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Original article

Evaluation of dysmorphic children according to echocardiographic findings: A single center experience

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ABSTRACT

Background: Abnormal echocardiographic findings are more common in dysmorphic children. In our study, dysmorphic child development and echocardiographic findings were presented according to prenatal, natal and postnatal periods.

Aim of the study: The aim of this study is to evaluate the frequency and distribution of cardiac anomalies in dysmorphic children. The other aim is to investigate the prenatal, natal and postnatal characteristics of dysmorphic childs according to echocardiography findings.

Design and setting: This study was carried out jointly by the Medical Genetics and Pediatric Cardiology Departments. The files and the genetic reports of the patients were examined and the hospital registry system scanned, retrospectively. The patients were followed up by the medical geneticist from 2012 to 2017. Their systemic physical examination was performed and recorded.

Methods: This is a retrospective study which contains 468 children (244 males and 224 females) who were referred to the department of medical genetics due to dysmorphic features.

Results: Abnormal echocardiography findings were detected in 157 dysmorphic children (33.4%). Atrial septal defect, patent foramen ovale and ventricular septal defect were the most commonly detected echocardiography findings in dysmorphic children. The number of male children in the abnormal echocardiography group was significantly higher than in the normal echocardiography group. The incidences of consanguineous marriage, polyhydramnios, intrauterine growth retardation (IUGR) and preterm delivery in the abnormal echocardiography group were significantly higher than in the normal echocardiography group. Chromosomal aneuploidy rate in the abnormal echocardiography group was significantly higher than in the normal echocardiography group (37.6% vs 1.0%; $p = 0.001$).

Conclusion: According to our study findings, abnormal echocardiography findings were significantly associated with neonatal sex, consanguineous marriage, polyhydramnios, IUGR, preterm delivery and chromosomal aneuploidies in dysmorphic children.

1. Introduction

The ‘dysmorphic’ therm is originated from the Greek words ‘dys’ (disordered, abnormal, painful) and ‘morph’ (shape, form). Dysmorphology is a clinical genetic discipline which examines and interprets patterns of human growth and structural defects. These structural defects include malformation (an intrinsic developmental anomaly, e.g., spina bifida), disruption (an event disrupting intrinsically normal development, e.g., amniotic bands), deformation (an external force altering the shape of development, e.g., face shape due to severe oligohydramnios) and dysplasia (abnormal growth and maturation of cells, e.g., achondroplasia).

Congenital heart disease (CHD) is defined as a gross structural abnormality of the heart or intrathoracic great vessels that is actually or potentially of functional significance. This definition excludes the dysfunction of the great arteries, hypertrophic or dilated cardiomyopathies and congenital arrhythmias such as the long QT and the Wolf-Parkinson-White syndromes, even if the disorders are based on abnormalities present at birth [1,2]. Congenital heart malformations are the most frequent (one-third) of all major birth defects.’ The incidence of CHD in the normal population is approximately 0.8% [1,2]. In our literature review, abnormal echocardiographic findings in dysmorphic children have not been

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Table 1
Echocardiography findings in dysmorphic children.

Echocardiography findings	Number (%)
Normal findings	311 (66.4%)
Atrial septal defect	63 (13.5%)
Patent foramen ovale	43 (9.2%)
Ventricular septal defect	32 (6.8%)
Mitral insufficiency	22 (4.7%)
Aortic stenosis	15 (3.2%)
Patent ductus arteriosus	13 (2.8%)
Aortic insufficiency	11 (2.3%)
Tricuspid insufficiency	11 (2.3%)
Other findings	54 (11.5%)

investigated before and this is a unique issue. The echocardiographic findings in dysmorphic children are seen at Table 1.

2. Aim of the study

The aim of this study is to evaluate the frequency and distribution of cardiac anomalies in dysmorphic children. The other aim is to investigate the prenatal, natal and postnatal characteristics of dysmorphic childs according to echocardiography findings.

3. Subject and methods

This is a retrospective study of 468 pediatric patients who were referred to the department of medical genetics due to dysmorphism. The study cohort consisted of 244 boys and 224 girls who are aged between 0 and 18 years.

Data related with the cohort were obtained from medical records and parent response questionnaires. The prenatal characteristics that were questioned included fetal malpresentation, polyhydramnios, oligohydramnios, intrauterine growth restriction (IUGR), placenta previa, abruptio placenta, premature rupture of membranes and preterm premature rupture of membranes. Delivery time (preterm or term), delivery type (vaginal or cesarean), and indication for cesarean delivery were the natal characteristics that were questioned. Additionally, neonatal sex and anthropometric measurements at birth (weight, length and head circumference) were recorded. As for the postnatal characteristics, the Apgar score and the history for hypoxic ischemic encephalopathy and jaundice were investigated. In addition, detailed pedigree analysis was performed for all patients so that consanguineous marriages, similar cases and genetic diseases in the family could be detected.

Denver Developmental Screening Test-ii (DDST-ii) was applied to evaluate the neurological development of the patients aged over 6 years. In order to assess the motor, social and language skills of the patients aged less than 6 years, the time span for breastfeeding, the time for head and neck control, smiling, rolling prone to supine, sitting without support, creeping, distinguishing individuals, walking, pronouncing simple words, fluent speaking and playing simple games were asked. Moreover, it was questioned whether the patient underwent any surgical operation and the patient had a seizure. If the patient had a history of seizure, it was specified when the first seizure happened, how long it lasted, what the type of seizure was and how many seizures occurred so in total.

Collected data were analyzed by Statistical Package for Social Sciences version 18.0 (SPSS Inc., SPSS IBM, Armonk, NY, USA). Continuous data were expressed as mean ± standard deviation (range: minimum-maximum) whereas categorical data were denoted as numbers or percentages where appropriate. Chi-square test was used for the statistical comparisons. Two-tailed p values less than 0.05 were accepted to be statistically significant.

This study was approved by the Ethics Committee of Non-Interventional Clinical Investigations are with the approval number 2016-KAEK2.

4. Results

There were abnormal echocardiography findings in 157 dysmorphic children (33.4%). Atrial septal defect, patent foramen ovale and ventricular septal defect were the most commonly encountered echocardiography findings in dysmorphic children (13.5%, 9.2% and 6.8% respectively). The number of male children was significantly higher in the abnormal echocardiography group than in the normal echocardiography group (66/157, 42.0% vs 178/311, 57.2%; p = 0.002).

There was consanguineous marriage between the parents of 221 dysmorphic children (47.2%). The incidence of consanguineous marriage was significantly higher in the abnormal echocardiography group than in the normal echocardiography group (91/157, 58% vs 130/311, 42%; p = 0.001). The parents of 153 dysmorphic children were first degree relatives (32.7%) whereas the parents of 68 dysmorphic children were second degree relatives (14.5%).

Polyhydramnios was detected during the prenatal period in 15 dysmorphic children (3.2%). Polyhydramnios was significantly more frequent in the abnormal echocardiography group than in the normal echocardiography group (9/157, 5.7% vs 6/311, 1.6%; p = 0.028).

IUGR was diagnosed during the prenatal period in 151 dysmorphic children. The rate of IUGR was significantly higher in the abnormal echocardiography group than in the normal echocardiography group (61/157, 39% vs 90/311, 29%; p = 0.034).

Fifty-five dysmorphic children had preterm delivery (11.8%). The incidence of preterm delivery was significantly higher in the abnormal echocardiography group than in the normal echocardiography group (30/157, 19.1% vs 25/311, 8.0%; p = 0.001).

Cesarean delivery rate was 50.6% in dysmorphic children (237 out of 468). Cesarean delivery rate was higher in children with abnormal echocardiography findings than in children with normal echocardiography but this difference was statistically insignificant (89/157, 56.7% vs 148/311, 47.6%; p = 0.074).

Eighty-eight children with dysmorphology had low Apgar scores (18.8%). The number of children with low Apgar score was higher in the abnormal echocardiography group than in the normal echocardiography group but this difference was statistically insignificant (35/157, 22.3% vs 53/311, 17.0%; p = 0.180).

Seizures occurred during the postnatal period in 23 children with dysmorphology (4.9%). The number of children who had seizures were higher in the abnormal echocardiography group than in the normal echocardiography group but this difference was statistically insignificant (12/157, 7.6% vs 11/311, 3.5%; p = 0.054).

There were chromosomal aneuploidies in 62 dysmorphic children (13.2%). Chromosomal aneuploidies included Down syndrome in 42 children, Edwards syndrome in 12 children and Patau syndrome in 8 children. Moreover, there were chromosomal abnormalities in eight children with DiGeorge syndrome and six children with Williams syndrome. All children with both dysmorphism and chromosomal abnormalities had abnormal echocardiography findings. Chromosomal aneuploidy rate was significantly higher in the abnormal echocardiography group than in the normal echocardiography group (59/157, 37.6% vs 3/311, 1.0%; p = 0.001).

5. Discussion

Congenital heart disease is defined as a gross structural abnormality of the heart or intrathoracic great vessels that is actually or potentially of functional significance. This definition excludes the dysfunction of the great arteries, hypertrophic or dilated cardiomy-