Congenital heart defects in Central Australia

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ABSTRACT

Objective: To determine the incidence of congenital heart defects (CHD) in Aboriginal and non-Aboriginal infants in Central Australia and to compare this with the incidence elsewhere in Australia.

Design and setting: Data on cases were obtained from patient records of the Alice Springs Hospital, Central Australia, the sole referral centre for paediatric and initial cardiac diagnostic services for the region.

Participants: Patients with CHD proven by echocardiography reported between 1 January 1993 and 30 June 2000.

Main outcome measures: Incidence of CHD using all live births in Central Australia as the denominator.

Results: 108 patients with CHD were detected among 6156 live births (incidence, 17.5 per 1000; 95% CI, 14.9–21.7 per 1000); 57 of 2991 were Aboriginal (19.0 per 1000; 95% CI, 14.4–24.6 per 1000) and 51 of 3165 were non-Aboriginal (16.1 per 1000; 95% CI, 12.0–21.1 per 1000). The difference between the two groups was not statistically significant (relative risk, 1.18; 95% CI, 0.81–1.72). CHD incidence in Central Australia was significantly higher than that reported for other parts of Australia (4.3 per 1000 live births in New South Wales and the Australian Capital Territory, 1981–1984; 7.65 and 12 per 1000 total births in Western Australia, 1980–1989, and South Australia, 1993–2000, respectively).

Conclusions: The high rates of CHD in Central Australia may partly reflect the high utilisation of echocardiography for assessing minor lesions. However, the incidence of both major and minor types of CHD was significantly higher than previously reported from other regions of Australia. The role of socioenvironmental factors in this high incidence should be explored.

MJA 2004; 180: 614–617

CONGENITAL HEART DEFECTS (CHD) are among the more common major malformations at birth.[1] Knowledge of the epidemiology of CHD is important in determining cause, allocating appropriate resources and planning effective prevention and management. In the past two decades, advances in the non-invasive diagnosis of CHD using echocardiography and colour Doppler ultrasonography may have improved case ascertainment.

The inland region of Central Australia encompasses over 1 million square kilometres but is sparsely populated, with a proportionately large Aboriginal population. Local experience suggests that the incidence of CHD in Central Australia is much higher than previously reported for Australia.[2-4] This study aimed to determine the incidence of CHD in infants of the two major population groups in the region (Aboriginal and non-Aboriginal) and to compare these incidences with those found in other parts of Australia.

METHODS

Study population

The study population comprised all infants born alive in Central Australia between 1 January 1993 and 30 June 2000. This region includes the Alice Springs, Tennant Creek and Barkly regions of the Northern Territory, and the Anangu Pitjantjatjara (AP) Lands in South Australia. It has a population of about 47 000 (Australian Bureau of Statistics 1996 Census), with an average of 820 births per year.

Data sources

Birth data were obtained for the Alice Springs, Tennant Creek and Barkly regions from the Epidemiology Unit in Darwin, and for the AP Lands (coded as unincorporated Far North region of South Australia) from the Pregnancy Outcome Unit, Department of Human Services, Adelaide, SA.

Patients with congenital heart defects were identified from echocardiography reports and case records of the Alice Springs Hospital. This is the sole referral centre for paediatric and initial cardiac diagnostic services for the region, and the site for a major proportion of births in Central Australia. Patients presenting with clinical signs or symptoms suggesting CHD undergo assessment and echocardiography at the hospital. All echocardiograms are performed by a single paediatrician (GW) with training in paediatric cardiology and echocardiography. Patients with clinically significant CHD are also reviewed by visiting paediatric cardiologists from the Women's and Children's Hospital, Adelaide, SA.

CHD was defined as a structural abnormality of the heart or great vessels. This included ventricular septal defects of all sizes and congenital heart block. Excluded were diagnoses based only on clinical findings; patent ductus arteriosus (PDA) in infants born before 37 weeks' gestation; asymptomatic PDA in the first 3 months of life in infants born at term; simple bicuspid aortic valves; isolated peripheral pulmonary artery stenosis with no clinical significance; and minor atrial shunts across

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1: Comparison of incidence of congenital heart defects between Central Australia and New South Wales and the Australian Capital Territory*

Type of defect	Central Australia: Number of cases (incidence†) Aboriginal	Non-Aboriginal	Total	Regional comparison: NSW/ACT incidence†	Relative risk (95% CI)‡
Left–right shunt lesions	40	42	82	1.23	10.77 (8.51–13.62)
Ventricular septal defect	30 (10)	32 (10.1)	62 (10)	0.99	10.21 (7.79–13.36)
Atrial septal defect	5 (1.7)	7 (2.2)	12 (1.9)	0.13	14.6 (7.72–27.47)
Patent ductus arteriosus	5 (1.7)	3 (0.9)	8 (1.3)	0.12	11.16 (5.23–23.83)
Other lesions					
Atrioventricular septal defect	1 (0.3)	0	1 (0.16)	0.21	0.76 (0.11–5.50)
Pulmonary stenosis	2 (0.7)	0	2 (0.32)	0.23	1.43 (0.35–5.82)
Aortic stenosis	1 (0.3)	1 (0.3)	2 (0.32)	0.10	3.28 (0.79–13.66)
Coarctation of aorta	1 (0.3)	1 (0.3)	2 (0.32)	0.27	1.20 (0.30–4.87)
Parachute mitral valve	0	1 (0.3)	1 (0.16)	0	0
Tetralogy of Fallot	1 (0.3)	1 (0.3)	2 (0.32)	0.27	1.21 (0.30–4.92)
Double-outlet right ventricle	4 (1.3)	1 (0.3)	5 (0.81)	0.07	11.63 (4.44–30.46)
Hypoplastic left heart syndrome	2 (0.7)	1 (0.3)	3 (0.49)	0.23	2.09 (0.66–6.62)
Pulmonary atresia	2 (0.7)	0	2 (0.32)	0.09	3.72 (0.89–15.56)
Tricuspid atresia	0	1 (0.3)	1 (0.16)	0.09	1.80 (0.25–13.18)
Transposition of great arteries	0	1 (0.3)	1 (0.16)	0.33	0.49 (0.07–3.54)
Total anomalous pulmonary venous drainage	0	1 (0.3)	1 (0.16)	0.07	2.23 (0.30–16.47)
Ebstein anomaly	1 (0.3)	0	1 (0.16)	0.02	9.30 (1.12–77.24)
Common atrium	1 (0.3)	0	1 (0.16)	0	0
Congenital heart block	1 (0.3)	0	1 (0.16)	0	0
Total	57 (19.0)	51 (16.1)	108 (17.5)	3.5	5.06 (4.17–6.16)

* National Perinatal Statistics Unit (NPSU) study.[2] † Incidence per 1000 live births.
‡ Relative risk in Central Australia compared with NSW/ACT.

defects measuring less than 5 mm which subsequently closed. In patients with more than one cardiac defect, a single diagnosis was assigned according to the most haemodynamically significant lesion.

For each identified patient, we reviewed both maternal and patient records for information on place of birth, sex, ethnicity, age at diagnosis, associated malformations or genetic conditions, perinatal data of significance, and follow-up until June 2000.

Statistical analyses

The overall incidence of CHD and the incidence of each type of defect were determined. The overall incidence was compared with South Australian data for 1993–2000 (Heather Scott, Senior Project Officer, South Australian Birth Defects Register, personal communication) and published figures from New South Wales and the Australian Capital Territory (National Perinatal Statistics Unit [NPSU] Study)[2] and Western Australia.[3] Comparative data on specific echo-proven defects were available only for NSW and the ACT.[2]

Chi-square analyses were carried out using the Epi Info statistical package, version 2000.[5]

The study was approved by the Central Australian Human Research Ethics Committee.

RESULTS

A total of 108 patients with CHD were identified, of whom 57 were Aboriginal and 51 non-Aboriginal. Of the latter group, one was of Asian origin, with the remainder of European descent. Forty-seven patients were reviewed by both a local (GW) and a visiting (BK) paediatric cardiologist, with diagnostic concurrence for 46 (98%).

During the study period, there were 6156 live births in the region, 2991 Aboriginal and 3165 non-Aboriginal. This gave an incidence of CHD of 17.5 per 1000 live births overall (95% CI, 14.9–21.7), with 19.0 per 1000 live births in the Aboriginal population (95% CI, 14.4–24.6) and 16.1 per 1000 live births in the non-Aboriginal population (95% CI, 12.0–21.1). Although the incidence of CHD was 18% higher in the Aboriginal population than in the non-Aboriginal population, this difference was not statistically significant (relative risk [RR], 1.18; 95% CI, 0.81–1.72).

There was also no significant difference in the distribution and incidence of specific defects between the two population groups (Box 1). Left–right shunt lesions (ventricular septal defects, atrial septal defects and PDA) predominated in both groups. There were more cases of double-outlet right ventricle in Aboriginal infants, but the difference was not statistically significant (RR, 5.78; 95% CI, 0.65–51.66). There was only one case of transposition of the great arteries, and this was in a non-Aboriginal infant.

Comparison with other regions

The population-based incidence of CHD is compared between different regions of Australia in Box 2. The incidence of CHD in South Australia for the period 1993–2000 was 12 per 1000 births, with little difference between Aboriginal and non-Aboriginal births (11.3 and 12.1 per 1000 total births, respectively) (South Australian Birth Defects Register). In comparison, live-born infants in Central Australia had a much higher incidence during the same period (17.5 per 1000; RR, 1.46; 95% CI, 1.21–1.77). Similarly, the incidence in Central Australia was significantly higher than the incidence in NSW and the ACT (4.3 per 1000 live births) and Western Australia (7.65 cases per 1000 total births).[2,3]

2: Comparison of population-based studies of incidence of congenital heart disease (CHD), Australia

Region	Period	Denominator	CHD cases (enumeration notes)	Incidence per 1000	Relative risk (95% CI) in Central Australia
Central Australia	1993–2000	6156 (live births)	108 (1.2% diagnosed after 1 year of age)	17.5	1.00
NSW and ACT[2]	1981–1984	343 521 (live births)	1479 (diagnosed in first year of life)	4.3	5.06 (4.17–6.16)
Western Australia[3]	1980–1989	233 502 (total births)	1787 (diagnosed antenatally or up to 6 years of age)	7.65	2.32 (1.89–2.83)
South Australia*	1993–2000	152 299 (total births)	1835 (diagnosed antenatally or up to 5 years of age)	12	1.46 (1.21–1.77)

*Heather Scott, Senior Project Officer, South Australian Birth Defects Register, personal communication.

The regional differences in specific types of echo-proven heart defects are shown in Box 1. Only NSW and ACT data from the NPSU study were available for this comparison.[2] The incidence of left–right shunt lesions was significantly higher in Central Australia (RR, 10.77; 95% CI, 8.51–13.62), with no significant change after excluding two cases diagnosed after 1 year of age. The combined incidence of heart defects other than left–right shunt lesions was also significantly higher in Central Australia, but the difference was less marked (RR, 2.02; 95% CI, 1.35–3.00). Double-outlet right ventricle was the most common primary cyanotic heart lesion in Central Australia, with an incidence about 11 times higher than that reported for NSW and the ACT (RR, 11.63; 95% CI, 4.44–30.46).

Patient characteristics

Demographic and clinical characteristics of the patients with CHD in Central Australia are shown in Box 3. One hundred and six cases (98%) were diagnosed within the first year of life. One case each of atrial septal defect and aortic stenosis were diagnosed at 2 and 4 years of age, respectively. Twenty-six patients (24%) underwent surgical correction of their CHD. Ten patients died (9%).

Eight infants (7%) had a known syndromic or chromosomal malformation (trisomy 18 [2], Pierre–Robin sequence [2], duplication of short arm of chromosome 8 [1], fetal alcohol syndrome [1], Leigh syndrome [1] and Diamond–Blackfan syndrome [1]). Eleven had extracardiac malformations with no identified syndromic or chromosomal abnormality.

Detailed maternal history was available for 106 patients (98%); 15 mothers (14%) had diabetes, while alcohol misuse was documented for seven (7%).

DISCUSSION

This is the first report on the incidence of CHD in Central Australia. We included only cases proven by echocardiography in live births. The overall incidence of echo-proven CHD of 17.5 per 1000 live births is significantly higher than the combined incidence of clinical and echo-proven defects reported from South Australia, NSW and the ACT, and Western Australia (Box 2).[2,3] We also found that the incidence of CHD was 18% higher in the Aboriginal population compared with the non-Aboriginal population, but the difference was not statistically significant, probably because of the small cohort size.

There are several possible explanations for the variation in incidence between the different regional studies, including errors in population denominator data, changing case ascertainment with evolving echocardiography technology, and inclusion of small left–right shunt lesions. In our study we used the best available source of population data to eliminate denominator error. We confirmed that all patients lived in Central Australia. However, as some patients from the northern and southern extremes of the region might have been referred directly to hospitals other than Alice Springs Hospital, the calculated incidence may be an underestimate.

We acknowledge that epidemiological studies on relatively small cohorts by a small and committed group of investigators may tend to find a higher incidence of CHD than studies in larger cohorts. An example is a US study from the 1970s, which found an incidence of around 10 per 1000 of definite plus possible congenital heart defects[6] — much greater than had been docu-

3: Demographic and clinical characteristics of patients with congenital heart disease, Central Australia

Characteristic	Aboriginal (*n*=2991)	Non-Aboriginal (*n*=3165)	Total (*n*=6156)
Maternal age < 30 years*	40 (71%)	35 (70%)	75 (71%)
Primigravida	16 (27%)	12 (24%)	28 (26%)
Female sex	36 (63%)	33 (65%)	69 (65%)
Age at diagnosis			
< 1 month	42 (74%)	37 (72%)	79 (73%)
1–12 months	14 (24%)	13 (25%)	27 (25%)
> 12 months	1 (2%)	1 (2%)	2 (1.2%)
Chromosomal or syndromic abnormality	5 (9%)	3 (6%)	8 (7%)
Birthweight < 2.5 kg*	14 (25%)	9 (18%)	23 (22%)
< 37 weeks' gestation at birth*	15 (27%)	9 (18%)	24 (23%)

*Details unknown for two patients, one Aboriginal and the other non-Aboriginal.

mented by most earlier studies based on much larger cohorts.

High case ascertainment in Central Australia is likely for several reasons. Echocardiography has been readily available in Alice Springs since the beginning of the study period in 1993, and colour Doppler ultrasonography since 1994. This probably enhanced diagnosis of minor left–right shunt lesions which might have been missed in the previous two Australian studies.[2,3]

Most deliveries in Central Australia occur at Alice Springs Hospital, and, as follow-up can be difficult, there is a low threshold for early investigation of neonates with murmurs. There also appears to be a low threshold for referral from primary health services of patients with incidental murmurs. High rates of hospital admission for Aboriginal children in the first 2 years of life also contribute further opportunities for detection of CHD.

Recent reports suggest a rapid increase in the rates of minor defects, such as small ventricular and atrial septal defects, through active case ascertainment and widespread use of echocardiography.[7,8] However, in our study, there was also a higher incidence of major heart defects, such as double-outlet right ventricle, compared with the 1980s studies. Some of these findings can be explained by changes in diagnostic and reporting practices, but risk factors for CHD may also have differed between the populations.

The proportions of Aboriginal and non-Aboriginal live births in Central Australia are similar, and this has been consistent over many years. We found no statistically significant difference in the incidence of CHD between the two major population groups. However, our study size has only 30% power to demonstrate a difference of 30% at the 5% level of significance in the incidence rates.

The influence of ethnicity on rates of CHD has been variously reported as significant and non-significant. A study in a tertiary hospital in the United Kingdom found that Asian infants had a higher incidence of CHD requiring hospital admission than non-Asian infants.[9] In Western Australia, CHD was 30% more common in Aboriginal compared with non-Aboriginal total births (prevalence ratio, 1.3; 95% CI, 1.1–1.6).[3] In contrast, studies from the United States show either no difference or a slightly increased prevalence of CHD in the white population compared with the non-white population.[8,10-13]

Ethnicity may also influence type of CHD. We found more cases of double-outlet right ventricle in the Aboriginal population, but no cases of transposition of the great arteries, although these differences were not statistically significant. Similarly, no cases of transposition of the great arteries were found in Aboriginal children in a Western Australian study from the 1970s[14] or in South Australian data for 1993–2000 (Heather Scott, Senior Project Officer, South Australian Birth Defects Register, personal communication). The apparent low incidence of this condition is unexplained. In the United States, the black population has also been observed to have a lower incidence of this condition than the white population.[8]

It is likely that high utilisation of echocardiography contributed to the high incidence of CHD found in our study. However, incidence was high for serious as well as minor types of CHD. Possible perinatal, socioeconomic and environmental factors should also be explored in the light of these findings.

ACKNOWLEDGEMENTS

We thank Dr Annabelle Chan, Pregnancy Outcome Unit, Women's and Children's Hospital, Adelaide, SA, for a critical review of the article.

COMPETING INTERESTS

None identified.

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(Received 22 Jul 2003, accepted 26 Mar 2004) ❑