serious anomalies such as spina bifida and Tetralogy of Fallot. Thus when considering the overall impact of congenital anomalies on children's subsequent developmental trajectories, or on future health service demand, it is necessary to consider the data presented on an anomaly by anomaly basis.

Note 4: In the New Zealand level analyses, large reductions in congenital anomaly rates are seen between 2007 and 2009, with rates then reverting to their pre-existing baseline by around 2012. It remains unclear however, whether these changes reflect real changes in the number of babies born with congenital anomalies, changes in the comprehensiveness of the recording of minor congenital anomalies by clinicians, or changes in the way in which congenital anomalies were coded in the hospital admission dataset.

New Zealand Distribution and Trends

New Zealand Distribution

In New Zealand during 2008–2012, a large number of congenital anomalies were identified at the time of birth, with these ranging in severity from minor skin conditions (e.g. skin tags, non-neoplastic nevus), through to anomalies which were incompatible with life (e.g. anencephaly). When interpreting the information in **Table 1** and **Table 2**, it must be remembered that the figures presented relate to the number of anomalies identified, rather than the number of babies, with many babies having more than one anomaly.

New Zealand Trends

In New Zealand, the number of babies with one or more congenital anomalies identified at birth increased gradually during the early 2000s, reached a peak in 2007 and then declined, with the most rapid declines occurring between 2007 and 2009. Rates then gradually increased again, reaching 5.5% of births by 2012 (**Figure 1**). It remains unclear however, whether the large declines seen between 2007 and 2009, and their subsequent rebound, reflect real changes in the number of babies born with congenital anomalies, changes in the comprehensiveness of the recording of minor congenital anomalies by clinicians, or changes in the way in which congenital anomalies were coded in the hospital admission dataset.

Distribution by Maternal Age

In New Zealand during 2008–2012, while the largest absolute numbers of babies with congenital anomalies were born to women aged 30–34 years, congenital anomaly rates rose steadily with increasing maternal age, with the highest rates being seen in the babies of mothers aged 40+ years. The babies of mothers aged 40+ years had congenital anomaly rates that were 1.32 (95% CI 1.20–1.44) times higher than the babies of teenage mothers (**Figure 2**, **Table 3**).

Table 1. Congenital Anomalies Evident at Birth, New Zealand 2008−2012 (Table 1 of 2)

Congenital Anomaly	Number: Total 2008−2012	Number: Annual Average	Anomalies per 100,000 Births*
Anencephaly	13	2.6	4.3
Encephalocele	13	2.6	4.3
Microcephaly	61	12.2	20.2
Congenital Hydrocephalus	77	15.4	25.5
Other Brain Malformations	190	38.0	62.9
Spina Bifida	53	10.6	17.5
Other Spinal Cord Malformations	14	2.8	4.6
Other CNS Malformations	24	4.8	7.9
Total Malformations of the Nervous System	445	89.0	147.2
Eyelid/Lacrimal/Eye/Orbit Malformations	105	21.0	34.7
Ear Malformations Impairing Hearing	19	3.8	6.3
Accessory Auricle	255	51.0	84.4
Other Ear Malformations	181	36.2	59.9
Other Face/Neck Malformations	62	12.4	20.5
Total Malformations of Eye, Ear, Face and Neck	622	124.4	205.7
Malformations Cardiac Chambers/Connections	190	38.0	62.9
Ventricular Septal Defect	533	106.6	176.3
Atrial Septal Defect	588	117.6	194.5
Atrioventricular Septal Defect	42	8.4	13.9
Tetralogy of Fallot	92	18.4	30.4
Pulmonary/Tricuspid Valve Malformations	185	37.0	61.2
Aortic/Mitral Valve Malformations	111	22.2	36.7
Other Heart Malformations	246	49.2	81.4
Patent Ductus Arteriosus (PDA)	1,490	298.0	492.9
Malformations Great Arteries (Excluding PDA)	313	62.6	103.5
Malformations Great Veins	52	10.4	17.2
Other Peripheral Vascular Malformations	165	33.0	54.6
Other Circulatory Malformations	15	3.0	5.0
Total Malformations of the Circulatory System	4,022	804.4	1,330.4
Nose Malformations	44	8.8	14.6
Larynx Malformations	105	21.0	34.7
Trachea/Bronchus Malformations	32	6.4	10.6
Lung Malformations	118	23.6	39.0
Other Respiratory Malformations	3	0.6	1.0
Total Malformations of the Respiratory System	302	60.4	99.9
Ankyloglossia (Tongue Tie)	3,699	739.8	1,223.6
Tongue/Mouth/Pharynx Malformations	79	15.8	26.1
Oesophagus/Upper Alimentary Malformations	80	16.0	26.5
Intestinal Malformations	270	54.0	89.3
Other Digestive Malformations	42	8.4	13.9
Total Malformations of the Digestive System	4,170	834.0	1,379.3

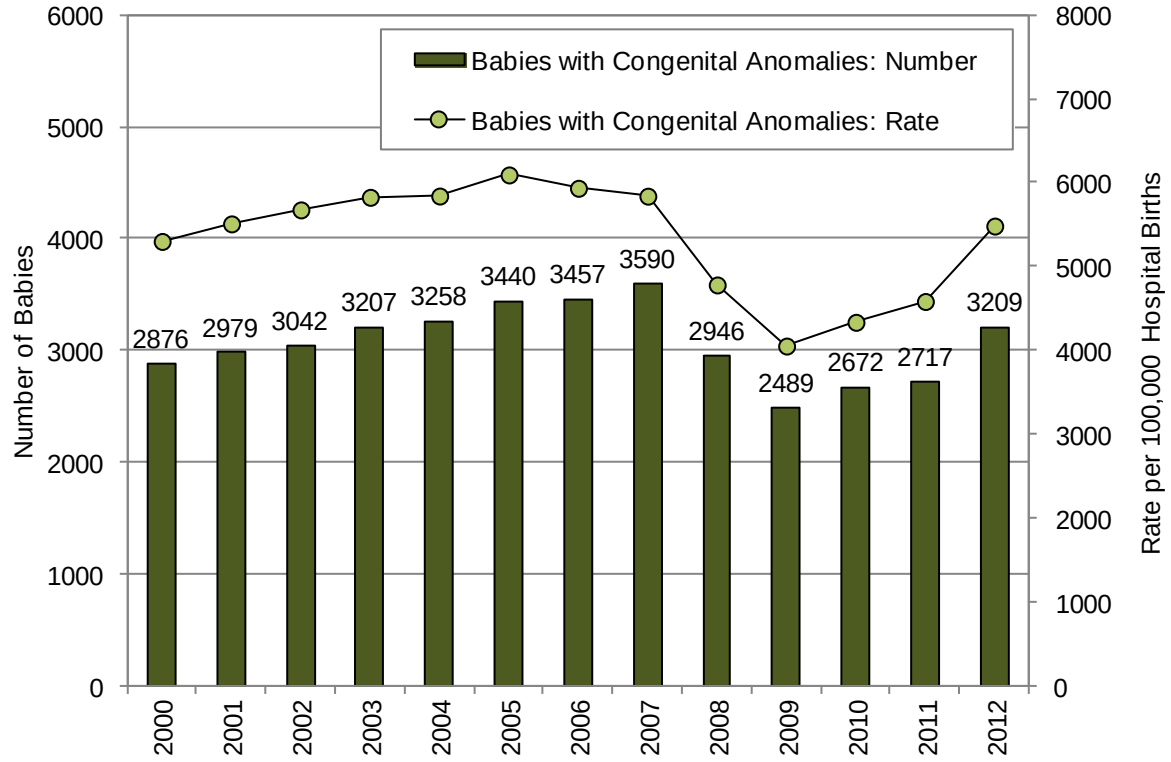
Source: National Minimum Dataset: Numerator: Hospitalisations with event type = birth and a congenital anomaly in any of first 15 diagnoses; Denominator: Hospitalisations with event type = birth; Note: *Anomalies per 100,000 births = number of anomalies rather than number of babies, as some babies have >1 anomaly

Table 2. Congenital Anomalies Evident at Birth, New Zealand 2008−2012 (Table 2 of 2)

Congenital Anomaly	Number: Total 2008−2012	Number: Annual Average	Anomalies per 100,000 Births*
Cleft Palate	186	37.2	61.5
Cleft Lip	68	13.6	22.5
Cleft Palate and Lip	116	23.2	38.4
Total Cleft Lip and Palate	370	74.0	122.4
Female Genital Malformations	64	12.8	21.2
Undescended Testicle	735	147.0	243.1
Hypospadias	576	115.2	190.5
Other Male Genital Malformations	157	31.4	51.9
Indeterminate Sex/Pseudohermaphrodism	24	4.8	7.9
Total Malformations of the Genital Organs	1,556	311.2	514.7
Renal Agenesis/Reduction Defects	94	18.8	31.1
Cystic Kidney Disease	144	28.8	47.6
Renal Pelvis Obstruction/Ureter Malformations	403	80.6	133.3
Other Kidney/Urinary Malformations	376	75.2	124.4
Total Malformations of the Urinary System	1,017	203.4	336.4
Congenital Dislocation/Subluxation Hip	60	12.0	19.9
Other Deformities Hip	461	92.2	152.5
Foot Deformities	1,516	303.2	501.5
Polydactyly	279	55.8	92.3
Syndactyly	106	21.2	35.1
Reduction Defects/Other Limb Malformations	177	35.4	58.6
Skull/Facial Bones/Spine/Thorax Malformations	214	42.8	70.8
Other Musculoskeletal Malformations	532	106.4	176.0
Osteochondrodysplasia	35	7.0	11.6
Total Malformations of the Musculoskeletal System	3,380	676.0	1,118.0
Ichthyosis	8	1.6	2.7
Non-Neoplastic Naevus	451	90.2	149.2
Epidermolysis Bullosa	11	2.2	3.6
Other Skin Malformations	450	90.0	148.9
Breast Malformations	15	3.0	5.0
Other Integument Malformations	434	86.8	143.6
Other Malformations	396	79.2	131.0
Total Other Congenital Malformations	1,765	353.0	583.8
Down Syndrome	257	51.4	85.0
Edwards and Patau Syndromes	40	8.0	13.2
Other Autosomal Trisomies	15	3.0	5.0
Turners Syndrome	10	2.0	3.3
Sex Chromosome Anomalies Male Phenotype	12	2.4	4.0
Monosomies/Autosomal Deletions/Rearrangements	25	5.0	8.3
Other Chromosome Anomalies	33	6.6	10.9
Total Chromosomal Anomalies	392	78.4	129.7

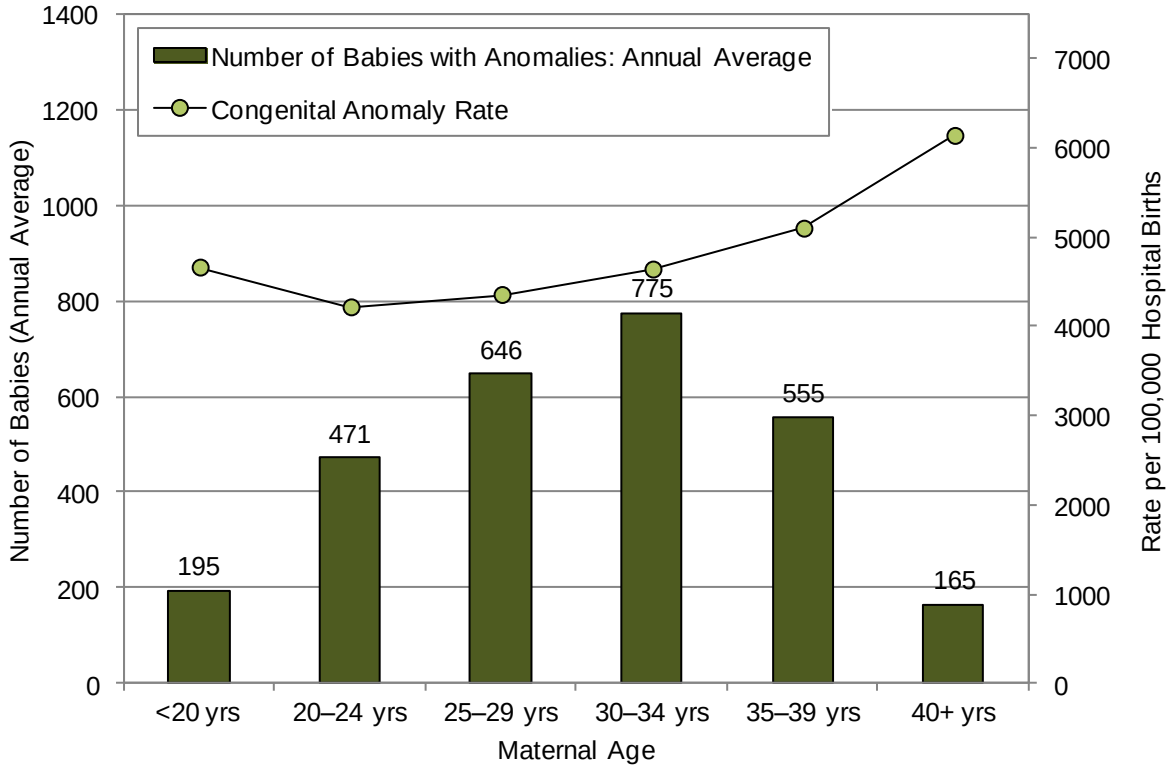
Source: National Minimum Dataset: Numerator: Hospitalisations with event type = birth and a congenital anomaly in any of first 15 diagnoses; Denominator: Hospitalisations with event type = birth; Note: *Anomalies per 100,000 births = number of anomalies rather than number of babies, as some babies have >1 anomaly

Figure 1. Babies with Congenital Anomalies Evident at Birth, New Zealand Hospital Births 2000–2012



Source: National Minimum Dataset: Numerator: Hospital admissions with event type = birth and a congenital anomaly listed in any of first 15 diagnoses; Denominator: All hospital admissions with event type = birth; Note: Rate per 100,000 refers to number of babies with one or more anomalies, rather than number of anomalies

Figure 2. Babies with Congenital Anomalies Evident at Birth by Maternal Age, New Zealand Hospital Births 2008–2012



Source: National Minimum Dataset: Numerator: Hospital admissions with event type = birth and a congenital anomaly listed in any of first 15 diagnoses; Denominator: All hospital admissions with event type = birth; Note: Rate per 100,000 refers to number of babies with one or more anomalies, rather than number of anomalies

Distribution by Prioritised Ethnicity, NZDep Index Decile and Gender

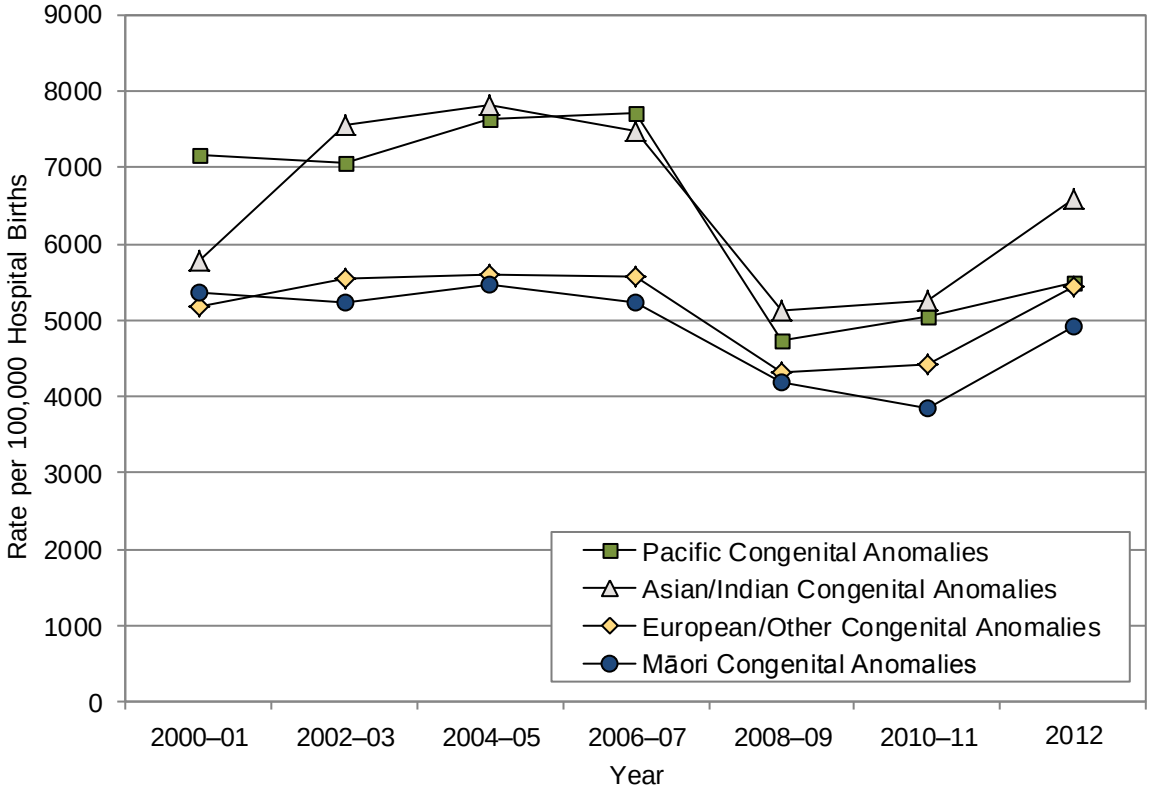
In New Zealand during 2008–2012, the proportion of babies with one or more congenital anomalies identified at birth was *significantly* higher for males, Asian/Indian and Pacific > European/Other > Māori babies, and those from the least deprived (NZDep06 deciles 1–2 vs. 5–10) areas (**Table 3**). Similarly, congenital anomaly rates were generally higher for Asian/Indian and Pacific babies, than for European/Other and Māori babies during 2000–2012 (**Figure 3**).

Table 3. Babies with Congenital Anomalies Evident at Birth by Ethnicity, NZ Deprivation Index Decile, Gender and Maternal Age, New Zealand Hospital Births 2008–2012

Variable	Number of Babies: Annual Average	Rate per 100,000 Births	Rate Ratio	95% CI
Babies with Congenital Anomalies				
Prioritised Ethnicity				
Māori	559	4,190	0.92	0.88–0.96
Pacific	339	5,003	1.09	1.04–1.15
European/Other	1,516	4,573	1.00	
Asian/Indian	378	5,528	1.21	1.15–1.27
NZ Deprivation Index				
Deciles 1–2	427	5,035	1.00	
Deciles 3–4	432	4,790	0.95	0.90–1.01
Deciles 5–6	516	4,584	0.91	0.86–0.96
Deciles 7–8	651	4,618	0.92	0.87–0.97
Deciles 9–10	774	4,420	0.88	0.83–0.92
Gender				
Female	1,116	3,797	1.00	
Male	1,690	5,441	1.43	1.39–1.48
Maternal Age				
<20 Years	195	4,662	1.00	
20–24 Years	471	4,216	0.90	0.84–0.97
25–29 Years	646	4,354	0.93	0.87–1.00
30–34 Years	775	4,640	1.00	0.93–1.07
35–39 Years	555	5,098	1.09	1.02–1.17
40+ Years	165	6,137	1.32	1.20–1.44

Source: National Minimum Dataset: Numerator: Hospital admissions with event type = birth and a congenital anomaly listed in any of first 15 diagnoses; Denominator: All hospital admissions with event type = birth; Note: Rate per 100,000 refers to number of babies with one or more anomalies, rather than number of anomalies; Rate Ratios are unadjusted

Figure 3. Babies with Congenital Anomalies Evident at Birth by Ethnicity, New Zealand Hospital Births 2000–2012



Source: National Minimum Dataset: Numerator: Hospital admissions with event type = birth and a congenital anomaly listed in any of first 15 diagnoses; Denominator: All hospital admissions with event type = birth; Note: Rate per 100,000 refers to number of babies with one or more anomalies, rather than number of anomalies

South Island DHBs Distribution and Trends

South Island DHBs Distribution

In the South Island DHBs during 2008–2012, a large number of congenital anomalies were identified at birth, with these ranging in severity from minor (e.g. tongue tie) through to serious (e.g. malformations of the great arteries) (**Table 4** to **Table 14**).

When the number of babies with one or more congenital anomalies, rather than the number of anomalies was considered, on average during 2008–2012, 61 Nelson Marlborough, 14 South Canterbury, 256 Canterbury, 17 West Coast, 96 Otago and 70 Southland babies per year (range 2.3%–5.0% of all births) had one or more congenital anomalies identified at birth. Rates in Nelson Marlborough, South Canterbury and Canterbury were *significantly* lower than the New Zealand rate, while rates in the West Coast, Otago and Southland were not *significantly* different (**Table 15**).

Note: It is unclear whether DHB vs. NZ differences in congenital anomaly rates reflect real differences in the underlying prevalence of congenital anomalies in the community, or differences in the thoroughness with which minor congenital anomalies are recorded in the clinical notes or the National Minimum Dataset.

Table 4. Congenital Anomalies Evident at Birth, Canterbury 2008–2012 (Table 1 of 2)

Congenital Anomaly	Number: Total 2008−2012	Number: Annual Average	Number of Anomalies per 100,000 Births*
Canterbury			
Anencephaly	3	0.6	9.9
Microcephaly	6	1.2	19.8
Congenital Hydrocephalus	15	3.0	49.4
Other Brain Malformations	34	6.8	112.1
Spina Bifida	<3	s	s
Other CNS Malformations	<3	s	s
Total Malformations of the Nervous System	61	12.2	201.0
Eyelid/Lacrimal/Eye/Orbit Malformations	10	2.0	33.0
Accessory Auricle	8	1.6	26.4
Other Ear Malformations	41	8.2	135.1
Other Face/Neck Malformations	12	2.4	39.6
Total Malformations of Eye, Ear, Face and Neck	71	14.2	234.0
Malformations Cardiac Chambers/Connections	21	4.2	69.2
Ventricular Septal Defect	99	19.8	326.3
Atrial Septal Defect	46	9.2	151.6
Atrioventricular Septal Defect	3	0.6	9.9
Tetralogy of Fallot	10	2.0	33.0
Pulmonary/Tricuspid Valve Malformations	25	5.0	82.4
Aortic/Mitral Valve Malformations	11	2.2	36.3
Other Heart Malformations	16	3.2	52.7
Patent Ductus Arteriosus (PDA)	188	37.6	619.6
Malformations Great Arteries (Excluding PDA)	39	7.8	128.5
Malformations Great Veins	7	1.4	23.1
Other Circulatory Malformations	3	0.6	9.9
Total Malformations of the Circulatory System	468	93.6	1,542.3
Nose Malformations	7	1.4	23.1
Larynx Malformations	16	3.2	52.7
Trachea/Bronchus Malformations	3	0.6	9.9
Lung Malformations	14	2.8	46.1
Total Malformations of the Respiratory System	40	8.0	131.8
Ankyloglossia (Tongue Tie)	389	77.8	1,282.0
Tongue/Mouth/Pharynx Malformations	13	2.6	42.8
Oesophagus/Upper Alimentary Malformations	7	1.4	23.1
Intestinal Malformations	28	5.6	92.3
Other Digestive Malformations	3	0.6	9.9
Total Malformations of the Digestive System	440	88.0	1,450.1

Source: National Minimum Dataset: Numerator: Hospitalisations with event type = birth and a congenital anomaly in any of first 15 diagnoses; Denominator: Hospitalisations with event type = birth; Note: *Anomalies per 100,000 births = number of anomalies rather than number of babies, as some babies have >1 anomaly; s: suppressed due to small numbers

Table 5. Congenital Anomalies Evident at Birth, Canterbury 2008–2012 (Table 2 of 2)

Congenital Anomaly	Number: Total 2008−2012	Number: Annual Average	Number of Anomalies per 100,000 Births*
Canterbury			
Cleft Palate	18	3.6	59.3
Cleft Lip	7	1.4	23.1
Cleft Palate and Lip	6	1.2	19.8
Total Cleft Lip and Palate	31	6.2	102.2
Female Genital Malformations	5	1.0	16.5
Undescended Testicle	22	4.4	72.5
Hypospadias	46	9.2	151.6
Other Male Genital Malformations	15	3.0	49.4
Other Genital Malformations	<3	s	s
Total Malformations of the Genital Organs	89	17.8	293.3
Renal Agenesis/Reduction Defects	11	2.2	36.3
Cystic Kidney Disease	16	3.2	52.7
Renal Pelvis Obstruction/Ureter Malformations	21	4.2	69.2
Other Kidney/Urinary Malformations	37	7.4	121.9
Total Malformations of the Urinary System	85	17.0	280.1
Skull/Facial Bones/Spine/Thorax Malformations	20	4.0	65.9
Foot Deformities	95	19.0	313.1
Polydactyly	14	2.8	46.1
Syndactyly	5	1.0	16.5
Congenital Dislocation Hip	4	0.8	13.2
Other Deformities Hip	30	6.0	98.9
Reduction Defects/Other Limb Malformations	26	5.2	85.7
Osteochondrodysplasia	8	1.6	26.4
Other Musculoskeletal Malformations	45	9.0	148.3
Total Malformations of the Musculoskeletal System	247	49.4	814.0
Non-Neoplastic Naevus	7	1.4	23.1
Epidermolysis Bullosa	4	0.8	13.2
Other Skin Malformations	21	4.2	69.2
Other Integument Malformations	37	7.4	121.9
Other Malformations	42	8.4	138.4
Total Other Congenital Malformations	111	22.2	365.8
Down Syndrome	23	4.6	75.8
Edwards and Patau Syndromes	3	0.6	9.9
Turners Syndrome/Other Female Chromosome Anomalies	4	0.8	13.2
Monosomies/Autosomal Deletions/Rearrangements	3	0.6	9.9
Other Chromosome Anomalies	5	1.0	16.5
Total Chromosomal Anomalies	38	7.6	125.2

Source: National Minimum Dataset: Numerator: Hospitalisations with event type = birth and a congenital anomaly in any of first 15 diagnoses; Denominator: Hospitalisations with event type = birth; Note: *Anomalies per 100,000 births = number of anomalies rather than number of babies, as some babies have >1 anomaly; s: suppressed due to small numbers

Table 6. Congenital Anomalies Evident at Birth, Nelson Marlborough 2008–2012 (Table 1 of 2)

Congenital Anomaly	Number: Total 2008−2012	Number: Annual Average	Number of Anomalies per 100,000 Births*
Nelson Marlborough			
Brain Malformations	6	1.2	76.5
Spina Bifida and Other CNS Malformations	<3	s	s
Total Malformations of the Nervous System	8	1.6	102.0
Accessory Auricle/Other Ear Malformations	4	0.8	51.0
Other Eye/Face/Neck Malformations	3	0.6	38.2
Total Malformations of Eye, Ear, Face and Neck	7	1.4	89.2
Malformations Cardiac Chambers/Connections	5	1.0	63.7
Ventricular Septal Defect	14	2.8	178.4
Atrial Septal Defect	14	2.8	178.4
Other Heart Malformations	10	2.0	127.5
Patent Ductus Arteriosus (PDA)	43	8.6	548.1
Malformations Great Arteries (Excluding PDA)	9	1.8	114.7
Other Peripheral Vascular Malformations	<3	s	s
Total Malformations of the Circulatory System	96	19.2	1,223.6
Lung Malformations	3	0.6	38.2
Other Respiratory Malformations	3	0.6	38.2
Total Malformations of the Respiratory System	6	1.2	76.5
Cleft Palate	4	0.8	51.0
Cleft Lip	<3	s	s
Cleft Palate and Lip	5	1.0	63.7
Total Cleft Lip and Palate	10	2.0	127.5
Ankyloglossia (Tongue Tie)	38	7.6	484.3
Oesophagus/Upper Alimentary Malformations	3	0.6	38.2
Intestinal Malformations	4	0.8	51.0
Other Digestive Malformations	<3	s	s
Total Malformations of the Digestive System	47	9.4	599.0
Undescended Testicle	9	1.8	114.7
Hypospadias	28	5.6	356.9
Other Male Genital Malformations	7	1.4	89.2
Other Genital Malformations	<3	s	s
Total Malformations of the Genital Organs	45	9.0	573.6
Cystic Kidney Disease	3	0.6	38.2
Renal Pelvis Obstruction/Ureter Malformations	9	1.8	114.7
Other Kidney/Urinary Malformations	9	1.8	114.7
Total Malformations of the Urinary System	21	4.2	267.7
Down Syndrome	4	0.8	51.0
Other Chromosome Anomalies	5	1.0	63.7
Total Chromosomal Anomalies	9	1.8	114.7

Source: National Minimum Dataset: Numerator: Hospitalisations with event type = birth and a congenital anomaly in any of first 15 diagnoses; Denominator: Hospitalisations with event type = birth; Note: *Anomalies per 100,000 births = number of anomalies rather than number of babies, as some babies have >1 anomaly; s: suppressed due to small numbers

Table 7. Congenital Anomalies Evident at Birth, Nelson Marlborough 2008–2012 (Table 2 of 2)

Congenital Anomaly	Number: Total 2008−2012	Number: Annual Average	Number of Anomalies per 100,000 Births*
Nelson Marlborough			
Skull/Facial Bones/Spine/Thorax Malformations	3	0.6	38.2
Foot Deformities	34	6.8	433.3
Polydactyly	10	2.0	127.5
Syndactyly	3	0.6	38.2
Other Deformities Hip	23	4.6	293.1
Reduction Defects/Other Limb Malformations	3	0.6	38.2
Other Musculoskeletal Malformations	14	2.8	178.4
Total Malformations of the Musculoskeletal System	90	18.0	1,147.1
Non-Neoplastic Naevus	9	1.8	114.7
Other Skin Malformations	16	3.2	203.9
Other Integument Malformations	7	1.4	89.2
Other Malformations	11	2.2	140.2
Total Other Congenital Malformations	43	8.6	548.1

Source: National Minimum Dataset: Numerator: Hospitalisations with event type = birth and a congenital anomaly in any of first 15 diagnoses; Denominator: Hospitalisations with event type = birth; Note: *Anomalies per 100,000 births = number of anomalies rather than number of babies, as some babies have >1 anomaly

Table 8. Congenital Anomalies Evident at Birth, West Coast 2008–2012 (Table 1 of 2)

Congenital Anomaly	Number: Total 2008−2012	Number: Annual Average	Number of Anomalies per 100,000 Births*
West Coast			
Congenital Hydrocephalus	<3	s	s
Other Brain Malformations	<3	s	s
Spina Bifida	<3	s	s
Total Malformations of the Nervous System	4	0.8	217.4
Malformations of Eye, Ear, Face and Neck	<3	s	s
Total Malformations of Eye, Ear, Face and Neck	<3	s	s
Malformations Cardiac Chambers/Connections	3	0.6	163.0
Atrial Septal Defect	5	1.0	271.7
Heart Valve Malformations	4	0.8	217.4
Other Heart Malformations	5	1.0	271.8
Patent Ductus Arteriosus (PDA)	11	2.2	597.8
Malformations Great Arteries (Excluding PDA)	3	0.6	163.0
Other Circulatory Malformations	<3	s	s
Total Malformations of the Circulatory System	32	6.4	1,739.1
Malformations of the Respiratory System	<3	s	s
Total Malformations of the Respiratory System	<3	s	s
Total Cleft Lip and Palate	<3	s	s
Total Cleft Lip and Palate	<3	s	s

Source: National Minimum Dataset: Numerator: Hospitalisations with event type = birth and a congenital anomaly in any of first 15 diagnoses; Denominator: Hospitalisations with event type = birth; Note: *Anomalies per 100,000 births = number of anomalies rather than number of babies, as some babies have >1 anomaly; s: suppressed due to small numbers

Table 9. Anomalies Evident at Birth, West Coast 2008–2012 (Table 2 of 2)

Congenital Anomaly	Number: Total 2008−2012	Number: Annual Average	Number of Anomalies per 100,000 Births*
West Coast			
Ankyloglossia (Tongue Tie)	30	6.0	1,630.4
Other Digestive Malformations	<3	s	s
Total Malformations of the Digestive System	32	6.4	1,739.1
Undescended Testicle	<3	s	s
Hypospadias	3	0.6	163.0
Other Male Genital Malformations	<3	s	s
Total Malformations of the Genital Organs	6	1.2	326.1
Malformations of the Urinary System	3	0.6	163.1
Total Malformations of the Urinary System	3	0.6	163.1
Skull/Facial Bones/Spine/Thorax Malformations	3	0.6	163.0
Foot Deformities	8	1.6	434.8
Polydactyly/Syndactyly	3	0.6	163.1
Other Deformities Hip	4	0.8	217.4
Other Musculoskeletal Malformations	3	0.6	163.0
Total Malformations of the Musculoskeletal System	21	4.2	1,141.3
Other Congenital Malformations	3	0.6	163.1
Total Other Congenital Malformations	3	0.6	163.1
Down Syndrome	4	0.8	217.4
Other Chromosomal Anomalies	<3	s	s
Total Chromosomal Anomalies	5	1.0	271.7

Source: National Minimum Dataset: Numerator: Hospitalisations with event type = birth and a congenital anomaly in any of first 15 diagnoses; Denominator: Hospitalisations with event type = birth; Note: *Anomalies per 100,000 births = number of anomalies rather than number of babies, as some babies have >1 anomaly; s: suppressed due to small numbers

Table 10. Congenital Anomalies Evident at Birth, South Canterbury 2008–2012

Congenital Anomaly	Number: Total 2008−2012	Number: Annual Average	Number of Anomalies per 100,000 Births*
South Canterbury			
Microcephaly	<3	s	s
Congenital Hydrocephalus	<3	s	s
Other Brain Malformations	4	0.8	128.7
Spina Bifida	<3	s	s
Other CNS Malformations	<3	s	s
Total Malformations of the Nervous System	11	2.2	354.0
Malformations of Eye, Ear, Face and Neck	<3	s	s
Total Malformations of Eye, Ear, Face and Neck	<3	s	s
Heart Malformations	9	1.8	289.7
Patent Ductus Arteriosus (PDA)	10	2.0	321.9
Malformations Great Arteries (Excluding PDA)	3	0.6	96.6
Total Malformations of the Circulatory System	22	4.4	708.1
Cleft Palate	3	0.6	96.6
Total Cleft Lip and Palate	3	0.6	96.6
Ankyloglossia (Tongue Tie)	22	4.4	708.1
Other Digestive Malformations	3	0.6	96.6
Total Malformations of the Digestive System	25	5.0	804.6
Hypospadias	4	0.8	128.7
Other Male Genital Malformations	4	0.8	128.8
Other Genital Malformations	<3	s	s
Total Malformations of the Genital Organs	9	1.8	289.7
Renal Pelvis Obstruction/Ureter Malformations	3	0.6	96.6
Other Kidney/Urinary Malformations	3	0.6	96.6
Total Malformations of the Urinary System	6	1.2	193.1
Foot Deformities	3	0.6	96.6
Deformities of Hip	4	0.8	128.8
Other Musculoskeletal Malformations	6	1.2	193.1
Total Malformations of the Musculoskeletal System	13	2.6	418.4
Other Congenital Malformations	5	1.0	160.9
Total Other Congenital Malformations	5	1.0	160.9
Down Syndrome	<3	s	s
Other Chromosomal Anomalies	<3	s	s
Total Chromosomal Anomalies	4	0.8	128.8

Source: National Minimum Dataset: Numerator: Hospitalisations with event type = birth and a congenital anomaly in any of first 15 diagnoses; Denominator: Hospitalisations with event type = birth; Note: *Anomalies per 100,000 births = number of anomalies rather than number of babies, as some babies have >1 anomaly; s: suppressed due to small numbers

Table 11. Congenital Anomalies Evident at Birth, Otago 2008–2012 (Table 1 of 2)

Congenital Anomaly	Number: Total 2008−2012	Number: Annual Average	Number of Anomalies per 100,000 Births*
Otago			
Microcephaly	<3	s	s
Congenital Hydrocephalus	<3	s	s
Other Brain Malformations	3	0.6	31.3
Spina Bifida	<3	s	s
Total Malformations of the Nervous System	7	1.4	73.1
Eyelid/Lacrimal/Eye/Orbit Malformations	4	0.8	41.7
Accessory Auricle	3	0.6	31.3
Other Ear Malformations	4	0.8	41.7
Total Malformations of Eye, Ear, Face and Neck	11	2.2	114.8
Malformations Cardiac Chambers/Connections	6	1.2	62.6
Ventricular Septal Defect	40	8.0	417.4
Atrial Septal Defect	56	11.2	584.4
Atrioventricular Septal Defect	3	0.6	31.3
Tetralogy of Fallot	3	0.6	31.3
Pulmonary/Tricuspid Valve Malformations	4	0.8	41.7
Aortic/Mitral Valve Malformations	7	1.4	73.1
Other Heart Malformations	15	3.0	156.5
Patent Ductus Arteriosus (PDA)	80	16.0	834.8
Malformations Great Arteries (Excluding PDA)	14	2.8	146.1
Other Circulatory Malformations	4	0.8	41.8
Total Malformations of the Circulatory System	232	46.4	2,421.0
Lung Malformations	3	0.6	31.3
Total Malformations of the Respiratory System	3	0.6	31.3
Cleft Palate	7	1.4	73.1
Cleft Lip	<3	s	s
Cleft Palate and Lip	<3	s	s
Total Cleft Lip and Palate	11	2.2	114.8
Ankyloglossia (Tongue Tie)	237	47.4	2,473.1
Oesophagus/Upper Alimentary Malformations	5	1.0	52.2
Intestinal Malformations	10	2.0	104.4
Other Digestive Malformations	<3	s	s
Total Malformations of the Digestive System	253	50.6	2,640.1
Hypospadias	13	2.6	135.7
Other Male Genital Malformations	3	0.6	31.3
Other Genital Malformations	<3	s	s
Total Malformations of the Genital Organs	17	3.4	177.4
Cystic Kidney Disease	3	0.6	31.3
Renal Pelvis Obstruction/Ureter Malformations	7	1.4	73.1
Other Kidney/Urinary Malformations	4	0.8	41.8
Total Malformations of the Urinary System	14	2.8	146.1

Source: National Minimum Dataset: Numerator: Hospitalisations with event type = birth and a congenital anomaly in any of first 15 diagnoses; Denominator: Hospitalisations with event type = birth; Note: *Anomalies per 100,000 births = number of anomalies rather than number of babies, as some babies have >1 anomaly; s: suppressed due to small numbers

Table 12. Congenital Anomalies Evident at Birth, Otago 2008–2012 (Table 2 of 2)

Congenital Anomaly	Number: Total 2008−2012	Number: Annual Average	Number of Anomalies per 100,000 Births*
Otago			
Skull/Facial Bones/Spine/Thorax Malformations	9	1.8	93.9
Foot Deformities	8	1.6	83.5
Polydactyly/Syndactyly	3	0.6	31.3
Congenital Dislocation Hip	3	0.6	31.3
Other Deformities Hip	16	3.2	167.0
Other Musculoskeletal Malformations	7	1.4	73.1
Total Malformations of the Musculoskeletal System	46	9.2	480.0
Skin Malformations	4	0.8	41.7
Other Malformations	14	2.8	146.1
Total Other Congenital Malformations	18	3.6	187.8
Down Syndrome	11	2.2	114.8
Other Chromosome Anomalies	5	1.0	52.2
Total Chromosomal Anomalies	16	3.2	167.0

Source: National Minimum Dataset: Numerator: Hospitalisations with event type = birth and a congenital anomaly in any of first 15 diagnoses; Denominator: Hospitalisations with event type = birth; Note: *Anomalies per 100,000 births = number of anomalies rather than number of babies, as some babies have >1 anomaly

Table 13. Congenital Anomalies Evident at Birth, Southland 2008–2012 (Table 1 of 2)

Congenital Anomaly	Number: Total 2008−2012	Number: Annual Average	Number of Anomalies per 100,000 Births*
Southland			
Encephalocele	<3	s	s
Congenital Hydrocephalus	5	1.0	65.4
Other Brain Malformations	3	0.6	39.2
Spinal Cord Malformations	<3	s	s
Total Malformations of the Nervous System	10	2.0	130.7
Eyelid/Lacrimal/Eye/Orbit Malformations	4	0.8	52.3
Ear Malformations	5	1.0	65.4
Other Face/Neck Malformations	<3	s	s
Total Malformations of Eye, Ear, Face and Neck	11	2.2	143.8
Ventricular Septal Defect	15	3.0	196.1
Atrial Septal Defect	30	6.0	392.2
Heart Valve Malformations	9	1.8	117.7
Other Heart Malformations	8	1.6	104.6
Patent Ductus Arteriosus (PDA)	40	8.0	522.9
Malformations Great Arteries (Excluding PDA)	5	1.0	65.4
Other Peripheral Vascular Malformations	<3	s	s
Total Malformations of the Circulatory System	109	21.8	1,425.0

Source: National Minimum Dataset: Numerator: Hospitalisations with event type = birth and a congenital anomaly in any of first 15 diagnoses; Denominator: Hospitalisations with event type = birth; Note: *Anomalies per 100,000 births = number of anomalies rather than number of babies, as some babies have >1 anomaly; s: suppressed due to small numbers

Table 14. Congenital Anomalies Evident at Birth, Southland 2008–2012 (Table 2 of 2)

Congenital Anomaly	Number: Total 2008−2012	Number: Annual Average	Number of Anomalies per 100,000 Births*
Southland			
Larynx Malformations	<3	s	s
Lung Malformations	3	0.6	39.2
Total Malformations of the Respiratory System	5	1.0	65.4
Cleft Palate	3	0.6	39.2
Cleft Lip	<3	s	s
Cleft Palate and Lip	<3	s	s
Total Cleft Lip and Palate	6	1.2	78.4
Ankyloglossia (Tongue Tie)	41	8.2	536.0
Tongue/Mouth/Pharynx Malformations	3	0.6	39.2
Oesophagus/Upper Alimentary Malformations	3	0.6	39.2
Intestinal Malformations	4	0.8	52.3
Total Malformations of the Digestive System	51	10.2	666.8
Undescended Testicle	5	1.0	65.4
Hypospadias	23	4.6	300.7
Other Genital Malformations	3	0.6	39.2
Total Malformations of the Genital Organs	31	6.2	405.3
Renal Agenesis/Reduction Defects	4	0.8	52.3
Cystic Kidney Disease	5	1.0	65.4
Renal Pelvis Obstruction/Ureter Malformations	82	16.4	1,072.0
Other Kidney/Urinary Malformations	4	0.8	52.3
Total Malformations of the Urinary System	95	19.0	1,242.0
Skull/Facial Bones/Spine/Thorax Malformations	4	0.8	52.3
Foot Deformities	32	6.4	418.4
Polydactyly	7	1.4	91.5
Syndactyly	3	0.6	39.2
Deformities of Hip	11	2.2	143.8
Other Musculoskeletal Malformations	14	2.8	183.0
Total Malformations of the Musculoskeletal System	71	14.2	928.2
Skin Malformations	10	2.0	130.7
Other Integument Malformations	7	1.4	91.5
Other Malformations	11	2.2	143.8
Total Other Congenital Malformations	28	5.6	366.1
Down Syndrome	8	1.6	104.6
Total Chromosomal Anomalies	8	1.6	104.6

Source: National Minimum Dataset: Numerator: Hospitalisations with event type = birth and a congenital anomaly in any of first 15 diagnoses; Denominator: Hospitalisations with event type = birth; Note: *Anomalies per 100,000 births = number of anomalies rather than number of babies, as some babies have >1 anomaly; s: suppressed due to small numbers

Table 15. Babies with Congenital Anomalies Evident at Birth, South Island DHBs vs. New Zealand Hospital Births 2008–2012

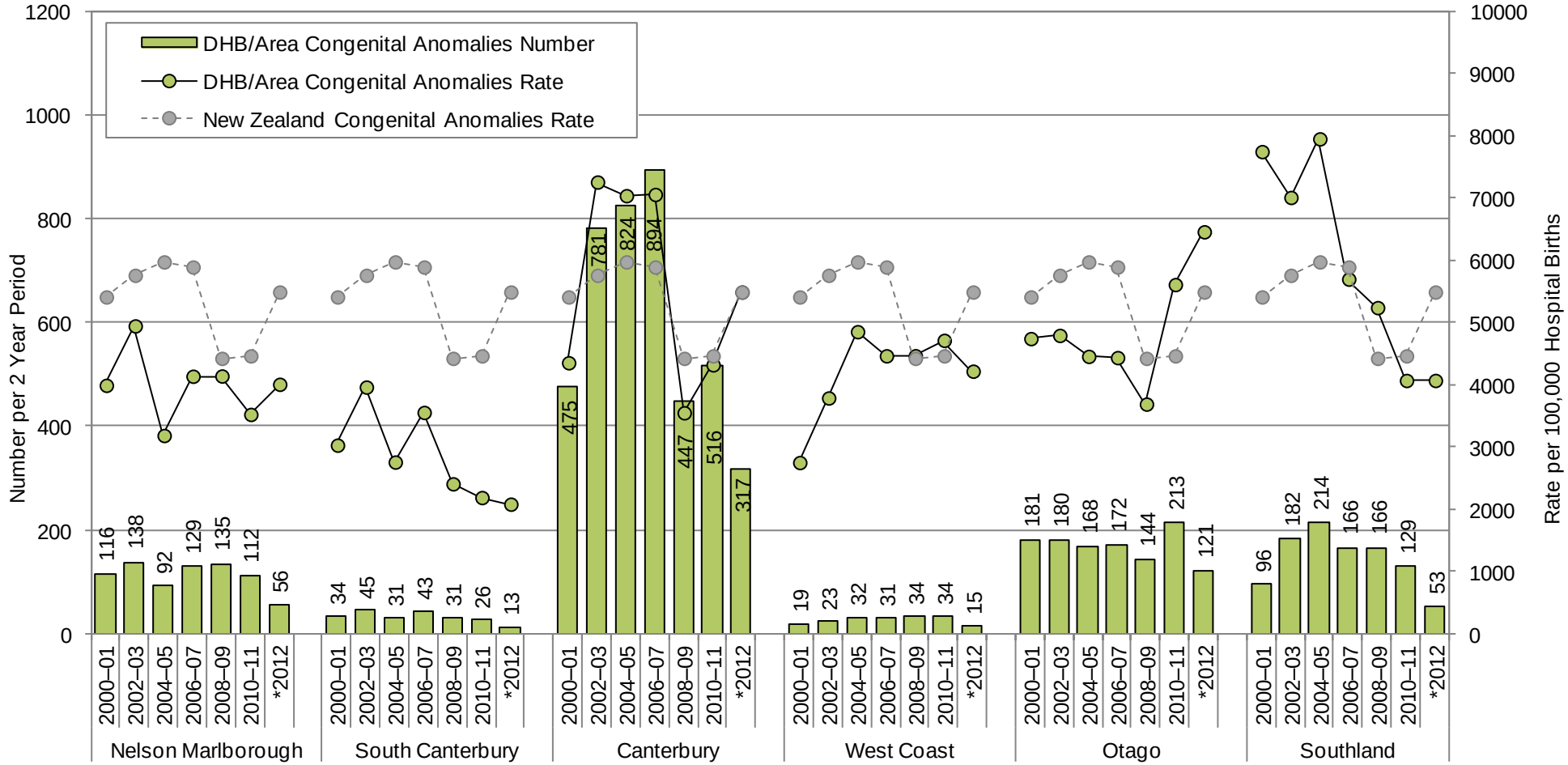
DHB/Area	Number: Total 2008−2012	Number: Annual Average	Rate per 100,000	Rate Ratio	95% CI
Babies with Congenital Anomalies					
Nelson Marlborough	303	61	3,862	0.83	0.74–0.93
South Canterbury	70	14	2,253	0.49	0.38–0.61
Canterbury	1,280	256	4,218	0.91	0.86–0.96
West Coast	83	17	4,511	0.97	0.79–1.20
Otago	478	96	4,988	1.07	0.98–1.17
Southland	348	70	4,550	0.98	0.88–1.09
New Zealand	14,033	2,807	4,642	1.00	

Source: National Minimum Dataset: Numerator: Hospital admissions with event type = birth and a congenital anomaly listed in any of first 15 diagnoses; Denominator: All hospital admissions with event type = birth; Note: Rate per 100,000 refers to number of babies with one or more anomalies, rather than number of anomalies; Rate Ratios are compared to New Zealand rates and have not been adjusted for population demographics

South Island DHBs Trends

In the South Island during 2000–2012, while trends for individual DHBs varied, the proportion of babies with congenital anomalies identified at birth in Nelson Marlborough and South Canterbury was consistently lower than the New Zealand rate (**Figure 4**). Again it is unclear whether individual DHB trends reflect real changes in prevalence, or merely changes in the way in which minor congenital anomalies were recorded in the hospital admission dataset over time.

Figure 4. Babies with Congenital Anomalies Evident at Birth, South Island DHBs vs. New Zealand Hospital Births 2000–2012



Source: National Minimum Dataset: Numerator: Hospital admissions with event type = birth and a congenital anomaly listed in any of first 15 diagnoses; Denominator: All hospital admissions with event type = birth; Note: Rate per 100,000 refers to number of babies with one or more anomalies, rather than number of anomalies; Note: *Numbers are per 2 year period, except for 2012 which is for a single year only

Local Policy Documents and Evidence-Based Reviews Relevant to Congenital Anomalies

In New Zealand there is a paucity of publications relevant to the prevention or management of congenital anomalies as a group. The publications that are available are summarised in **Table 16** below along with a range of reviews which consider these issues in the overseas context.

In addition, Table 16 (Local Policy Documents and Evidence-Based Reviews Relevant to Antenatal and Newborn Screening) on Page 81 considers publications relevant to antenatal and newborn screening, while Table 38 (Policy Documents and Evidence-Based Reviews Relevant to Cardiovascular Anomalies) on Page 118 considers cardiovascular anomalies, Table 44 (Local Policy Documents and Evidence-Based Reviews Relevant to Down Syndrome and Other Chromosomal Anomalies) on Page 128 considers Down syndrome and other chromosomal anomalies, and Table 49 (Policy Documents and Evidence-Based Reviews Relevant to Neural Tube Defects) on Page 138 considers neural tube defects. Finally Table 56 (Local Policy Documents and Evidence-Based Reviews Relevant to the Early Detection and Management of Permanent Hearing Loss in Children) on Page 154 provides a brief overview of publications relevant to newborn screening for congenital hearing loss.

Table 16. Local Policy Documents and Evidence-Based Reviews Relevant to Congenital Anomalies

Ministry of Health Documents
Ministry of Health. 2011. **Immunisation Handbook 2011**. Wellington: Ministry of Health. http://www.health.govt.nz/system/files/documents/publications/immunisationhandbook2011-v3.pdf Chapter 12 of the Immunisation handbook deals with rubella. It states that maternal infection with rubella in the first eight weeks of pregnancy leads to fetal damage in 85% of infants and that multiple defects are common. The current immunisation schedule contains two doses of measles, mumps and rubella vaccine, offered at ages 15 months and four years. It is recommended that all women of childbearing age, particularly immigrants from less developed countries, be offered screening to check their rubella immunity in their early reproductive years, when they are planning a pregnancy and when they are pregnant. Non-pregnant non-immune women should be offered vaccination immediately, and pregnant women should be offered vaccination after delivery. Women should not be vaccinated if they are pregnant and they should avoid pregnancy for four weeks after immunisation.
International Guidelines and Systematic and Other Reviews
Di Mario S, Basevi V, Gagliotti C, et al. 2013. **Prenatal education for congenital toxoplasmosis**. Cochrane Database of Systematic Reviews doi:10.1002/14651858.CD006171.pub3 http://onlinelibrary.wiley.com/doi/10.1002/14651858.CD006171.pub3/abstract Toxoplasmosis is caused by an animal parasite, *Toxoplasma gondii*. Infection occurs by ingesting oocysts excreted by cats, contaminated soil or water, or by eating the undercooked meat of infected animals. Infection in pregnancy can result in transmission of infection to the fetus. Congenital toxoplasmosis can lead to intrauterine death or stillbirth, mental retardation, malformations and deafness and blindness of the infant. Both prenatal screening of women and postnatal screening of babies are possible but such screening has a number of limitations and may not be effective for improving fetal outcomes. Prenatal education of pregnant women encourages them to adopt preventive measures including not eating insufficiently cooked meat, washing hands after gardening or handling raw meat, and avoiding contact with cats' faeces, either directly or indirectly through soil or contaminated fruit or vegetables. This review assessed the effects of prenatal education for preventing congenital toxoplasmosis. It included two RCTs, both of low methodological quality, one conducted in Canada (432 women) and one in France (5023 women). Both trials had high losses to follow up. The authors of the Canadian trial reported only p values (< 0.05 for all outcomes), not measures of association. They concluded that prenatal education can change women's behaviour as it increased personal, food and pet hygiene. The authors of the French trial concluded that prenatal education significantly improved women's knowledge but had no effect on behaviour. The French trial measured sero-conversion rates and found no difference between the intervention group (13/2591) and the control group (4/1358).The authors concluded that although evidence from observational studies suggests that prenatal education is effective in reducing rates of congenital toxoplasmosis there is little evidence from RCTs to support the practice.

Hackshaw A, Rodeck C, Boniface S. 2011. **Maternal smoking in pregnancy and birth defects: a systematic review based on 173 687 malformed cases and 11.7 million controls**. Human Reproduction Update, 17(5), 589-604. http://humupd.oxfordjournals.org/content/17/5/589

This open-access systematic review reports on a meta-analysis of observational studies published in 1959–2010 (172 publications in total). There were significant positive associations found between maternal smoking and the following birth defects: cardiovascular/heart defects (Odds Ratio 1.09); musculoskeletal defects (OR 1.16); limb reduction defects (OR 1.26); missing/extra digits (OR 1.18); clubfoot (OR 1.28); craniosynostosis (OR 1.33); facial defects (OR 1.19); eye defects (OR 1.25); orofacial clefts (OR 1.28); gastrointestinal defects (OR 1.27); gastroschisis (OR 1.50); anal atresia (OR 1.20); hernia (OR 1.40); and undescended testes (OR 1.13). There was a reduced risk for hypospadias (OR 0.90) and skin defects (OR 0.82). For all defects combined the OR was 1.01 (0.96–1.07). This was non-significant due to including defects with a reduced risk and those with no association (including chromosomal defects). The authors stated that information on the specific defects that are associated with smoking in pregnancy should be included in public health educational information to encourage more women to stop smoking before or early on in pregnancy and that such information should be targeted at the groups who have the highest prevalence of smoking: younger women and those from lower socio-economic groups.

McCarthy FP, Giles ML, Rowlands S, et al. 2011. **Antenatal interventions for preventing the transmission of cytomegalovirus (CMV) from the mother to fetus during pregnancy and adverse outcomes in the congenitally infected infant**. Cochrane Database of Systematic Reviews doi:10.1002/14651858.CD008371.pub2 http://onlinelibrary.wiley.com/doi/10.1002/14651858.CD008371.pub2/abstract

Cytomegalovirus (CMV) is the most common cause of congenital infection in developed countries. The virus is ubiquitous in the general population. Once a person is infected, the virus can remain dormant in the body for life. There is small risk of transmission of the virus from mother to fetus when the mother has acquired the infection before becoming pregnant (0.2 to 2%) but around 40% of women who acquire infection with CMV during pregnancy transmit the infection to their fetus. Congenital infection may result in mental retardation and sensorineural deafness. It is difficult to diagnose infection in pregnant women on the basis of symptoms and signs. Those infected are often asymptomatic (>90% of individuals) and clinical signs, if present, are non-specific. There are laboratory tests for CMV infection but the detection of CMV IgG and IgM antibodies does not reliably distinguish recent infections from those in the distant past. The aim of this review was to assess the benefits and harms of interventions in pregnancy for the prevention of mother to fetus transmission of CMV infection. The authors sought RCTs and quasi RCTs investigating antenatal preventive interventions but none were available. They stated that further research is needed on this topic.

A recent review of CMV prevention issues, intended for obstetricians, can be found in the following open-access article:

Johnson J, Anderson B, Pass RF. 2012. **Prevention of maternal and congenital cytomegalovirus infection**. Clinical Obstetrics & Gynecology, 55(2), 521-30 http://journals.lww.com/clinicalobgyn/toc/2012/06000

Shorter D, Hong T, Osborn DA. 2011. **Screening programmes for developmental dysplasia of the hip in newborn infants**. Cochrane Database of Systematic Reviews: Reviews, DOI: 10.1002/14651858.CD004595.pub2.http://onlinelibrary.wiley.com/doi/10.1002/14651858.CD004595.pub2/abstract

Developmental dysplasia of the hip (DDH), is a condition in which the femoral head and the acetabulum are in improper alignment or grow abnormally, or both. The term DDH is used in preference to the older term of Congenital Dislocation of the Hip since it is recognised that the disorder represents a continuum on which the abnormality ranges from mild to severe. Most cases of DDH resolve untreated in the first two months of life but those that persist beyond 6–8 weeks without being recognised and treated are associated with significant long term problems including gait abnormalities, chronic pain and premature degenerative arthritis. This review assessed the effect of different screening programmes (clinical examination, ultrasound (either universal or targeted), or a combination of both) on the incidence of late presentation of DDH. The usual treatment for DDH diagnosed early, hip abduction splinting, has risks including parental anxiety and pressure sores and, rarely, avascular necrosis of the femoral head and femoral nerve palsy so it is important that infants are not subjected to unnecessary treatment. The reviewers found no study examining the effect of screening and early treatment compared to no screening and later treatment. Five studies were found which fulfilled the review criteria by being RCTs, quasi-RCTs or cluster trials. These compared different types of screening and different timings for screening and splinting of infants with unstable hips or mild hip dysplasia. The authors found that there was insufficient evidence to provide clear recommendations on screening practice. There was inconsistent evidence as to whether or not universal ultrasound results in a significant increase in treatment compared to the use of targeted ultrasound or clinical examination alone. Neither universal nor targeted ultrasound has been shown to improve clinical outcomes, including rates of late diagnosis and surgery. Much larger studies (100,000+ infants) are needed to detect significant clinically important differences given the rarity of late detection and surgery.

Tieu J, Middleton P, Crowther CA. 2010. **Preconception care for diabetic women for improving maternal and infant health**. Cochrane Database of Systematic Reviews doi:10.1002/14651858.CD007776.pub2 http://onlinelibrary.wiley.com/doi/10.1002/14651858.CD007776.pub2/abstract

There is an increased risk of congenital anomalies, still birth and neonatal mortality in babies born to women with pre-existing type 1 or type 2 diabetes. There is an association between adequate pre-conception glycaemic control and a reduced incidence of congenital anomalies therefore clinical guidelines stress the importance of multidisciplinary care and education for pregnant women about the risks diabetes poses to their own and their infant's health and the need to achieve glycaemic targets (as measured by HbA_{1c} levels) before attempting pregnancy. The authors identified only one small RCT (53 women) and this did not report on the review's pre-specified health outcomes. Accordingly they stated that there was little evidence from RCTs regarding the health effects of pre-conception care for women with diabetes.

Stothard KJ, Tennant PW, Bell R, et al. 2009. **Maternal overweight and obesity and the risk of congenital anomalies: a systematic review and meta-analysis**. JAMA, 301(6), 636-50 http://jama.jamanetwork.com/article.aspx?articleid=183375

This systematic review included 39 articles reporting on 29 case-control studies and 12 cohort studies. The results from 18 articles which contained sufficient data to compare overweight or obese mothers (defined by BMI) with mothers with recommended BMI were combined in a meta-analysis. Many articles used the World Health Organization BMI categories but across all the studies ranges recommended BMI ranged from 18.1 to 28.3, overweight from 22.8 to 30 and obesity from less than 26 to greater than 30. Where the number of cases included in the risk and comparison groups together was greater than 150, pooled ORs for overweight and obesity were calculated for 16 and 15 anomaly groups and subtypes, respectively. Compared with mothers of recommended BMI, obese mothers had statistically significant greater odds of pregnancies affected by neural tube defects (Odds ratio 1.87), spina bifida (OR 2.24), cardiovascular anomalies (OR 1.30), septal anomalies (OR 1.20), cleft palate (OR, 1.23), cleft lip and palate (OR, 1.20), anorectal atresia (OR 1.48), hydrocephaly (OR 1.68), and limb reduction anomalies (OR 1.34). The risk of gastroschisis among obese mothers was significantly reduced (OR 0.17). The authors concluded that maternal obesity is associated with an increased risk of structural anomalies but the absolute increase is likely to be small. They stated that further studies are needed to determine whether maternal overweight is also associated with an increased risk of structural anomalies.

Gagnon A, Wilson RD, Allen VM, et al. 2009. **Evaluation of prenatally diagnosed structural congenital anomalies**. J Obstet Gynaecol Can, 31(9), 875-81, 82-9. http://www.sogc.org/guidelines/documents/gui234CO0909.pdf

These concise evidence-based Canadian guidelines are intended for genetic counsellors, midwives, nurses, and physicians who may be involved in care for a pregnant woman whose fetus that has been prenatally diagnosed with isolated or multiple structural congenital anomalies. (Around 1% of prenatal ultrasound examinations reveal a fetal structural anomaly.) The evidence is graded, and the recommendations are classified, using criteria adapted from those of The Canadian Task Force on Preventive Health Care.

Whitworth M, Dowswell T. 2009. **Routine pre-pregnancy health promotion for improving pregnancy outcomes**. Cochrane Database of Systematic Reviews doi:10.1002/14651858.CD007536.pub2 http://onlinelibrary.wiley.com/doi/10.1002/14651858.CD007536.pub2/abstract

There are a number of potentially modifiable risk factors for poor pregnancy outcomes (including congenital anomalies). These include poor nutrition, smoking and drinking excess alcohol. This review assessed the effectiveness of routine pre-pregnancy health promotion (compared to usual care or no pre-pregnancy care) for improving pregnancy outcomes. It included four trials (2300 women) but only one of them involved follow up of pregnancy outcomes. In this study there was no strong evidence of a difference between the intervention and control groups for preterm birth, congenital anomalies or weight for gestational age. There was a statistically significant difference in mean birthweight: mean difference (intervention group – control group) -97.00g, 95% confidence interval -168.05g to -25.95g but this finding was based on outcome data for only half of the randomised women. There was some evidence that health promotion interventions were associated with positive behaviour change including reduced binge drinking (risk ratio 1.24, 95% CI 1.06 to 1.44). The authors concluded that, overall there was little evidence on the effects of pre-pregnancy health promotion on pregnancy outcomes and that more research in this area is needed.

Tekgül S, Riedmiller H, Gerharz E, et al. 2008. **Paediatric Urology**. European Society for Paediatric Urology and the European Association of Urology. http://www.uroweb.org/fileadmin/user_upload/Guidelines/Paediatric%20Urology.pdf

These concise and well-referenced guidelines include guidance on the management of congenital urological anomalies. The authors stated that, where possible statements in the guidelines have been classified in terms of level of evidence and grade of recommendation but that because there have been a limited number of RCTs in this field and a considerable number of the treatment options involve surgical interventions on a wide spectrum of different congenital problems, most of the recommendations are based on consensus.

Walker Godfrey JA. 2001. **Antibiotics for syphilis diagnosed during pregnancy**. Cochrane Database of Systematic Reviews doi:10.1002/14651858.CD001143 http://onlinelibrary.wiley.com/doi/10.1002/14651858.CD001143/abstract

A pregnant woman who is infected with syphilis can pass the infection to her fetus. When a woman acquires the infection close to the time she becomes pregnant or during pregnancy the fetus is often aborted or stillborn. Pregnancies in an untreated woman later in the course of her disease may lead to a baby born with congenital syphilis. These babies may be born prematurely, small for gestational age and with low birthweight. They may have visible signs of the disease but most are asymptomatic at birth, developing signs of infection after a few weeks or months. Without treatment they may develop blindness, deafness, and facial, dental, skeletal and neurological abnormalities later in life. In developed countries pregnant women are screened for syphilis infection and congenital syphilis is rare. The standard treatment for syphilis in adults is long-acting penicillin by injection but there have been concerns raised that the standard treatment regimen may not be optimal, particularly in women who also have HIV. The authors of this review assessed RCTs and quasi RCTs evaluating treatment regimens (in terms of dose, length of treatment course, and mode of administration) for pregnant women with syphilis, with and without concomitant HIV infection. They were unable to identify any studies that met their criteria and they stated that more research is needed on this issue.

Peyron F, Wallon M, Liou C, et al. 1999. **Treatments for toxoplasmosis in pregnancy**. Cochrane Database of Systematic Reviews doi:10.1002/14651858.CD001684
http://onlinelibrary.wiley.com/doi/10.1002/14651858.CD001684/abstract

Congenital infection with toxoplasmosis may lead to an infant being with mental retardation and blindness. The authors of this review assessed whether treating toxoplasmosis infection in pregnancy reduces the risk of congenital toxoplasmosis infection. They did not identify any RCTs addressing treatment for pregnant women who had serological evidence of recent toxoplasma infection so they concluded that it is unknown whether such treatment is effective. They noted that some countries (France and Austria) have introduced toxoplasmosis screening for pregnant women. They stated that since screening is expensive, the benefits of treatment are uncertain and there may be adverse effects from treatment drugs, screening should not be introduced in other countries other than as part of a carefully controlled trial.

International guidelines and systematic reviews concerning the management of congenital anomalies

Guidelines from the National Institute of Health and Care Excellence:

National Institute of Health and Clinical Excellence. 2011. **Thoracoscopic repair of congenital diaphragmatic hernia in neonates (IPG379)**. London: National Institute of Health and Clinical Excellence.
http://publications.nice.org.uk/thoracoscopic-repair-of-congenital-diaphragmatic-hernia-in-neonates-ipg379

National Institute for Health and Clinical Excellence. 2006. **Percutaneous laser therapy for fetal tumours (IPG180)**. London: National Institute for Health and Clinical Excellence. http://publications.nice.org.uk/percutaneous-laser-therapy-for-fetal-tumours-ipg180

National Institute for Health and Clinical Excellence. 2009. **Placement of pectus bar for pectus excavatum (also known as MIRPE or the Nuss procedure) (IPG310)**. London: National Institute for Health and Clinical Excellence. http://publications.nice.org.uk/placement-of-pectus-bar-for-pectus-excavatum-also-known-as-mirpe-or-the-nuss-procedure-ipg310

National Institute for Clinical Excellence. 2004. **Intralesional photocoagulation of subcutaneous congenital vascular disorders (IPG90)**. London: National Institute for Clinical Excellence.
http://publications.nice.org.uk/intralesional-photocoagulation-of-subcutaneous-congenital-vascular-disorders-ipg90/the-procedure

National Institute for Health and Clinical Excellence. 2006. **Foker technique for long-gap oesophageal atresia (IPG153)**. London: National Institute for Health and Clinical Excellence. http://publications.nice.org.uk/foker-technique-for-long-gap-oesophageal-atresia-ipg153

National Institute for Health and Clinical Excellence. 2007. **Thoracoscopic aortopexy for severe primary tracheomalacia (IPG243)**. London: National Institute for Health and Clinical Excellence.
http://publications.nice.org.uk/thoracoscopic-aortopexy-for-severe-primary-tracheomalacia-ipg243/the-procedure

Cochrane Reviews:

Cedin AC, Atallah ÁN, Andriolo Régis B, et al. 2012. **Surgery for congenital choanal atresia**. Cochrane Database of Systematic Reviews doi:10.1002/14651858.CD008993.pub2
http://onlinelibrary.wiley.com/doi/10.1002/14651858.CD008993.pub2/abstract

Gray K, Pacey V, Gibbons P, et al. 2012. **Interventions for congenital talipes equinovarus (clubfoot)**. Cochrane Database of Systematic Reviews doi:10.1002/14651858.CD008602.pub2
http://onlinelibrary.wiley.com/doi/10.1002/14651858.CD008602.pub2/abstract

Pádua LMS, Garcia LC, Rubira CJ, et al. 2012. **Stent placement versus surgery for coarctation of the thoracic aorta**. Cochrane Database of Systematic Reviews doi:10.1002/14651858.CD008204.pub2
http://onlinelibrary.wiley.com/doi/10.1002/14651858.CD008204.pub2/abstract

Wagner Jennifer VE, Moe-Byrne T, Grover Z, et al. 2012. **Glutamine supplementation for young infants with severe gastrointestinal disease**. Cochrane Database of Systematic Reviews doi:10.1002/14651858.CD005947.pub3
http://onlinelibrary.wiley.com/doi/10.1002/14651858.CD005947.pub3/abstract

Leonardi-Bee J, Batta K, O'Brien C, et al. 2011. **Interventions for infantile haemangiomas (strawberry birthmarks) of the skin**. Cochrane Database of Systematic Reviews doi:10.1002/14651858.CD006545.pub2
http://onlinelibrary.wiley.com/doi/10.1002/14651858.CD006545.pub2/abstract

Das A, Shah Prakeshkumar S. 2010. **Octreotide for the treatment of chylothorax in neonates**. Cochrane Database of Systematic Reviews doi:10.1002/14651858.CD006388.pub2
http://onlinelibrary.wiley.com/doi/10.1002/14651858.CD006388.pub2/abstract

Frobel A-K, Hulpke-Wette M, Schmidt Klaus G, et al. 2009. **Beta-blockers for congestive heart failure in children**. Cochrane Database of Systematic Reviews doi:10.1002/14651858.CD007037.pub2
http://onlinelibrary.wiley.com/doi/10.1002/14651858.CD007037.pub2/abstract

Lee ASY, Law J, Gibbon FE. 2009. **Electropalatography for articulation disorders associated with cleft palate**. Cochrane Database of Systematic Reviews doi:10.1002/14651858.CD006854.pub2

http://onlinelibrary.wiley.com/doi/10.1002/14651858.CD006854.pub2/abstract

Nasser M, Fedorowicz Z, Newton T, et al. 2008. **Interventions for the management of submucous cleft palate**. Cochrane Database of Systematic Reviews doi:10.1002/14651858.CD006703.pub2 http://onlinelibrary.wiley.com/doi/10.1002/14651858.CD006703.pub2/abstract

Long V, Chen S, Hatt SR. 2006. **Surgical interventions for bilateral congenital cataract**. Cochrane Database of Systematic Reviews doi:10.1002/14651858.CD003171.pub2 http://onlinelibrary.wiley.com/doi/10.1002/14651858.CD003171.pub2/abstract

Smeulders MJC, Coester A, Kreulen M. 2005. **Surgical treatment for the thumb-in-palm deformity in patients with cerebral palsy**. Cochrane Database of Systematic Reviews doi:10.1002/14651858.CD004093.pub2 http://onlinelibrary.wiley.com/doi/10.1002/14651858.CD004093.pub2/abstract

Davies Mark W, Kimble Roy M, Woodgate Paul G. 2002. **Ward reduction without general anaesthesia versus reduction and repair under general anaesthesia for gastroschisis in newborn infants**. Cochrane Database of Systematic Reviews doi:10.1002/14651858.CD003671 http://onlinelibrary.wiley.com/doi/10.1002/14651858.CD003671/abstract

Moyer VA, Moya FR, Tibboel D, et al. 2000. **Late versus early surgical correction for congenital diaphragmatic hernia in newborn infants**. Cochrane Database of Systematic Reviews doi:10.1002/14651858.CD001695 http://onlinelibrary.wiley.com/doi/10.1002/14651858.CD001695/abstract

Other Relevant Publications and Websites

Medsafe. http://medsafe.govt.nz/ accessed April 2013.

There are a number of commonly prescribed drugs that have been associated with increased risks of congenital anomalies. The following webpages on the Medsafe website provide information on this topic:

The use of antidepressants in pregnancy. (2012)
http://medsafe.govt.nz/profs/puarticles/theuseofantidepressantssept10.htm

Anticonvulsants and congenital malformations (2009)
http://medsafe.govt.nz/profs/puarticles/anticonvulsants-feb09.htm

SSRI Use in Pregnancy - Collaborative Decision-Making is Key (2008)
http://medsafe.govt.nz/profs/puarticles/ssripreg.htm

Isotretinoin - indications and teratogenicity (2009) http://medsafe.govt.nz/profs/puarticles/isotretinoin-may09.htm

Avoiding Teratogenicity with Isotretinoin (2002) http://medsafe.govt.nz/profs/puarticles/teratogen.htm

ACE Inhibitors in Early Pregnancy (1998) http://medsafe.govt.nz/profs/puarticles/aceinhibitors.htm

Centre for Public Health Research, Massey University. 2013. **New Zealand Birth Defects Registry**. http://nzbdr.ac.nz/ accessed April 2013.

The New Zealand Birth Defects Registry runs a birth defects monitoring programme under contract to the Ministry of Health. The website contains data on birth defects among live births (born or treated) at public hospitals 2000–2009: http://nzbdr.ac.nz/assets/FILES/Final%20published%20table%202000-2009.pdf . It also has links to other publications associated with the NZBDR and international birth defects related websites.

EUROCAT (European surveillance of congenital anomalies). 2012. **Primary Prevention**. http://www.eurocat-network.eu/preventionandriskfactors/primaryprevention accessed May 2013.

This webpage has links to the EUROCAT/EUROPLAN recommendations on policies to be considered for the primary prevention of congenital anomalies in National Plans and Strategies on Rare Diseases. It also contains links to publications relating to risk factors, EUROCAT's review of environmental risk factors for congenital anomalies, and systematic reviews relating to congenital anomalies.

National Collaborating Centre for Women's and Children's Health (Commissioned by the National Institute for Health and Clinical Excellence). 2011. **Hypertension in pregnancy (NICE Clinical Guideline CG107)**. London: Royal College of Obstetricians and Gynaecologists. http://www.nice.org.uk/guidance/CG107

(full guideline: http://www.nice.org.uk/nicemedia/live/13098/50475/50475.pdf)

These guidelines state that women who have chronic hypertension and who may become pregnant should be advised that taking angiotensin-converting enzyme (ACE) inhibitors or angiotensin II receptor blockers (ARBs) during pregnancy is associated with an increased risk of congenital abnormalities and that if they are planning to become pregnant they should discuss alternative anti-hypertensive medication with the healthcare professional responsible for managing their hypertension. They should also be advised that there may be an increased risk of congenital abnormalities associated with chlorothiazide. Treatment with any of these drugs should be stopped promptly and alternatives offered if a woman taking them becomes aware that she is pregnant.

Kyle PM, Coghlan P, Matthews J, et al. 2009. **Accuracy of prenatal diagnosis in a tertiary fetal medicine unit**. New Zealand Medical Journal, 122(1288), 50-61. http://journal.nzma.org.nz/journal/122-1288/3441/

This study reports on the accuracy of prenatal diagnosis in the fetal medicine unit in Christchurch. It involved a review 681 cases seen over an 18 month period from 1 June 2004 to 30 November 2005 in which prenatal diagnoses made via ultrasound were compared those made postnatally (or at post-mortem in cases where there was a termination or an intrauterine or postnatal death. For the live born babies 93.6% had their prenatal diagnosis confirmed, 5.1% had an issue which had resolved by the time of birth, and 1.3% had an additional major abnormality that had a significant clinical effect. Fifty two percent of the fetal or neonatal deaths with a normal karyotype were followed by a post-mortem examination and there was only one new finding that changed the prenatal diagnosis significantly. The authors stated that parents and staff need to be informed that, although not all abnormalities will be detected prenatally, inaccurate prenatal diagnosis is rare.

International Clearinghouse for Birth Defects Surveillance and Research. 2010. **Annual report 2010 with data for 2008**. Rome: The International Centre on Birth Defects - ICBDSR Centre. http://www.icbdsr.org/filebank/documents/ar2005/Report2010.pdf

This publication reports on two collaborative research projects involving data from several countries: one on multiple congenital anomalies and one on prenatal diagnosis and Down syndrome. It provides surveillance data on 39 birth defects that are monitored by 44 member programmes around the world. It includes data from the New Zealand Birth Defects Registry.

Note: The publications listed were identified using the search methodology outlined in Appendix 1 (Search Methods for Policy Documents and Evidence-based Reviews)

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7. Riley M, Halliday J. 2008. Birth Defects in Victoria 2005–2006. Melbourne: Victorian Perinatal Data Collection Unit, Victorian Government Department of Human Services http://docs.health.vic.gov.au/docs/doc/B22440E8FCBCBA25CA257A060020B967/$FILE/bd_05-06.pdf
8. Rynn L, Cragan J, Correa A, et al. 2008. Update on overall prevalence of major birth defects — Atlanta, Georgia, 1978–2005. *MMWR* 57(01) 1–5.
9. EUROCAT (European surveillance of congenital anomalies). 2012. Special Report: Congenital anomalies are a major group of mainly rare diseases. Newtownabbey, Co Antrim, Northern Ireland: EUROCAT Central Registry, University of Ulster http://www.eurocat-network.eu/content/Special-Report-Major-Group-of-Mainly-Rare-Diseases.pdf