TO IDENTIFY PATTERN OF CONGENITAL HEART DISEASES IN A NEWLY DEVELOPED TERTIARY CARE UNIT

Tayyaba Sehar, Abdul Malik Sheikh, Asma Kanwal

Rawalpindi Institute of Cardiology, Rawalpindi Pakistan

ABSTRACT

Objective: To determine the age distribution, gender and pattern of congenital heart diseases in pediatric and adult population in a newly developed tertiary care centre.

Study Design: Retrospective observational study.

Place and Duration of Study: Rawalpindi Institute of Cardiology, Rawalpindi, from Jan 2016 to Dec 2016.

Patients and Methods: Data of patients presenting for the first time in pediatric unit between Jan 2016 to Dec 2016 was collected. It consisted of 5128 echocardiographies, all done by consultant pediatric cardiologist. Demographical profile, gender predisposition and various types of heart defects were analyzed using SPSS version 17.

Results: Among enrolled patients 2461 (48%) were females and 2666 (52%) were male patients. The ages of patient ranged from newborn babies to adults upto 60 years of age. Out of five thousand one hundred twenty eight (5128) echocardiographies done, 82.9% had congenital heart defects and two hundred fifty five patients (5%) had acquired heart defects. Acyanotic heart defects were more common (72%). Among acyanotic heart defects Ventricular septal defect (VSD) was the most common lesion 1172 (27%) of all congenital heart defects. In cyanotic heart defects Tetrology of Fallot (TOF) is most common 468 (10.9%). 632 (54%) of VSD cases were male. Pulmonary stenosis was most common obstructive lesion 170 (4%) follow by Aortic stenosis 63 (1.5%). Mital valve prolapse was most common congenital valvular lesion 89 (2.1%). Dilated cardiomyopathy was most common congenital cardiomyopathy 129 (3%). Sixteen case of Eisenmenger syndrome were diagnosed with female preponderance (F:M 1.6:1).

Conclusion: Late presentation of VSD with complications and TOF were the common phenomenon in our setup. Identifying the pattern of diseases and their presentations can help to devise effective programme for early diagnosis and interventions in congenital and pediatric heart diseases.

Keywords: Acyanotic heart defects, Atrial septal defect, Congenital heart disease, Patent ductus arteriosus, Ventricular septal defect.

INTRODUCTION

Congenital heart diseases are the most common congenital defects present at birth. The exact incidence in our country is not known but universally it is considered to be 8/1000 live births excluding the congenital bicuspid aortic valve which can remain asymptomatic throughout life[1-4]. Congenital heart defects are seen in isolation or in association with syndromes. The defects can be broadly classified into cyanotic and acyanotic heart defects depending upon their physiology[5,6]. Delayed presentation of congenital heart defects and appearance of complications significantly increases the mortality and morbidity in pediatric age group[7,8]. Another important group of heart defects seen in pediatric age group is of acquired heart defects which require long term medical and surgical follow up.

Pediatric cardiology is a relatively newer field in a developing country like Pakistan. Many cases of congenital heart disease are diagnosed only incidentally as they comes across medical facility for some other issue. There is no national data registry in Pakistan so exact incidence is yet to be known. A meta-analysis by Linde *at al*[6] established that highest total birth prevalence of congenital heart defect was seen in Asia (9.3/1000)[9]. Rizvi *et al*[7] conducted a study in three

Correspondence: Dr Tayyaba Sehar, Senior Registrar, Rawalpindi Institute of Cardiology, Rawalpindi Pakistan
Email: drtayyabasehar@yahoo.com
Received: 04 May 2018; revised received: 08 Sep 2018; accepted: 18 Oct 2018

provinces of Pakistan and concluded the incidence of congenital heart defects in rural areas was 3.4/1000 population.

We started services of pediatric cardiology at RIC as an outpatient unit. It is the only public sector centre unit in this area catering patients from Potohar region of Punjab province (it forms the northern part of Punjab, bordering the western parts of Azad Kashmir and the southern parts of Khyber Pakhtunkhwa (fig-1). The population of these areas is 17M, 4.6.million and 26.4 million respectively. This is the only public sector hospital in this area for a population above 35 Million which provides diagnostic, interventional and surgical facilities for pediatric cardiac patients. Our catchment area is about 42,490 square miles. In our outdoor clinic, patients not only present for cardiac evaluation but we also receive referrals of admitted pediatric patients from tertiary care hospitals of twin cities for cardiac evaluation on regular basis. Limited data is available regarding spectrum of congenital heart diseases in this area. Our objective was to identify the spectrum of congenital heart defects presenting in pediatric cardiology outdoor along with age of patients at presentation and their gender distribution.

MATERIAL AND METHODS

Retrospective observational study done using data of patients presenting in pediatric cardiology department of Rawalpindi Institute of Cardiology. Data of patients presenting for the first time in pediatric unit between Jan 2016 to Dec 2016 was collected. It consisted of 5128 echocardiographies, all done by consultant pediatric cardiologist.

Patients visiting or referred to pediatric cardiology outdoor for echocardio-graphy were enrolled after taking informed consent. Non-probability consecutive sampling was used.

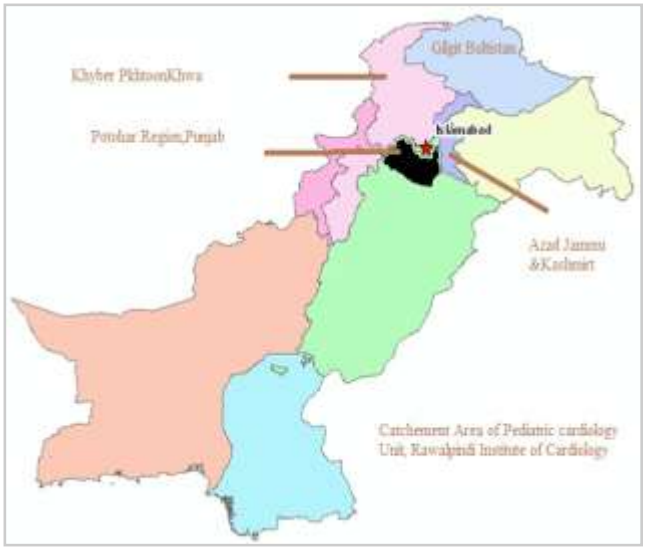
The inclusion of this study was Patient of any age with trans-thoracic echocardiography done by pediatric cardiologist at Rawalpindi Institute of cardiology confirming congenital heart defect.

Patients diagnosed as having acquired heart disease were exlusion of this study.

Demographical profile, gender predisposition and various types of heart defects were analyzed using SPSS 17 statistical software. Descriptive statistics including frequencies, mean and percentages were calculated.

RESULTS

Among enrolled patients 48% were females and 52% were male patients. The ages of patient ranged from newborn babies to adults upto 60 years of age. Out of five thousand one hundred twenty eight (5128) echocardiographies done, 82.9% had congenital heart defects and two hundred fifty five patients (5%) had acquired heart defects. 615 (12%) echocardiographies turned out normal and were excluded from the study. Acyanotic heart defects were more common (72%).



Punjab, Balochistan, Sindh, Khyber Pkhtoonkhwa.

Figure-1: Catchment area of pediatric cardiology unit, Rawalpindi institute of cardiology.

Among acyanotic heart defects Ventricular septal defect (VSD) was the most common lesion 27.5% (1172 cases) of all congenital heart defects followed by Patent ductus arteriosus (PDA) 13.4% (554 cases) and Atrial septal defect (ASD) 12.2% (511 cases) (table). Median age of presentation for ventricualr septal defect was 5 years (S.D ± 7.2 yrs). 54% (633) of VSD cases were male.

Our 18% (210) pateints had ventricular septal defect in isolation, while others had VSD in combination with another left to right shunts or cardiac lesion. 3.5% had ventricular septal defect with PDA, 0.6% has ASD, VSD, and PDA. Seven cases (0.2%) had VSD with ruptured sinus of valsalva, 0.2% (7) has VSD with ASD, 1.2% (51cases) of VSD with pulmonary stenosis, 0.3% (14) had coarctation with VSD were seen. Certain complications of VSD were also seen. Two cases of VSD presented with infective endocarditis, 59 cases (1.4%) had VSD with right ventricular outflow tract obstruction, 2 cases has left ventricular outflow tract obstruction with VSD, 73 cases (1.7%) had VSD with aortic regurgitation due to coronary cusp prolapse as shown in table.

Patent ductus arteriosus (PDA) was found second most common congenital cardiac lesion 13.4% (554) followed by Atrial septal defect (ASD) 12.2% (fig-2). PDA was found more common in females (F: M 57:43)The mean age of diagnosis was 5 years (SD ± 8.1), while maximum age of diagnosis was as late as 50 years of age.

Complete Arioventricualr septal defect was seen in 2.9% (123) cases with equal male: Female preponderance. In cyanotic heart defects Tetrology of Fallot (TOF) is most common 10.9% (468) followed by pulmonary atresia transposition of great arteries (TGA) and pulmonary atresia 1.5% (63). The age of presentation ranged from infancy to as late as 45 years. Mean age of presentation for Tetrology of Fallot was 5 years. Fifteen cases had TOF with absent pulmonary syndrome. 1.1% (46) cases had TGA with VSD, while 32 cases had TGA intact septum. Complex congenital heart diseases constitute around 2% of the cases. 0.9% (38 cases) had Univentricular heart, 0.9% had tricuspid atrsia, 0.2% were diagnosed as Mitral atresia and 2 cases of Hypoplastic left heart syndrome were diagnosed. 1.3% cases has anamolous pulmonaary venous drainage, 22 cases (0.5%) had total anamolous pulmonry veins while 0.8% had partial anamolous pulmonary veins. Supracardiac TAPVC was most common type of TAPVC seen as shown in fig-2.

Mitral valve prolpse was most common congenital valvular lesion seen in 2.1% (89 cases).

Table: Patient mean age and diagnosis.

Patient Mean Age and Diagnosis	Patient age in years
Diagnosis	Mean
ASD	7.5
ASD, PDA	0.1
ASD, VSD	0.1
ASD, VSD, PDA	0.1
ASD, VSD, PS	0.2
CAVSD, DILV, DOLV, PS	1.5
CAVSD, DORV, PA	1.0
CAVSD,DORV, Subpulmonic PS	3.0
CCTGA.CAVSD.PH	0.1
Coarctation	0.1
coarctation, PDA	0.3
DCMP	9.0
Dilated LV, sev dysfunction, PDA	4.0
Dilated LV, Severe dysfunction	2.0
DORV, CAVSD, PS	1.5
DORV, VSD, PH	3.0
Mild MR	12.0
Mitral atresia, ASD, VSD, DORV	0.1
Multiple ASDs	33.0
Non compacted LV	7.0
PA, VSD	13.0
PAVSD, MR	7.0
PDA	4.9
PDA, VSD	3.0
PDA. Coarctation	12.0
PH	3.4
PS	12.8
Rastelli (TGA, VSD, PS)	10.0
Septal hypertrophy	0.5
Subaortic ridge,AR	13.0
SV ASD, PAPVC	7.0
TGA, ASD	8.0
TGA, intact septum	0.1
TOF	9.3
TOF with APV	0.9
Trivial MR	12.0
Univenricular heart, DIRV	0.2
VSD	5.6
VSD, AR	11.0
VSD, PDA	0.1
VSD, PDA, AR, RVOT	8.0
VSD, PS	0.8
VSD, RVOT	7.9
VSDs	0.1

In 0.3% cases it was associated with Atrial septal defect. Pulmonary stenosis was most common

obstructive lesion accounting for 4% of cases. Isolated PS was seen in 2.7% cases with slight female preponderance (56.1:43.9). In 1.3% cases pulmonary stenosis was associated with atrial septal defect. Aortic stenosis was seen in 1.5% cases with male: Female ratio (3:1). 0.1% cases had supravalvular aortic stenosis. 1.2% patients had coarctation of the aorta with M:F 1:0.3. The age of diagnosis ranged from neonate to 28 years of age.

132 (3.1%) cases had dilated cardiomyopathy with non compacted LV. It was found more common in male patients (M:F 1:0.38) while 1.1% cases of resrtictive cardiomyopathy were diagnosed.

Sixteen case of Eisenmenger syndrome were diagnosed with female preponderance (fig-3) (F:M 1.6:1).

DISCUSSION

Congenital heart defects are problems with the heart's structure that are present at birth. These defects can involve the chamber or valves inside the heart or the blood vessels which carry blood to and from the heart. The defects can be in isolation or there combination different defects[10,11]. Physiologically they can be divided broadly into cyanotic and acyanotic heart defects. Acyanotic heart defects can remain asymptomatic throughout life but some of them warrant early intervention[12-15].

Congenital heart defects vary from simple defects with no symptoms to complex lesions that present with severe, life-threatening symptoms[16].

Congenital heart defects are the most common type of birth defect. They affect 8 out of every 1,000 newborns[17].

In developed world remarkable improvements are seen in terms of life expectancy and quality of life of children with congenital heart defects[18]. Better diagnostic facilities, early intervention and improved surgical outcomes have led to a change in spectrum of patients with congenital heart defects. This improved survival has also led to development of new specialities like GUCHD (Grown up with congenital heart defects). In recent years, a significant proportion of children born with CHD are expected to lead a normal, productive life[3,19]. Fetal echocardiography is also an important tool for early identification and referral of babies with congenital heart defects[20]. Antenatal detection of

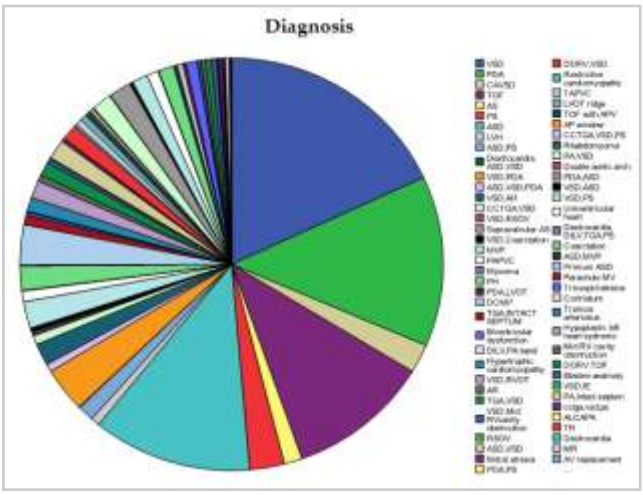


Figure-2: Pie chart of different congenital heart diseases.

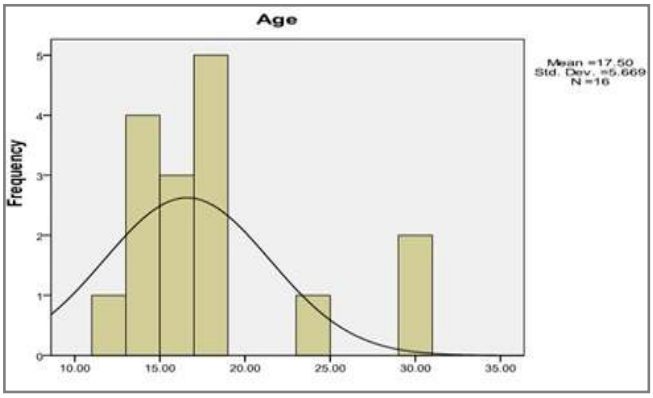


Figure-3: Age Vs frequency for eisenmenger syndrome.

CHD is now considered standard of care in developed countries, improving the final outcome. In developing countries like Pakistan, the diagnostic and intervention facilities for congenital heart defects are very scarce. Most of the patients remain undiagnosed and present later on with complications of the disease. The diagnosed cases only represent the tip of ice berg as less than 15 centers in the country are providing pediatric cardiac care for population of

above 200 million. The poor economic condition and suboptimal health coverage adds further to the problem. Majority of patients diagnosed are unable to bear cost of management leading to further delay in the treatment and contributing further in mor-bidity and mortality due to congenital heart defects. Co-morbidities especially undernutrition seriously compromise the results of any intervention in these children[6,21].

We lack any data registry at national level to exactly identify the burden of disease and make policies accordingly. Therefore exact incidence of congenital heart defects is unknown in our country. The only reported incidence is 4/1000 live births[11]. Considering high rate of consanguineous marriages, lack of antenatal detection, high neonatal mortality rate, and overburdened health care facilities, the incidence of congenital heart defects should be considered much higher than reported.

The pattern of congenital heart defects encountered in our study is in line with most f the published data. Left to right shunts constitute the major share of the disease. Ventricular septal defect remains the most common defect (fig-2). Majority of different studies at other tertiary care hospitals of the country have also concluded ventricular septal defects as most common congenital heart defect. However one study by Najma *et al*[19] showed Tetrology of Fallot as the most common congenital cardiac lesion. In our study Ventricular septal defect (VSD) was the most common lesion 27.5% of all congenital heart defects. Among these 18% pateints had ventricular septal defect only, while others had VSD in combination with another left to right shunts or cardiac lesion. Complications of VSD are also seen. Two cases of VSD presented with infective endocarditis, 59 cases (1.4%) had VSD with right ventricular outflow tract obstruction, 2 cases has left ventricular outflow tract obstruction with VSD, 73 cases (1.7%) had VSD with aortic regurgitation due to coronary cusp prolapse. This ratio is very high as it makes 10% of the total cases diagnosed as ventricular septal defect. Ijaz *et al*[21] reported Aortic valve prolpse in 6.7% cases in a study done at Khyber teaching hospital. Brauner *et al*[22], Lue *et al*[23], and Ando *et al*[24], reported aortic cusp prolapse in about 5% with isolated congenital VSD, 12%, 16% cases, respectively.

Patent ductus arteriosus (PDA) was found second most common congenital cardiac lesion 13.4% followed by Atrial septal defect (ASD) 12.2%. PDA was found more common in females (F : M 57.5 : 42.5) The mean age of diagnosis was 5 years, while maximum age of diagnosis was as late as 50 years of age (table-I). These findings are consistent with other studies showing left to right shunts are more prevalent in our population. In contrast to our study Sadiq *et al*[16] and Ahmad *et al*[18] reported Atrial septal defect is more common than PDA.

1.2% had coarctation of the aorta. This comparable to study in Taiwan by Mary K, Shan M which reported 1.1% patients with congenital heart defect had coarctation.

Among cyanotic heart defects Tetrology of Fallots is most common defect (10.9%) followed by TGA 2% and pulmonary atresia 1.5%. Mean age of presentation for Tetrology of Fallots was 5 years, while one patient presented for the first time at the age of 45 years with RV failure. This shows that delayed diagnosis and surgery can lead to life threatening co morbid conditions. Transposition of great arteries was second most common cyanotic heart defect seen. This is consistent with the study by Ijaz *et al* in 2015 as they reported transposition most common cyanotic defect (19% of cyanotic cardiac lesion) after TOF. Another study by Khaled Amro in Jordan also reported TGA accounting for 3.5% cases of Congenital heart defects[23]. In our study Transposition of great arteries with intact septum (0.8%) was found more common in male babies (M:F 24:8) while most of them presented after 20 days of life due to delayed referral.

We have seen 16 cases (0.4%) of Eisenmenger syndrome over a period of one year (fig-3), ages ranging from 13 to 35 years, most of them either presented for the first time at medical facility or didn't opt for surgery despite medical advice.

Fourteen patients had large VSDs now shunting right to left while in two cases patent ductus arteriosus led to irreversible pulmonary hypertension. A study by Ijaz *et al* showed 2% of patients presenting with cyanotic congenital heart defect had Eisemenger syndrome. This shows the unfortunate state of affairs. Thus delayed presentation not only contributes to poor quality of life but also leads to increase in mortality[24,25].

CONCLUSION

Congenital heart diseases are common in our society and often missed due to lack of health care facilities. Late diagnosis not only increase the morbidity and mortality but it is also associated with increased cost of treatment extra burden over already exhausted health system. Identification of incidence, prevalence and pattern of congenital heart defects is of fundamental importance to devise long term effective policies for their ealry diagnosis and prompt intervention.

LIMITATION OF STUDY

Study was limited to one tertiary care centre so it does not give the true incidence or prevalence of congenital heart diseases. Due to lack of awarenes for antenatal screening, many neonatal critical cardiac lesions often cdo not reach to the health care facilitiy and missed; yet, results are comparable with other international studies.

CONFLICT OF INTEREST

This study has no conflict of interest to be declared by any author.

REFERENCES

1. Fyler DC, Buckley LP, Hellenbrand WE, Cohn HE. Report of the New England regional 217 infant care program. Pediatrics 1980; 65: 375-461.
2. Abdullah R. what is the prevalence of Congenital Heart Disease? Ped Cardiol 1997; 18: 268.
3. Vaidyanathan B, Nair SB, Sundaram KR, Babu UK, Shivaprakasha K, Rao SG, et al. Malnutrition in children with congenital heart disease (CHD) determinants and short-term impact of corrective intervention. Indian Pediatr 2008; 45(7): 541-46.
4. Panni RZ. Earlier surgical intervention in congenital heart disease results in better outcome and resource utilization. BMC Health Services Res 2011; 11(1): 353-58.
5. Shrivastava S. Timing of surgery/catheter intervention in common congenital cardiac defects. Indian J Pediatr 2000; 67: 273-77.
6. Linde D, Konings EEM, Stager MA, Witsenburg, M, Helbing WA, Takkenberg JJM, et al. Birth prevalence of congenital heart diseases worldwide. A systematic Review and Meta analysis. J Am Coll Cardiol 2015; 58(21): 2241-47.
7. Rizvi SF, Mustafa G, Kundi A, Khan MA. Prevalence of congenital heart disease in rural 230 communities of Pakistan. J Ayub Med Coll Abbottabad 2015: 27(1): 124-27.
8. Thiene G. Anatomical and patho-physiological classification of congenital heart disease. Cardiovasc Physiol 2010; 19(5): 259-74.
9. Rychik J, Ayres N, Cuneo B, Gotteiner N, Hornberger L, Spevak PJ, et al. American Society of Echocardiography guidelines and standards for performance of the fetal echocardiogram. J Am Soc Echocardiogr 2004; 17(7): 803-10.
10. Brown KL, Ridout DA, Hoskote A, Verhulst L, Ricci M, Bull C. Delayed diagnosis of congenital heart disease worsens preoperative condition and outcome of surgery in neonates. Heart 2006; 92(9): 1298-1302.
11. Peterson C, Dawson A, Glidewell J, Garg LF, Braun KVN, Knapp MM, et al. Hospitalizations, costs, and mortality among infants with critical congenital heart disease: How important is timely detection? Birth Defects Res A Clin Mol Teratol 2013; 97: 664-72.
12. Rashid U, Qureshi AU, Hyder SN, Sadiq M. Pattern of congenital heart disease in a developing country tertiary care center: Factors associated with delayed diagnosis. Ann Pediatr Cardiol 2016; 9(3): 210-15.
13. Shirazi H, Haider N, Hassan M. Pattern of heart diseases in children. Ann Pak Inst Med Sci 2008; 4: 50-5.
14. Hassan I, Haleem AA, Bhutta ZA, Profile and risk factors for Congenital heart disease. J Pak Med Asoc 1997; 47(3): 78-81.
15. Ibrahim SA, Abdelrehman MH, Elshazali OH. Pattern and diagnosis of congenital heart disease in patients attending Ahmed Gasim Cardiac Centre. Sudan J Paediatr 2017; 17(2): 49-55.
16. Sadiq M, Roshan B, Khan A, Latif F, Bashir I, Sheikh SA. Pattern of paediatric heart disease in Pakistan. J Coll Physicians Surg Pak 2002; 12: 149-53.
17. Rahim F, Younas M, Gandapur AJ, Talat A. Pattern of congenital heart Diseases in children at tertiary care center in Peshawar. Pak J Med Sci 2003; 19(1): 19-22.
18. Ahmad R, Awan ZA, Bukshi F. A prevalence study of congenital heart disease in NWFP. Pak J Med. Sci 2002; 18(2): 95-98.
19. Patel N, Jawed S, Nigar N, Junaid F, Wadood AA, Abdullah F. Frequency and pattern of congenital heart defects in a tertiary care cardiac hospital of Karachi Pakistan. Pak J Med Sci 2002; 18: 95-98.
20. Kazmi U, Sadiq M, Hyder SN. Pattern of ventricular septal defects and associated complications. J Coll Physicians Surg Pak 2009: 19(6): 342-45.
21. Hussain I, Zeb S, Shah ST, Ghaffar A, Hisar, Irfan M. Spectrum of ventricular septal defects in patients with congenital heart disease. Pak Heart J 2014: 47(1): 28-33.
22. Brauner R, Birk E, Blieden L, Sahar G, Vidne BA. Surgical management of ventricular septal defect with aortic valve prolapse: Clinical considerations and results. Eur J Cardio-thoracic Surg 1995; 9: 315-19.
23. Lue HC, Sung TC, Hou SH, Wu MH, Cheng SJ, Chu SH, et al. Ventricular septal defect in Chinese with aortic valve prolapse and aortic regurgitation. Heart Vessels 1986; 2(2): 111-16.
24. Ando M, Takao A. Pathological anatomy of ventricular septal defect associated with aortic valve prolapse and regurgitation. Heart Vessels 1986; 2(2): 117-26.
25. Schneider DJ, Moore JW. Patent ductus arteriosus. Circulation 2006; 114(7): 1873-82.