

## FETAL ULTRASOUND AND NEONATAL DIAGNOSIS OF CONGENITAL HEART DEFECTS

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### Abstract

*The congenital heart defect is one of the major causes of neonatal and pediatric mortality. A retrospective study of all the patients with singleton pregnancies, admitted in our hospital between 2010-2012 was performed. The data collected included information referring to the age of the patients, the gestational age, the cardiac diagnosis, the extracardiac anomalies, the prenatal and postnatal management and evolution. Out of 7,195 patients, 23 living newborns had CHDs (congenital heart defects). The mean gestational age was 34.12 weeks (range 30-39 weeks). We recorded VSD (ventricular septal defect) in 47.8% newborns, ASD (atrial septal defect) in 26.1% cases, TGA (transposition of great arteries) in 8.7% cases, coarctation of the aortic artery (COA) in 4.3% cases, TOF (tetralogy of Fallot) in 8.7% cases and HLHS (hypoplastic left heart syndrome) in 4.3% cases. The ultrasound findings in utero were VSD (30.4%), ASD (39.1%), TGA (4.3%), coarctation of the aortic artery (4.3%), TOF (4.3%) and HLHS (4.3%). In our study there was a strong correlation between the antenatal ultrasound findings and the neonatal diagnosis.*

**Keywords:** congenital heart defect, ultrasound, diagnosis.

**Abbreviations:** CHD-congenital heart defect; VSD - ventricular septal defect; ASD - atrial septal defect; TGA - transposition of the great arteries; COA - coarctation of the aortic artery; TOF - tetralogy of Fallot; HLHS - hypoplastic left heart syndrome.

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### Introduction

The congenital heart defect (CHD) is defined as a structural abnormality of the heart or of the intrathoracic great vessels [1]. It occurs in approximately 8 per every 1,000 live births and one quarter of these are considered to be severe congenital heart defects (CHD), which require heart surgery or catheterization before the age of 1 year [2]. Approximately 25 years ago, the four-chamber ultrasound view was introduced as a screening method for the prenatal detection of heart anomalies [3]. The initial reports were

promising and suggested that the four-chamber ultrasound view could detect 70–90% of the fetuses with congenital heart anomalies [4].

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### Materials and Methods

A retrospective study of all the patients with singleton pregnancies, admitted in the Bucur Maternity of Bucharest between 2010-2012 was performed. The data collected included information referring to the age of the patients, the gestational age, the cardiac diagnosis, the

extracardiac anomalies, the prenatal and postnatal management and evolution.

The data were analyzed with the Statistical Package for the Social Sciences (SPSS for Windows, version 19.0) and Microsoft Office Excel. The descriptive statistics included the mean and the standard deviation for numerical variables, and the percentage of the different categories for the categorical variables. A P-value < 0,05 was considered statistically significant.

## Results

Out of a total of 7,195 patients admitted in our hospital, a number of 23 newborns with CHD were included in this study. We also analyzed the stillbirths and the miscarriages. The average age of the mother was 29.3 (16 to 38 years). The mean gestational age was 34.96 weeks (range 30-39 weeks). We mention that the manner of delivery was vaginal delivery in 56.5% of the cases (n=13 cases) and Cesarean section in 43.5% (n=10 cases) of the cases. In our cases, there was no significant correlation between CHD and the manner of delivery.

The newborns with CHD had different socioeconomic backgrounds, 56.5% being from the urban area. Even if the percentages were similar, there was no connection between the manner of delivery and the socioeconomic status. Smoking as a normal risk factor for mothers was recorded in 65.2% (n=15 subjects), but without any statistical significance in relation to the CHD. A total of 39% (n=9) of the births had no ultrasound examination before the last gestational week. The most common associated maternal pathology was the uterine malformation in 8.7% of the cases (2 cases). This study included 3 cases of newborns without any antepartum ultrasound.

We recorded VSD (ventricular septal defect) (Figure 1.) in 47.8% of the newborns, ASD (atrial septal defect) (Figure 2.) in 26.1% cases, TGA (transposition of the great arteries) (Figure 3 and Figure 4.) in 8.7% cases, coarctation of the aortic artery (COA) in 4.3% cases, TOF (tetralogy of Fallot) in 8.7% cases and HLHS (hypoplastic left heart syndrome) in 4.3% cases. We also noticed 3 cases of newborns with combined cardiac defects (ASD with VSD, ASD with TGA and TGA with VSD). (Table 1).



Figure 1 - Ventricular septal defect



Figure 2 - Atrial septal defect



Figure 3 - Transposition of the great arteries



Figure 4 - Transposition of the great arteries

CHD in newborns	Percent (number)
ventricular septal defect	47.8% (n=11)
atrial septal defect	26.1% (n=6)
transposition of the great arteries	8.7% (n=2)
coarctation of the aortic artery	4.3% (n=1)
tetralogy of Fallot	8.7% (n=2)
hypoplastic left heart syndrome	4.3% (n=1)

Table 1: CHD in newborns

The obstetric ultrasound showed VSD (30.4%), ASD (39.1%), TGA (4.3%), coarctation of the aortic artery (4.3%), TOF (4.3%) and HLHS (4.3%). (Table 2). There is also a strong correlation between the cardiac diagnosis in newborns and the antepartum ultrasound (p=0.001).

Antepartum ultrasound findings	Percent (number)
ventricular septal defect	30.4 % (n=7)
atrial septal defect	39.1% (n=9)
transposition of the great arteries	4.3% (n=1)
coarctation of the aortic artery	4.3% (n=1)
tetralogy of Fallot	4.3% (n=1)
hypoplastic left heart syndrome	4.3% (n=1)
No ultrasound	13% (n=3)

Table 2: Antepartum ultrasound findings

The abdominal ultrasound was also used in the diagnosis of the extracardiac anomalies. We recorded 8 cases of such situations (hydrocephaly, cheiloschisis, esophageal atresia, abdominal situs inversus, encephalocele and renal malformation).

This study also considered the stillbirths and miscarriages with CHD. We followed the evolution of all the births with CHD ultrasound finding, even if in the end they were stillbirths or miscarriages. Thus, we report a total of 5 stillbirth cases with the mean gestational age

of 35 weeks and 4 miscarriages between 12 and 24 weeks with CHD associated with extracardiac anomalies. The postmortem examinations revealed that the stillbirths had the following CHD: 2 cases with ASD and VSD, two cases with ASD , one with hydrocephaly, and one case with TGA. Among the miscarriages the most interesting case was that of a body stalk anomaly diagnosed by ultrasound (Figure 5 and 6) and pathological anatomy examination (Figure 7, 8) and one case with complex CHD (ASD, VSD and TGA), and associated palate defect and encephalocele

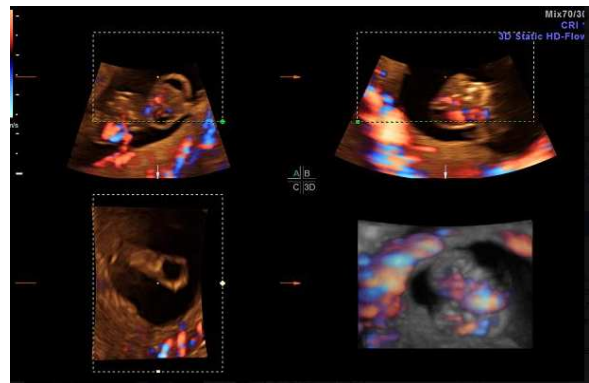


Figure 5 - Body stalk anomaly



Figure 6 - Body stalk anomaly



Figure 7 - Body stalk anomaly



Figure 8 - Body stalk

## Discussions

The interventional and surgical techniques have been improved, but congenital heart defects in children continue to be an important cause of morbidity and mortality [5]. There are several interrelated factors which can cause birth defects (BD) [6]. Congenital heart malformations are the leading organ-specific birth defect in the United States [7].

In the case of congenital heart defects, the social and economic factors have been proven to have an impact upon the rate of stillbirths as well as on neonatal and infant mortality [8]. There were no significant differences between the rural and urban areas in our study, but other publications indicate that there is a slightly increased prevalence in the rural area [9]. The advanced maternal age (over 35) was correlated with a higher prevalence of congenital defects, but with no significant differences in comparison with the younger patients, this aspect being also noticed in the current study. This study also shows an insignificant association between the smoking status of the mother and the presence of CHD, although in literature there is an association between smoking and congenital defects detected at birth, particularly orofacial defects [10].

The countries which have introduced well-established prenatal diagnostic techniques, as well as access to abortion due to congenital defects have showed a reduction in neonatal mortality [11], in comparison with the countries

with restrictive policies regarding therapeutic abortion. [12]. The prenatal detection of major heart defects varies from 5% to 75% in low-risk populations [13,14]. The socioeconomic variations regarding abortion following the antenatal diagnosis of a malformation cause inequalities in the development and postpartum mortality of liveborn infants with heart defects [15].

In the 1990s the examination of the four-chamber view was introduced as the standard of fetal heart scanning. However, sometimes this examination fails to detect some defects as: tetralogy of Fallot, truncus arteriosus communis, and interruption of the aortic arch. In our study there was a strong correlation between the antepartum ultrasound findings and the neonatal diagnosis.

The International Society of Ultrasound in Obstetrics and Gynecology (ISUOG) has published guidelines for the “basic” and the “extended basic” fetal cardiac ultrasound and echocardiography examination. [16] The use of color Doppler is mandatory in the examination of the fetuses with suspected heart defects [17]. However, the Doppler examination in low-risk populations is still controversial [18]. The isolated VSDs are the most common form of congenital heart malformation described in literature [19], this aspect being also illustrated in the current study.

Some authors have described that CHDs decrease the incidence of spontaneous birth in such situations, but it has been demonstrated that the manner of delivery does not have any impact on the neonatal evolution [20], this aspect being also shown in our study. The results of our study are consistent with the literature on associated extracardiac malformations, thus encephalocele and complex heart defects being rarely associated [21], and body stalk anomaly occurs with a prevalence of 0.12 per 10.000 liveborn or stillborn infants [22].

Other studies maintain that the Doppler examination may be helpful in the detection of major heart defects in low-risk populations, but there are still severe malformations which are undiagnosed prenatally [23].

## Conclusions

- In our study there was a strong correlation between the antenatal ultrasound findings and the neonatal diagnosis.
- Even if there are some undiagnosed prenatal malformations, there is no doubt that their detection has increased, and the ultrasound remains a feasible method for the detection of CHD even at an early gestational age.

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