Scientific Research Publishing

World Journal of Cardiovascular Diseases, 2024, 14, 465-479
https://www.scirp.org/journal/wjcd
ISSN Online: 2164-5337
ISSN Print: 2164-5329

Isolated Ventricular Septal Defect: Ultrasound, Therapeutic and Evolutionary Aspects of 85 Cases in the Cardiology Department of the Ignace Deen National Hospital in Conakry

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How to cite this paper: Bah, M.B., Doumbouya, A.D., Balde, E.Y., Balde, M.A., Kone, A., Sylla, I.S., Bah, M.D., Bah, A., Diallo, M., Balde, T.S., Camara, A., Soumaro, M., Barry, I.S., Diakité, S. and Balde, M.D. (2024) Isolated Ventricular Septal Defect: Ultrasound, Therapeutic and Evolutionary Aspects of 85 Cases in the Cardiology Department of the Ignace Deen National Hospital in Conakry. *World Journal of Cardiovascular Diseases*, **14**, 465-479.
https://doi.org/10.4236/wjcd.2024.148040

Received: June 30, 2024
Accepted: August 19, 2024
Published: August 22, 2024

Abstract

Introduction: Ventricular septal defect (VSD) is the most common congenital heart disease of all congenital heart defects. The aim of this study was to investigate the echographic, therapeutic and evolutionary aspects of ventricular septal defects (VSD) in the general cardiology department of the Hôpital National Ignace Deen. Methods: A retrospective data collection was carried out from January 2018 to December 2023 including 85 cases of isolated IVC was performed. The variables studied were epidemiological, clinical, paraclinical, therapeutic and evolutionary. Results: Of the 320 patients seen during the study period for congenital heart disease, 85 (26.556%) were isolated IVCs. Age at diagnosis ranged from 3 months to 16 years, with an average age of 3.59 years. The most represented ethnic group was the Fulani (50.58%). The 8.24% came from consanguineous marriage versus 22.35%. 91.76% of children had a history of bronchitis. The most common clinical signs found were systolic murmur (90.58%), growth retardation (51.76%). Only 4 cases (4.70%) had a malformation associated with IVC represented by DiGeorges disease (2.35%) and trisomy 21 (2.35%). Nearly half the patients had type IIb VIC (44.71%). The other half were represented by type 1 (18.82%), type IIa (20%), type III (10.59%) and type IV (5.88%). According to site more than two-thirds of VICs (71.64%) were perimembranous in location, followed by infundibular (16.47%) and muscular (11.76%) VICs. In our study 55.29% presented an indication for both surgical intervention and medical treatment, while 16.47% required only medical treatment. In contrast, 28.23% were

placed under exclusive surveillance. Of the 47 patients for whom surgery was indicated, 29 (61.17%) underwent surgical repair, while 18 (38.83%) were awaiting confirmation for surgery. Conclusion: VIC is the most common congenital heart disease. An early detection strategy and the establishment of specialized centers could improve the outcome of these children.

Keywords

Ventricular Septal Defect, Congenital Heart Disease, Ignace Deen

1. Introduction

Congenital heart disease (CHD) is the most common birth defect in the world, affecting millions of newborns every year. They are defined as a structural abnormality of the heart and/or large vessels present at birth [1]. Ventricular septal defect (VSD) is the most common congenital heart defect, accounting for 30% of all congenital heart defects [2].

VSD results from dysfunctional cardiac development, possibly including failure of alignment or fusion of the atrioventricular beads during formation of the interventricular septum [3].

VSDs may occur in isolation or coexist with other types of congenital heart disease [4].

Color Doppler echocardiography combined with two-dimensional (2D) echocardiography facilitates the diagnosis of VSD, measuring the size and position of the defect, and assessing the anatomical relationship with the surrounding tissue. The rate of spontaneous closure of VSD remains controversial, but VSD generally close spontaneously in 12% to 84% of cases. This wide range can be attributed to age, size and position of the defect, diagnostic methods and follow-up period [5] [6].

Hemodynamically, small defects may be inconsequential, but the presence of a large left-right shunt can lead to left ventricular (LV) overload, pulmonary hypertension, ventricular dysfunction and arrhythmias. Surgical closure at an early age remains the treatment of choice, with good medium- and long-term results in terms of survival, morbidity and quality of life [7].

The prevalence of childhood heart disease in sub-Saharan Africa is estimated at around 8 per thousand live births for congenital heart disease [8]. However, they have little or no access to treatment of any kind. Around half of all cases die within a few years of birth, and a third within the first month of life [9].

In Cameroon, a survey covering the period from January 2012 to January 2018 identified 45 cases of VSD out of the total number of congenital heart diseases, *i.e.* 42.9% [10].

In Senegal, Mbaye A *et al.* reported a 12% prevalence of VSD in congenital heart disease from 2003 to 2012 [11].

In Guinea, 30 cases of congenital heart disease were reported from 2017 to

2019 in adolescents and adults by Baldé MD [12].

VICs can lead to serious hemodynamic complications that require early and appropriate management, either by surgical closure or percutaneous closure with a device [13].

Like most countries in sub-Saharan Africa, Guinea has no specialized center for congenital heart surgery.

It is therefore important to carry out a study of VSD in Guinea, in order to describe its epidemiology, clinical and paraclinical profile, and to propose strategies to improve its screening, diagnosis and treatment.

2. Methods

2.1. Study Framework

This study took place in the Republic of Guinea, a West African country in its capital Conakry. The city of Conakry comprises 5 communes (Kaloum, Matam, Dixinn, Matoto and Ratoma). It has a population of 2,317,376 and a density of 5150 inh./km^2 for an area of 450 km^2.

Figure 1. Map of Guinea and neighbouring countries [14].

This study is being carried out at the Ignace Deen National Hospital, in the cardiology department. This department is the country's benchmark for general cardiology.

2.2. Materials

All patients in the CIV cohort during the study period were included in this study. Collection media: for data collection we used a survey form, patient records and operative reports.

2.3. Methods

2.3.1. Type and Duration of Study

This was a retrospective longitudinal study of the cohort of children followed up for IVC, during the period from January 1, 2018 to December 31, 2022.

2.3.2. Inclusion Criteria

We included children under 18 years of age during the study period with an ultrasound-confirmed diagnosis of isolated IVC, whether operated or not.

2.3.3. Non-Inclusion Criteria

The following are excluded from our study:

- VSD within the framework of the atrioventricular canal,
- VSD in tetralogy of Fallot,
- VSD classified as complex heart disease.

2.3.4. Sampling and Collection Procedure

All children meeting the selection criteria were exhaustively recruited. All those included had a medical record that included the entire clinical history, paraclinical data (electrocardiogram, chest X-ray, transthoracic echography) and data on the evolution of the disease. The information contained in the files was collected using a survey form.

2.3.5. Study Variables: The Variables Studied Were

- Sociodemographic: age, gender, education, ethnicity, origin
- Clinical: symptoms, circumstances of discovery, physical signs
- Paraclinical:

- ECG to check for enlarged chambers, rhythm or conduction disorders, or calculation of cardiac axis.
- Chest X-ray for cardiomegaly, pulmonary hypervascularity, pleural effusion, parenchymal condensation…
- Cardiac Doppler ultrasound was used to identify any cardiac anomalies (valvular, cavitary, segmental…) and/or vascular anomalies.
- Biology: look for anemia, ionic disorders, …

- Therapeutic: includes drug treatments (iron, ACE inhibitors, diuretics, etc.) and surgery
- Evolutionary: the evolution of VSD after surgery (residual VSD, pericardial effusion, pleural effusion, etc.).

2.3.6. Data Management and Analysis

Data analysis and processing were carried out using EPI info software version 7.2.3 and Microsoft 365 for data entry, table and figure design. References were

managed by Mendeley.

2.3.7. Consent and Ethics

We have obtained authorization from the ethics committee of the Ignace DEEN national hospital to access patient records.

3. Results

Patient ages ranged from 3 months to 16 years, with an average age of 3.59 years. The most common age group was between 2 and 5 years. There were more boys (54.12%) than girls (45.88%). The most represented ethnic group in our cohort was the Peulh (50.58%), followed by the Malinké (25.88%). Foresters (9.41%) and Sousous were poorly represented (4.70%). In our cohort, 8.24% were from consanguineous marriages, compared with 22.35%. However, 64.41% of the respondents had no information on consanguinity in their records (Table 1). All children from consanguineous marriage in this study belonged to the Peulh ethnic group. In this study 91.76% of children had a history of bronchitis versus 8.24% (Figure 1). The most common clinical signs found were systolic murmur (90.58%), growth retardation (51.76%), difficulty feeding (42.35%), dyspnea (24.70%) and cyanosis (5.88%) (Table 2). Only 4 cases (4.70%) had a congenital malformation associated with IVC represented by DiGeorges disease (2.35%) and trisomy 21 (2.35%) (Table 3).

Table 1. Presentation of children received for VSD at the cardiology department of Ignace Deen National Hospital according to sociodemographic data (1st January 2018 to December 31, 2023, Conakry, Guinea).

Variables	Number	Pourcentage
Age		
[1 - 2 years[	22	25.88
[2 - 5 years[	25	29.41
Average age	3.59 ± 3.58	Extreme 3 months and 16 years
Schooling		
Yes	51	60.00
No	34	40.00
Gender		
Male	46	54.12
Female	39	45.88
Ethnic origin		
Foresters	8	9.41%
Mandingo	22	25.88%
Fulani	43	50.58%
Sousous	4	4.70%
Consanguinity		
Yes	7	8.24%
No	19	22.35%
Not specified	59	64.41%

Table 2. Distribution of children received for VSD at the cardiology department of Ignace Deen National Hospital according to clinical signs (1st January 2018 to December 31, 2023, Conakry, Guinea).

Clinical signs	Number	Pour cent
Systolic murmur	77	90.58
Growth retardation	44	51.76
Feeding difficulties	36	42.35
Dyspnea	21	24.70
Cyanosis	5	5.88
Thoracic deformity	4	4.70
Malformation	4	4.70
Hepatomegaly	3	3.52
Digital Hippocratism	1	1.17

Table 3. Distribution of children received for VSD at the cardiology department of Ignace Deen National Hospital according to associated malformations (1st January 2018 to December 31, 2023, Conakry, Guinea).

Variables	Number	Percentage
Congenital malformation		
Yes	4	4.70
No	81	95.29
If yes		
DiGeorges	2	2.35
Trisomy 21	2	2.35

Almost half the patients had type IIb VSD (44.71%). The other half were represented by type 1 (18.82%), type IIa (20%), type III (10.59%) and type IV (5.88%) (Table 4).

Table 4. Distribution of children received for VSD at the cardiology department of Ignace Deen National Hospital according to hemodynamic classification (1st January 2018 to December 31, 2023, Conakry, Guinea).

Classification	Number	Pour cent
Type 1	16	18.82%
Type IIa	17	20.00%
Type IIb	38	44.71%
Type III	9	10.59%
Type IV	5	5.88%
TOTAL	**85**	**100.00%**

According to site more than two-thirds of VSDs (71.64%) were perimembranous in location, followed by infundibular (16.47%) and muscular (11.76%) (Table 5).

Table 5. Distribution of children received for VSD at the cardiology department of Ignace Deen National Hospital according to anatomical characteristics (1st January 2018 to December 31, 2023, Conakry, Guinea).

VSD characteristic	Number	Pour cent
VSD headquarters		
Infundibular	14	16.47%
Peri membranous	61	71.64%
Muscular	10	11.76%
LV-RV gradient		
Average gradient	48.61 ± 17.96	Extreme 3 et 160
VSD diameter		
Average diameter	12 ± 3.64	Extreme 4 et 22

In our study 55.29% presented an indication for both surgical intervention and medical treatment, while 16.47% required only medical treatment. In contrast, 28.23% were placed under exclusive surveillance (Table 6).

Table 6. Distribution of children received for VSD at the cardiology department of Ignace Deen National Hospital according to therapeutic approach (1st January 2018 to December 31, 2023, Conakry, Guinea).

Therapeutic approach	Number	Pourcentage
Monitoring only	24	28.23
Medical treatment alone	14	16.47
Medical + surgical treatment	**47**	**55.29**
Total	85	100

Of the 47 patients for whom surgery was indicated, 29 (61.17%) underwent surgical repair, while 18 (38.83%) were awaiting confirmation for surgery (Figure 2).

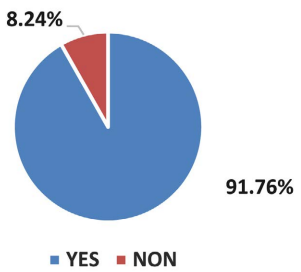

Figure 2. Distribution of children received for VSD at the cardiology department of Ignace Deen National Hospital according to history of bronchitis (1st January 2018 to December 31, 2023, Conakry, Guinea).

Of the 29 patients who underwent surgical repair, 11 (37.93%) were treated in Île-de-France, 8 (27.58%) in Bordeaux, 5 (17.26%) in Switzerland and 3 (10.34%) in Toulouse 2 (6.89%) in Nantes (Figure 3 & Figure 4).

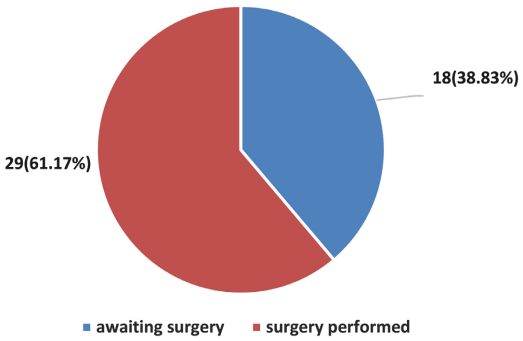

Figure 3. Presentation of children received for IVC at the cardiology department of Ignace Deen National Hospital with an indication for surgical repair (1st January 2018 to December 31, 2023, Conakry, Guinea).

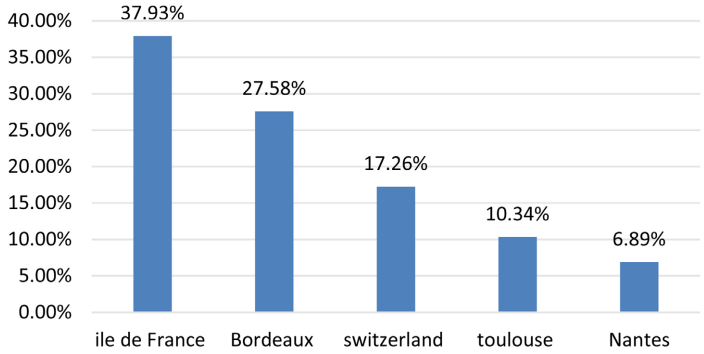

Figure 4. Distribution of the 18 children operated on for IVC at the cardiology department of Ignace Deen National Hospital according to the site that performed the procedure (1st January 2018 to December 31, 2023, Conakry, Guinea).

Complications encountered during follow-up were pericardial effusion (4 cases), residual VSD (3 cases), pleural effusion (3 cases), pulmonary hypertension (2 case) (Table 7).

Table 7. Distribution of children received for IVC at the cardiology department of Ignace Deen National Hospital according to hemodynamic classification (1st January 2018 to December 31, 2023, Conakry, Guinea).

*TTU de suivie	Number N = 29	Pour cent
Pericardial effusion	4	13.79
*VSD residual	3	10.34
Pleural effusion	3	10.34
*PH	2	6.89

*TTU: trans thoracic ultrasound, VSD: ventricular septal defect, PH: pulmonary hypertension.

4. Discussion

Coarctation of the descending and/or abdominal aorta is a rare vascular anomaly, accounting for only 0.5% to 2% of all coarctations of the aorta [4] [5]. This form, discovered in adulthood, is rarely described.

The present study focuses on isolated ventricular septal defects diagnosed in the cardiology department of Ignace Deen.

Firstly, retrospective studies rely heavily on medical records and available data, which can lead to gaps in information collection and variability in data quality.

In addition, only children with suspected congenital heart disease underwent cardiac Doppler echocardiography to confirm VSD. There is therefore a risk of underestimating the prevalence of IVC due to this selection bias.

Despite these challenges, this study provides crucial insight into the disease burden, epidemiological characteristics and challenges encountered in the management of isolated ventricular septal defects in Guinea. It also underlines the need to develop innovative strategies to improve access to specialist healthcare and to strengthen paediatric heart disease management capabilities in resource-limited regions.

The incidence of IVC in children with congenital heart disease in Africa ranges from 17.1% to 67.3% [15] [16]. According to a meta-analysis and systematic review looking at the prevalence of congenital septal defects in patients with congenital heart defects 36.04% in Ethiopia, 37% in Djibouti and at 32.59% in Sudan [17].

The average age of screening may vary according to health policies and medical practices in different regions. In Europe, 92% of congenital heart defects are detected before birth, with an average screening age of 22.2 weeks [18]. In our study, the mean age of detection was 3.59 years. A later diagnosis was found in other studies, notably that of Ba Ngouala GA *et al.* in Dakar, which reported an average age of 8 years [19]. Late diagnosis of congenital heart disease in Africa is a major concern. Limited resources, a lack of personnel qualified in fetal heart ultrasound and the absence of systematic screening programs explain this late diagnosis. These late discoveries have a dreadful prognosis, given the potential for VSD to progress to obstructive pulmonary vascular disease.

VSDs were more frequent in boys than in girls. Male predominance reported in other studies [2] [20]. However, the exact reasons for this sex difference are complex and not fully understood.

The mean age of the mothers in our study was 30.71. According to a recent systematic review and meta-analysis, there is no significant correlation between advanced maternal age and susceptibility to congenital heart disease in the child [21]. However, the study revealed that other maternal factors, such as obesity during pregnancy, smoking during pregnancy, maternal diabetes, and exposure of pregnant women to solvents were significantly associated with an increased risk of congenital heart disease in the child [21].

The Peulh ethnic group was the most represented among the children, and all the children from consanguineous marriages were Peulh. Consanguineous marriage is a common practice in many cultures throughout the world, including the Peulh, where the choice of wife is strongly influenced by the family, which holds the real decision-making power, and social and religious endogamy is virtually established as a rule [22].

The results of a study of consanguinity and congenital heart disease risk suggest that the risk of congenital heart disease is increased in consanguineous unions in the populations studied, mainly at first cousin level [23] [24]. A factor that should be taken into account in empirical estimates of risk in genetic counselling.

Almost all patients in our study had a systolic murmur. A Senegalese study also showed 100% systolic murmur, 40.9% recurrent bronchitis and 86.3% dyspnoea [25].

The intense nature of the VIC murmur prompted echocardiography, which partly explains its high prevalence.

Trisomy 21 or Down's syndrome is one of the most common genetic anomalies in newborns, and the most common chromosomal disorder associated with congenital heart disease [26]. In our study we found 4 cases of genetic malformation, including 2 cases of Trisomy 21.

In this study, 75% of VSD cases were peri-membranous. Similar observations were made by Tougouma S *et al*. [27] with a frequency of 56.8%, and Elmarsawy *et al*. [2] with a frequency of 52%. The reasons why perimembranous VSDs are more frequent than other types are not fully understood, but may be related to the following factors: firstly, it is an anatomically fragile zone since it is the meeting point between different parts of the septum that must fuse during embryonic development; secondly, there is a delayed closure of this zone, making it more susceptible to closure anomalies; and thirdly, perimembranous VSDs are easy to diagnose due to their location.

Type 2a and 2b VSDs were the most common, accounting for 64.71% of cases. A similar observation was made in Burkina Faso [28] where these types of VSD accounted for 64.5% of cases. This high prevalence could be due to late diagnosis and pronounced symptoms of VSD at these stages, prompting consultation at primary care centers. Medical treatment is aimed at infants with large VICs, responsible for congestive heart failure while awaiting surgery or spontaneous improvement [17].

Closure of IVCs is sometimes necessary because spontaneous closure may not occur, with potential complications later on. Surgical closure remains the mainstay of treatment, particularly for large defects [29] as in our study, in which the mean diameter of the defect was 12 mm. However, some defects are more accessible percutaneously [2]. Surgical indication was retained in 46 (54.11%) patients. Despite the absence of a cardiac surgery center, more than half, *i.e*. 29 out of 47 (61.17%), benefited from corrective surgery. However, most African studies report a low percentage of cases operated on, ranging from 0% to 21%

[30]-[32]. The high rate (61.17%) observed in our study could be attributed to the significant commitment of humanitarian NGOs and the proactivity of the referring cardiologists of these NGOs in Guinea.

Residual muscular interventricular septal defects are a surgical challenge, particularly after repair of complex congenital heart defects [29]. An Egyptian study showed 14.7% residual IVC (2) compared with 10.30% in our study. Residual IVC closure is recommended in cases of pulmonary flow over/systemic flow > 1.5 or significant hemodynamic instability (size defect ≥ 2 mm) [29]. None of our patients met these criteria.

In view of the above, to optimize care for children with heart disease, it would be wise to consider setting up a specialized cardiac surgery center. This would not only provide faster care, but also reduce dependence on humanitarian NGOs and treatment abroad. It would also be beneficial to promote the training of local cardiologists and cardiac surgeons in collaboration with foreign institutions, and to train general cardiologists in antenatal detection for early intervention. These proposals could greatly improve access to cardiac care in Guinea. However, it is crucial to emphasize that the realization of these recommendations will require careful planning, adequate resources and sustained commitment.

5. Conclusion

Coarctation

Ventricular septal defect (VSD) is the most common type of cardiac malformation identified at the Ignace Deen National Hospital. Its late detection and the lack of resources for adequate surgical intervention mean that the vital prognosis for children with this congenital condition is uncertain. Developing multidisciplinary strategies and pooling resources in sub-Saharan African countries could help improve the management of this heart condition.

Conflicts of Interest

The authors declare no conflicts of interest regarding the publication of this paper.

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Appendix

1) General information:

- Rhythm: 1[first] consultation // known in cardiology //.
- Age (year): ……..
- Sex: Male // Female //
- In school: Yes // No //
- Mother's age //, maternal pathology: ……
- Ethnic origin: Peulh // Malinké // Soussou // Forestiers //

2) Clinical parameters

- **Medical history:** repeated bronchitis. //
- Circumstances of discovery: Systematic discovery // Dyspnea // Cyanosis // Staturoponderal delay // Other: ……

Age of discovery: 0 - 30 days // 1 month - 12 months // 1 year - 2 years // >2 years.

- Clinical signs: Dyspnea // feeding difficulties // Cyanosis // Staturoponderal retardation // Other: …….
- Physical examination: sytolic sulk // hepatomegaly // cyanosis// thoracic deformity //

3) Sign paraclinical parameters

- **ECG**: RSR// BBD// BBG// HVG// HVD// WPW// others: ……;

a) Biology: Hb, NT proBNP, GB:

b) **Pulmonary x-ray**: normal // cardiomegaly // pulmonary overload // Other: .....

- **Transthoracic ultrasound:**
- VSD location: infundibular // muscular // membranous // admission // subarticular // other: …….
- Classification: Type I // Type Iia // Type Iib // Type III // Type IV //
- Right venticular hypertrophy // Gradient LV-RV: …….
- Left venricular hypertrophy //
- **ETO:** ……..
- **Scanner cardiaque**:
- **KT droit**

4) Care

- Therapeutic approach: Surveillance //Medical only // Surgical only // Medical and surgical //
- Medical treatment: Diuretic // converting enzyme inhibitor // Iron // beta blocker // Other: .....
- Surgical treatment