Comparative Study Between Echocardiography and Autopsy Results of Congenital Heart Defects

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Background: Congenital heart disease is currently the world's leading birth defect, with incidence estimated at 8 per 1000 live births. Confidence in the images obtained using echocardiography has continued to increase, with many patients referred for corrective or palliative surgery on the basis of echocardiographic imaging alone. This review outlines the manner in which echocardiography is used to plan and guide congenital heart surgery or intervention, along with some of the advantages and disadvantages of which to be aware.
Aim: To assess the degree of harmonization of echocardiography with postmortem diagnosis in congenital heart disease.
Material and method: We examined the morphology of congenital heart diseases by autopsy in the Morphopathology Department of the County Hospital of Tîrgu Mureș in 2008 and 2009. We analyzed the components of the congenital heart disease and we compared the echocardiographic results with the autopsy results also.
Results: In 2008 and 2009 621 necropsies were carried out, from which 49, meaning 7.89% were diagnosed with simple or complex cardiac malformations; the male-female ration was 1:0.9. We found a few differences between the echocardiography and autopsy results on the following malformations: total anomalous pulmonary venous connection, double outlet ventricle, univentricular heart.
Conclusions: Echocardiography is accepted as the first-line imaging modality for diagnosing most types of congenital heart defects but in some difficult cases surgeons must always keep in mind the possibility of the presence of other heart malformation too.

Keywords: congenital heart disease, echocardiography, autopsy results

Introduction

Congenital heart diseases are the most common single group of congenital abnormalities accounting for about 30% of the total abnormalities [1]. In most patients, congenital heart disease occurs as an isolated malformation, but also about 33% have associated anomalies [2]. Worldwide congenital heart disease in children continues to be a major public problem with it's incidence in different studies varying from 1–17.5/1000 live births and 10% of spontaneously aborted fetus [3].

Echocardiography is well established as the first-line imaging technique for the diagnosis of all forms of congenital heart disease, most institutions continue to perform cardiac catheterization prior to complete repair of more complex defects.

Confidence in the images obtained using echocardiography has continued to increase, with many patients referred for corrective or palliative surgery on the basis of echocardiographic imaging alone [4].

This review outlines the manner in which echocardiography is used to plan and guide congenital heart surgery or intervention, along with some of the advantages and disadvantages (pitfalls) of which to be aware.

Material and methods

We examined the morphology of congenital heart diseases by autopsy in the Morphopathology Department of the County Hospital of Tîrgu Mureş in 2008 and 2009. We summarized cases of complex congenital heart disease, and split them in to their component parts. We analyzed components of congenital heart disease on their own and we also compared the echocardiographic results with the autopsy results. The echocardiographic results were obtained from the Pediatric Cardiology Department of the County Hospital of Tîrgu Mureş. The study meets the ethical requirements.

Results

In 2008 and 2009, 621 necropsies were carried out, from which 49, meaning 7.89% were diagnosed with simple or complex cardiac malformations; the male-female ratio was 1:0.9. The distribution of the patients with congenital heart disease on age groups was the following: in 2008, 63.6% of the patients were babies, 18.1% newborns, 18.3 children, in 2009 it was relatively homogeneous, the highest occurrence being in the case of babies (40.7%) and newborns (40.7%) (Table I).

By autopsy various type of congenital heart disease were diagnosed as mirrored in Table II. The most common congenital malformations were septal defects (either atrial septal defect (24.8%) or ventricular septal defect (13.5%)), transposition of the great arteries (9.0%), and patent ductus arteriosus (14%).

We found no differences between the echocardiographic results and autopsy results on the following malformations: dextrocardia, mezocardia, atrial septal defect, com-

Table I. Distribution on age groups of the patients with congenital heart disease in 2008 and 2009

AGE GROUP	2008	2009
New-born (under 1 month)	4	11
Baby (1 month-1 year)	14	11
1-3 years	2	3
3-7 years	1	1
Over 7 years	1	1

Table II. Patients' distribution according to the type of congenital heart disease

TYPE OF CONGENITAL HEART MALFORMATION	NUMBER OF CONGENITAL HEART DISEASE
PERSISTENCY OF ARTERIAL DUCT	16
ATRIAL SEPTAL DEFECT	33
VENTRICULAR SEPTAL DEFECT	18
AORTIC HYPOPLASIA	3
MITRAL ATRESIA	1
AORTIC ATRESIA	1
TRICUSPID ATRESIA	1
COMMON ATRIOVENTRICULAR CANAL	5
DOUBLE OUTLET RIGHT VENTRICLE	5
DOUBLE OUTLET LEFT VENTRICLE	3
SINGLE ATRIUM	1
TRANSPOSITION OF THE GREAT VESSELS	12
COMMON ARTERIAL TRUNK	1
SHONE'S SYNDROME	1
TETRALOGY OF FALLOT	4
PULMONARY STENOSIS	11
DEXTROCARDIA	2
MEZOCARDIA	1
DEXTROPOSITION OF THE AORTA	2
TOTAL ANOMALOUS PULMONARY VENOUS CONNECTION	1
CORD TRIATRIATUM	1
AORTIC COARCTATION	3
UNIVENTRICULAR HEART	1
TRICUSPID STENOSIS	1
PULMONARY ATRESIA	2
TRICUSPID VALVE AGENESIS	1
BICUSPID PULMONARY VALVE	1
ABERRANT MITRAL VALVE	1

mon atrioventricular canal, cor triatriatum, single atrium, transposition of the great vessels, common arterial trunk, aortic coarctation, tetralogy of Fallot, aortic hypoplasia, dextroposition of the aorta, mitral atresia, aortic atresia, tricuspid atresia, pulmonary atresia, pulmonary stenosis, tricuspid stenosis, tricuspid valve agenesis bicuspid pulmonary valve, aberrant mitral valve, Shone's syndrome.

We found some cases with partially different clinical and morphopathologic diagnosis:

- One 4 month old, male patient, diagnosed by echocardiography and clinical examination with atrial septal defect, cor triatriatum and a total anomalous pulmonary venous connection, supracardiac type. By autopsy releved that the left-sides pulmonary veins drained through a left-sided venous collector into the superior vena cava; the right-sided pulmonary veins drained through the right-sided venous collector directly into the hepatic vein. Our diagnosis was: situs solitus, atrial septal defect, cor triatriatum and total anomalous pulmonary venous connection, mixed type (Figures 1 and 2).
- In the case of a 5 years and 11 month old female patient the cardiologist diagnosed by echocardiography double inlet left ventricle with a rudimentary right ventricle type second chamber, situated anteriorly and to the left, transposition of great vessels, ventricular septal defect; the left-sided aorta arising from the outlet chamber without any obstruction and a right-sided pulmonary artery connected to the morphologically left ventricle. By autopsy morphologic findings included situs solitus of the viscera and atria; normal systemic and pulmonary venous connection, two atrioventricular valves, an indeterminated type of univentricular heart; both great vessels arose from the single ventricle; the only septal structure was a muscular outlet septum interposed between the subarterial outlet. Also we found transposition of the great vessels (Figure 3).
- In another case of a female newborn by echocardiography double outlet right ventricle, pulmonary stenosis, hypoplasia of pulmonary trunk and atrial septal defect was diagnosed. Pathologic examination identified situs solitus, double outlet left ventricle and both great arteries originate entirely or predominantly from the

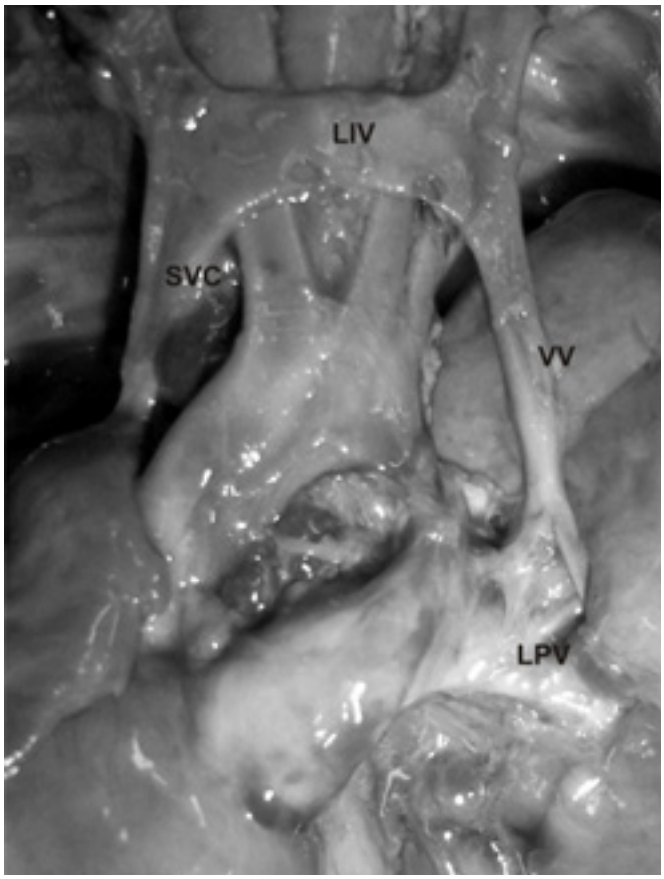

Fig. 1. Total anomalous pulmonary venous connection mixed type: the left-sides pulmonary veins drained through a left-sided venous collector into the superior vena cava (LIV – left inominate vein, VV – verticalis vein, LPV – left pulmonary vein, SVC – superior vena cava)

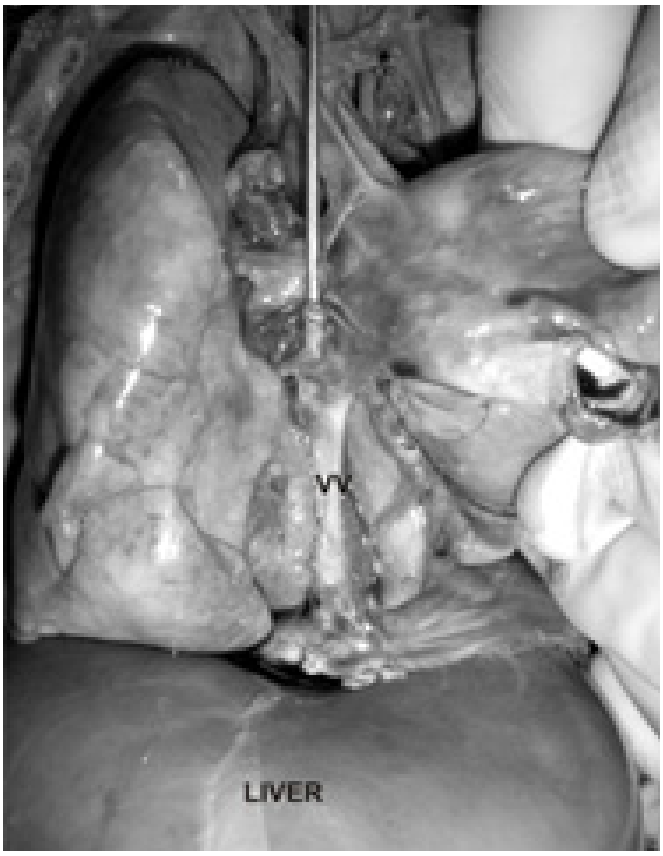

Fig. 2. Total anomalous pulmonary venous connection mixed type: the right-sided pulmonary veins drained through the right-sided venous collector into the vena hepatis (RPV – right pulmonary vein, VV – verticalis vein).

morphologic left ventricle. Pulmonary hypoplasia and atrial septal were observed too.

Discussions

The incidence of congenital heart disease in different studies varies from about 4/1,000 to 50/1,000 live births [5,6].

The relative frequency of different major forms of congenital heart disease also differs greatly from study to study. During a child's first year of life, the most common defects that are symptomatic include ventricular septal defect, transposition of the great vessels, tetralogy of Fallot, coarctation of the aorta, and hypoplastic left heart syndrome. Premature infants have an increased presentation of ventricular septal defect and patent ductus arteriosus [6,7,8].

In 2008-2009 in the Pathology Department of the County Hospital of Tîrgu Mureş 49 cases with simple or complex cardiac malformations were autopsied, the male-female ration was 1:0.9. As compared with other studies we found no differences related to the most common forms of congenital heart disease.

Echocardiography is accepted as the first-line imaging modality for diagnosis of both simple and complex congenital heart defects [9,10,11,12].

We found three different results between the echocardiography and autopsy examinations on the following malformations: total anomalous pulmonary venous connection, double outlet ventricle, univentricular heart.

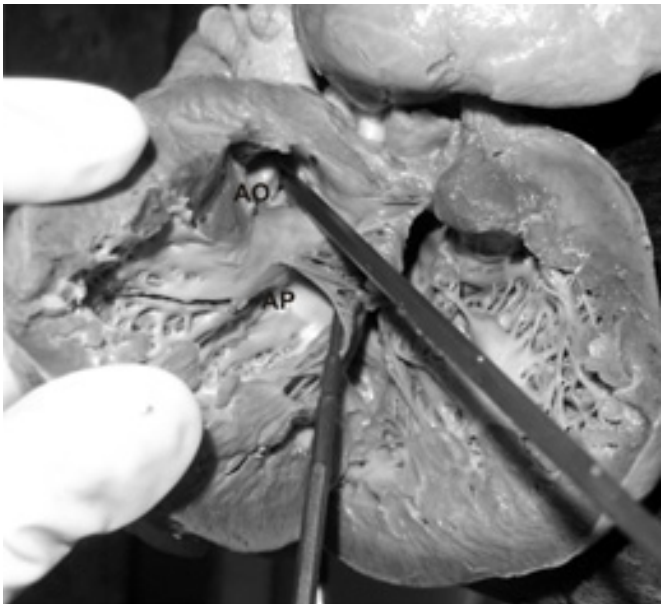

Fig. 3. Univentricular heart, indeterminate type: both great vessels arose from the single ventricle (AO – aorta, AP – pulmonary artery).

Conclusions

Echocardiography is currently the only preoperative diagnostic study performed in most cases of total anomalous pulmonary venous connection; however, surgeons must always keep in mind the possibility of a mixed type of total anomalous pulmonary venous connection.

The truly anatomically univentricular heart is a very rare circumstance. Prior to pathologically diagnosing the presence of an anatomically univentricular heart, it is also necessary to exclude the possibility of a huge ventricular septal defect. A muscular outlet septum could imitate the presence of the rudimentary second chamber.

It is very difficult to distinguish the morphology of the double outlet ventricle by echocardiography. Two dimensional echocardiographic findings for the diagnosis of double outlet left ventricle are: both great arteries origins from the left ventricle, absence of the right outlet, absence of the right ventricular outflow, absence of apical trabeculation.

Echocardiography provides sufficient preoperative anatomic and physiologic information for planning the repair of defects such as patent ductus arteriosus, and atrial and ventricular septal defects.

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