Acta Clin Croat 2024; 63: 101-105
doi: 10.20471/acc. 2024.63.01.12

Original Scientific Paper

THE SUCCESS RATE OF INTRAUTERINE CONGENITAL HEART DISEASE DETECTION AT DEPARTMENT OF GYNECOLOGY AND OBSTETRICS, SVETI DUH UNIVERSITY HOSPITAL FROM ZAGREB, 2016-2020

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SUMMARY – The aim was to assess the frequency and success rate of prenatal diagnosis of congenital heart diseases (CHDs) in live-born neonates using ultrasound, as well as the occurrence of different types of CHD in the tertiary medical care institution in the period from January 1, 2016 till December 31, 2020. In this retrospective study, data were collected by reviewing medical history of both mothers and newborns. During the study period, there were 13,932 childbirths, and 42 newborns were diagnosed with CHD. Excluding the diagnoses of persistent duct of Botallo (n=4) and CHDs arising as a consequence of maternal condition (n=2), out of 36 newborns, 31 (86%) were diagnosed prenatally and 5 (14%) postnatally. The prevalence of CHDs was 30 *per* 10,000 births (3.0‰). In conclusion, the rate of prenatal diagnosis of CHDs was very high. The prevalence of CHDs was substantially lower than the European average published in the EUROCAT study for the 2016-2019 period. Additional education and examination algorithms need to be implemented to achieve more successful rates of prenatally diagnosed CHDs recorded in the best centers across Europe and the world.

Key words: *Prenatal diagnostics; Congenital heart disease; Ultrasound*

Introduction

Congenital diseases and genetic disorders are among the leading causes of neonatal and infant mortality, with a global prevalence of 3%-5%[1]. Congenital heart diseases (CHDs) are one of the most common congenital anomalies in neonates, accounting for about one third of all congenital anomalies[2]. The prevalence varies across different countries and regions of the same country, which can be attributed to differences in the availability of medical care.

The aim of this study was to assess the prevalence of CHDs and the rate of their prenatal detection by ultrasound in neonates whose mothers were monitored during pregnancy.

Methods

Data were collected by reviewing medical history of both mothers and neonates. The study included all pregnancies regularly monitored at the Department of Gynecology and Obstetrics, Sveti Duh University Hospital, Zagreb, Croatia. Pregnancies that were not

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Received June 1, 2023, accepted July 19, 2023

observed but resulted in delivery of newborns with CHDs in the same hospital were excluded from the study (three newborns in total). The screening program for CHDs included fetal echocardiography performed by an algorithm that includes four-chamber view, which has 60.3% sensitivity of CHD detection[3] and specificity of 100%[4], three-vessel view, which has 91% sensitivity of detection of large vessel error[5], and outflow tract view. Ethical approval was required and obtained from the institutional Ethics Committee.

Results

During the 5-year period, 13,932 pregnant women gave birth at our hospital, of which 10,838 (77.8%) deliveries were performed vaginally, and 3094 (22.2%) by cesarean section. There were 53 (0.38%) stillborn neonates, while 36 neonates were diagnosed with CHD, out of which 18 (50%) were female and 18 (50%) male. In 31 (86%) neonates, the diagnosis was made prenatally and in 5 (14%) neonates postnatally. There were 29 full term neonates and seven preterm neonates. The prevalence of particular forms of CHD is shown in Table 1.

Table 1. Distribution of congenital heart diseases by type

	n	%
Complex heart disease*	21	50
Persistent duct of Botallo	4	9.5
Cardiomyopathy	4	9.5
Coarctation of the aorta	3	7.2
Ventricular septal defect	2	4.7
Tetralogy of Fallot	2	4.7
Transposition of great arteries	1	2.4
Hypoplastic left heart syndrome	1	2.4
Common arterial trunk	1	2.4
Hypoplastic aortic arch	1	2.4
Triatrial heart	1	2.4
Dextrocardia	1	2.4
Total	42	100

*except for the types which often occur as individual congenital heart disease (tetralogy of Fallot, hypoplastic left heart syndrome)

The share of preterm newborns was 19% (n=7), with an average gestational age of 36 weeks. Three (43%) newborns were female, and four (57%) were male. Six (86%) out of seven preterm newborns were diagnosed prenatally, and only one (14%) postnatally. Two preterm newborns were diagnosed with defects on the exit of large blood vessels, three with defects in the heart itself, and two with complex CHD. Two preterm newborns were also diagnosed with chromosomal abnormalities, one with Down syndrome, and one with *cri du chat* syndrome. Both were diagnosed prenatally.

In full term newborns, defects in the heart itself were detected in three neonates, two (67%) prenatally and one (33%) diagnosed with both perimembranous and muscular ventricular septal defect and *cri du chat* syndrome postnatally. Defects on the exit of large blood vessels were detected in four newborns. Three (75%) newborns were diagnosed prenatally, and one (25%) newborn diagnosed with common arterial trunk postnatally. Complex CHDs were detected in 22 newborns, 21 (95%) of which were diagnosed prenatally and one (5%) postnatally. Five newborns were diagnosed with CHD as part of a syndrome. Four newborns were diagnosed prenatally (as part of Down syndrome, Edwards syndrome, osteochondrodysplasia, and tetrasomy 9p one each). One newborn with ventricular septal defect as part of the *cri du chat* syndrome was diagnosed postnatally.

Four newborns were postnatally diagnosed with persistent duct of Botallo. Two newborns were diagnosed with cardiomyopathic changes of the heart due to maternal condition, including one due to maternal diabetes and one due to maternal viral infection. One (50%) newborn was detected prenatally and one (50%) postnatally. One newborn was diagnosed with a heart rhythm disorder (second-degree AV block).

A positive family history of CHD was detected in five (13%) mothers, of which only one had already given birth to a child with CHD.

In four mothers, increased nuchal translucency was detected in the first trimester, although fetal chromosomal abnormality was confirmed in none of their newborns. In two mothers, chromosomal abnormality of the fetus was suspected with a noninvasive prenatal test. Both of them were confirmed with amniocentesis (trisomy 21 and tetrasomy 9p), and both mothers were referred for fetal echocardiography, where the diagnosis of CHD was made.

Discussion

The total percentage of 86% of prenatally detected CHDs in the five-year period, from 2016 to 2020, indicates a very high success rate of their timely detection. The results of studies including Europe show an average rate of prenatal CHD detection of 25%[6,7]. The worst results were achieved in the countries of Eastern Europe[8], while the average results of Western European countries ranged from 19% to 48%[8], with the rates in certain areas of up to 87%[9]. Studies from France show a success rate of prenatal CHD detection around 71%[10,11]. In western Sweden, the percentage of prenatally detected CHDs was 53%[12]. At the same time, a study from The Netherlands shows the percentage of prenatally detected CHDs of 59.7%[13] after introducing the national screening program. Spanish studies show an average percentage of prenatally detected CHDs of 42.8%[14] and 65.7%[15]. An Italian study shows a success rate of prenatal CHD detection of 75.4%[16], while Polish research shows 76.2%[17]. Results from the two studies in the USA show it to range from 26% to 42%[18] and 61%[19]. Studies from China show distinct differences in the rates of prenatally detected CHDs, from the average of 22.2%[20] to up to 90.5%-91.66% and 98.68%[21].

Despite high results, additional efforts are needed in order to analyze other factors contributing to the efficacy of prenatal diagnostics of CHDs. Additional education and implementation of the latest ultrasound algorithms for prenatal diagnostics in primary and secondary medical centers in Croatia is required in order to increase the success rate of prenatal diagnostics in the whole Republic of Croatia.

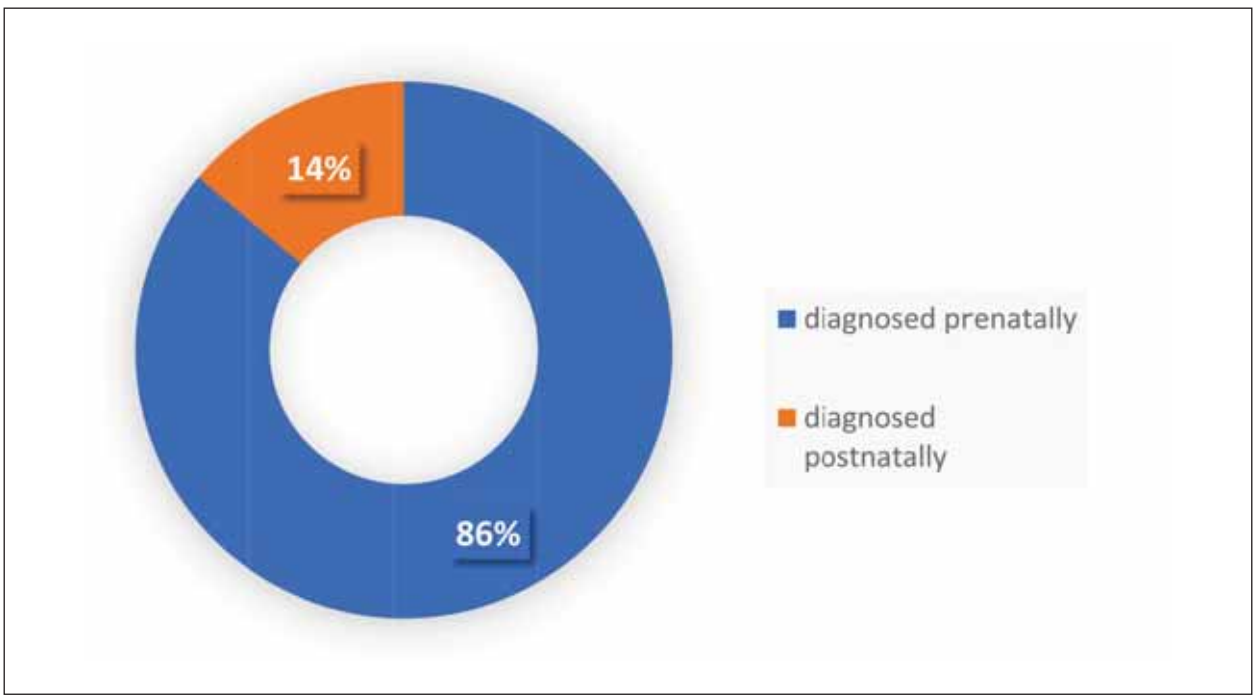

Fig. 1. Timing of congenital heart disease diagnosis.

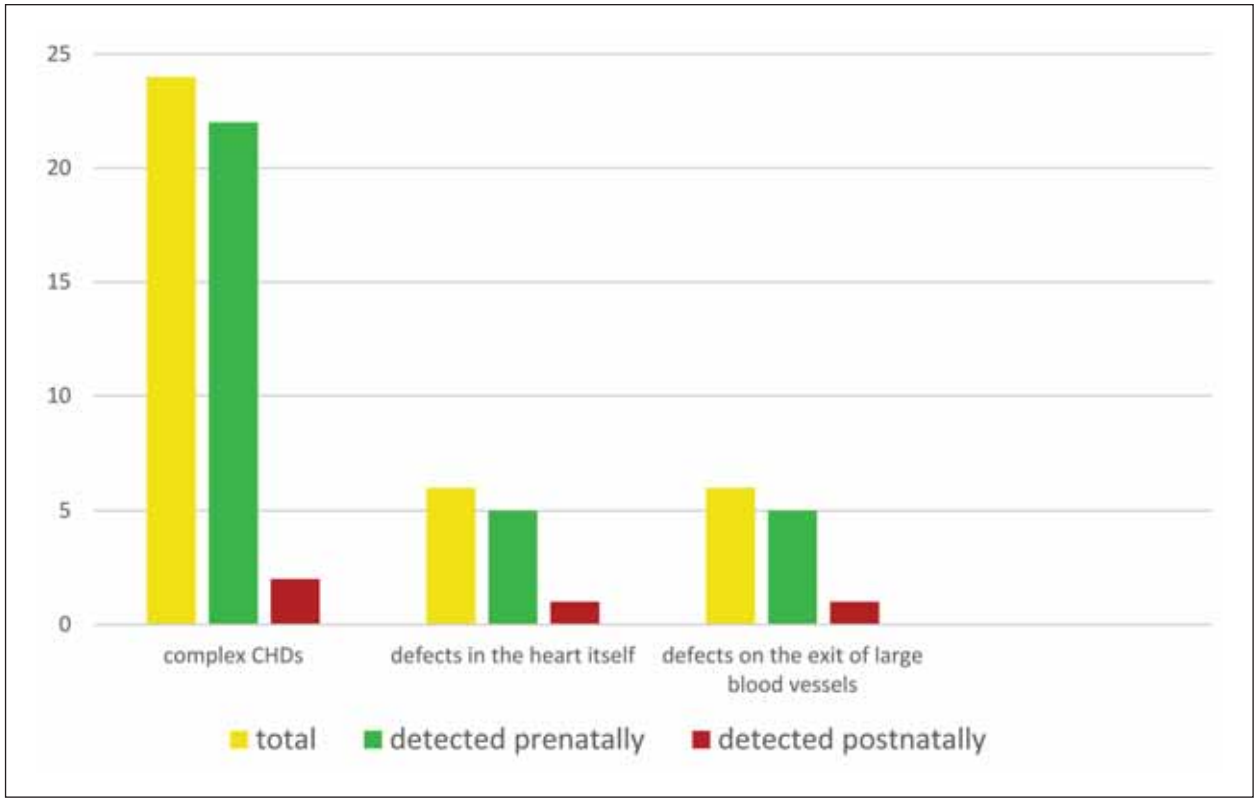

Fig. 2. Prevalence of different types of congenital heart disease and timing of their detection.

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Sažetak

USPJEŠNOST INTRAUTERINOG OTKRIVANJA PRIROĐENIH SRČANIH BOLESTI NA KLINICI ZA GINEKOLOGIJU I PORODNIŠTVO OPĆE BOLNICE "SVETI DUH", ZAGREB, 2016.-2020.

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Cilj ovog istraživanja bio je utvrditi učestalost i stopu prenetalnog postavljanja dijagnoze prirođenih srčanih bolesti u živorođene novorođenčadi uz pomoć ultrazvuka (UZV) u tercijarnom centru zdravstvene zaštite, kao i učestalost različitih oblika prirođenih srčanih bolesti u razdoblju od 1. siječnja 2016. do 31. prosinca 2020. godine. Provedeno je retrospektivno istraživanje, a podaci su skupljeni uvidom u povijesti bolesti majka i novorođenčadi. Od ukupno 13.932 poroda kod 42 novorođenčeta dijagnosticirana je prirođena srčana bolest. Ako isključimo dijagnozu perzistentnog Botallijeva duktusa (n=4) te srčane bolesti nastale kao posljedica stanja majke (n=2), od 36 novorođenčadi u 31 (86%) novorođenčeta bolest je dijagnosticirana prenatalno, a u 5 (14%) novorođečadi postnatalno. Učestalost je iznosila 30 na 10.000 poroda (3,0‰). Stopa postavljanja dijagnoze je značajno veća u usporedbi sa stopom otkrivanja u drugim zemljama Europe. Učestalost prirođenih srčanih grešaka značajno je manja od učestalosti na razini Europe objavljene u studiji EUROCAT za razdoblje od 2016. do 2019. godine. Treba provesti dodatne mjere izobrazbe i algoritme pregleda kako bi uspješnost prenatalnog otkrivanja prirođenih srčanih bolesti dostigla one u najboljim centrima Europe i svijeta.

Ključne riječi: *Prenatalna dijagnostika; Prirođena srčana bolest; Ultrazvuk*