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Congenital heart diseases with neonatal onset: Prevalence and circumstances of diagnosis in the newborn

Ouahiba Benrabah [1], Tahar Ait Mouheb [2], Rachida Dahmoune [1, 2], Sihem Bellouti [2], Lynda Ait Mokhtar [2, *], Zakaria Amine [2] and Abdelmadjid Maoudj [1]

[1] *Department of pediatric medicine, Faculty of medicine Algiers, Algeria.*
[2] *Department of anesthesia and resuscitation Faculty of medicine Algiers, Algeria.*

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Abstract

Introduction: Congenital heart disease (CHD) is the most common congenital malformation. Their prevalence remains underestimated in our country despite the improvement in diagnostic means.

Objective: determine the prevalence and circumstances of diagnosis of CHD in a neonatology department.

Materials and methods: We carried out a descriptive cross-sectional study which included all live births with congenital heart disease, in the neonatology department of Hussein Dey University Hospital over a period of seven years (2016-2022). Data collection was done as the diagnosis progressed.

We didn't include non-malformative cardiac abnormalities.

Results: We collected 231 cases of congenital heart disease (CHD) out of 66,520 live births (LV), i.e. a prevalence of 3.41‰ LV. Antenatal diagnosis was made in 89 cases (45%). Ultrasound diagnosis was made in 70% of cases in the first 3 days with 46% of cases in the first 24 days of life. Physical signs were dominated by cyanosis in 38%. Congenital heart disease with a high neonatal risk level was in 15 cases (6.5%), including 14 cases of TGV and a single case of total RVPA. Heart disease with a medium to high neonatal risk level was represented by ductal-dependent heart disease in 95 cases (41.3%) of which 30% of cases were straight obstacles including 10.8% T4F. Cardiac diseases with low neonatal risk were in 94 cases (40.8%), of which 38% of cases represented by IVC followed in 29% by AVC and 10.6% by IAC.

Conclusions: Given the insufficiency of antenatal diagnosis in our cohort, congenital heart diseases were mainly diagnosed after birth in the face of suggestive clinical symptoms, thus ,The best ways to improve early diagnosis of congenital heart disease would be antenatal screening using fetal echocardiography and the introduction of systematic postnatal screening by the systematic measurement of pre- and post-ductal saturation and the generalization of echocardiography in neonatology departments.

Keywords: Congenital heart diseases; Newborns; Prevalence; Patient

1. Introduction

Congenital heart defects are the most common congenital malformations in newborns. Their prevalence varies between 8 and 17.9 per 1000 births [1-2]. The clinical presentation varies between minor forms, often asymptomatic, to severe critical forms, discovered early in the first days of life and endangering life. The prognosis is variable and depends on

* Corresponding author: Lynda Ait Mokhtar

the type of heart disease, the delay in diagnosis and treatment. The prevalence of congenital heart disease remains underestimated in our country despite improvements in diagnostic methods.

The goal of our study was to determine, the prevalence and circumstances of diagnosis of congenital heart disease in a neonatology department.

2. Materials and methods

This was a single-center, descriptive cross-sectional study carried out in the neonatology department of the Hussein Dey University Hospital in Algiers over a period of seven years from January 1, 2016 to December 31, 2022. We included all births with congenital heart disease confirmed by postnatal ultrasound. We didn't include newborns with non-malformative cardiac anomalies (hypertrophic cardiomyopathy, cardiac tumors, rhythm disturbance, patent foramen oval and patent ductus arteriosus (PDA) in premature infants).

2.1. Data collection

Data collection was carried out, retrospectively, by consulting the files of the patients selected. For each observation, we noted family history of congenital heart disease, consanguinity, neonatal data at birth (gestational age in weeks of amenorrhea (SA), birth weight (PN), gender). The circumstances of diagnosis (antenatal diagnosis, clinical signs, age of onset of symptoms). The type of heart disease: heart diseases were classified according to the level of neonatal risk.

2.2. Statistical study

All data were entered and analyzed using SPSS 25.0 software. For qualitative variables, we calculated frequencies and relative frequencies (percentages). For quantitative variables, we calculated means, medians and standard deviations and determined the range (extreme values: minimum and maximum).

We declare that we have no conflicts of interest in relation to this study.

3. Results

During the study period, we collected 231 cases of congenital heart disease out of 66,520 live births (LV), i.e. a prevalence of 3.4% LV. (table No. I)

Table 1 Annual prevalence of congenital heart disease

	2016	2017	2018	2019	2020	2021	2022	2016-2022
Live births (N)	10870	10330	9386	10848	8354	8637	8095	66520
Heart disease Congenital (N)	33	26	27	33	35	38	37	231
Prevalence(‰ Live births)	3.03	2.51	2.87	3.04	4.20	4.40	4.60	3.47
Antenatal diagnosis (%)	25	32	31	52	55	58	62	45

(N: number, %: percentage)

A family history of congenital heart disease was found in 8.2% of cases (n=19). The main pregnancy pathologies were diabetes (n=70; 30%), pre-eclampsia (n=39; 17%). and dysthyroidism (n=9; 4%). The average gestational age was 36.5±3.2 weeks with extremes ranging from 28 weeks to 42 weeks. The average PN was 3045±675 g [830-4450g]. The sex ratio was 1.4. The overall antenatal diagnosis was in 45% of cases, it was increasing significantly over the years, estimated at 25% in 2016 compared to 62% in 2022. Antenatal ultrasounds showed associated anomalies in 20 fetuses. These were intrauterine growth retardation (n=10), renal malformation (n=5), cerebral malformation (n=2), omphalocele (n=2) and laparoschisis (n=1). The main physical signs suggestive of heart disease were cyanosis in 38%, respiratory distress in 4%, signs of heart failure in 5% of cases, an abnormality in isolated cardiac auscultation 35%, a reduction or abolition of pulses. femoral joints were in 2% of cases). (Figure No. 1)

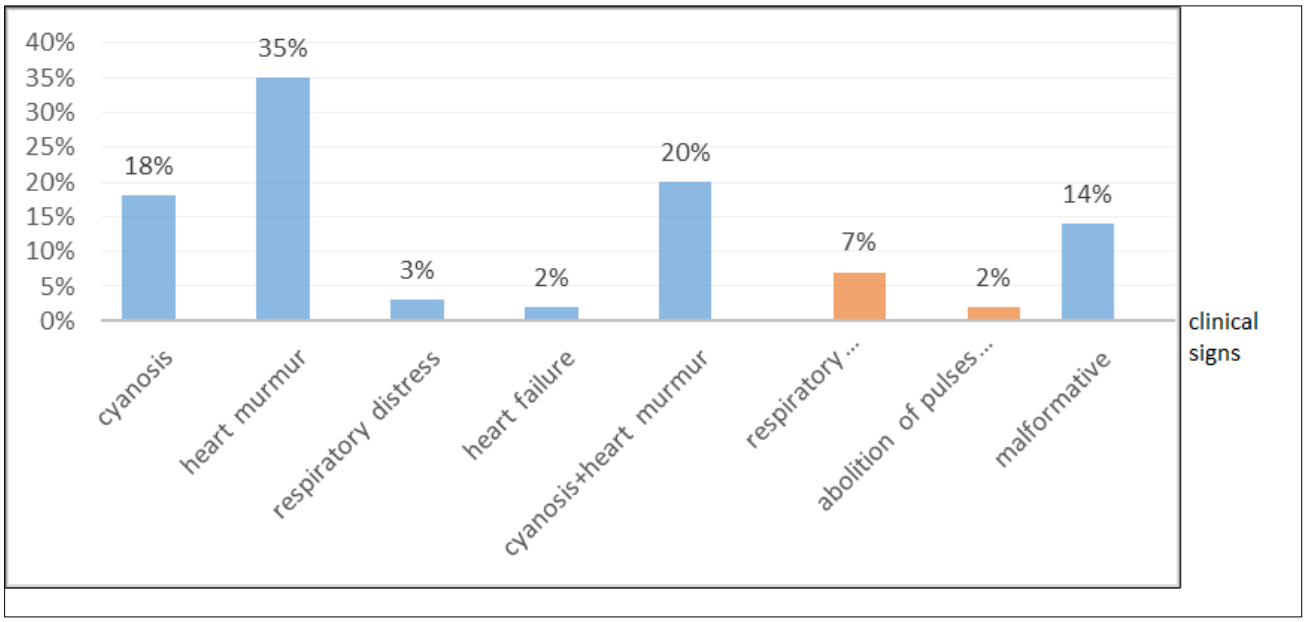

Figure 1 The main physical signs suggestive of congenital heart disease

A chance discovery during a malformative assessment in 14% or targeted screening given the family history of heart disease (n=8).

The onset of symptoms was in the first week in 80% of cases including more than 50% in the first 24 hours of life**.** Ultrasound diagnosis was made in 70% of cases in the first 3 days with 46% of cases in the first 24 days of life. (Figure No. 2).

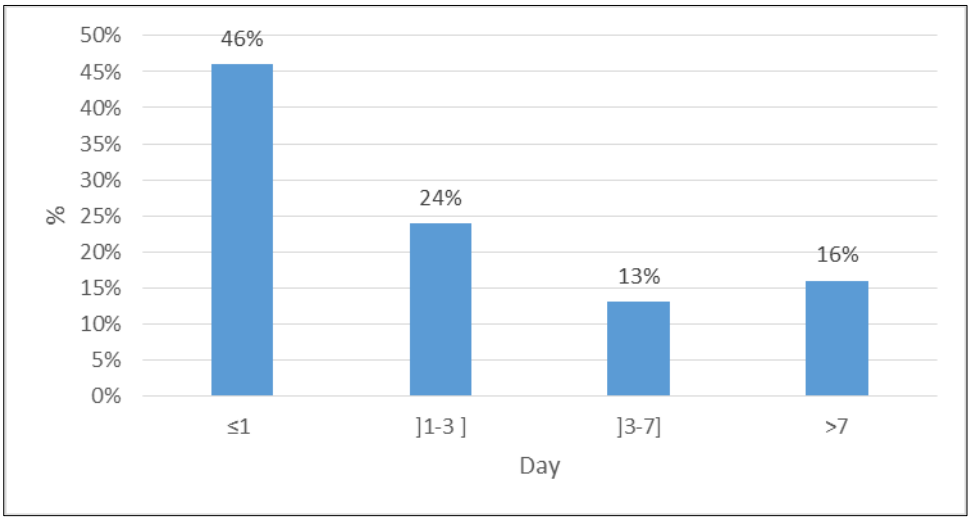

Figure 2 Age of ultrasound diagnosis of congenital heart disease

Congenital heart disease with a high neonatal risk level was in 16 cases (18%), including 15 cases of TGV and a single case of total RVPA. Heart disease with a medium to high neonatal risk level was represented by ductal-dependent heart disease in 95 cases. (41.3%) which 30% of cases represented by right obstacles, Low neonatal risk heart disease was in 94 cases (40.8%) of which 38% of cases represented by IVC followed in 29% by AVC and 10 .6% of IAC. (Table No. 2). The two main malformations associated with congenital heart disease were renal malformation (n=5) and brain malformation (n=4). The heart disease was part of a poly-malformative syndrome in 32 cases. These were trisomy 21 (n=19) and trisomy 18 (n=5), VACTERL association (n=2), Di-George syndrome (n=1). The etiology was undetermined in five cases.

Table 2 Types of congenital heart diseases

Neonatal Risk Level	Type of heart disease		Number (N)	Percentage (%)
Pupil	TGV		14	5.2
	RVPAT		1	0.4
Medium to high	Obstacle Right	T4F	23	10.8
		Critical PS	11	4.7
		PAIS	12	5.2
		OSPA	13	5.6
		SEEN	13	5.6
		Major Ebstein's disease	4	1.7
		Tricuspid atresia	4	1.7
	Obstacle LEFT	Hypo VG	12	5.2
		CoAo	3	1.3
		Aortic hypoplasia	4	1.7
		TAC	4	1.7
		CAI	1	0.4
		DORV +CoAo	3	1.3
Weak	IVC		36	15.6
	IAC		10	4.2
	IVC+IAC		12	5.2
	Complete AVC		14	6
	Partial AVC		13	5.6
	PDA		10	4.2
others	IVC + hypoplasia of the aorta		3	1.3
	Single earpiece		3	1.3
	DORV		3	1.3
Total			231	100

IAC: interatrial communication; **VSP**: ventricular septal defect; **AVC**: atrioventricular canal; **TGV:** transposition of the great vessels; **DORV:** double outlet right ventricle; **CoA**: coarctation of the aorta; **IAA**: interruption of the aortic arch; **PDA:** Patent ductus arteriosus; **PAIS:** pulmonary atresia with intact septum; **OPSA**: open septum pulmonary atresia; **PS**: pulmonary stenosis **IVC** interventricular communications

4. Discussion

In our study, the prevalence of congenital heart disease was 3.4% LV. This prevalence was lower than that reported by Dj .Chighem in his thesis which was 28.6% explained by postnatal echocardiographic screening carried out in all births [3] and was close to that reported by the team from Farhat hospital Hashed from Sousse (2.77% in 2009) [4]. In the literature, the prevalence of congenital heart disease varied between 8 and 17.9% [1-2]. The availability of antenatal diagnosis reference centers on the one hand, and the establishment of systematic neonatal screening programs on the other, would also explain the higher prevalence of heart disease in developed countries [5]. In our series, the diagnosis was made antenatally in only 45% of cases. In a study conducted in China, Zhang et al reported an antenatal diagnosis rate of 22.2% for congenital heart disease and 90% for critical congenital heart disease [6]. Several factors would explain the large variation in the antenatal diagnosis rate. The skill of the operator, the performance of the ultrasound machine (3D, 4D technique, etc.) and the conditions of the examination. The diagnosis was made after birth in 55% of cases (n=127).

The importance of systematic postnatal screening for heart disease by systematic measurement of preductal and post ductal pulsed oxygen saturation must also be emphasized. [7-8] The 2018 Cochrane systematic review concluded that pulse oximetry is a highly specific and moderately sensitive test for the detection of critical congenital heart disease with very low false positive rates. The authors therefore recommended the introduction of systematic screening for critical congenital heart disease in asymptomatic newborns before leaving the maternity ward [9]. It's a very rarely used tool in our hospital.

In our series, heart disease was at low neonatal risk in 94 cases (40.8%), at medium to high neonatal risk level in 41.3% of cases and at high neonatal risk level in 18% of cases. These results were similar to data in the literature [2-7-10]. In a study published by the pediatric cardiology team in Tunis, 20.3% of congenital heart diseases diagnosed antenatally were complex heart diseases [11]. In the literature, the percentage of critical congenital heart diseases varied between 10% to 26.3% of all diagnosed heart diseases [6-14].

5. Conclusion

Given the insufficiency of antenatal diagnosis in our cohort, congenital heart diseases were mainly diagnosed after birth in the face of suggestive clinical symptoms, thus, the best ways to improve early diagnosis of congenital heart disease would be antenatal screening using fetal echocardiography and the introduction of systematic postnatal screening by the systematic measurement of pre- and post-ductal, pulse oximetry and the generalization of echocardiography in neonatology departments.

Compliance with ethical standards

Disclosure of conflict of interest

No conflict of interest to be disclosed.

Statement of informed consent

Informed consent was obtained from all individual participants included in the study.

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