RESEARCH ARTICLE

The Congenital Cardiovascular Abnormalities

Mobisson Samuel Kelechi[1], Wopara Iheanyichukwu[2], Nwachukwu Christian C[3] , Madu Emmanuel Chibuike[4]

[1]Department of Human Physiology, Faculty of Basic Medical Sciences, University of Calabar, Cross River State, Nigeria, [2]Department of Biochemistry, Faculty of Sciences, University of Port Harcourt, Rivers State, Nigeria,[3]Institute of Tropical Medicine and International Health, Charite - University Medicine Berlin, Germany, [4]Department of human physiology, faculty of basic medical sciences, Madonna University, Nigeria

ABSTRACT

The cardiovascular system (CVS) which includes the heart and blood vessels is a wide area for medical research and discussion in the world of science. The purpose of this review is to summarize the congenital anomalies associated with cardiovascular system. The congenital anomaly (birth defect) of cardiovascular system occur during embryonic development or during birth and may likely be due to the presence of teratogenic agents or may be as a result of genetic malformation. They are various kinds of cardiovascular congenital anomalies, ranging from mild to severe. The major cause of these have not really be ascertained. Though, scientists have attributed these anomalies to genetics, abnormal drugs intake during gestation, obesity and dirtiness during pregnancy, malnutrition and the heart malformation itself. Congenital anomalies of cardiovascular system also affect blood vessels and its treatment is dependent on the type or symptom that appears. Many scientists, anatomists, physiologists and physicians have taken critical study in this regard because it is very crucial as long as life is concern. Various treatments including surgery have been discovered. The symptoms of congenital anomalies of CVS differ in some individuals which may vary from mild to severe and even life threatening. In some children this defect may present no signs whereas others may exhibit shortness of breath, cyanosis, fainting, heart murmur, under-development of limbs and muscles, poor feeding or growth and respiratory infections. Congenital heart defects results to abnormal heart structure resulting in production of certain sounds called heart murmur. These can sometimes be detected by auscultation; however, not all heart murmurs are caused by congenital heart defects.

Key words: Cardiovascular System, Congenital Anomaly, Heart, Teratogenic Agents, Blood Vessels.

INTRODUCTION

The Cardiovascular system involves the heart and blood vessels which function to pump and circulate blood all through the body. The heart consists of two pumps (right and left) arranged in series for normal blood flows through the body. The left pump has a high pressure pump and refers to as systemic circulation (I.e. it pumps blood through the tissues except the lungs). It is done through the left ventricle. The right pump is a low pressure pump and is pulmonary circulation which consists of the right ventricle pumping blood to the lungs for oxygenation, [8].

Address for correspondence:
Mobisson, Samuel Kelechi, Department of Human Physiology, Faculty of Basic Medical Sciences, University of Calabar, Cross River State, Nigeria.

DOI: 10.33309/2638-7719.050102

The congenital cardiovascular system abnormalities make up the largest category of human birth defects, accounting for 1% of malformations among live born infants. They are diseases or defects associated with the structure of the heart and vessels which may result to changes in the normal flow of blood through the heart. Congenital heart defects are the most common type of birth defect [14]. There are many types of congenital heart defects (table 1). The most common defects involve the inside walls of the heart, the valves of the heart, or the large blood vessels that carry blood to and from the heart. Simple defects, such as a trial septal defect and ventricular septal defects may have no symptoms and may not require surgery [21]. Complex or critical defects such as hypoplastic left heart syndrome may have severe, life-threatening symptoms. Babies born with a critical congenital heart defect typically have low levels of oxygen soon after birth and need surgery within the first year of life [17]. The incidence among still births is 10 times as high. It is estimated that 8% of cardiac malformations are due to genetic factors, 2% are due to environmental agents and most are due to complex interplay between genetic and environmental influences [17]. Chromosomal abnormalities are associated with heart malformations, with 6% to 10% of newborns with this defect. Furthermore, 33% of children with chromosomal abnormalities have a congenital heart defect. Finally, cardiac malformations are associated with a number of genetic syndromes, including craniofacial abnormalities, such as Turner syndrome, Marfan syndrome, DiGeorge, Goldenhar and Down syndrome [17].

The cause of a congenital heart defects have not been fully ascertain but certain cases may be due to infections during pregnancy such as rubella virus, use of certain medications or drugs such as thalidomide, retinoic acid, alcohol or tobacco, parents being closely related, or poor nutritional status or obesity in the mother. Having a parent with a congenital heart defect is also a risk factor. Maternal diseases such as hypertension and insulin-dependent diabetes could as well be linked to cardiovascular defects [21].

Congenital heart defects are divided into two main groups: cyanotic heart defects and non-cyanotic heart defects [21]. This depends on whether the child has the potential to turn bluish in color. The problems may involve the interior walls of the heart, the heart valves, or the large blood vessels that lead to and from the heart. Congenital heart defects are partly preventable through rubella vaccination. Addition of iodine to salt and folic acid to certain food products has helped to minimize the defects [21]. Some defects do not need treatment. Others may be effectively treated with catheter based procedures or heart surgery. Occasionally a number of operations may be needed, or a heart transplant may be required. With appropriate treatment, outcomes are generally good, even with complex problems [13].

The purpose of this review is to summarize the causes, effects and possible symptoms and treatment of different kinds of congenital abnormalities of the cardiovascular system.

The development of the heart

The development of the heart is first noticed in the embryo around the eighteenth or nineteenth day of the intra-embryonic life in the cardiogenic area from where the mesenchymal cells aggregate themselves side by side as two longitudinal cellular strands known as cardiogenic cords. These cellular cords become canalized to form two thin walls of endothelial tubes called endothelial heart tubes. As lateral folds of the embryo progresses, the heart tubes gradually fuse together to form a single heart tube. This fusion begins at the cranial end of the tube and extends caudally to form a single heart tube. As the heart fuses, the external layer of the embryonic heart called the "primordial myocardium" is formed. At this stage, the developing heart is composed of a thin endothelial tube separated from a thick primordial myocardium by gelatinous connective tissues called cardiac jelly. The endothelial heart tube forms the endocardium, while the primordial myocardium forms the musculature of the heart (myocardium) and the epicardium (viscera epicardium), [17].

As the cephalocaudal folding progress, the heart and the pericardial cavity come to lie ventral to the foregut and caudal to the oropharyngeal membrane. At the same time, the single heart tube thus formed shows a series of dilations and constrictions to form bulbous cordis, primitive ventricle, primitive atrium and sinus venosus. The ventricle and the atrium communicate through a narrow atrioventricular canal. The sinus venosus prolongs to form right and left horns. The bulbous cordis lies at the arterial end of the heart dividing into dilated proximal one-third that forms part of the right ventricle, [17]. The middle third is called the "conus cordis" while the distal third is called the "truncus arteriosus". The truncus arteriosus forms the root of the proximal portion of the aorta and pulmonary artery. The sinus venosus lies at the venous end of the heart that receives the umbilical vein-vitelline vein and the common cardinal veins from the corium which serves as the primitive placenta. The bulbous cordis and ventricle bend upon itself forming a U-shaped bulbo-ventricular loop, and later an S-shaped heart is formed. As the primitive heart tube continues to grow in size, it tends to bend on itself such that the sinus venosus come to lie dorsal to the bulbous cordis, truncus arteriosus and ventricle. At this stage, the sinus venosus develops into lateral expansions called the right and left horns of sinus venosus, [17].

Congenital Abnormalities of the Cardiovascular System

Congenital abnormalities of the cardiovascular system affect about 6 to 8 babies in every 1000 live births [20]. It accounts for numerous numbers of deaths from a congenital structural

abnormality in newborns in the Western world, and is mostly associated with loss of fetus. In Australia, over 2000 babies are born with congenital heart defect (CHD) each year, with about half of these requiring surgery or catheter interventions. The other half has minor abnormalities (negligible valve lesions or extremely little ventricular or atrial septal defects) devoid of useful impact and barely affects wellbeing or necessitates intervention [11]. More patients with CHD need treatment each year than those with other significant conditions like childhood cancer or cystic fibrosis [11]. About a half of the patients that need treatment will require surgery within the first year of life. Majority of the infants and children that requires single interventions may expect to lead a near normal life. A little number of infants with multifaceted lesions requires multiple surgical procedures, intensive support and close monitoring during the first few years of life, although their quality of life may still be good. Many patients with complex lesions are reaching adult life if the surgical procedures are successful and the population of adults' with CHD now exceeds the number of children with structural heart abnormality [11].

Causes of Congenital Cardiovascular Abnormalities

The major cause of congenital heart defects is unknown. In recent development, the complex genetic and inheritance of CHD remains partly understood. Previously, the conditions were extremely worst since a lot of children with CHD did not live to age of reproduction and fetal echocardiography was not available. Majority of the cases then were multi-factorial in origin and is due to both genetic predisposition and environmental factors. Known genetic causes of heart disease includes inherited chromosomal abnormalities such as trisomy 21, 13 and 18, in addition to variety of recently identified genetic point mutations, point deletions and additional genetic deformities as implicated in syndromes like CATCH 22, familial ASD with heart block, Alagille syndrome, Noonan syndrome and lots of more. Known ante-natal environmental factors involve maternal infections (*Rubella*), drugs (alcohol, hydantoin, lithium and thalidomide) and maternal illness (diabetes mellitus, phenylketonuria and systemic lupus erythematosus), [1].An increased level of corticotrophin-releasing hormone (CRH) in the blood has been shown in elevated stressful conditions. Furthermore, it has been documented that there is an association between increased serum concentration of CRH and preterm labor or congenital heart defect in the baby. It was opined that the elevated stress-linked hormones may likely have effect on both fetal growth and development and maternal blood pressure, [2]. The parts of cardiac mal-development that sustain intrauterine circulation compromise the range of congenital heart defect (CHD). The anatomic attributes of many CHD in humans have been analyzed carefully. Genetic abnormalities and null mutations have targeted the cardiovascular system and identified the abnormalities in cardiovascular ontogeny as a principal cause of embryonic disorder, (table 1), [19]. Presently, it is known through ultrasound and fetal echocardiographic studies that the occurrence is very much higher than that normally speculated previously. Prenatal ultrasound assessment has also identified cardiac arrhythmias and myocardial dysfunction as major causes of fetal morbidity and mortality. The cardiac anomalies and arrhythmias are the most common causes of non-immune hydrops-fetalis. About 50% of deaths associated with congenital heart defects occur within 6 months and 80% by 1 year of age [9]. Therefore, it is indicated that neonate should be close monitored on the risk of heart disease, [7].

Table 1. Classification of different kinds of congenital cardiovascular abnormalities based on Pathogenetic mechanisms [5]

1. Abnormalities of Mesenchymal Tissue Migration (conotruncal defects)
Subarterial ventricular septal defects
Aortopulmonary window(AP window)
Double-outlet right ventricle(DORV)
Tetralogy of Fallot (TOF)
D- Transposition of the great vessels (DTGA)
Truncusarteriosus communis
Interrupted aortic arch, type B
Pulmonary atresia with ventricular septal defect (VSD)
2. Altered Cardiac Hemodynamics
Coarctation of aorto with intact ventricular septum
Hypoplastic left heart syndrome (HLHS)
Aortic valvular stenosis (AS)
Interrupted aortic arch, type A
Atrial septal defect, secundum type (20ASD)
Pulmonary atresia without ventricular septal defect
Perimembranous ventricular septal defect (pmVSD)
3. Abnormalities in Programmed Cell Death
Muscular ventricular septal defect
Ebstein's anomaly
4. Abnormalities of Extracellular Matrix
Endocardial cushion defects (AV canal defects)
5. Targeted Growth Defects
Total anomalous pulmonary venous return (TAPVC)
Partial anomalous pulmonary venous return Single atrium

Presented in table 1 above are different kinds of congenital cardiovascular abnormalities based on Pathogenetic mechanisms

Diagnosis of Congenital Cardiovascular Abnormalities

Family History

An in-depth history of the family covering no less than three generations must be gathered during the examination of any child with CHD. The likelihood of consanguinity must be ascertained because it is an indication for autosomal recessive inheritance and a known cause for elevated risk of

occurring again. The chances of increase risk of recurrence may happen if close family members are also affected, [3]. Family members of patients with CHD syndrome should be thoroughly examined for negligible manifestations of the disorder. It is principally essential in people with mandolin syndromes that are identified to have an elevated degree of variability of expression (e.g. Kartagener syndrome, Noonan syndrome, hypertelorism-hypospadias syndrome and Waardenburg syndrome), [18].

Clinical Features of Congenital Cardiovascular Abnormalities

Clinical manifestations of congenital cardiovascular defect differ based on the kind and severity of the defect, [15]. The presenting characteristics of CHD in neonate are cyanosis, heart failure, survival failure and an abnormal clinical sign noticed on regular test, [4]. The normal presenting characteristics in infancy and childhood include, cyanosis, clubbing of the digits, heart murmur, syncope, squatting episodes, heart failure, arrhythmia and failure to survive **[5]**. In newborn cyanosis is the key clinical sign. It is the major symptom of the most common types of the cyanotic congenital cardiac ailment which is symptomatic in the newborn. If cyanosis due to congenital cardiac illness is not indicated, the newborn may experience quick and severe cardiovascular decomposition. If the newborn has arterial oxygen saturation over 85%, cyanosis may be relatively difficult to notice through visual examination. Pulse oximetry should be used to measure oxygen saturation. The Pulse oximetry should be carried out on the right hand, because the aortic arch usually obtains blood through the ascending aorta and on any foot, they obtains blood via the descending aorta. Many conditions are related through diverse relationships in oxygen saturation between the upper and lower body and knowing this relationship aids to recognize the exact disorder that resulted to cyanosis[16].A pulse oximetry screen is done on an infant within the first 24 h of life and can simply and rapidly verify if an infant has CHD. This method was developed in the early 1970's, and is reliant on the diverse absorption spectrum that exists between oxygenated and deoxygenated hemoglobin, [16].

Management of congenital cardiovascular abnormalities

The first step of managing congenital cardiovascular abnormalities is through prevention. The most possible ways of preventing these conditions is via abolishment of various risk factors such as indulging in harmful substances like drugs and other diets that may cause defects in development of fetus. Expectant mothers should receive Vaccines, indulge in exercise and should be placed on folic acid and other vitamins before conceiving. There should be identification of subclinical disease and immediately treat of previous clinical presentations to prevent progression of disease [10].

Treatment of congenital cardiovascular abnormalities

Commencement of medical treatment in a newborn with a severe congenital cardiovascular disorder is required to avert and repeal clinical deterioration and complications. The common approach must follow the standard guiding principle for management of a seriously ill newborn, such as; oxygen supplementation in cyanotic newborn and mechanical ventilation and sedation in newborn with decrease systemic perfusion [12].

Close monitoring of fluid levels and urine production is vital when managing newborns with a severe congenital heart disorder**.** Normally, within 24 or 48 hours of extra-uterine life, a newborn with CHD, show similar fluid, glucose, and electrolyte requirements as infants with no congenital heart defect. Although, based on the type of defect. Nevertheless, fluid and electrolyte management may vary considerably in the neonatal stage. Signs and symptoms of heart failure may develop as a result of rising pulmonary blood flow, decreased systemic output and compensatory sodium and water retention. Free water restriction and diuretic treatment are needed to limit total body water and sodium [10].

Surgery

Normally, prostaglandin E1 (PGE1) is given to a cyanotic newborn with duct dependent condition in addition to further supportive measure. Surgery should be planned on a semi-elective basis after comprehensive evaluation is concluded. The only efficient treatment of obstructed total anomalous pulmonary venous return in neonates is surgery and must be done immediately the diagnosis is ascertained**.** Patients with d-transposition of the great arteries who have restrictive patent foramen ovale mostly require emergent balloon atrial septostomy due to profound hypoxemia. After successful septostomy these newborn are relatively stable and curative surgery should be postponed pending any end-organic damage resolves. A newborn that show congestive heart failure and shock due to left heart obstructive lesions (e.g., interrupted aortic arch) also can be stabilized by the giving PGE1 and other supportive care. Surgery is performed for more complex defects or when catheterization cannot correct the defect. But some defects may require a combination of surgery and catheterization. The kind of surgery will depend on what defect the child has. Some congenital heart defects can be completely repaired with one surgery. Defects that are more complex often require staged surgeries over time [6].

CONCLUSION

Congenital Heart Defect is one of the leading causes of hospitalization and eventual death in early childhood. Recent advances in cardiology has really reduced the complications and unfortunate death associated with this abnormalities.

Today, the pediatric cardiology discipline takes care of pediatric cardiac patients from infant to adult congenital. But the laboratory cardiac catheterization has obviously changed from a diagnostic facility to a place for rendering perfect treatment to almost 65% of uncomplicated CHDs with no scar on the chest. Since improved medical interventions have extensively elevated the rate of survival for infants and children, there has been an elevated focus on the quality of life of these persons and their relatives. Current improvements in the field of Pediatric cardiac surgery and Pediatric Cardiology particularly non-surgical trans-catheter interventions have improved the pre-operative or pre-procedure diagnostic workup of the cardiac malformations. Therefore, the physicians and surgeons taking care of Pediatric Cardiac patients should harmonize with one another in the care and wellbeing of the sick babies and children.

REFERENCES

1. Anderson, R. H. *et al* (1987). "Fetal circulation and circulatory changes at birth". Paediatric Cardiology. Churchill Livingstone; 2:109.
2. Artman, M., *et al*. (2002b). "Neonatal Cardiology". The McGraw-Hill Companies Medical Publishing Division.
3. Bosi, G. (2003). "IMER Working Party". Temporal variability in birth prevalence of congenital heart defects as recorded by a general birth defects registry. *Journal of Pediatrics*; 142:690-8.
4. Camm, A. J. (2005). Cardiovascular Disease (6th edition). Kumar & Clark Clinical Medicine. Edinburgh Elsevier Saunders; 725-872.
5. Clark, E. B. (1986). "Mechanisms in the pathogenisis of congenital cardiac malformations". Genetics of cardiovascular disease; 3-11.
6. Freda D. K., *et al*. (2004). "Psychoneuroimmunology in Critically Ill Patients". American Association of critical care Nurses. 121-126.
7. Friedman, W. F., *et al*. (2001). "Congenital Heart Disease in Infancy and Childhood", Heart Disease (6th edition). W. B Saunders Company Philadelphia London, New York, St Louis Sydney Toronto; 1505-91.
8. Guyton, A. C. & Hall, J. E. (Eds.). *Textbook of Medical Physiology (11th edition)*. Philadelphia W. B. Saunders Publishers, 2004; 543- 603.
9. Hoffman, J.L. & Kaplan, S. (2002). "*The incidence of congenital heart disease"*.
10. Johnson, K. *et al*. (2006). "Recommendations to improve preconception health and health care" in the United States. MMWR Recomm Rep. 1-23.
11. Khairy, P., Ionescu-Ittu, R.&Mackie, A. S. (2010). Changing mortality in congenital heart disease. *Journal of American College of Cardiology*; 56: 1149-1157.
12. Lewis, A. B., *et al*. (1981). "Side effect of prostaglandin E1 in infants with critical congenital heart disease". Circulation. 64:893-8.
13. Milunsky, A. (2011). *Genetic Disorders and the Fetus: Diagnosis, Prevention and Treatment. John Wiley & Sons.*
14. Mitchell, S. C. *et al*., (1971). "Congenital heart disease in 56,109 births" Incidence and natural history. Circulation. 43(3):323-32.
15. Noonan, J. A. (1878). "Association of congenital heart disease with syndromes or other defects". Pediatric Clinical American; 25:797-816.
16. Sadeghian, H. & Savand-Rooni Z. (2015). Fetal heart echocardiography of twins, Echocardiographic Atlas of adults Congenital Heart Dieses. *Springer International Publishing*. 461-463.
17. Sadler, T.W. (2010). *"Medical embryology, 11th edition"*.
18. Stephensen, S. S. (2004). Congenital cardiac malformations in Iceland from 1990 through 1999. Cardiol Young; 14:396-401.
19. Sullivan, I. D. (2002). Prenatal diagnosis of structural heart disease: does it make a difference to survival. The Heart; 87:405-6.
20. Wessels, M. W. & Willems, P. J. (2010). Genetic factors in non-syndromic congenital heart malformations. *Clinical Genetic;* 78: 103-123.
21. World Health Organization (2011). Global Atlas on Cardiovascular Disease Prevention and Control (PDF). World Health Organization in collaboration with the World Heart Federation and the World Stroke Organization. P p. 3, 60.

How to cite this article: Kelechi M S, Iheanyichukwu W, Christian N C, Chibuike M E. The Congenital Cardiovascular Abnormalities. J Community Prev Med 2022;5(1):06-10.
DOI: 10.33309/2638-7719.050102