

## Original Article

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# Early diagnosis and screening of congenital cardiac anomalies

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**Abstract** Considerable numbers of congenital cardiac anomalies are missed at the time of delivery. Study reports show that congenital cardiac anomalies are the second most common birth defect in many countries. Despite this fact, our previous study showed that the prevalence of congenital cardiac anomalies is the fifth most common one, indicating that many of these defects might not be properly diagnosed at the time of delivery and birth. The aim of this study was to estimate the missing frequency of congenital cardiac anomalies at the time of delivery and birth. The population of the study was 185,650 births in the Northwest region of Iran covered by the Tabriz Registry of Congenital Anomalies. A total of 451 children with congenital cardiac anomalies were studied in the region from 2000 to 2009. The expected prevalence of congenital cardiac anomalies at birth was estimated to be 24.2 per 10,000 births while a prevalence of 9.2 per 10,000 births was observed at the same time and place. This indicated that 59.1% of children with congenital cardiac anomalies were not identified at birth (p-value less than 0.05). This proportion increased by 13% over the study period from 2000 to 2009 (p-value greater than 0.1). Our findings indicated that a remarkable frequency of congenital cardiac anomalies was not diagnosed at birth because there was no paediatric cardiologist available at the time of birth in the gynaecology and obstetrics wards.

Keywords: Cardiac birth defect; paediatric cardiology; prevalence

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CONGENITAL CARDIAC ANOMALIES REFER TO A massive structural and functional malformation of the heart. This may accompany other additional malformation syndromes.<sup>1,2</sup> The universal prevalence of congenital cardiac anomalies is estimated at 8 per 1000 live born infants.<sup>3</sup> The study reports show that congenital cardiac anomalies are the second most common birth defect in many countries.<sup>4,5</sup> Despite this fact, our previous study showed that the prevalence of congenital cardiac anomalies is the fifth most common one, indicating that many of the newborns with life-threatening cardiac anomalies are missed at the time of delivery or diagnosed later after birth.<sup>6,7</sup>

Early diagnosis and screening of congenital cardiac anomalies is vital because the lack of proper diagnosis of congenital cardiac malformation will result in a high risk of mortality in children and decrease life expectancy of newborn infants.<sup>8,9</sup> The study of missing frequency of congenital cardiac anomalies will be important in planning for prevention strategies, medical care, and surgical treatment of newborn babies.<sup>10,3</sup> Infants with congenital cardiac diseases may not even be diagnosed until a paediatric cardiologist visits them for some other medical reasons.<sup>11,12</sup> The aim of this study was to estimate the missing frequency of congenital cardiac anomalies at the time of delivery or after birth.

## Method

The study population was 185,650 births – 183,579 live births and 2071 stillbirths – in the Northwest

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region of Iran covered by the Tabriz Registry of Congenital Anomalies<sup>13</sup> under the Tabriz University of Medical Sciences. A total of 451 children with congenital cardiac anomalies were studied in the region from 2000 to 2009. University hospitals in the area provide the study population with advanced facilities and equipments, medical care, surgical operation, and clinical follow-up.

Neonatal examinations consist of echocardiography, pulse oxymetry, and Doppler colour echocardiography. Newborn babies are routinely examined with common neonatal tests before their first discharge from the hospital.

The anomalies in this study were classified using the International Classification of Diseases coding system (Version 10). Two midwives and a nurse were recruited to carry out this classification in the Tabriz Registry of Congenital Anomalies programme. Hospital records, common hospital discharge forms, and paediatric discharge records were used as the data sources in this study.

The expected prevalence of congenital cardiac anomalies at birth was estimated for the study population using the Tabriz Registry of Congenital Anomalies data. This was then compared to the observed prevalence of congenital cardiac anomalies at the same place and time; 95% confidence intervals were calculated to assess the statistical significance of the data.

## Results

A total of 185,650 births were recorded in the study area, including 183,579 live births (99.96%) and 2071 stillbirths (1.11%). During the study period, 451 cases with a primary diagnosis of congenital cardiac disease were identified and ascertained.

The main characteristics of study subjects, including gender, type of delivery, and type of birth, are presented in Table 1. Most of the cases were male (57.4%). The majority of infants were born live (89.1%). Caesarean section was the main type of delivery (49.4%) in children born with congenital cardiac disease.

Total prevalence of congenital cardiac anomalies at birth was 9.2 per 10,000 births (95% confidence intervals: 7.8–10.6). Patent arterial duct (15%), atrial septal defect (12%), and ventricular septal defect (5%) accounted proportionally for about one-third of anomalies as the most common cardiac defects identified and recorded in the area. The expected total prevalence of congenital cardiac anomalies at birth was estimated at 24.2 per 10,000 births (95% confidence intervals: 22.1–26.5) at the same time and place. A comparison of the observed and expected prevalence rates indicated that 59.1% of children

Table 1. Characteristic of the study subjects.

	n	%
Gender		
Female	190	42.12
Male	259	57.42
Unknown	2	0.44
Type of delivery		
Vaginal	190	42.12
Caesarean section	223	49.44
Unknown	38	8.42
Type of birth		
Live	403	89.1
Abortion	3	0.66
Stillbirth	4	0.88
Unknown	4	9.09

with congenital cardiac anomalies were not identified at birth (p-value less than 0.05). This proportion increased by 13% over the study period from 2000 to 2009 (p-value greater than 0.1).

## Discussion

This was a cross-sectional study aimed at identifying the missing rate of congenital cardiac anomalies at birth in the Northwest region of Iran. We found a high proportion of children born with congenital cardiac anomalies with no diagnosis at birth because there was no paediatric cardiologist available in the gynaecology and obstetrics wards as a routine. If those infants with congenital cardiac anomalies are properly diagnosed soon after birth, we will be able to provide them with proper surgery and the essential medical care that they need. This will then result in longer survival for children born with congenital cardiac anomalies.

According to the study of Wren et al,<sup>11</sup> only 45% of infants with cardiac defects are identified as abnormal in neonatal examination and only 16% of infants with congenital cardiac abnormality could be recognised before discharge from the hospital.

This study had some advantages. First, it included a very large population consisting of 185,650 births during a decade. Second, it was carried out in three university hospitals, with advanced facilities and equipment, run by professional staff. Third, the data for this study came from the registry-based information of the Tabriz Registry of Congenital Anomalies system established in the region. Fourth, the data for this study included stillbirths as well as live births.

This study had some limitations too. First, it might have been affected by inter-observational bias. In some cases, for instance, there might be inconsistencies in identifying cases of congenital

cardiac anomalies in the Tabriz Registry of Congenital Anomalies registry system. Second, we were unable to follow up the study subjects for more than 1 year. Third, since many cases of abnormalities and congenital cardiac diseases are usually recognised over the next few years after birth, there might have been an underestimation of the prevalence of congenital cardiac anomalies in the region. Fourth, some patients with undetermined anomalies might have been placed in a wrong category in the classifications of anomalies, resulting again in an underestimation of the prevalence of congenital cardiac anomalies.

Examination of infants born in the local hospitals is performed by neonatologists. As mentioned previously, it is not routine that the paediatric cardiologists attend in the delivery room. The role of neonatologists is therefore essential in identifying congenital cardiac anomalies at the time of birth.<sup>14</sup> The proper use of colour Doppler cardiography will provide a good chance for identification of minor cardiac defects by trained neonatologists.<sup>15</sup> Moreover, since the pulse oxymetry technique is not very complicated, it can be considered as a reliable method for these purposes by neonatologists too.<sup>16,17</sup>

While there is no paediatric cardiologist attending at the time of birth, the application of these techniques and procedures by neonatologists may eventually result in longer survival and suitable medical care for infants born with congenital cardiac defects.<sup>18,19</sup>

It is concluded that in order to diagnose and treat cardiac defects at an early stage and to enhance the life span of the patients, it is recommended that suitable training programmes and fellowships are offered for paediatric cardiologists and neonatologists. It is also recommended that paediatric cardiologists are present in obstetrics wards of the hospitals to minimise the missing cases of congenital cardiac anomalies at birth.

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