CONGENITAL HEART DISEASE

Introduction

Congenital heart disease (CHD) is the most common congenital disorder in newborns [1]. Due to definitional issues, there are large variations in prevalence estimates. In 2002, Hoffman et al. [2] estimated the birth prevalence of severe CHD (e.g. transposition of the great arteries, Tetralogy of Fallot) requiring expert cardiological care to be 2.5–3.0 per 1,000 live births, with moderately severe forms of CHD (e.g. large atrial septal defects, complex forms of ventricular septal defects) accounting for another 3 per 1,000 live births. The overall prevalence increased to 75 per 1,000 if minor anomalies (e.g. small ventricular septal defects, atrial septal defects, or patent ductus arteriosus) were included. Subsequent reviews, however, have yielded much lower prevalence estimates, ranging from 3–6 per 1,000 live births [1].

The aetiology of CHD remains largely unknown. Only around 15% of cases can be traced to a known cause. These causes include chromosomal anomalies, such as Down syndrome and trisomies 13 and 18, which account for around 8–10% of cases and single gene defects, which account for a further 3–5%. The causes of non-syndromal CHD are less clear, with only around 2% of cases being attributable to known environmental factors such as maternal diabetes, obesity, alcohol use, rubella, febrile illnesses and use of certain drugs [1]. Genetic factors may play a role, as the risk of recurrence is 1–6% if one sibling is affected, and up to 10% if two siblings are affected [1].

While prevention may need to await a better understanding of causal pathways, for affective treatment early detection and timely management are crucial. In this context, research has suggested that up to 25% of babies with severe forms of congenital heart disease may be discharged from hospital undiagnosed. Recent studies have suggested that pulse oximetry, if used in conjunction with a clinical examination prior to discharge, may improve the detection rate of some forms of CHD [3]. However, it is likely that a number of babies with serious cardiovascular anomalies will still be missed in the neonatal period and so antenatal screening has become established practice in many centres [3].

In tertiary centres dealing with the diagnosis and management of fetal cardiac anomalies a high degree of diagnostic accuracy is possible, and most (but not all) major forms of CHD can be detected antenatally [3]. Most cases of congenital heart disease, however, occur in low risk groups and will only be detected antenatally if a detailed examination of the fetal heart (using a four chamber view of the fetal heart) is included as part of routine obstetric ultrasound screening. In such cases detection rates are likely to depend on the level of sonographer training and experience, the adequacy of the equipment available, and the time allowed for sonographers to undertake routine examinations [3]. Investing in such resources is important, as early detection provides an opportunity to exclude associated extra-cardiac and chromosomal abnormalities, discuss pregnancy options, prepare parents, and plan for delivery in a tertiary centre [3].

The following section uses data from the National Minimum Dataset to review the number of babies with cardiovascular anomalies evident at the time of birth.

Data Source and Methods

Definition

1. Number of cardiovascular anomalies evident at birth (by anomaly type)

2. Number of babies with one or more cardiovascular anomalies evident at birth (by anomaly type)

Data Source

1. National Minimum Dataset

Numerator: Hospital Admissions with event type = birth and a cardiovascular anomaly (ICD-10 Q20-Q28) listed in any of the first 15 diagnoses.

Denominator: All hospital admissions with event type = birth

Notes on Interpretation

Note 1: The analysis includes all admissions in the National Minimum Dataset where the Event Type was listed as a Birth (Note: Only one birth event is allowed per NHI number, with babies being born prior to hospital admission, or being readmitted shortly after discharge being listed as a routine inpatient event). Thus the analysis does not include babies born prior to hospital admission, babies born at home, or babies whose cardiovascular anomaly was overlooked at the time of initial hospital discharge, but who re-presented shortly thereafter. Further, the methodology used may significantly undercount those cardiovascular anomalies which are difficult to detect on routine newborn examination.

Note 2: In the analysis which follows, 64.2% of patent ductus arteriosus (PDA) identified during 2008–2012 were in preterm babies (<37 weeks gestation) who had no other cardiovascular anomalies. Prematurity is known to increase the risk of PDA as a result of increased exposure to hypoxia and underdeveloped heart and lungs. During 2008–12, 23.8% of all cardiovascular anomalies were isolated PDAs in preterm infants, many of whom would not have had a PDA had they been born at term. As the possibility that any analysis of risk factors for cardiovascular anomalies may be inadvertently distorted by the risk factor profile of those babies being born prematurely, preterm (<37 weeks) babies with isolated PDAs (i.e. a PDA with no other cardiovascular anomaly) have been excluded from rate calculations after the initial overview table.

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

New Zealand Distribution and Trends

Cardiovascular Anomalies Evident at Birth

In New Zealand during 2008–2012, patent ductus arteriosus (PDA) was the most frequent cardiovascular anomaly identified at birth, with 64.2% of PDAs being in preterm babies (<37 weeks gestation) who had no other cardiovascular anomalies (see Methods section for rationale for exclusion of these cases from subsequent analyses). Atrial septal and ventricular septal defects were the next most frequent causes of cardiovascular anomalies, followed by other malformations of the great arteries (**Table 1**).

New Zealand Trends

In New Zealand during the mid-2000s the number of babies born with one or more cardiovascular anomalies was relatively stable. Rates declined between 2007 and 2009 and then remained static until 2012, when a small upswing was evident (**Figure 1**). It remains unclear however, whether the decline in rates during 2007–2009, and the subsequent rebound, reflects real changes in the number of babies born with cardiovascular anomalies, changes in the thoroughness of the recording of cardiovascular anomalies by clinicians, or changes in the way these anomalies were coded in the hospital admission dataset.

Distribution by Maternal Age

While in numerical terms, the largest number of babies born with cardiovascular anomalies during 2008–2012 had mothers who were aged 30–34 years, the risk of cardiovascular anomalies rose progressively with increasing maternal age. Thus babies whose mothers were aged 40+ years had cardiovascular anomaly rates which were 1.81 (95% CI 1.42–2.32) times higher than those whose mothers gave birth in their teens (**Figure 2**).

Distribution by Prioritised Ethnicity, NZDep Index Decile and Gender

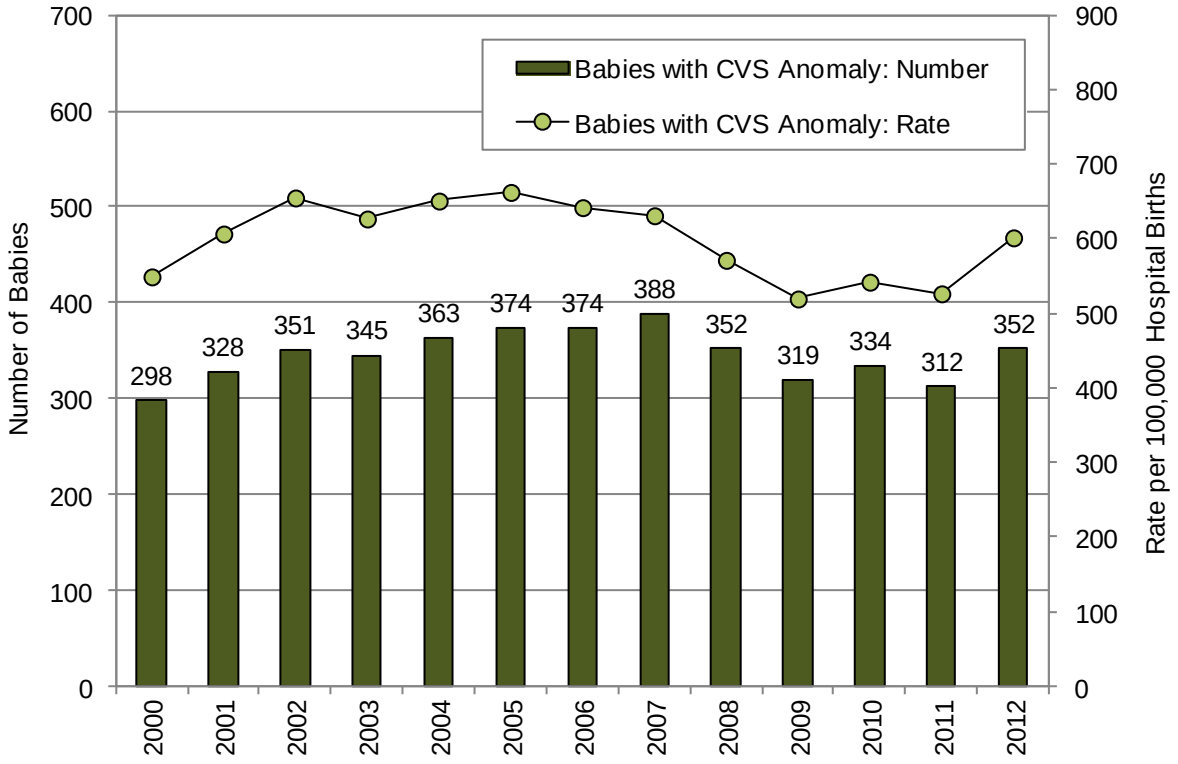
In New Zealand during 2008–2012, there were no *significant* ethnic differences in the proportion of babies born with cardiovascular anomalies. Rates were *significantly* higher however, for babies from the least deprived (NZDep06 deciles 1–2 vs. deciles 5–10) areas, for males, and for those with older (40+ years vs. <20 years) mothers (**Table 2**). While cardiovascular anomaly rates were higher for European babies than for Māori babies during the early 2000s, rates became similar from 2010–11 onwards. Differences between European, Pacific and Asian/Indian babies however, were less consistent during 2000–2012 (**Figure 3**).

Table 1. Cardiovascular Anomalies Evident at Birth, New Zealand Hospital Births 2008–2012

Cardiovascular Anomaly	Number: Total 2008−2012	Number: Annual Average	Anomalies per 100,000 Births*
New Zealand			
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

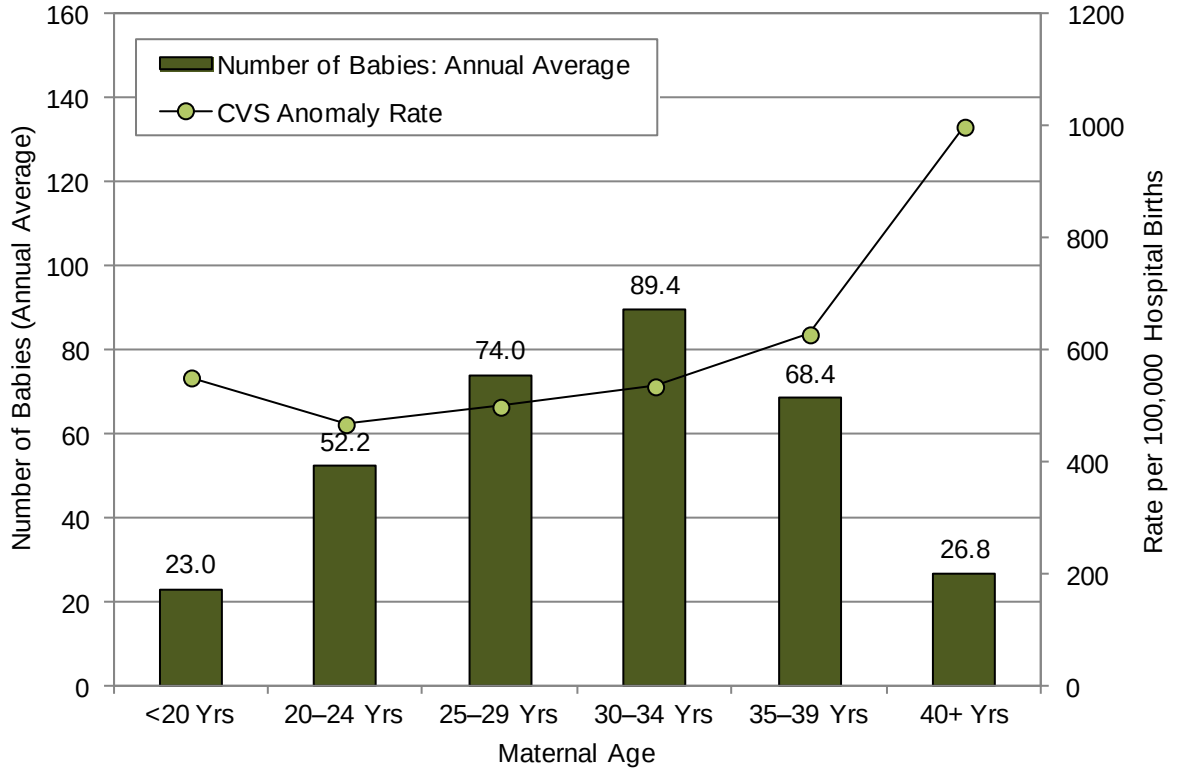
Source: National Minimum Dataset: Numerator: Hospital Admissions with event type = birth and a cardiovascular anomaly in any of the first 15 diagnoses; Denominator: All hospital admissions with event type = birth; Note: *Anomalies per 100,000 births refers to total number of anomalies rather than number of babies, as some babies have more than one anomaly; [+]Patent Ductus Arteriosus includes 957 cases of Isolated PDA in preterm infants (<37 weeks), which have been excluded in subsequent analyses

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



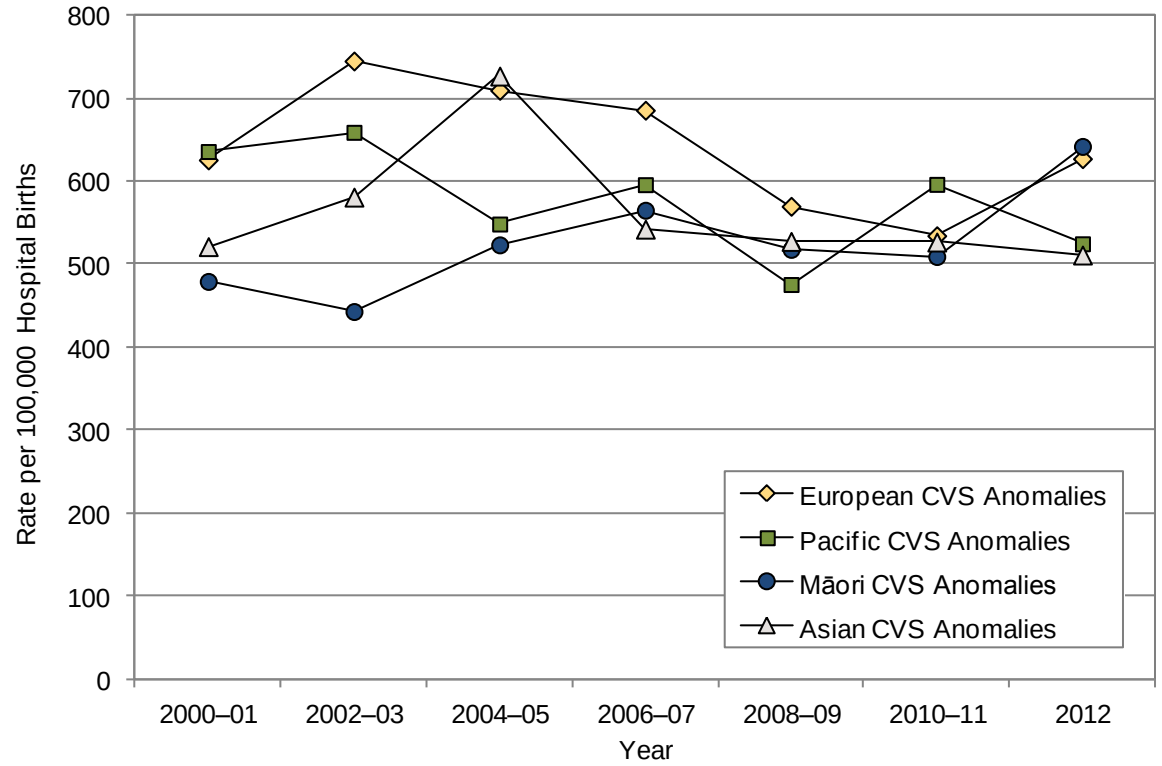
Source: National Minimum Dataset: Numerator: Hospital Admissions with event type = birth and a cardiovascular anomaly in any of the first 15 diagnoses; Denominator: All hospital admissions with event type = birth; Note: Rate per 100,000 refers to number of babies with one or more CVS anomalies, rather than the number of anomalies; Preterm (<37 weeks) babies with Isolated Patent Ductus Arteriosus have been excluded

Figure 2. Babies with Cardiovascular 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 Cardiovascular 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 CVS anomalies, rather than the number of anomalies; Preterm (<37 weeks) babies with Isolated Patent Ductus Arteriosus excluded

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



Source: National Minimum Dataset: Numerator: Hospital Admissions with event type = birth and a cardiovascular anomaly in any of the 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 CVS anomalies, rather than the number of anomalies; Preterm (<37 weeks) babies with Isolated Patent Ductus Arteriosus excluded

Table 2. Distribution of Babies with Cardiovascular Anomalies Evident at Birth by Prioritised 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
Any Cardiovascular Anomaly				
Prioritised Ethnicity				
Māori	71.8	538.2	0.95	0.84–1.07
Pacific	36.2	533.9	0.94	0.80–1.10
European/Other	187.8	566.4	1.00	
Asian/Indian	35.8	523.2	0.92	0.79–1.08
NZ Deprivation Index				
Deciles 1–2	55.6	655.3	1.00	
Deciles 3–4	53.4	592.1	0.90	0.76–1.07
Deciles 5–6	60.4	536.2	0.82	0.70–0.96
Deciles 7–8	75.8	537.4	0.82	0.70–0.96
Deciles 9–10	88.4	504.8	0.77	0.66–0.89
Gender				
Female	151.6	515.7	1.00	
Male	182.2	586.5	1.14	1.03–1.25
Maternal Age				
<20 Years	23.0	551.0	1.00	
20–24 Years	52.2	467.4	0.85	0.68–1.06
25–29 Years	74.0	498.6	0.91	0.73–1.12
30–34 Years	89.4	535.1	0.97	0.79–1.19
35–39 Years	68.4	628.0	1.14	0.92–1.41
40+ Years	26.8	999.3	1.81	1.42–2.32

Source: National Minimum Dataset: Numerator: Hospital Admissions with event type = birth and a cardiovascular anomaly in any of the 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 CVS anomalies, rather than the number of anomalies; Preterm (<37 weeks) babies with Isolated Patent Ductus Arteriosus excluded; Decile is NZDep06

South Island DHBs Distribution and Trends

In the South Island DHBs during 2008–2012, patent ductus arteriosus was the most frequent cardiovascular anomaly identified at birth, although a large proportion (range 45.5%–80.0%) were in preterm babies with no other cardiovascular anomalies. Ventricular and atrial septal defects were the next most frequent anomalies identified (**Table 3**, **Table 4**).

When the number of babies with one or more CVS anomalies (rather than the number of CVS anomalies) was considered, on average 7.6 Nelson Marlborough, 1.6 South Canterbury, 38.2 Canterbury, 2.6 West Coast, 20.0 Otago and 10.6 Southland babies each year (excluding isolated preterm PDAs) were born with one or more CVS anomalies, with rates in Nelson Marlborough, Canterbury, the West Coast and Southland not being *significantly* different from the New Zealand rate. In contrast, rates in South Canterbury were *significantly* lower than the New Zealand rate, while in Otago rates were *significantly* higher (**Table 5**) (**Figure 4**).

Table 3. Cardiovascular Anomalies Evident at Birth, Nelson Marlborough, South Canterbury, Canterbury and the West Coast Hospital Births 2008–2012

Cardiovascular Anomaly	Number: Total 2008−2012	Number: Annual Average	Number of Anomalies per 100,000 Births*
Nelson Marlborough			
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
South Canterbury			
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
Canterbury			
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
West Coast			
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

Source: National Minimum Dataset: Numerator: Hospital Admissions with event type = birth and a cardiovascular anomaly in any of the first 15 diagnoses; Denominator: All hospital admissions with event type = birth; Note: *Anomalies per 100,000 refers to total number of anomalies rather than number of babies, as some babies have more than one anomaly; [+]Patent Ductus Arteriosus includes 29 Nelson Marlborough, 8 South Canterbury, 126 Canterbury and 5 West Coast cases of isolated PDA in preterm (<37 weeks) infants, which have been excluded from subsequent analyses; s: suppressed due to small numbers

Table 4. Cardiovascular Anomalies Evident at Birth, Southern DHB Hospital Births 2008–2012

Cardiovascular Anomaly	Number: Total 2008−2012	Number: Annual Average	Number of Anomalies per 100,000 Births*
Otago			
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
Southland			
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

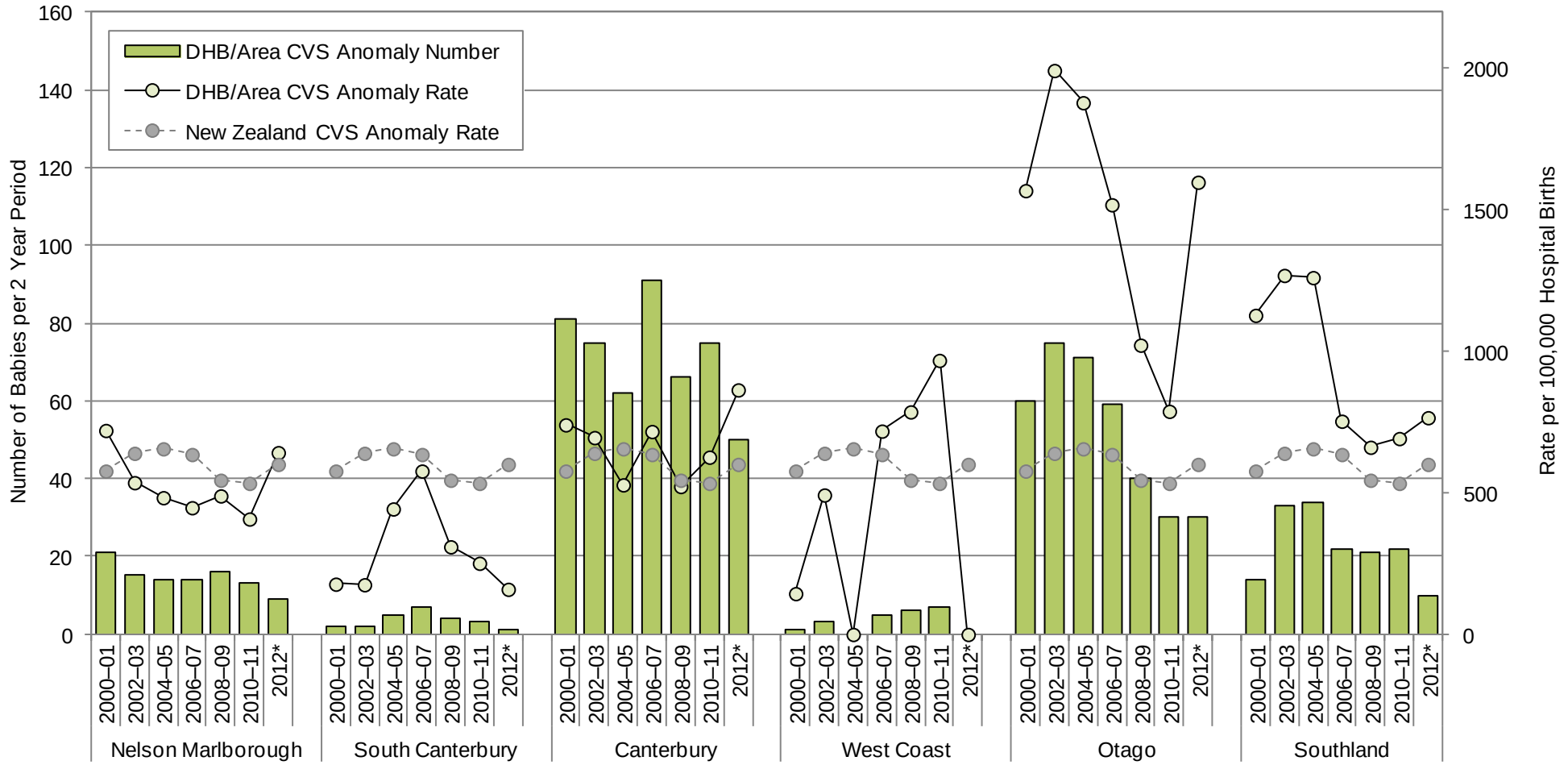
Source: National Minimum Dataset: Numerator: Hospital Admissions with event type = birth and a CVS anomaly in any of the first 15 diagnoses; Denominator: All hospital admissions with event type = birth; Note: *Anomalies per 100,000 refers to total number of anomalies rather than number of babies, as some babies have >1 anomaly; [+]PDA includes 45 Otago and 19 Southland cases of isolated PDA in preterm (<37 weeks) infants, which have been excluded from subsequent analyses; s: suppressed due to small numbers

Table 5. Number of Babies with One or More Cardiovascular 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
Cardiovascular Anomalies					
Nelson Marlborough	38	7.6	484.3	0.88	0.64–1.21
South Canterbury	8	1.6	257.5	0.47	0.23–0.93
Canterbury	191	38.2	629.5	1.14	0.98–1.32
West Coast	13	2.6	706.5	1.28	0.74–2.20
Otago	100	20.0	1,043.5	1.89	1.55–2.31
Southland	53	10.6	692.9	1.26	0.96–1.65
New Zealand	1,669	333.8	552.1	1.00	

Source: National Minimum Dataset: Numerator: Hospital Admissions with event type = birth and a CVS anomaly in any of the first 15 diagnoses; Denominator: All hospital admissions with event type = birth; Note: Rate per 100,000 refers to number of babies with 1+ CVS anomalies, rather than the number of anomalies; Preterm (<37 weeks) babies with Isolated PDA excluded; Rate Ratios are compared to New Zealand rates

Figure 4. Babies with Cardiovascular 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 cardiovascular anomaly in any of the 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 CVS anomalies, rather than the number of anomalies; Preterm (<37 weeks) babies with Isolated Patent Ductus Arteriosus excluded; *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 Heart Disease

In New Zealand there is a paucity of publications relevant to the prevention or management of congenital heart disease. **Table 6** summarises a number 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 32 (Local Policy Documents and Evidence-Based Reviews Relevant to Congenital Anomalies) on Page 105 considers congenital anomalies collectively and Table 44 (Local Policy Documents and Evidence-Based Reviews Relevant to Down Syndrome and Other Chromosomal Anomalies) on Page 128 considers Down syndrome (which is of relevance as a high proportion of babies with Down syndrome also have congenital heart disease).

Table 6. Policy Documents and Evidence-Based Reviews Relevant to Cardiovascular Anomalies

International Guidelines and Systematic and Other Reviews
Ewer AK, Furmston AT, Middleton LJ, et al. 2012. **Pulse oximetry as a screening test for congenital heart defects in newborn infants: a test accuracy study with evaluation of acceptability and cost-effectiveness**. Health Technol Assess 2012*,* 16(2).http://www.hta.ac.uk/fullmono/mon1602.pdf In 2007, the National Institute of Health Research Heath Technology Assessment Programme in the U.K. commissioned a project to evaluate pulse oximetry as a screening procedure to detect major congenital heart defects (CHDs) in newborn infants. (Major CHDs are defined as those resulting in death or requiring invasive intervention in infancy and they are subdivided into critical defects which usually present in the first few days or weeks of life and serious CHDs which tend to present after 28 days of life.) The project comprised three distinct pieces of work which assessed the accuracy, acceptability (to parents and healthcare staff) and cost-effectiveness of pulse oximetry testing compared to existing strategies for detecting CHDs (antenatal ultrasound and newborn clinical examination). The test accuracy study included 20,055 newborns. Sensitivity of pulse oximetry was found to be 75.0% for critical cases and 49.1% for critical plus serious cases combined. The false positive rate was 0.84 % giving a specificity of 99.16%. Of those babies with a false positive test result, 3.5% had a significant but non-major heart defect and 24% had a respiratory or infective illness requiring hospital treatment. Overall, 37% of babies who had a positive test result had a condition requiring medical intervention. Parents and staff were predominantly satisfied with pulse oximetry screening and satisfaction was predicted by greater understanding of heart disease and the possibilities for treatment, and lower anxiety, stress and depression. The average time taken for pulse oximetry testing was 6.9 minutes (median 5 minutes, range 1 to 30 minutes) and each test was estimated to cost £6.24. Pulse oximetry plus clinical examination was twice as costly as clinical examination alone. The project concluded that pulse oximetry is a feasible, safe, simple, non-invasive and reasonably accurate test that is more sensitive than antenatal ultrasound screening and clinical examination and is likely to identify cases of critical CHD that would otherwise have been missed at an estimated cost of £24,000 per additional case. Pulse oximetry, like the other screening methods, misses some cases. Most of these are associated with obstruction of the aortic arch which may not produce hypoxaemia.
Thangaratinam S, Brown K, Zamora J, et al. 2012. **Pulse oximetry screening for critical congenital heart defects in asymptomatic newborn babies: a systematic review and meta-analysis**. Lancet*,* 379(2459–64). This systematic review included data from 13 studies (229,421 babies) assessing the accuracy of pulse oximetry for the detection of critical congenital heart defects (CHDs) in newborn babies. The reviewers calculated sensitivity and specificity (with associated 95% CIs) for the individual studies and used a hierarchical summary receiver operating characteristics model to obtain summary estimates of sensitivity and specificity and also to assess whether the variability in test accuracy between studies was related to timing of test (< 24 hours vs. ≥ 24 hours after birth), method of testing (right hand and foot vs. foot only), oxygen saturation (functional vs. fractional) or antenatal screening (previously identified cases excluded vs. included). Overall sensitivity for the detection of critical CHDs was found to be 76.5% (95% CI 67.7–83.5). The specificity was 99.9% (95% CI 99.7–99.9) and the false positive rate 0.14% (95% CI 0.06%–0.33%). It was especially noteworthy that the false positive rate was significantly lower when pulse oximetry was done more than 24 hours after birth than less than 24 hours (0·05% [95% CI 0·02–0·12] *vs.* 0·50 [95% CI 0·29–0·86]; p=0·0017). The reviewers concluded that pulse oximetry meets criteria for universal screening by being highly specific and moderately sensitive. This review was reviewed by the Centre for Reviews and Dissemination (CRD) which stated that it was generally a well-conducted review and that the authors' conclusions were likely to be reliable. The CRD review is available at: http://www.crd.york.ac.uk/CRDWeb/ShowRecord.asp?ID=12012020804

Papatheodorou SI, Evangelou E, Makrydimas G, et al. 2011. **First-trimester ductus venosus screening for cardiac defects: a meta-analysis**. BJOG. An International Journal of Obstetrics and Gynaecology, 118(12), 1438-45.

Abnormal flow in the ductus venosus (DV) can be detected via Doppler ultrasound in the first trimester of pregnancy, when nuchal translucency (NT) is measured, and it has been associated with adverse perinatal outcomes, chromosomal abnormalities and congenital heart disease (CHD). This review assessed the diagnostic performance of DV waveform examination as a screening technique for CHD in chromosomally normal fetuses with normal and also abnormal NT status. The review included seven studies assessing DV performance regardless of NT status (50,354 fetuses), nine where there was increased NT (n= 2908) and seven (n= 47,610) where NT was normal. Overall the summary sensitivity of DV for CHD detection was 50% (0.50, 95% CI 0.27–0.73) and the specificity 93% (95% CI 0.88–0.96). In participants with increased NT the sensitivity and specificity were 83% and 80% respectively and in those with normal NT they were 19% and 96%, respectively.

Chelemen T, Syngelaki A, Maiz N, et al. 2011. **Contribution of Ductus Venosus Doppler in First-Trimester Screening for Major Cardiac Defects**. Fetal Diagnosis and Therapy, 29(2), 127–34.

This study from the U.K. was by far the largest of those included in the above meta-analysis. The aim of the study was to determine whether assessment of ductus venosus (DV) flow at 11–13 weeks could be used to improve the rate of detection of cardiac defects beyond that achieved by nuchal translucency (NT) measurements. The study population of chromosomally normal fetuses included 40,905 fetuses without heart defects and 85 with major heart defects. Of the fetuses with major heart defects, 35.3% had a NT above the 95th percentile and 21.2% above the 99th. By comparison, of the fetuses without major heart defects, 4.8% had a NT above the 95th percentile and 0.7% above the 99th. Reversed A wave on DV Doppler was seen in 28.2% of the fetuses with cardiac defects and 2.1% of those without. The authors state that providing specialist echocardiography to those with NT above the 99th percentile and those with reversed A wave on DV Doppler, regardless of NT value, would detect 38.8% of major cardiac defects at an overall false positive rate of 2.7%.They concluded that assessment of DV flow improves the performance of NT screening for cardiac defects by about 10% and they suggest that, ideally, fetal echocardiography should be offered to all those with reversed A wave and those with NT above the 95th percentile. Where there are insufficient resources to achieve this, specialist examination could be limited to those with NT above the 99th percentile and those with abnormal DV Doppler.

National Collaborating Centre for Women's and Children's Health. **Antenatal care: Routine care for the healthy pregnant woman**. London: RCOG Press, 2008. URL: http://www.nice.org.uk/nicemedia/live/11947/40145/40145.pdf

Section 9.1 of this publication covers screening for structural anomalies and reviews studies relating to the use of ultrasound and serum screening (alpha-fetoprotein, AFP) for this purpose. One systematic review (of 5 studies) and two other studies looked at foetal echocardiography. There was a wide range of reported values for sensitivity by centre and condition but the reported specificity was generally high. There was some evidence from 2 uncontrolled observational studies that babies with transposition of the great arteries (and possibly hypoplastic left heart syndrome) diagnosed prenatally had reduced mortality compared with those diagnosed postnatally. Nuchal translucency measurement seemed to have poor diagnostic value for detecting cardiac anomalies.

A short version of this guideline can be found at: http://www.nice.org.uk/nicemedia/live/11947/40115/40115.pdf

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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3. Sharland G. 2010. Fetal Cardiac Screening: Why Bother? *Archives of Disease in Childhood Fetal & Neonatal Edition* 95 F64-F68.