Ultrasound in the Service of Early Diagnosis and Treatment of Congenital Heart Defects: Bosnian and Herzegovinian Experience

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ABSTRACT

Introduction: Congenital heart defects (CHDs) are the most common congenital anomalies in the pediatric population, representing about one-third of all congenital anomalies in infants. Modern pediatric cardiology deals mainly with CHD. Echocardiography is a well-established diagnostic method in fetal and pediatric cardiology. The paper aimed to show the importance of early diagnosis of CHD in Bosnia and Herzegovina by fetal and pediatric echocardiography to foster the treatment if necessary and to present the outcome of children with early prenatal screening and postnatal diagnosis of CHD.

Material and methods: In a retrospective study, we investigated all patients with prenatally detected CHDs at the Department of Obstetrics and Gynecology, University Clinical Center Sarajevo, Bosnia and Herzegovina, in the 8 year period (2012–2020).

Results: During this 8 year period, out of 2,620 pregnant women in whom fetal echocardiography was performed as a screening, 64 (2.4%) fetuses were suspected for CHD of which the diagnosis was confirmed postnatally. Out of 64 patients with CHDs, hemodynamically significant CHDs were diagnosed in 22 (34.4%), while in 42 (65.6%) CHDs were mild and hemodynamically nonsignificant. In 16 (25.0%) patients, the CHD was associated with other congenital malformations (syndrome, chromosomopathy). In 28 (43.8%), atrial septal defect (ASD) and ventricular septal defect (VSD) were isolated CHDs, while in 14 they were syndromic. Atrioventricular septal defect (AVSD) was isolated in 1, while in 2 patients it was part of trisomy 21. Other extracardiac congenital anomalies were trisomy 21 in 5 infants, as well as Holt Oram syndrome in 1 infant with VSD. A total of 5 (7.8%) out of 64 patients died (one termination of pregnancy after 18 weeks of gestation, four postnatal deaths).

Conclusion: The importance of fetal screening and prenatal diagnosis of patients with CHDs, as well as the postnatal confirmation thereof, enables the timely planning of an intervention in accordance with the needs and better communication and better understanding with the parents. This approach is important for healthcare providers as well and could be proposed as the universal approach at the national level in entire Bosnia and Herzegovina.

Keywords: Congenital heart defects, Echocardiography, Screening.

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INTRODUCTION

Congenital heart defects (CHDs) are the most common congenital anomalies of the pediatric population, representing about one-third of all congenital anomalies in infants. It is estimated that 1.3 million children are born worldwide with this condition with an incidence of 7–12 per 1,000 newborns. The development of fetal echocardiography as well as pediatric cardiology is crucial to timely screen for and detect these defects. Prenatal diagnosis provides prognostic information, and possible therapeutic modalities either pre- or postnatally. Timely diagnosis enables proper and life-saving intervention especially in those 25% of neonates with severe CHD, who are at great risk to die in the first week of life.^{1–5} Early fetal echocardiography (use began in 1972; Winsberg F.) is increasingly recommended in weeks 11–14, although it is often done in the period from 18 to 22 weeks (as this is the time when detailed evaluation of fetal cardiac anatomy is possible).^{6–10} Today, under the guidance of a transvaginal probe, it is possible to perform the first fetal heart analysis between 11th and the 14th week of gestation.^{10,11} Additionally, the time of examination depends on the experience of the gynecologist and pediatric cardiologist. Fetal echocardiography is a diagnostic noninvasive procedure that performs an analysis of cardiac anatomy, large blood vessels of the heart, as well as an analysis of heart rhythm.^{10–13}

The aim of the paper was to present the importance of early prenatal and postnatal diagnosis of CHD by echocardiography

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in Bosnia and Herzegovina in order to increase the treatment efficiency and improve the prognosis.

MATERIALS AND METHODS

In the retrospective study, we investigated all patients with prenatally detected CHDs at the Department of Obstetrics and Gynecology, University Hospital Center Sarajevo, Bosnia and Herzegovina, in the 8 year period (2012–2020). The data were derived from the medical records after approval by the local ethical committee.

RESULTS

Out of 2,620 patients in whom fetal echocardiography screening was performed, 64 (2.4%) were diagnosed with CHDs, all of which were confirmed postnatally by a pediatric cardiologist. The number of patients diagnosed with isolated CHDs or CHDs accompanied by other congenital anomalies and their outcome is shown in Table 1. Out of 64 patients with CHDs, hemodynamically significant CHDs were diagnosed in 22 (34.4%), while in 42 (65.6%) CHDs were mild and hemodynamically nonsignificant. In 16 (25.0%) patients, the CHD was associated with other congenital malformations (syndrome, chromosomopathy). In 28 (43.8%), atrial septal defect (ASD) and ventricular septal defect (VSD) were diagnosed as isolated CHDs without extracardiac malformations, while in 14 they were syndromic. Atrioventricular septal defect (AVSD) was the only malformation in 1, while in 2 patients it was part of trisomy 21. Other extracardiac congenital anomalies were trisomy 21 in 5 infants, Holt Oram syndrome in 1 infant with VSD. A total of 5 (7.8%) out of 64 patients died (one termination of pregnancy after 18 weeks of gestation, four postnatal deaths). Termination of pregnancy was performed at 18 weeks of gestation of the fetus diagnosed with mitral atresia and VSD who developed nonimmune fetal hydrops. In four children, postnatal death occurred as follows: two patients with Ebstein's anomaly died in the first month of life, the infant with cardiac tumor died in the second month of life, while the infant with transposition of the great arteries underwent cardiac palliative surgery and died due to severe sepsis after surgery at 2 months.

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DISCUSSION

Fetal echocardiography should be part of the screening of each pregnant woman, although due to the small number of staff doing this, it is difficult.¹⁰ The use of Color Doppler enhances the visualization of large blood vessels.¹⁰ Decreased mortality and morbidity due to antenatal diagnosis have already been reported for the entity of aortic coarctation, left heart hypoplasia, and transposition of large blood vessels.^{11,14–16} Sometimes it is also necessary to perform the examination several times, especially if the fetal echocardiography is performed before 18 weeks, and if there is something suspicious about that finding. Indications for fetal echocardiography are divided into indications by the mother, indications by the fetus, and indications by the family. Indications by the mother can be as follows: metabolic maternal diseases (diabetes mellitus, phenylketonuria), congenital heart disease (congenital heart abnormalities, cardiomyopathies), maternal exposure by cardiac teratogens (alcohol, valproic acid), vitamin A, lithium, or lysine (scleroderma, systemic lupus erythematosus). Indications by the fetus can be as follows: suspected CHD, fetal arrhythmias, or anomalies of other organs with or without pathological karyotype. Indications by the family can be Mendel syndromes (such as Holt-Oram syndrome, DiGeorge syndrome, Noonan syndrome, Williams-Beuren syndrome, Ellis van Creveld syndrome, or cardiomyopathies).¹⁷ Choi et al. divide all detected anomalies by fetal echocardiography into normal, minor abnormalities, simple, moderate, and complex cardiac anomalies (Figs 1 to 7).¹⁸

By introduction of fetal echocardiography at the Clinic of Obstetrics and Gynecology, University Hospital Center Sarajevo, Bosnia and Herzegovina, in 2012, a new era of diagnosis of CHD in Bosnia and Herzegovina was introduced. The most common CHD in our sample was VSD and ASD. Charafeddin et al. most commonly

Table 1: Prenatal and postnatal diagnosis of isolated or syndromic congenital heart defects (CHDs) and their outcome at the Department of Obstetrics and Gynecology, University Hospital Center Sarajevo, Bosnia and Herzegovina, in the 8-year period (2012–2020)

CHD detected prenatally and confirmed postnatally	Number of fetuses/infants with CHDs		Outcome	Total (%)
	Isolated (%)	Syndromic or with other extracardiac congenital defects (%)		
Ventricular septal defect	13 (59.1)	9 (40.9)	Alive	22 (34.4)
Atrial septal defect	15 (75.0)	5 (25.0)	Alive	20 (31.3)
Atrioventricular septal defect	1 (33.3)	2 (66.7)	Alive	3 (4.6)
Hypoplastic left heart syndrome	6	—	Alive	6 (9.4)
Hypoplastic right heart syndrome	2	—	Alive	2 (3.1)
Single ventricle	2	—		2 (3.1)
Transposition of the great arteries	1	—	Died after surgery at 2 months	1 (1.6)
Tetralogy of Fallot	1	—	Alive	1 (1.6)
Mitral atresia with VSD	3	—	1 patient 18 weeks' termination of pregnancy	3 (4.6)
Pulmonary stenosis	1	—	Alive	1 (1.6)
Ebstein anomaly	2	—	2 postnatal deaths	2 (3.1)
Cardiac rhabdomyoma	1	—	Postnatal death	1 (1.6)
Total	48 (75.0)	16 (25.0)	5 (7.8) deaths	64 (100.0)

Fig. 1: Hypoplastic left heart associated with mitral valve atresia

Fig. 2: Hypoplastic left heart associated with mitral valve atresia

Fig. 3: Single ventricle

Fig. 4: Single ventricle

Fig. 5: Ebstein anomaly

Fig. 6: Ductus venosus

detected VSD in the Lebanese sample, while Ozkutlu et al. also point VSD as most common detected CHD.^{19,20} Echocardiography in the pediatric population was introduced in the Clinical Center University of Sarajevo (CCUS) 37 years ago with around 4,000

check-ups per year. The introduction of echocardiography and diagnosis of severe cardiac conditions stimulated the development of cardiac surgery at CCUS. Over the past 20 years, from April 1997 to 2020, 771 patients with CHDs were operated at CCUS, of which

Fig. 7: Ventricular septal defect

205 (26.6%) were cyanotic CHD, while 106 (13.8%) were obstructive CHDs. Postoperative mortality was 7.8% (60 patients). All of these cyanotic defects could easily be prenatally diagnosed with fetal echocardiography. In childhood, the treatment of CHDs ranges from the possibility of surgical, radical, or palliative treatment to interventional and hybrid operations, but also to the treatment of postoperative sequelae with highly differentiated therapy. Of particular issues are the grown-up congenital heart diseases (GUCH) that are relatively common (because of unrecognized or untimely treatment in childhood, but also because of the sequelae after cardiac surgery).

Each pregnant woman who carried a fetus with a suspected heart anomaly was given a clear plan, and even in the case of an unexpected birth, the pediatric cardiologist had the correct guidelines on neonatal treatment, the need for additional supervision, and transfer to the intensive care unit. Patient screening was performed only at the tertiary level of health care, for those patients referred from lower levels of health care of pregnant women. A large number of patients were followed up on the primary and secondary level of health care and therefore were unable to undergo evaluation by fetal echocardiography. The absence of fetal echocardiography, especially in the segment of primary health care of pregnant women, is an objectively big problem in Bosnia and Herzegovina. The number of unrecognized heart defects indicates the need to establish fetal echocardiography as a standard procedure for fetal screening. Given the severity of individual anomalies, in some situations the clinical psychologist was involved in supporting the parents during the further treatment of the child. The decision to terminate a pregnancy has to be approved by the ethics committee of the institution. Fetal echocardiography requires faith in what we do, responsibility, teamwork primarily by gynecologists and pediatric cardiologists, adequate timing for examination, and correlation with an increase in the nuchal fold, with a constant reflection on the evolution of CHD and repeated examination depending on the complexity of the anomaly. Fetal echocardiography is an indispensable part of the daily work of gynecologists/obstetricians, together with pediatric cardiologists. It is important for the possible early detection of CHD, possible fetal interventions, and in our country for the preparation for early surgical correction (eventual relocation out of Bosnia and

Herzegovina). The most important determinants of CHD are that they are in principle a genetic disease, presented individually and are evolutionary. The segment of fetal echocardiography should be inseparable from the segment of pediatric cardiology/cardiac surgery as well as cardiology/cardiac surgery of an adult with CHD. Not only the better technical capabilities of gynecology but also the advancement of surgical intervention for the care of children and adults could produce better results in the future. Combination of well-developed fetal echocardiography with well-developed pediatric cardiac surgery would lead to good results.

CONCLUSION

The data on fetal echocardiography with regard to the possibility of further treatment are encouraging, especially as a continuation of treatment for childhood CHD. The future treatment of CHD could be comprehensively viewed only through complementary diagnostics as well as treatment in the antenatal period, childhood, and adulthood. The urgent establishment of fetal echocardiography in Bosnia and Herzegovina is imperative.

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