Congenital anomalies — why bother?

Carol I Bower, David Lester-Smith and Elizabeth J Elliott

The challenge of convincing governments of the value of a nationally comprehensive data collection

Congenital anomalies are worth bothering about — they affect around one in 20 births in Australia.[1-3] They are the second most common cause of perinatal and infant mortality and the fourth commonest cause of mortality in 1–14-year-olds in Australia.[4,5] They are major contributors to hospital admissions[6] and often result in lifetime disability. They are costly to our health system, including the considerable expense of providing programs to screen for, diagnose and terminate pregnancies affected by major congenital anomalies (Down syndrome and neural tube defects in particular). Importantly, some anomalies are preventable, including neural tube defects (70% preventable with adequate periconceptional folic acid[7]) and anomalies resulting from exposure to teratogens (eg, by avoiding alcohol during pregnancy). For many congenital anomalies, early identification allows interventions to decrease the risk of secondary disabilities.

We need good data to monitor trends in congenital anomalies, to identify clusters of cases that may require investigation for possible environmental causes, and to evaluate the effectiveness of interventions for screening, treatment and prevention.

Nationally, the Australian Institute of Health and Welfare (AIHW) National Perinatal Statistics Unit (NPSU) collates information on congenital anomalies that is supplied voluntarily by health departments in the states and territories. However, there remains considerable variability between jurisdictions in the scope and quality of the data collected. This variability — which includes the sources of case ascertainment, the upper age limit for inclusion, definitions and classifications used, methods of operation, and resources available for collecting, updating, validating and using the information — limits the utility of the collection.

National data published by the NPSU can only be as complete as the data provided by individual states and territories. Lack of completeness is evident in two recent AIHW reports. The first, *Congenital anomalies in Australia 2002–2003*,[8] does not include data from the Northern Territory, because data were not available. This may change, as the NT is reviewing its perinatal data needs. In addition, data were only available from four states on terminations of pregnancy at less than 20 weeks' gestation for congenital anomalies.[8]

The absence of information on early terminations is also evident in the second report, *Neural tube defects in Australia*.[9] The prevalence of neural tube defects at birth for the period 1998–2005 was similar for all states included in the report, at around 5 per 10 000 births, but the total prevalence (including early terminations of pregnancy from the four states collecting such information) was more than twice as high, at 10.1 per 10 000 pregnancies. Furthermore, of the four states collecting data on early terminations, the total prevalence in 2005 in South Australia, Victoria and Western Australia was 13.3 per 10 000, double that in New South Wales (6.2 per 10 000), suggesting incomplete ascertainment of terminations in NSW.

One of the purposes of the report on neural tube defects[9] was to provide baseline data against which to monitor the effect of the introduction of mandatory fortification of bread-making wheat flour with folic acid, in place nationally by 13 September 2009. Because such a high proportion of neural tube defects are diagnosed prenatally and affected pregnancies terminated, post-intervention monitoring in Australia will be restricted to the three states where there is complete ascertainment of such terminations.

The inclusion of terminations of pregnancy is essential for a national data collection on congenital anomalies — not only for evaluating interventions such as folic acid fortification, but also for evaluating and monitoring the safety and quality of prenatal screening programs and diagnostic tests, and the associated health and psychosocial impacts.

In response to the limitations in national data collection, a program was commenced in 2007 to develop a national minimum dataset on congenital anomalies. Members of a committee representative of the states and territories reached consensus on collecting good data on a limited number of conditions, particularly those with important clinical, social or health care impacts; and on the use of internationally agreed definitions for congenital anomalies. However, because of existing data limitations in some jurisdictions, commitment to a national minimum dataset is not currently possible. In addition, the scope of the proposed national collection has been limited to the perinatal period, which means terminations of pregnancy for congenital anomalies before 20 weeks' gestation will not be included. These decisions are very disappointing and suggest that Australian policymakers and governments are still to be convinced of the value of monitoring congenital anomalies, which, despite their magnitude and importance as a cause of mortality and morbidity, are clearly not seen as a public health priority.

Historically, data collection for congenital anomalies has been unfunded or under-resourced in Australia. Apart from mandatory folic acid fortification, there has been no national policy on the surveillance, prevention and management of congenital anomalies. There has also been no consumer involvement in deliberations on the societal impact of these anomalies, the need to collect national data, and the ways in which these data should be collected and used. The challenge remains to convince governments of the value of a nationally comprehensive collection that can be used to monitor trends, identify clusters that may require investigation, evaluate the effectiveness of screening and interventions for treatment and prevention, and allow research into the prevention of congenital anomalies.

Author details

Carol I Bower, MB BS, PhD, FAFPHM, Epidemiologist and Senior Principal Research Fellow[1]
David Lester-Smith, BM BS, FRACP, MPH, General Paediatrician and Research Fellow[2,3,4]
Elizabeth J Elliott, MD, MPhil, FRACP, Professor of Paediatrics and Child Health,[2,3] and Director[4]

1 Telethon Institute for Child Health Research, University of Western Australia, Perth, WA.
2 Sydney Medical School, University of Sydney, Sydney, NSW.
3 The Children's Hospital at Westmead, Sydney, NSW.
4 Australian Paediatric Surveillance Unit, Sydney, NSW.
Correspondence: carolb@ichr.uwa.edu.au

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