



**Incidence of congenital heart disease among patients  
Referred to Echocardiography Unit at Cairo  
University Children Hospital (CUCH)**

*Thesis*

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بسم الله الرحمن الرحيم

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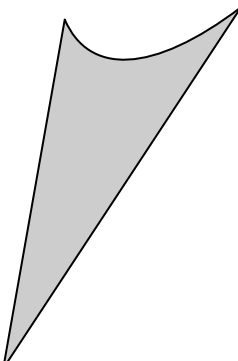
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# *Abstract*

**Key Words:** congenital heart disease— echocardiography

Observational study was conducted to determine the Incidence and relative frequency of congenital heart diseasediagnosed by echocardiography Unit in pediatric Hospital, Cairo University Children Hospital (CUCH) over a period of 1 year (1 January 2013- 31 December 2013).

Congenital heart diseases were the most frequent cardiac abnormalities (41.9%).

Majority of the cases (69.7%) were Acyanotic, remaining(30.3%) were cyanotic. Among the Acyanotic VSD (14.87 %\*) was the commonest 2<sup>nd</sup> was ASD (13.98 %\*) while in acyanoticgroup D-TGA (6.65 %\*) was the commonest followed by TOF (6.43 %\*).

Male preponderance,the most clinical presentation was Breathlessness and poor weight gain,majority of case was sever CHD are important observation made in the study.

\*, %: % of CHD.

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## List of Abbreviations

<b>ACHD</b>	Acyanotic congenital heart disease
<b>ASD</b>	Atrial septal defect
<b>APV</b>	Absent pulmonary valve
<b>AS</b>	Aortic stenosis
<b>AVSD</b>	Atrioventricular septal defect
<b>AVC</b>	Atrioventricular Canal
<b>BAV</b>	Bicuspid aortic valve
<b>BCAV</b>	Bicuspid aortic valve
<b>BT shunt</b>	Blalock–Taussig shunt
<b>CAD</b>	Coronary artery disease
<b>CCHD</b>	Cyanotic congenital heart disease
<b>ccTGA</b>	Congenitally corrected TGA
<b>CHD</b>	Congenital heart disease
<b>COA</b>	Coarctation of aorta
<b>CW</b>	Continuous wave
<b>DORV</b>	Double outlet right ventricle
<b>D-TGA</b>	Dextro-Transposition of the great arteries
<b>ECHDO</b>	European congenital heart disease organization
<b>EA</b>	Ebstein anomaly
<b>E/A ratio</b>	The ratio of the early (E) to late (A) ventricular filling velocities
<b>ED</b>	End diastolic
<b>ESPAP</b>	Estimated systolic pulmonary artery pressure
<b>EF</b>	Ejection fraction
<b>ET</b>	Ejection time
<b>FISH</b>	Fluorescence in situ hybridization
<b>ES</b>	End systolic
<b>FS</b>	Fractional shortening
<b>HLHS</b>	Hypoplastic left heart syndrome
<b>HR</b>	Heart rate
<b>ICT</b>	Isovolumic contraction time
<b>IRT</b>	Isovolumic relaxation time
<b>IVS</b>	Inter ventricular septum
<b>LA</b>	Left atrium
<b>LVEDD</b>	Left ventricle end-diastolic diameter
<b>LVEF</b>	Left ventricular ejection fraction
<b>LVESD</b>	Left ventricle end-systolic diameter
<b>LVIDd</b>	Left ventricle internal diameter in diastole
<b>LVIDs</b>	Left ventricle internal diameter in systole
<b>LV</b>	Left ventricle
<b>LVPW</b>	Left ventricle posterior wall
<b>LVOT</b>	Left ventricular outlet
<b>MDM</b>	Maternal diabetes mellitus
<b>MMHG</b>	MilliMeters of Mercury
<b>MPI</b>	Myocardial performance index
<b>MV</b>	Mitral valve
<b>NYHA</b>	New York Heart Association Classification
<b>PA</b>	Pulmonary artery

<b>PA</b>	Pulmonary atresia
<b>PAPVD</b>	Partial anomalous pulmonary venous drainage
<b>PBF</b>	Pulmonary blood flow
<b>PBS</b>	Pulmonary branch stenosis
<b>PDA</b>	Patent ductus arteriosus
<b>PFO</b>	Patent foramen ovale
<b>PGE<sub>1</sub></b>	Prostaglandin E1
<b>PH</b>	Pulmonary hypertension
<b>PPHN</b>	Persistent pulmonary hypertension
<b>PRF</b>	Pulse repetition frequency
<b>PS</b>	Pulmonary stenosis
<b>PTA</b>	Persistent truncus arteriosus
<b>PW</b>	Pulsed wave
<b>Qp:Qs</b>	Pulmonary-Systemic Flow Ratio
<b>RV</b>	Right ventricular
<b>RVOT</b>	Right ventricular outflow tract
<b>RHD</b>	Rheumatic heart disease
<b>TA</b>	Tricusped atresia
<b>TAPVR</b>	Total anomalies pulmonary venous return
<b>TCAV</b>	Tricuspid aortic valve
<b>TGA</b>	Transposition of great arteries
<b>TOGV</b>	Transposition of the great vessels
<b>TOF</b>	Tetralogy of Fallot
<b>TV</b>	Tricuspid valve
<b>TYPE 1a</b>	Normally related great vessels with pulmonary atresia
<b>TYPE 1b</b>	Normally related great vessels with pulmonary stenosis
<b>TYPE 2a</b>	D-transposed of the great arteries with pulmonary atresia
<b>TYPE 2b</b>	D-transposed of the great arteries with pulmonary stenosis
<b>TYPE 3a</b>	L-transposed of the great arteries with pulmonary atresia
<b>TYPE 1c</b>	Normally related great vessels
<b>TYPE 3c</b>	L-transposed of the great arteries
<b>UCAV</b>	Unicuspid aortic valve
<b>VPS</b>	Valvular pulmonary stenosis
<b>VSD</b>	Ventricular septal defect
<b>SAM</b>	Subaortic membrane
<b>SV</b>	Single ventricle
<b>SVD</b>	Single ventricle defect

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## INTRODUCTION

The normal heart is like a three store building (atria, ventricles, and great arteries): the atria are the platform, the ventricles are the first floor, and the great arteries are the second floor, connected to each other through valve orifices and totally separated by septa. The segments are disposed in such a way as to allow deoxygenated venous blood to go to the lungs through the pulmonary artery, and oxygenated venous blood to go to the systemic organs through the aorta. Small and great circulations are in sequence, with no communication with each other apart from the lung capillary network(*Thiene and Frescura, 2010*).

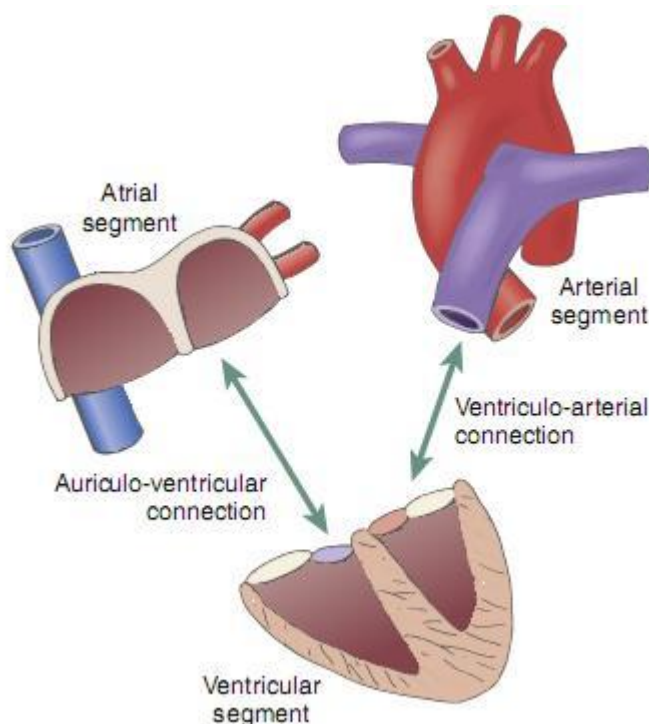


Figure {1}: Schema of segmental approach: heart is divided into three segments (atria, ventricles, and atreial trunks) and two junctions (atrio-ventricular and ventriculo-arterial junctions) (*Bettex&Chassot, 2011*).

Congenital is derived from the Latin con, together, and genitus, born. However the implication that CHD simply means "present at birth" require elaboration. The natural history begins before birth. The majority of congenital anomalies of the heart are present six week after conception, and most anomalies compatible with six months of intrauterine life permit live offspring at term. A given malformation may exist in relative harmony with the fetal circulation only to be modified considerably, at least physiologically, by the dramatic circulatory adjustments at birth. Weeks, months or years may elapse before the anomaly reveals itself as the "typical" clinical picture. Pathophysiological and structural changes continue or, conversely, the malformation may "vanish" (*Perloff, 1998*).

Congenital heart disease (CHD) is a complex problem which can affect the patient, the family, the society and the country. CHD describes a wide range of conditions resulting from abnormalities of the heart structure and/or function that is present from birth. The severities of conditions are classified as complex, moderate, and simple(*Marelli et al, 2007*). CHD is the most common birth defect, with approximately 9 in 1,000 new borns being affected(*Van der Linde et al, 2011*).Although CHD is the most common survivable birth defect, the etiology of most CHD remains unclear(*Yuan et al, 2013*).It represents one of the most common causes of death of children at different ages starting from abortion of the fetus, stillbirth, death of the patient at birth, during infancy, during early childhood, during late childhood and may at adulthood(*Ferencz et al, 2008*).

From a surgeon's perspective, CHD consists of shunts, obstructions, valvular regurgitation, or combinations of these. Operative interventions are ingenious and numerous, and they encompass complete or partial repairs, single and multistage palliative procedures, and cardiac transplantation(*Edwards, 2010*).

Cardiac imaging plays an important role in establish the diagnosis, interventional management;follow up after palliative or corrective surgery. Various imaging modalities for the diagnosis of CHD progress from plain chest radiograph, 2-dimensional echocardiography and conventional cardiac catheterization to noninvasive advanced cardiac imaging, 3- and 4- dimensional echocardiography, transesophagealechocardiography, cardiac MRI (cMRI), andmultidetector CT (MDCT). Echocardiography remains a first line noninvasive imaging tool for establishing the diagnosis and follow up in most patients. However, echocardiography has inherent limitations, including a limited acoustic window. This is particularly the case with postsurgical sternal wires and mediastinal scar tissue, and extracardiac vascular structures. Echocardiography is also an operator dependent imaging tool with poorer spatial resolution than CT or CT angiography(CTA)(*Bailliard et al,2008*).

CHD was considered a predominantly pediatric condition, as patients with complex forms seldom survived beyond childhood. However, patient care markedly improved, resulting in a growing and aging population. Indeed, adults now outnumber children with both severe and other types of CHD(*Marelli et al, 2007*).

Ultimately, the goals of pediatric research and clinical care are to maximize health and minimize symptomatology, disability, and dysfunction that may impact the lives of children with acute and chronic disease processes. Over the last several decades, new surgical techniques and advances in cardiopulmonary bypass, intensive care, interventional cardiac catheterization, non-invasive imaging, and medical therapies have significantly lowered neonatal mortality rates for children with the most complex congenital heart disease (CHD) (e.g. hypoplastic left heart disease) to less than 10% (*Tweddell et al, 2002*).

## **CARDIOVASCULAR EMBRYOLOGY**

Commitment to the cardiogenic cell lineage occurs early in development soon after gastrulation (the embryonic state following the blastula), approximately 48h following fertilization (*Khan, 2006*).

Heart is formed in a fashion where components are added in a sequence to an initial primary structure, the cardiac crescent. The first step in the heart development is the formation of two heart fields of precardiac mesoderm, which are situated on opposite sides of the primitive foregut situated at the embryonic midline and contain endocardial as well as myocardial precursor cells. The first or the posterior heart field contributes to the bulk of the left ventricle and the secondary or the anterior heart field forms the right ventricle, outflow tracts, sinus venosus and atrial chambers (*Bruneau, 2008*). The first evident assembly of the mesoderm-derived myocardial (or cardiogenic) plate is seen at 18 days, followed by initiation of heart tube formation at 22 days (*Mitchell et al, 2007*) which connects at the cephalic end with the developing aortic arch system, and caudally with the vitelline and umbilical venous systems (*Reller et al, 1991*).