İnönü Üniversitesi Tıp Fakültesi Dergisi
9(4) 229-232 (2002)

Congenital Heart Disease Associated With Gastrointestinal System Malformations In The Newborn: Is It Sufficiently Evaluated?

Sema Uğuralp*, Feyza Ayşenur Paç**, Necla Gürbüz*, Mehmet Demircan*,

Inonu University, Medical Faculty, *Department of Pediatric Surgery, Malatya, Turkey
Inonu University, Medical Faculty, **Department of Pediatric Cardiology, Malatya, Turkey

Purpose: We aimed to reveal the difference in the rate of congenital heart disease (CHD) presence between the neonates with and without cardiac murmurs in neonates with congenital gastrointestinal malformations (CGM).
Patients and methods: We report on 115 infants treated between 1995-2000 with CGM. In group A, echocardiographies were performed only to those with murmurs. Group B, all patients had undergone echocardiography (ECHO) examination.
Results: CHD diagnosis was established in 6% and 18% of group A and B patients, respectively (x^2: 3.95, $p<0.05$).
Conclusion: It has been shown that physical examination itself is not reliable in diagnosing CHD. We recommend routine ECHO evaluation with or without murmur in all neonates with CGM.

Key Words : Congenital Heart Disease, Gastrointestinal Malformation, Echocardiography, Neonate.

Yenidoğanlarda Konjenital Gastrointestinal Sistem Malformasyonlari İle Birlikte Görülen Konjenital Kalp Hastaliğini Yeterince Değerlendirebiliyor muyuz?

Amaç: Bu çalışmanın amacı kardiak üfürümü olan ve olmayan konjenital gastrointestinal sistem malformasyonlu yenidoğanlar arasındaki konjenital kalp hastalığı (KKH) oranında fark olup olmadığını ortaya çıkarmaktır.
Hastalar ve metot: 1995-2000 yılları arasında konjenital gastrointestinal malformasyon (KGM) tanısı ile tedavi edilen 115 infant rapor edildi. A grubunda sadece üfürümü olan yenidoğanlara Ekokardiografi (EKO) yapılırken, B grubunda üfürüm gözetmeksizin tüm olgulara EKO yapıldı.
Bulgular: A ve B grubundaki yenidoğanlarda KKH tanısı sırası ile %6 ve %18 idi. (x^2: 3.95, $p<0.05$).
Sonuç: KKH tanısında tek başına fizik muayenenin güvenilir olmadığı gösterilmiştir. Konjenital gastrointestinal sistem malformasyonu olan tüm yenidoğanlara üfürüm olsun ya da olmasın rutin EKO yapılmasını öneriyoruz.

Anahtar Kelimeler: Konjenital Kalp Hastalığı, Gastrointestinal Malformasyon, Ekokardiografi, Yenidoğan

Diagnosis of congenital heart disease (CHD) in the neonate is difficult. Genitourinary, skeletal, cardiovascular, and chromosomal anomalies are frequently associated with congenital gastrointestinal malformations (CGM). The prevalence of CHD during the neonatal period have been reported as 3.7 - 5.5 in 1000 live births in different series.[1-3] The incidence of congenital heart disease is more common in infants with CGM than the normal population. In esophageal atresia, the rate of cardiovascular anomalies was reported as 38%, whereas other system anomalies were in the range of 64%.[4] Additionally, association of CHD was reported in 35% of omphalocele, 12% of gastroschisis, 6% of Hirschsprung's disease (HD), and 18.5% of anal atresia (AA) cases.[5-7]

PATIENTS AND METHODS

In the 5 year study period between 1995-2000, CHD associations have been evaluated in 115 infants with upper and lower CGM. Patients were assessed in 2 groups. Group A consisted of 65 CGM patients who admitted between 1995 and 1997, and echocardiography (ECHO) was performed only when they have murmurs. Group B consisted of

50 CGM patients who admitted between 1998 and 2000, and all had undergone echocendiographic (ECHO) examination, prospectively.

During the hospitalization period, each patient has been fully investigated for the presence of other coexisting system anomalies. The diagnosis of CHD was established by patient history, physical examination, teleradiography (TELE), electrocardiography (ECG), and ECHO findings.

Chi-square test was used for the statistical analysis.

RESULTS

Congenital heart disease was present in; 4/14 (28.5%) of esophageal atresia and tracheoesophageal fistula (EA+TEF), 2 patients associated with and/or duodenal atresia, anal atresia, Down syndrome; 1/6 (16.6%) of pyloric atresia (PA); 2/6 (33.3%) of duodenal atresia (DA), 1 patient associated with EA+TEF; 1/9 (11%) of intestinal atresia (IA); 1/4 (25%) of omphalocele; 1/8 (12.5%) of gastroschisis; and 5/46 (10.8%) of anal atresia (AA), 1 patient associated with EA+TEF+DA. There were no CHD in 22 patients with HD (Table 1). All 4 patients diagnosed as Down syndrome with chromosome analysis had CHD.

Murmurs were present in 9/65 of group A. Two of these 9 patients had cardiomegaly. While 4 patients were diagnosed as CHD by ECHO, 4 were evaluated as normal (periferic pulmonary stenosis), and remaining one patient had tricuspid and pulmonic valve insufficiencies which were in normal physiologic limits, and sustained during follow up, this condition was regarded as physiologic. As a result, 5/9 patients in the group A were classified as normal, and remaining 4 neonates had CHD in group A (4/65, 6%) (Table 2).

Table 1. Distribution of CHD cases in patients with different CGM types

Diagnosis	No	CHD
EA+TEF	12	3
PA	6	1
DA	4	1
IA	9	1
Gastroschisis	8	1
Omphalocele	4	1
ARM	46	4
EA+TEF+DA+ARM	1	1
EA+TEF+DA	1	-
Persistant Cloaca	1	-
Cloacal extrophy	1	-
HD	22	-
Total	**115**	**13**

EA+TEF: Esophageal Atresia and Tracheoesophageal Fistula, PA: Pyloric Atresia, DA: Duodenal Atresia, IA: Intestinal Atresia, ARM: Anorectal Malformation, HD: Hirschsprung's Disease.

In group B, the diagnosis of CHD in 9/50 (18%) were established by routine ECHO examination for all patients. Five of 50 patients had murmurs; 3 of them had cardiomegaly and all also had CHD diagnosis by ECHO, and remaining 2 had tricuspid and/or pulmonic valve insufficiencies, and pulmonary hypertension. Pulmonary hypertension was also excluded from CHD classification. During follow up, tricuspid and pulmonic valve insufficiencies sustained in physiologic limits. Pulmonary hypertension decreased to normal levels in 3 months. Other 6 patients with the dizgnosis of CHD had no murmur (Table 3).

CHD dizgnosis of was established in 4/65 (6%) of group A, and 9/50 (18%) of group B patients. The difference in the rate of diagnosing CHD in groups A and B was statistically significant (x^2: 3.95, $p<0.05$) (Table 4).

Table 2. Group A patients with murmur, and their TELE, ECG, and ECHO findings.

Case No	Age (day)	Diagnosis	Murmur	TELE	*ECG*	ECHO
A1	2	Anal atresia	Ejection	N	N	PS
A2	3	EA+TEF+Down syndrome	Continuous	Cardiomegaly	N	PDA
A3	1	EA+TEF	Systolic	N	N	ASD
A4	1	EA+TEF+DA	Systolic	N	N	TI,PI (physiologic)
A5	1	Omphalocele	Systolic	N	N	N
A6	20	DA+Down syndrome	Continuous	Cardiomegaly	N	PDA, TI
A7	2	HD	Systolic	N	N	N
A8	2	Anal atresia	Systolic	N	N	N
A9	2	EA+TEF	Systolic	N	N	N

Abbreviations: N: Normal; PS: Pulmonary Stenosis, EA+TEF: Esophageal Atresia and Tracheoesophageal Fistula, PDA: Patent Ductus Arteriosus; ASD: Atrioseptal Defect, DA: Duodenal Atresia, TI: Tricuspit Insufficiency, PI: Pulmonary Insufficiency, HD: Hirschsprung's Disease.

Table 3. Six patients of group B without murmurs but diagnosed as CHD by ECHO, and 5 patients with murmurs and their ECG, TELE, and ECHO findings.

Case No	Age (day)	Diagnosis	Murmur	TELE	ECG	ECHO
B1	2	EA+TEF+Dudenal atresia+Anal atresia	-	**N**	**N**	MI
B2	1	EA+TEF+Down syndrome	-	N	N	ASD
B3	4	**Intestinal atresia**	-	N	N	IMC
B4	1	Gastroschisis	-	N	**N**	IMC
B5	1	Omphalocele	-	N	N	ASD,VSD
B6	9	Pyloric atresia	-	N	N	MI, PI
B7	28	Anal atresia	Continuous	Cardiomegaly	N	VSD,PDA
B8	25	Anal atresia	Systolic	Cardiomegaly	N	TI,PI,PH
B9	1	Anal atresia+ Down syndrome	Ejection	N	N	PS
B10	1	Omphalocele	Systolic	N	N	TI,PI,PH
B11	1	Anal atresia	Systolic	Cardimegaly	N	IMC, PI

Abbreviations: EA+TEF: Esophageal Atresia and Tracheoesophageal Fistula, MI: Mitral Insufficiency, ASD: Atrioseptal Defect, IMC: Isolated Mitral Cleft; VSD: Ventricular Septal Defect, PI: Pulmonary insufficiency, VSD: Ventricular Septal Defect, PDA: Patent Ductus Arteriosus, TI: Tricuspit Insufficiency, PH: Pulmonary Hypertension, PS: Pulmonary Hypertension.

Table 4. Comparison of the groups according to presence of CHD by ECHO.

	CHD (+)	%	CHD (-)	%	Total	%
Group A	4	6.0	61	94.0	65	100.0
Group B	9	18.0	41	82.0	50	100.0
Total	**13**	**11.3**	**102**	**88.7**	**115**	**100.0**

x^2: 3.95, df: 1, $p<0.05$, CHD: Congenital Heart Disease

ECHO findings were present in 10 of 14 patients with murmurs in both groups. 7/14 (50%) of patients with murmurs had diagnosis of CHD by ECHO, other 3 patients had no CHD.
The findings in ECG were right axial QRS deviation and right ventricular hypertrophy, which were in the normal physiological limits for the newborns. TELE revealed cardiomegaly in 2 patients of group A, and 3 patients of group B as depicted in Tables 2 and 3.

DISCUSSION

CHD is a significant cause of neonatal deaths [3], and requires special consideration for diagnosis in the neonatal period.[8] In a clinical study with 7204 newborn babies, it has been reported that routine physical examination can only detect 44% of cardiac malformations, and when a murmur is heard, there is a 54% chance of estzblishing an underlying cardiac malformation.[9] Also the incidence of congenital heart disease (CHD) is more common in infants with CGM than the normal population. Therefore, one should be aware that a normal neonatal examination does not preclude a clinically significant cardiac malformation.[1] In our patients; ECHO findings were present in 10 of 14 patients with murmurs in both groups, and 3/10 of the patients had tricuspid and/or pulmonic valve insufficiencies, and/or pulmonary hypertension, and subsequently pulmonary hypertension decreased to normal levels, whereas tricuspid and pulmonic valve insufficiencies sustained in physiologic limits. As a result, only 7/14 (50%) of patients with murmurs had diagnosis of CHD by ECHO.

One baby with ventricular septal defect (VSD) and patent ductus arteriosus in group B had undergone cardiac catheterization and angiography, and subsequently operated on. All 4 babies (2 EA+TEF, 1 DA, 1 AA) with Down syndrome diagnosed as CHD were died because of extracardiac reasons in the newborn period. All other remaining patients in both groups are still on follow-up by Pediatric Cardiology Department.

The comparison of diseases when classified according to their types of malformations, the frequency of CHD association with CGM was found to correlate with the literature. In group A, one patient diagnosed HD in the neonatal period had no murmur and was not performed ECHO. The same patient was hospitalized for definitive operation while 15 months age. At physical examination, there was systolic murmur and she had VSD diagnosis by ECHO. This patient was excluded from off the study because of was not able to diagnosed CHD in the neonatal period.

ECG may reveal specific information in only a few types of cardiovascular diseases in the newborn period. In our patients, we did not observe findings other than right axial deviation and right ventricular hypertrophies, which may normally be present in the

newborns. TELE showed cardiomegaly in some patients and all also had CHD.

There were 6 patients in group B without murmur but diagnosed as CHD by ECHO; 4 with mitral insufficiency (MI), 1 with atrioseptal defect (ASD) and VSD (color-Doppler showed small-sized muscular VSD), and 1 with ASD. Continuous-wave Doppler spectrum with increased intensity found that the pansystolic configuration was showing typical characteristics in patients with MI. Mitral regurgitation color-flow jet was towards the posterior wall of the left atrium. In 2 of MI patients, complex of anomalous mitral arcade or "hammock" valves have been demonstrated. In one patient, although the mitral valve was in normal configuration, continuous-wave and color-Doppler detected MI. Systolic function was in low normal limit, and speculated that resulting MI may be due to a previous attack of myocarditis. Remaining one patient had MI because of isolated mitral cleft. Interestingly, the cardiac pathology in patients without murmurs detected by ECHO was mainly MI, which was independent of the type of CGM. We were not able to support this noteworthy association with literature reports.

Echocardiography has been the primary method for imaging the hearts of infants and children with congenital and acquired heart disease for the past 25 years. [10] With %91,6 sensitivity and %99,9 specifity, ECHO has been proved to be a reliable method in diagnosis CHD in antenatal, neonatal and childhood periods.[11-13]

While the ratio of CHD diagnosis was being found as 18% in group B patients that underwent routine ECHO examination, group A ratio remained as low as 6%. The difference between the two groups was statistically significant ($p<0.05$). With these results, it may easily be speculated that group A patients would have had a higher ratio if routine ECHO examination was performed.

It has been shown that physical examination itself is not reliable in diagnosing CHD.[9] ECG or TELE may produce nonspecific findings in the newborns. ECHO can achieve prompt and specific diagnosis of CHD associated with CGM, which is noninvasive and high in specifity and sensitivity.

In conclusion, we strongly recommend a non-invasive and effective method, ECHO, as a routine preoperative procedure in neonates with CGM even if they have normal cardiovascular system findings in physical examination, in order to reduce their pre- and postoperative mortality rates.

REFERENCES

1- Ferencz C, Rubin JD, Mc Carter RJ, et al. Congenital heart disease: prevalance at live birth. Am J Epidemiol 1985; 121: 31-36.

2- Grabitz RG, Joffres MR, Collins-Nakai RL. Congenital heart disease: incidence in the first year of life. Am J Epidemol 1988; 128: 381-388.

3- Hassan I, Haleem AA, Bhutta ZA. Profile and risk factors for congenital heart disease. J Pak Med Assoc 1997; 47: 78-81.

4- Engum SA, Grosfeld JL, West KW, et al. Analysis of morbidity and mortality in 227 cases of esophageal atresia and/or tracheoesophageal fistula over two decades. Arch Surg 1995; 130: 502-508.

5- Fogel M, Copel JA, Cullen MT, et al. Congenital heart disease and fetal thoracoabdominal anomalies: associations in utero and the importance of cytogenetic analysis. Am J Perinatol 1991; 8: 411-416.

6- Holschneider A, Ure BM. Hırschsprung's disease. In: Ashcraft KW, Murphy JP, Sharp RJ, Sigalet DL, Snyder CL (eds) Pediatric Surgery 3rd ed. WB. Saunders Company, Philadelphia, Pennsylvania, 2000 pp 453-472.

7- Chen CJ. The treatment of imperforate anus: experience with 108 patients. J Pediatr Surg 1999; 34: 1728-1732.

8- Kuehl KS, Loffredo CA, Ferencz C. Failure to diagnose congenital heart disease in infancy. Pediatrics 1999; 103: 743-747.

9- Ainsworth S, Wyllie JP, Wren C. Prevalance and clinical significance of cardiac murmurs in neonates. Arch Dis Child Fetal Neonatal Ed 1999; 80: 43-45.

10- Rice MJ, McDonald RW, Reller MD. Pediatric echocardiography: Current role and a review of technical advances. J Pediatr 1996; 128: 1-14.

11- Narchi H. Neonatal ECG screening for congenital heart disease in Down syndrome. Ann Trop Paediatr 1999; 19: 51-54.

12- Saxena A, Shrivastava S, Kothari SS. Value of antenatal echocardiography in high risk patients to diagnose congenital cardiac defects in fetus. Indian J Pediatr 1995; 62: 575-582.

13- Sarıoğlu A, Batmaz G, Levent İ, et al. Fallot tetralojili çocuklarda koroner anomali ve Ekokardiyografik tesbiti. Türk Kardiol Dern Arş 1996, 24 : 474-479

Corresponding author
Sema Uguralp, M.D.
Inonu University Medical Faculty
Derpartment of Pediatric Surgery
44069, Malatya, Turkey
Phone :90 422 212 5202
Fax : 90 422 341 0728
E-mail : suguralp@inonu.edu.tr