# Evaluating the overt extracardiac malformations in children with congenital heart disease in Khuzestan Province

*Original Article*


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## Abstract:

***Background:*** Extracardiac malformations can be seen in 20-45% of infants with congenital heart disease (CHD). Chromosomal abnormalities exist in 5-10% of patients with CHD. The aim of this study was to assess the frequency of overt extra cardiac malformations in children with CHD.

***Methods:*** This descriptive epidemiologic study was conducted on 720 patients with CHD referred to the pediatric cardiac clinic of the most important south west referral center of Iran in 2014. Data were collected by studying patients' files during this time.

***Results:*** Totally, 401(55.7%) and 319 (44.3%) of patients were male and female, respectively. Extracardiac malformations were diagnosed in 53 cases (7.36%) and multiple extracardiac malformations involving more than one anomaly were present in 10 cases (1.38%). The most frequent extracardiac malformations in the current study included skeletal and visceral abnormalities and then genitourinary was seen. Of all patients, 33 (4.58%) cases were affected by syndromic features of which Down syndrome was the most common.

***Conclusions:*** Investigation for extra cardiac abnormalities in patients with CHD is very important, since proper diagnosis and early treatment of these abnormalities can improve the patients' outcomes. On the other hand all the patients with overt and syndromic abnormalities require investigation for a possible CHD.



***Keywords:*** Extracardiac Malformation, Congenital Heart Disease, Children

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## Introduction:

Congenital heart disease (CHD) can be seen in 0.5-0.8% of live births. Congenital defects have a wide range in severity in children [1]. The initial assessment in cases that has been suspected congenital heart disease is a systematic progression. Congenital heart defects based on the presence or absence of cyanosis are divided into two main categories which can be determined by physical examination and help of pulse oximetry. The final diagnosis is confirmed by echocardiography [1]. CHD may be present with a syndrome containing known anomalies with specific physical findings or can be present with manifestations of a systemic defect which affects the heart and other systems. To assess the possible systemic disease or congenital malformation syndromes, all systems of the body should be examined. Extracardiac abnormalities are found in 20-45% of children with CHD. 5-10% of patients have a known chromosomal abnormality [2].


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The aim of this study was to evaluate the prevalence of obvious extra cardiac malformations in children with CHD.

## Methods:

This analytic epidemiologic study was conducted on children with CHD referred to the pediatric cardiac clinic of Golestan Hospitalin Ahvaz in 2014. The population included all patients with CHD referred to this center. Patients with patent ductus arteriosus in preterm neonates, bicuspid aortic valve, mitral valve prolapse and patent foramen oval were excluded from the present study.

Required information was collected from the patients' records in their files and pre-prepared questionnaires. Study variables were age, sex, type of congenital heart disease, type of heart abnormalities, type of malformations and type of syndrome. The study population was patients who had CHD confirmed with echocardiography.

All patients were evaluated for gross malformations divided into subgroups (skeletal, visceral, neurological, respiratory, genitourinary and miscellaneous). No X-ray or ultrasonography was done for all the patients, we just checked the one who carried the results of paraclinical data during the examinations sessions. Patients were categorized into different subgroups according to physical examinations and paraclinical data which had been previously performed.

The patients with syndromic features were referred to a genetic specialist for definite diagnosis and genetic tests were performed on some patients. The obtaining data were analyzed using SPSS 22.

## Results:

This study was conducted on 720 patients with CHD aged less than 18 years using census methods in Golestan Hospital of Ahvaz. Based on the findings, of all patients, 595 patients (82.74%) were acyanotic (45.8 % male and 36.8% female) and 125 (17.36%) were cyanotic (9.8% male and 7.6% female).

The total number of male patients was more than that of female patients. Most types of CHD based on the prevalence were: VSD (Ventricular Septal Defect) (35.3%), ASD (Atrial Septal Defect) (15.4%), TOF (Tetralogy of Fallot) (9.2%), PS (pulmonary valve stenosis) (7.9%) and PDA (Patent Ductus Arteriosus) (6.7%). Moreover, 55 patients (7.63%) had multiple congenital heart defects.

Among 720 patients, 667 cases had no apparent extracardiac malformations. 53 patients (7.36%) had extracardiac malformations that 38 cases (71.7%) were acyanotic and 15 cases (28.3%) were cyanotic. The highest number of malformations in acyanotic patients was skeletal and visceral type, and the highest number of malformations in cyanotic patients was skeletal and then visceral type. In both groups, the most prevalent malformation was skeletal.

Of the 720 patients examined in this study, 687 patients had no systemic anomalies, 33 patients (4.6%) had syndromic features of which Down syndrome was the most common. Distributions of extracardiac malformations are shown in Table 1 and the distributions of syndromes in children with CHD are illustrated in table 2.

**Table 1: Distribution of extra cardiac malformations in patients with CHD**

| Type of malformation | Number (%) |
|---|---|
| normal | 667 (92.6) |
| Skeletal | 13 (1.8) |
| Visceral | 12 (1.7) |
| Neurological | 2 (0.3) |
| Respiratory | 1 (0.1) |
| Genitourinary | 7 (1) |
| Miscellaneous | 8 (1.1) |
| 1,2,6 | 2 (0.3) |
| 1,3,5 | 1 (0.1) |
| 1,5 | 1 (0.1) |
| 1,6 | 3 (0.4) |
| 2,5 | 2 (0.3) |
| 5,6 | 1 (0.1) |
| **Total** | **720 (100)** |

1: skeletal, 2: visceral, 3: neurological, 4: respiratory, 5: Genitourinary, 6: Miscellaneous

**Table 2: Frequency distribution of syndromes in patients with CHD**

| Type of Syndrome | Number (%) |
|---|---|
| normal | 687 (95.4) |
| Down | 25 (3.5) |
| Allagile | 1 (0.1) |
| Turner | 1 (0.1) |
| Williams | 1 (0.1) |
| De Georges | 1 (0.1) |
| VACTER | 1 (0.1) |
| Unknown | 3 (0.4) |
| **Total** | **720 (100)** |

1: skeletal, 2: visceral, 3: neurological, 4: respiratory, 5: Genitourinary, 6: Miscellaneous

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## Discussion:

The most common type of CHD in this study was VSD with 35.3%. Khuzestan province is one of the greatest provincial health centers, according to the old and experienced physicians and fitness equipment, covering the southern and western provinces of the country.

Based on the findings in this study, 82.7% of patients were acyanotic and 17.3% of patients were cyanotic. The total number of male patients was more than that of female patients that is consistent with the findings of previous studies [3-5].

The most common type of CHD in this study was VSD with 35.3%, which is similar to the studies of Goucher et al.'s with 15.3%, Tennstedt et al.'s with 28% and Thompson et al.'s with 35%[3,4,6].

In a study conducted by Nahvi et al.'s on patients with facial skeletal problem, ASD with 28.3% was the most common CHD [7]. Cardoso et al.'s showed that CHDs were responsible for 40% of birth defects, being one of the most common and severe malformations [8]. Ferencz et al.'s performed a study in Baltimore-Washington during 1981-1989 and demonstrated an incidence of 27.71% of extracardiac anomalies in live-born infants with congenital cardiac malformations compared to only 3.4% in the control population [9]. Khalilian et al.'s found that the prevalence of heart murmur in neonates was 1.92% of which 50.8% were pathologic that the most common types of this disease were ventricular septal defect (25.4%) and atrial septal defect (16.9 %) [10].

In the present study, the prevalence of extracardiac anomalies was 7.36% (53 cases) of whom 10 patients (18.86%) had more than one extracardiac anomaly. Most cases of extracardiac malformations were skeletal, visceral and urogenital types with the prevalence of 24.45%, 22.55% and 13.17%, respectively. In the studies of Alabdulgader et al.'s 28.5% of patients had extracardiac anomalies [5] and Goucher et al.'s 15% of patients had extracardiac anomalies that over 45.9% of them had more than one anomaly and the highest number of anomalies was craniocephalous anomaly [3]. Jaiyesimi et al.'s found in their study that 13% of patients had extracardiac anomaly of which 27% were skeletal [11]. Tennstedt et al.'s suggested that 66% of patients had extracardiac anomaly that its highest number was syndromic [4].

Greenwood et al.'s revealed that the prevalence of CHD in patients with diaphragmatic hernia, imperforated anus, omphalocele and tracheoesophageal fistula was 23%, 12%, 11% and 15%, respectively [12]. Therefore, the results of the current study are confirmed by the mentioned results.

Today, the fetal echocardiography and sonography are performed during pregnancy for further evaluation. Tennstedt et al.'s showed that the fetal congenital heart malformations are common. These defects are often associated with other cardiovascular and extracardiac malformations, as well as with chromosome anomalies. Complex heart defects are frequent in fetuses, as they often lead to spontaneous abortion or stillbirth or, after prenatal diagnosis, to deliberate termination of pregnancy [13].

It is proposed to conduct exact research and the training centers should have a responsible person for recording all information completely. In addition, all findings, test results, clinical improvement and death of patients have to record exactly. Moreover, these centers should link together so that they build a Statistical Center in the entire country. By doing so, the areas where have more incidence of CHD can be recognized, the reasons of high incidence of CHD in some places can be found and the major step can be taken for the prevention and even treatment of CHD.

In conclusion, Determination of the prevalence of extracardiac abnormalities in patients with CHD, proper diagnosis and early treatment of them can improve the patients' outcomes. On the other hand, all patients with overt and syndromic abnormalities require investigation for a possible CHD.


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**Conflict of Interest:** There was no conflict of interest.


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