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Congenital Heart Defects and its Types

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Abstract

Context: In general, congenital heart defects are classified as single, complex and undifferentiated. *Aim:* The present study is reporting the types of congenital heart defects that were observed in 65 patients. *Methods and Material:* There were 33 males and 32 females and their age ranged from neonate to 16 years. For the gathered information, percentage analysis was calculated. *Results:* It is seen that congenital heart defects as single entity was present in 36 (55.5%) cases; complex in 14 (21.5%) and undifferentiated in 15 (23%). In 32 female patients, congenital heart defects as single were present in 21 (65.6%), complex in 5 (15.7%) and undifferentiated in 6 (18.7%); whereas they were 15 (45.4%), 9 (27.3%) and 9 (27.3%) respectively in male patients. In female, it is observed, that atrial and ventricular septal defects were 6 and 5; but in male they were 3 and 4. Patent foramen ovale and atrio ventricular canal defects were not observed in female and in the males, dextrocardia and coarctation of aorta were not seen. *Conclusion:* In the present study, even though for the total, the association could be elicited between the female patients and CHDs; in male patients the occurrence of the complex CHDs were associated. Genetic counseling for the diagnosis, recurrence risk and medical management are provided.

Keywords: Congenital; Atrial; Ventricular Septal; Dextrocardia.

Introduction

Congenital Heart Defects (CHDs) include all structural malformations of the heart and intrathoracic great vessels resulting from errors in morphogenesis. Among all births the reported incidence is 8.1 per 1000 (Mitchell et al 1971). Among live births, the prevalence of CHDs is 3.7 to 7.7 per 1000 (Ferencz et al 1985).The incidence of CHDs is estimated to be similar for all major ethnic groups (Mitchell et al1971) and for male and female (Richards et al 1955) although a differential sex ratio for certain types of CHDs exist. CHDs could occur isolated; for example not associated with non-cardiac malformations. 73% of live births with CHDs do not have serious non-cardiac malformations (Ferencz et al 1987). CHDs could be undifferentiated or single or multiple (20%); for example ventricular septal defects (VSD), mild tricuspid regurgitation and cleft mitral valve (Richards et al 1955). In this paper, it is aimed to report the types of CHDs in the referred patients to Division of Human Genetics for genetic counseling.

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Material and Method

65 patients who were referred consecutively were selected for the study. There were 32 female and 33 male patients with the age range from neonate to 16 years. Percentage analysis was calculated.

Results

The findings were tabulated in the next page.

Table 1 shows that congenital heart defects as single entity was present in 36 (55.5%) cases; complex in 14 (21.5%) and undifferentiated systolic murmur in 15 (23%).

Table 1: Congenital Heart Defects in female and male pateients

Serial No.	CHDs
1	Systolic Murmur
2	Ventricular septal defects (VSD), mild tricuspid regurgitation, cleft mitral valve
3	Atrial septal defects (ASD)
4	Systolic Murmur
5	Murmur
6	VSD
7	VSD, pulmonary stenosis, patent foramen ovale
8	VSD
9	Mitral & aortic valve stenosis
10	Sub aortic VSD
11	Systolic murmur
12	Patent foramen ovale
13	Pulmonary systolic murmur
14	Dextrocardia
15	Patent ductus arteriosus
16	Atrio ventricular septal defects (AVSD)
17	Mitral valve prolapsed
18	Systolic Murmur
19	VSD
20	ASD, patent foramen ovale
21	ASD
22	Coarctation of aorta, bicuspid aortic valve
23	AVSD
24	Tetralogy of Fallot with hypoplastic pulmonary artery
25	Patent ductus arteriosus
26	ASD
27	Systolic Murmur
28	AVSD
29	Systolic Murmur
30	VSD
31	Dextrocardia
32	ASD
33	VSD
34	ASD
35	VSD
36	ASD
37	VSD
38	ASD
39	AVSD
40	Coarctation of aorta
41	VSD
42	Murmur
43	Murmur
44	VSD,ASD
45	Murmur
46	Mitral valve prolapsed
47	Dextrocardia
48	VSD, pulmonary artery hypoplasia
49	VSD
50	Systolic Murmur
51	ASD
52	Mitral valve prolapse with mitral regurgitation
53	ASD
54	Double outlet right ventricle, VSD, pulmonary artery hypoplasia
55	Pulmonary valve stenosis, VSD
56	Mitral valve prolapsed
57	Tetralogy of Fallot
58	Transposition of great arteries, multiple VSD, single coronary artery
59	Tetralogy of Fallot, patent ductus arteriosus, pulmonary atresia
60	Patent ductus arteriosus
61	Atrio ventricular canal defect
62	Pulmonary tricuspid stenosis, VSD, patent ductus arteriosus
63	Pulmonary atresia,VSD
64	Tetralogy of Fallot
65	Pan systolic murmur

Table 2: Congenital Heart Defects in Female

Serial No.	CHDs
1	Ventricular septal defects (VSD), mild tricuspid regurgitation, cleft mitral valve
2	Atrial septal defects (ASD)
3	Murmur
4	VSD
5	Systolic murmur
6	Pulmonary systolic murmur
7	Dextrocardia
8	Patent ductus arteriosus
9	VSD
10	Coarctation of aorta, bicuspid aortic valve
11	AVSD
12	Tetralogy of Fallot with hypoplastic pulmonary artery
13	Patent ductus arteriosus
14	ASD
15	Systolic Murmur
16	VSD
17	Dextrocardia
18	ASD
19	VSD
20	ASD
21	ASD
22	AVSD
23	Coarctation of aorta
24	VSD
25	VSD,ASD
26	Murmur
27	Dextrocardia
28	VSD, pulmonary artey hypoplasia
29	Mitral valve prolapse with mitral regurgitation
30	ASD
31	Mitral valve prolapsed
32	Tetralogy of Fallot
	Congenital Heart Defects in Male
Serial No	**CHDs**
1	Systolic Murmur
2	Systolic Murmur
3	VSD, pulmonary stenosis, patent foramen ovale
4	VSD
5	Mitral & aortic valve stenosis
6	Sub aortic VSD
7	Patent foramen ovale
8	Atrio ventricular septal defects (AVSD)
9	Mitral valve prolapsed
10	Systolic Murmur
11	Atrial septal defects(ASD), patent foramen ovale
12	ASD
13	AVSD
14	Systolic Murmur
15	ASD
16	VSD
17	VSD
18	Murmur
19	Murmur
20	MVP
21	VSD
22	Systolic Murmur
23	ASD
24	Double outlet right ventricle, VSD, pulmonary artey hypoplasia
25	Pulmonary valve stenosis, VSD
26	Tetralogy of Fallot
27	Transposition of great arteries, multiple VSD, single coronary artery
28	Tetralogy of Fallot, patent ductus arteriosus, pulmonary atresia
29	Patent ductus arteriosus
30	Atrio ventricular canal defect
31	Pulmonary tricuspid stenosis, VSD, patent ductus arteriosus
32	Pulmonary atresia,VSD
33	Pan systolic murmur

The breakdown of the single entity were: VSD and ASD 9 each; AVSD in 4;, dextrocardia, patent ductus arteiosus and mitral valve prolapse 3 in each, Tetralogy of Fallo in 2 and coarctation of aorta, atrio ventricular canal defect and patent foramen ovale one in each.

13 out of 65 cases had (20%) undifferentiated CHDS. Then in 52 cases, because of the overlap of CHDS in the complex group, it becomes 73 CHDs, The breakdown of the 73 CHDs in the 52 cases were: VSD 19/ ASD 11/ patent ductus arteriosus and AVSD 5 in each/ mitral valve prolapse and Tetralogy of Fallot 4 in each/ dextrocardia, patent foramen ovale, pulmonary artery hypoplasia and pulmonary valve stenosis 3 in each/ coarctation of aorta and pulmonary atresia 2 in each/ tricuspid regurgitation, cleft mitral valve, mitral valve stenosis, aortic valve stenosis, bicuspid aortic valve, double outlet right ventricle, single coronary artey, transposition of great vessels and tricuspid valve stenosis one in each.

Among the 19 with VSD, 10 were simple and 9 were complex and in 11 with ASD, 10 were simple and 1 was complex.

Table 2 shows that out of 32 female patients, congenital heart defects as single was present in 21 (65.6%), complex in 5 (15.7%) and murmur in 6 (18.7%); whereas they were 15 (45.4%), 9 (27.3%) and 9 (27.3%) respectively in male patients.

In female, it is observed that atrial and ventricular septal defects were 6 and 5; but in male they were 3 and 4. Patent foramen ovale and atrio ventricular

Table 3: CHDs-Types-Single and Complex-Female & male patients

Serial No.	CHDs-Female-Single	CHDs-Male-Single
1	ASD	VSD
2	VSD	Patent foramen ovale
3	Dextrocardia	AVSD
4	Patent ductus arteriosus	Mitral valve prolapsed
5	VSD	ASD
6	AVSD	AVSD
7	Patent ductus arteriosus	ASD
8	ASD	VSD
9	VSD	VSD
10	Dextrocardia	MVP
11	ASD	VSD
12	VSD	ASD
13	ASD	Tetralogy of Fallot
14	ASD	Patent ductus arteriosus
15	AVSD	Atrio ventricular canal defect
16	Coarctation of aorta	-
17	VSD	-
18	Dextrocardia	-
19	ASD	-
20	Mitral valve prolapse	-
21	Tetralogy of Fallot	-
Serial No	**CHDs-Complex-Female**	
1	Ventricular septal defects (VSD), mild tricuspid regurgitation, cleft mitral valve	
2	Coarctation of aorta, bicuspid aortic valve	
3	Tetralogy of Fallot with hypoplastic pulmonary artery	
4	VSD, pulmonary artey hypoplasia	
5	Mitral valve prolapse with mitral regurgitation	
Serial No	**CHDs-Complex-Male**	
1	VSD, pulmonary stenosis, patent foramen ovale	
2	Mitral & aortic valve stenosis	
3	Sub aortic VSD	
4	Atrial septal defects(ASD), patent foramen ovale	
5	Double outlet right ventricle, VSD, pulmonary artey hypoplasia	
6	Pulmonary valve stenosis, VSD	
7	Transposition of great arteries, multiple VSD, single coronary artery	
8	Tetralogy of Fallot, patent ductus arteriosus, pulmonary atresia	
9	Pulmonary tricuspid stenosis, VSD, patent ductus arteriosus	

canal defects were not observed in female and in the males, dextrocardia and coarctation of aorta were not seen.

Discussion

From review, it is seen, that the incidence of CHDs is similar for all major ethnic groups (Mitchell et al 1971) and for males and females (Richards et al 1955); although a differential sex ratio for certain types of CHDs may exist. The reported sex ratio for CHD is 1:1 (Turnpenny and Ellard 2012). Preponderance of particular sex for certain types of CHDs such as ASD and Tetralogy of Fallot in females and coarctation of aorta and transposition of great vessels in males exist (Samanek 1994). An increase in the prevalence of CHDs in male has also been reported (Chadha et al 2001). In the present study, the sex ratio was 1.03:1 (33 male versus 32 female). The CHDs are found to be prevalent in female patients. The reason may be because of the types of CHDs or female conceptions might have survived. In the present study, ASD has occurred in 6 females versus 3 males. Tetralogy of Fallot was seen in one male and one female. Coarctation of aorta was seen in 2 Turner syndrome female and it may be because of its association to the chromosomal abnormality. Transposition of great vessels was present in a male. *VSD:* VSDs are the most common congenital cardiac anomalies in 30 to 60% of all newborns with a CHD or about 2 to 6 per 1000 births (Meberg et al 1994). The cause of VSD includes the incomplete looping of the heart during 24 to 28 days of development and errors in the NKX2.5 gene could also be the cause.VSD, in the present study was observed as single entity in 5 female and 4 male; when its occurrence in complex CHDs were included, then it was observed in 9 female and 10 male. The percentage occurrence was found to be 26% (19/73). ***ASD:*** As a group, ASDs are detected in 1 child per 1500 live births and are 30 to 40% of all CHDs in adults (Kaplan 1993). The ostium secundum ASD defect accounts for 7% of all CHDs and it shows a female preponderance, with a male:female ratio of 1:2. (Feldt et al 1971). A common genetic variation near a gene called MSX1 is strongly associated with the risk of ASD and the discovery of the particular gene is an important step forward; as it may lead to the better understanding of ASD (Rhodes 2013). In the present study, ASD as single entity has occurred in 6 female to 3 male and the ratio becomes 2:1 as mentioned in literature. As part of complex entity its occurrence was 7 female to 4 male. The percentage occurrence was 15.1% (15/73).

Patent foramen ovale: In approximately 25% of adults, the foramen ovale does not entirely seal and it is known as PFO, a type of ASD (Kumar 2007). The percentage occurrence in the present study was 4.1% (3/73).

AVSD or Atrioventricular Canal Defect (AVCD)

TBX, a T-box transcription factor is usually expressed during various areas of embryogenesis and is important in the development of proper chamber differentiation (Harrison 2004). The percentage occurrence in the present study was 6.8% (5/73).

Tetralogy of Fallot

It accounts for 7 to 10% of CHDs (CDC 2006). Tetralogy of Fallot occurs in approximately 400 per million live births. Its cause is thought to be due to environmental or genetic factors or a combination. It is associated with chromosome 22 deletions and DiGeorge syndrome. Specific genetic associations include: JAG 1, NKX2.5, ZFPM2, VEGF. It occurs slightly more often in males than in females. The percentage occurrence in the present study was 5.5% (4/73) and was present in 2 male and 2 female respectively.

Dextrocardia

The estimated incidence is approximately 1 in 12,000 people. The percentage occurrence in the present study was 4.1% (3/73).

Double outlet right ventricle

It affects between 1 to 3% of people born with CHD. Among the 73 CHDs, in the 52 cases were: VSD in 19/ ASD 11/ patent ductus arteriosus and AVSD 5 in each/ mitral valve prolapse and Tetralogy of Fallot 4 in each/ dextrocardia, patent foramen ovale, pulmonary artery hypoplasia and pulmonary valve stenosis 3 in each/ coarctation of aorta and pulmonary atresia 2 in each/ tricuspid regurgitation, cleft mitral valve, mitral valve stenosis, aortic valve stenosis, bicuspid aortic valve, double outlet right ventricle, single coronary artey, transposition of great vessels and tricuspid valve stenosis one in each. The percentage occurrence in the present study was 1.4% (3/73).

In the present study, even though for the total, the association could be elicited between the female patients and CHDs; in male patients the occurrence of the complex CHDs were associated.

The observed differences may be because of the sample size or sample selection.

Genetic counseling

CHDs are mostly under the category of multi-factorial disorders until and unless it has a definite cause such as single gene or chromosomal abnormality. The estimated empiric recurrence risk for congenital heart defects is 1 to 4% to unaffected parents having a second affected child and affected parent having affected child is 2% if father is affected and 6% if mother is affected (Turnpenny and Ellard 2012). Genetic counseling is a communication process on diagnosis, prognosis, risk of recurrence and medical management. Patients' families were referred to Cardiology for appropriate medical management and treatment.

Conclusion

The paper has reported the classified CHDs for the 65 patients. They were: single entity in 36 (55.5%) cases; complex in 14 (21.5%) and undifferentiated systolic murmur in 15 (23%). In the 32 female patients, congenital heart defects as single was present in 21 (65.6%), complex in 5 (15.7%) and murmur in 6 (18.7%); whereas they were 15 (45.4%), 9 (27.3%) and 9 (27.3%) respectively in male patients. In the present study, female patients and CHDs were associated; likewise male patients were associated to the complex CHDs.

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