

## A Study of Congenital anomalies of Heart

C Kishan Reddy<sup>1</sup>, M Dasarada Rami Reddy<sup>2</sup>, Mohd Inayatulla Khan<sup>3</sup>

1. Department of Anatomy, Prathima Institute of Medical Sciences, Karimnagar, Telangana.
2. Lecturer, Dept of Anatomy, Fathima Institute of Medical Sciences Kadapa, Andhra Pradesh.
3. Department of Physiology, Rajiv Gandhi Institute of Medical Sciences (RIMS) Adilabad.

### Abstract

**Background:** Congenital Heart Disease [CHD] is the commonest congenital disorder in newborns. It is an important cause of infant death from birth defects. Prevalence of congenital heart diseases varies in different regions of the world owing to several environmental and genetic factors. With this background we tried to evaluate the occurrence of CHD in human cadavers.

**Methods:** A total of 76 cases [60 foetus and 16 Adults] human cadavers were studied for congenital heart defects. **Results:** Out of 60 foetus live born were 18 (30%) and still born were 42 (70%) congenital anomalies were found in 9 (12%) of the specimen out of which Congenital Heart Disease were found in 7 (9.34%) out of 75 specimens. The occurrence of CHD in male was 5 (71.43%) and female 2 (28.57%) out of 7 cases. The commonest congenital defect found was Atrial Septal Defect [ASD] 6 (85.71%). One case was found with isolated small ventricular septal defect with dextrocardia. **Conclusions:** Greater incidences of Congenital Heart diseases were found in still born infants. We should try to reduce the mortality rate by giving proper health education and prenatal screening of all pregnant women.

**Keywords:** Congenital Heart Disease [CHD], Anomalies, Atrial Septal Defect [ASD]

**Address for correspondence:** Dr C Kishan Reddy, Department of Anatomy, Prathima Institute of Medical Sciences, Nagunur Karimnagar. E mail: [kishanreddy132@gmail.com](mailto:kishanreddy132@gmail.com). Mobile: +919885810645.

Received on :05/09/2016 Revised :20/09/2016 Accepted : 22/09/2016

### Introduction

The definition of congenital heart diseases varies widely according to several epidemiological reports. Mitchell et al; [1] defined congenital heart disease as “A gross structural abnormality of the Heart or Intrathoracic great vessels that is actually of possibly of functional significance”. Accordingly, abnormalities of systemic veins such as persistent left superior vena cava or inferior vena cava-azygos continuity, Marfans syndrome, cardiomyopathies and congenital arrhythmias are not included. The etiology of congenital heart disease is largely unknown. Only 15% of cases of congenital heart disease can be traced to known cause [2] Several researches have shown that the occurrence of congenital heart disease is associated with human genes such as TBX5, NKX2.5, GATA-4, which have been confirmed in the pedigree of congenital heart disease by genetic testing.

[3,4] Several congenital heart diseases form a part of well recognized chromosomal aneuploidies which cause malformation syndromes. The prevalence of congenital heart disease in Down syndrome is 45%. [5] Other syndromes with congenital heart diseases include Trisomy 13, Trisomy 18, Turner syndrome, and DiGeorge syndrome. [2] Only 2 % of all cases of congenital heart disease can be attributed to known environmental factors. [6]

Maternal diseases such as diabetes mellitus and phenylketoneuria are now accepted as major risk factors for congenital heart disease. Other risk factors include obesity, alcohol use, rubella infections, use of drugs and exposure to organic solvents. Heart basically originates from the mesodermal germ layer appearing in the middle of 3<sup>rd</sup> week of development. All major septa develop between 27<sup>th</sup> -37<sup>th</sup> day of intrauterine life. By 4<sup>th</sup> -7<sup>th</sup> week the heart divides into a four chambered organ. [7] All major septa may be exposed to teratogens between 5<sup>th</sup> -8<sup>th</sup> week.

Septal anomalies may be isolated or a complex one, 20% causes may be associated with extra cardiac defects. [8] Atrial Septal Defect is common in females while coarctation of aorta is more common in males. [9] In 2001 the ACC Bethesda conference Task force 1 first group attempted to estimate the burden of congenital heart disease 3.51 cases of all forms of congenital heart disease per 1,000 adults and 0.52 cases of severe congenital heart disease per 1,000 adults. [4] Marelli et al from Canada found in 2000, a prevalence of congenital heart disease of 11.89 cases per 1000 children and 4.09 cases per 1000 adults. The prevalence of severe congenital heart disease was reported to be 1.45 cases per 1000 children and 0.38 cases per 1000 adults. [10] Worldwide research works are focused on understanding important functional biological mechanisms that are essential for normal embryogenesis and foetal maturation. The knowledge about the type of anomaly helps in estimating the problem and planning, not only in minimizing but also managing by investigations and treatment. The present work was undertaken to study the distribution of congenital anomalies of heart along with external anomalies, if any.

## Materials & Methods

The present study was carried out in the Department of Anatomy, Prathima Institute of Medical Sciences, Karimnagar during period from 2007-2015. The fresh cadavers for study were received from the Department of Obstetrics and Gynecology and Pediatrics of Prathima Institute of Medical Sciences.

Ethical permission was obtained from institutional ethical committee for the research. A total of 76 cases [60 foetus and 16 Adults] human cadavers were studied for congenital heart defects. A longitudinal parasternal incision was made from the suprasternal notch to xiphoid process, then extending up to pubic symphysis encircling the umbilicus. The two lateral incisions were made along the subcostal margin extending from the first incision. Other system anomalies were noted down, if present.

The thoracic cavity was opened by cutting the ribs at costochondral junction. The position of the heart and great vessels were observed in situ and recorded. Then the heart along with lungs and thymus are separated from the internal

surface of the body of the sternum by cutting the sternopericardial ligament. The Inferior vena cava, the descending aorta and the oesophagus are cut at their openings into the diaphragm. Then the specimen was lifted up in between the fingers and the trachea was cut just above the thymus to take out the specimen. Next, the specimen was squeezed and thoroughly washed under running water, to remove the blood clots and debris present. Then it was put in a tray containing 10% formalin for minimum 24 hours for fixation.

After fixation, the specimen was cleaned. The heart with great vessels was separated out by removing the other organs. Further dissection was done by following incisions.

- 1) The first coronal section was made anterior to the ascending aorta up to diaphragmatic surface.
- 2) Second slice through the heart by cutting through the superior vena cava and aorta towards inferior surface. The following structures were observed and findings are recorded accordingly –

- a) Interatrial septum,
- b) Interventricular septum
- c) Tricuspid and mitral valves
- d) Aortic and pulmonary valves

Any associated external anomalies, if present, were recorded.

## Results

Total numbers of cases studied during the period were 76 cases. Foetus 60 (78.95%) and 16 Adults (21.34%) out of 60 foetus live born were 18 (30%) and still born were 42 (70%). There were 21 (35%) preterm and 39 (65%) term cadavers used in the present study. The number of adult cadavers used in the study were 16 (21.33%) and male were 61 (81.33%) and female were 14 (18.67%) given in table 1.

Congenital anomalies were found in 9 (12%) of the specimen out of which Congenital Heart Disease were found in 7 (9.34%) out of 75 specimens. The occurrence of CHD in male was 5 (71.43%) and female 2 (28.57%) out of 7 cases. All the congenital anomalies were found in foetus none were found to exist in the Adults. In foetus also all the congenital heart anomalies were found in still born infants (16.67%) of the

total 42 still born cases under the study details given in the table 2.

The commonest congenital defect found was Atrial Septal Defect [ASD] 6 (85.71%). Out of 6 cases 4 cases were found in male and 2 were found in female. One case was found with isolated small Ventricular Septal Defect with

Dextrocardia including an associated congenital abnormality called Dandy Walker syndrome which includes rudimentary cerebellar hemispheres and abnormal middle and posterior cranial fossa given in table 3.

Table 1: showing the characteristics of the specimen included in the study

	Liveborn	Stillborn	Preterm	Term	Adult	Male	Female
No. of Cases	18	42	21	39	16	61	14
Total	18	42	21	39	16	61	14
Percent	30	70	35	65	21.33	81.33	18.67

Table -2 Frequency of congenital anomalies of heart

Births	No. of cases	No. of CHD	Percentage (%)
Total	75	7	9.3
Live born	18	-	Nil
Still born	42	7	16.67
Adult	16	Nil	Nil

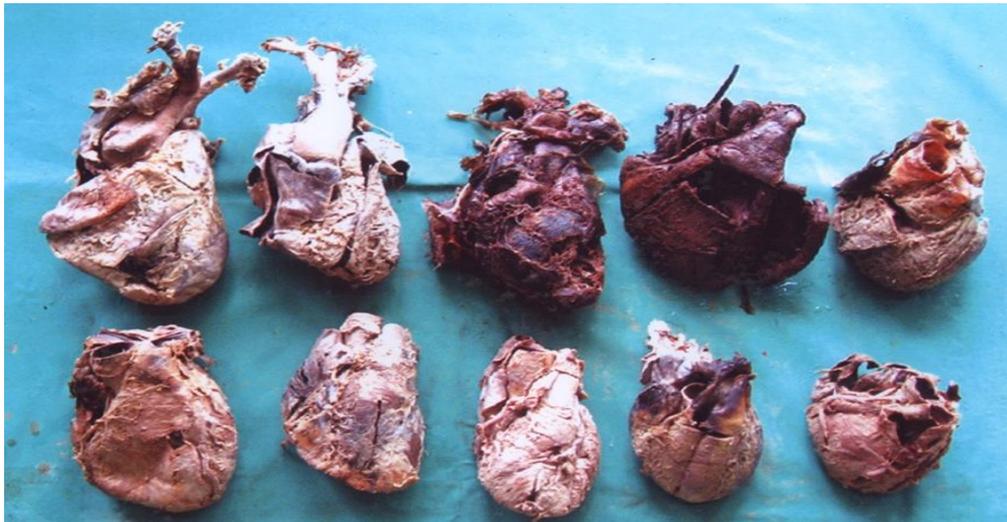
Table 3: Types of congenital heart diseases and associated defects

CHD	No. of cases	Associated defects	Male	Female	Percentage
ASD (Atrial septal Defect)	6	-	4	2	85.71
Dextrocardia with small ventricular septal defect	1	Dandy walker syndrome	1	-	14.28

Figure 1: showing some collected foetal hearts.



**Figure 2:** showing some collected adult hearts



## Discussion

Any disruption in the natural process of organogenesis occurring before birth leads to various malformations of heart. CHD signifies structural and functional heart diseases that are present at birth, even if it is discovered much later. The first 2-8 weeks of gestation is considered as a critical period for embryo heart development and it is a high-risk period for development of cardiac malformations. If exposure to risk factors for Congenital Heart Disease occurs during the first trimester of pregnancy and 3 months before pregnancy the foetus may suffer from cardiovascular malformations<sup>[11, 12]</sup> This study tried to evaluate whether the cause of death in still born and pre-term infants was Congenital Heart Disease. The prevalence of congenital heart disease is common in our population. In a study done by S L Chadha et al;<sup>[13]</sup> in which they conducted a community based survey of congenital heart disease in Delhi, India. They found overall prevalence of 4.2/1000 (4.6/1000 in boys and 3.7/1000 in girls). The prevalence rate was slightly higher than other studies carried out in the country. The most common lesion was Ventricular Septal Defect [VSD] (46%), Atrial Septal Defect (18%), Patent Ductus Arteriosus (14%), Fallot's Tetralogy (10%), Aortic Stenosis (4%), and Pulmonary Stenosis (4%) where as in our present study we found the commonest lesion were the Atrial Septal Defect however, our study involved only collected

samples. One study De Galan-Roosen AE et al<sup>[14]</sup> found that congenital defects contributed to 17.8% of perinatal mortality. Ravi Kumar et al<sup>[15]</sup> found that in India congenital heart diseases caused perinatal mortality up to 60% in still born infants. The present study shows that 16.67% of the still born babies had Congenital Heart Defects. Estimates of birth prevalence of moderate to severe congenital heart diseases are considerable more consistent than the estimates reported on the total birth prevalence. Most studies report of 1.5 cases per 1000 live births for severe congenital heart disease and similar number for moderately congenital heart disease.<sup>[16-18]</sup> Racial and ethnic variations in birth prevalence of congenital heart disease have been seen but factors needs to be elucidated<sup>[19,20]</sup> Geographical differences in birth prevalence are slightly difficult to evaluate. Differences in quality and access to health care could lead to large sampling bias, especially in less developed countries<sup>[18,20]</sup> Some congenital heart anomalies occur more often in newborn boys than in girls<sup>[3]</sup> In our present study we found CHD in male was 5 (71.43%) and female 2 (28.57%) out of 7 cases. Clearly more number of cases found in male as compared to females, but in female occurrence of secundum Atrial Septal Defect, Mitral Valve prolapsed, Persistent Ductus Arteriosus whereas male predominance is seen in cases of transposition of great arteries, Aortic Valve Stenosis, Coarctation, Tetralogy of Fallot and other defects which involve cardiac outflow.<sup>[21-23]</sup> In our study we found Atrial Septal Defect

[ASD] 6 (85.71%) out of 6 cases 4 cases were found in male and 2 were found in female. ASD was the commonest congenital anomaly seen by some other studies Atrial Septal Defect is often isolated, although may be associated with Downs Syndrome, anomalous Right Pulmonary Vein, pulmonary valvular stenosis VSD, persistent left superior venacava or mitral valve prolapse. [24] Among the foetus studies one case was found with isolated small ventricular septal defect with dextrocardia including an associated congenital abnormality called Dandy Walker syndrome. According to Fauci et al [9] lone dextrocardia is usually associated with complex multiple anomalies.

## Conclusion

Occurrences of Congenital anomalies are now increasing in our society. Among the congenital anomalies, Congenital Heart diseases are commonly found and they are also an important cause of infant mortality. Greater incidence of Congenital Heart diseases were found in still born infants. We should try to reduce the mortality rate by giving proper health education and prenatal screening of all pregnant women. Counseling should be done to the parents regarding avoidance of consanguineous marriages.

**Conflict of Interest:** None declared

**Source of Support:** Nil

**Ethical Permission:** Obtained

## References

- Mitchell, S. C., Korones, S. B., Berendes, H. W. Congenital heart disease in 56,109 births. Incidence and natural history. *Circulation* 1971; 43: 323–32.
- Nora, J., Berg, K. Nora, A. *Cardiovascular Diseases Genetics, Epidemiology and Prevention* 1991; Oxford University Press, New York.
- Samánek, M. Children with congenital heart disease: probability of natural survival. *Pediatr Cardiol* 1992; 13:152–58.
- Warnes, C. A. The adult with congenital heart disease: born to be bad? *J Am Coll Cardiol* 2005; 46:1–8.
- Vis, J. C. et al. Down syndrome: a cardiovascular perspective. *J Intellect Disabil. Res* 2009; 53: 419–25.
- Kuciene, R., Dulskiene, V. Selected environmental risk factors and congenital heart defects. *Medicina Kaunas* 2008; 44: 827–32.
- Sadler T.W. *Langman's Medical Embryology*, 9<sup>th</sup> Ed. Williams and Wilkins, Baltimore, Hongkong, London Sydney 2004; 224-29.
- Kasturi L, Kulakarni A.V., congenital heart disease clinical spectrum. *Indian Pediatr* 1999; 36:953.
- Fauci, Braunwald, Isselbacher, Wilsum, MastinKesper, Hauser, Longo Harrison, *Principles of Internal Medicine* Mc. Graw Hill, Newyork 14<sup>th</sup> Ed Mc; 1998.
- Marelli, A. J., Mackie, A. S., Ionescu-Iltu, R., Rahme, E. Pilote, L. Congenital heart disease in the general population: changing prevalence and age distribution. *Circulation* 2007; 115: 163–72.
- Carmichael SL, Shaw GM. Maternal life event stress and congenital anomalies. *Epidemiology* 2000; 11: 30–35.
- Jenkins KJ, Correa A, Feinstein JA, et al. Non inherited risk factors and congenital cardiovascular defects: current knowledge, a scientific statement from the American Heart Association Council on Cardiovascular Disease in the Young endorsed by the American Academy of Pediatrics. *Circulation* 2007; 115: 2995–3014.
- Chadha, S.L., Singh, N. Shukla, D.K. Epidemiological study of congenital heart disease. *Indian J Pediatr* 2001; 68: 507-510.
- De Galan-Roosen AE, Kuijper JC, Meershoek AP, Van Velzen D Contribution of congenital malformations to perinatal mortality. A 10 years prospective regional study in The Netherlands. *Eur J obstet Gynecol Reprod Biol* 1998; 80(1): 55-61.
- Ravi kumar M, Bhat BV., Early neonatal mortality in an intramural birth cohort at a tertiary care hospital *Indian J Pediatr* 1996;63:785-789.
- Mason, C. A., Kirby, R. S., Sever, L. E. Langlois, P. H. Prevalence is the preferred measure of frequency of birth defects *Birth Defects Res. Part A Clin Mol Teratol* 2005; 73: 690–692.
- Hoffman, J. I. & Kaplan, S. The incidence of congenital heart disease. *J Am Coll Cardiol* 2002; 39: 1890–1900.
- Bernier, P. L., Stefanescu, A., Samoukovic, G. Tchervenkov, C. I. The challenge of congenital heart disease worldwide epidemiologic and demographic facts. *Semin Thorac Cardiovasc Surg Pediatr Card Surg Annu* 2010; 13:26–34.
- Pradat P., Francannet C., Harris J. A. Robert, E. The epidemiology of cardiovascular defects, part I: a study based on data from three large registries of congenital malformations. *PediatrCardiol*2003; 24: 195–221.
- Nembhard, W. N., wang, T., Loscalzo, M. L. Salemi, J. L. variation in the prevalence of congenital heart defects by maternal race/ethnicity and infant sex. *J Pediatr*2010; 156:259–264.
- Wu, M. H. et al. Prevalence of congenital heart disease at live birth in Taiwan. *J Pediatr*;156:782–785.
- Robida A, Folger, G. M Hajar, H. A. Incidence of congenital heart disease in Qatari children. *Int J Cardiol* 1997;60: 19–22.
- Roguin, N. et al. High prevalence of muscular ventricular septal defect in neonates. *J Am Coll Cardiol* 1995;26: 1545–48.
- Bhupinder Singh, Malleth Kariyappa, Manjunath C. Halt-Oram Syndrome associated with double outlet right ventricle a rare association. *Ann Pediatr Cardiol* 2013; 6(1): 90-92.