CASE REPORT

Subaortic ventricular septal defect associated with discrete subaortic membrane and severe aortic regurgitation

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The following work aims to present a rare case of ventricular septal defect diagnosed lately in the childhood, its anatomophysiology, its clinic and its surgical treatment. The necessary criteria that the patient must present for surgical treatment are mentioned in this report. There is also, in some cases, the possibility to perform conservative treatment depending on the classification of the ventricular septal defect, including minimally invasive procedures to repair the subaortic membrane, without the risks of the classic open chest cardiac surgery. It is also emphasized the risk of worsening if the discrete subaortic membrane is left untreated at the time of the closure of the ventricular septal defect.

Key words: **Congenital heart disease; Subaortic stenosis, membrane; Ventricular Septal Defect.**

El presente estudio tiene la finalidad de presentar un caso poco común, de comunicación interventricular subaórtica diagnosticado de forma tardía en la infancia; así como su fisiopatología, clínica y tratamiento quirúrgico. Se mencionan aquí criterios diagnósticos que deben ser cumplidos para ser candidato a tratamiento quirúrgico. En algunos casos existe la posibilidad de un tratamiento conservador, pero esto dependerá del tipo de defecto interventricular y de los procedimientos mínimamente invasivos usados para la reparación de la estenosis subaórtica membranosa, sin el riesgo de la clásica cirugía tradicional abierta. También enfatiza el riesgo de empeorar si se deja sin tratar la membrana subaórtica al momento del cierre quirúrgico de la comunicación interventricular.

***Palabras clave*: Enfermedad congénita del corazón; Estenosis subaórtica; Comunicación interventricular.**

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Congenital heart diseases can be classified as acyanotic and cyanotic. The first include communications (interatrial, interventicular, persistent ductus arteriosus), and valve stenosis. The latter include Tetralogy of Fallot, transposition of the great arteries, among many others [1]. Congenital heart defects may result from changes in the embryonic development of normal structures or from the non-full development of these structures [2]. These defects account for about 8 to 30% of neonatal deaths. In addition, there is a prevalence of congenital heart diseases in 7 per 1000 live births. Birth defects discovered in childhood are the most frequent causes of emergency in pediatric cardiology [3].

Ventricular septal defect (VSD) is the most frequent congenital heart disease, affecting 20 to 30% of the total number of cases [3]. This problem generates pulmonary hyperflow and possible pulmonary venocapillary hypertension, which contributes to the symptoms, such as dyspnea.

VSD may be associated with other heart problems, such as subaortic stenosis. This pathology is included in the subvalvular aortic stenosis group (SAS), which account for about 1% of all congenital heart diseases. Subaortic membrane stenosis has progressive evolution with increased gradient in the left ventricle (LV) outflow tract and also aortic valve insufficiency. This progressive potential is due to the possibility of muscle extension of the lesion, intensifying and making more complex the obstructive process, and worsening the aortic regurgitation. This results from the jet injury produced by eccentric blood flow through the stenotic orifice, and it can lead to aortic insufficiency in rare cases [4].

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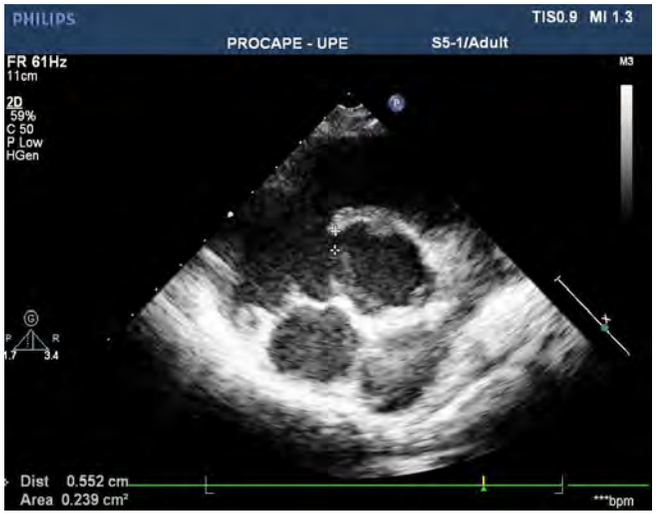

Figure 1. Echocardiography showing a subaortic ventricular septal defect of 5.5 mm in size.

This case report aims to highlight a case of ventricular septal defect associated with subaortic membrane with late diagnosis in childhood, consequences, highlighting semiological and therapeutic aspects.

CLINICAL CASE

A seven-year-old girl from the rural side of Brazil's northeast state of Pernambuco was referred from unit of family care to major cardiac hospital due to systolic murmur (4+/6+) in low left sternal lip with cruciform irradiation, and diastolic murmur in aortic focus (4+/6+). The patient reported dyspnea to great efforts (playing bicycles, running) and three episodes of chest pain in the last year. No cyanosis, lipothymia, or syncope were observed. The physical examination confirmed the murmurs and regular heart rhythm, wide pulses, heart rate of 110 bpm, blood pressure of 100/45 mmHg and no edemas.

Echocardiography showed subaortic VSD (5.5 mm with E-D shunt and maximum VE-VD gradient 97 mmHg); presence of subaortic membrane with maximum and mean systolic gradient in VSVE of 39 mmHg and 21 mmHg, respectively; and major regurgitation through the aortic valve (**Fig 1**).

Chest X-ray in PA showed signs of left atrial enlargement (Double density sign and splaying of the carina) and left ventricular dilatation (left heart border is displaced leftward, inferiorly and posteriorly and increased transverse diameter of the heart) (**Fig. 2**).

With all these preoperative findings in mind, the patient was operated on for VSD closure, subaortic membrane resection and aortic valve repair (**Fig. 3**). The postoperative course of the patient was uneventful. Length of stay in pediatric ICU of 5 days. In-hospital discharge was also free from complications.

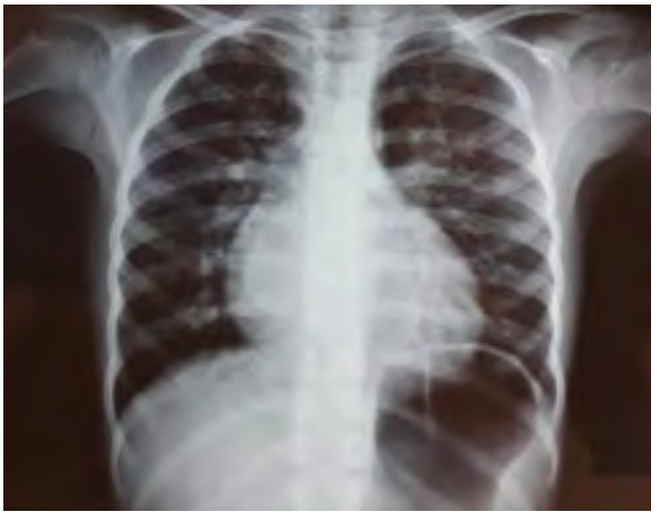

Figure 2. Chest X-ray in PA showing signs of left atrial enlargement (double density sign and displaying of the carina) and left ventricular dilatation (left heart border is displaced leftward, inferiorly and posteriorly and increased transverse diameter of the heart).

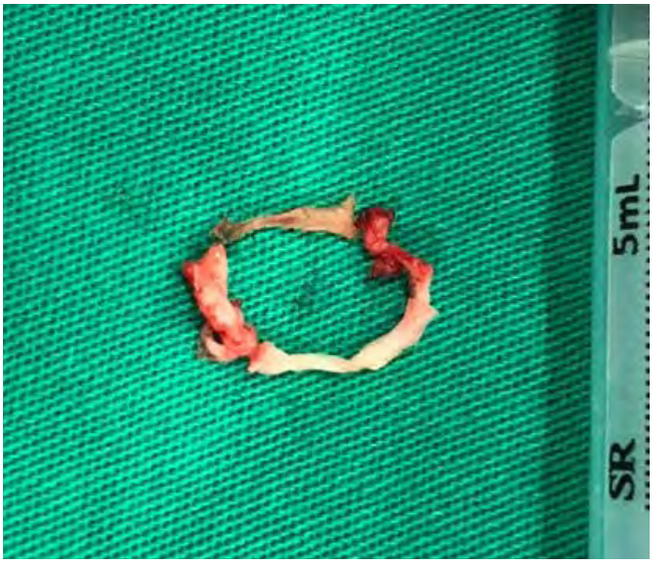

Figure 3. Excised subaortic membrane.

COMMENT

VSD has a congenital character and in the vast majority of cases has no definite cause. However, there are studies that point to a possible correlation between alcohol, phenobarbital, carbamazepine or amiptriline use during prenatal care. In addition, studies also suggest that VSD may be related to the following genetic alterations/congenital malformations: Trisomy of pairs 13,18 and 21; Pierre-Robin syndrome; VACTERL syndrome; diaphragmatic hernia; tracheoesophageal fistula/esophageal atresia [5].

Systolic murmur that may present as holosystolic or as

protosystolic, depending on the patient's VSD. This murmur is produced through the whirlwind of blood generated in passing through the defect. Therefore, the lower the magnitude of the shunt, the greater the intensity of the murmur [6].

Thus, as in the case described herein, valvular heart diseases may be associated with VSD, mainly aortic stenosis and aortic regurgitation causing different murmurs [7]. The murmur in the aortic stenosis is a systolic diamond-shaped murmur characterized by having an ejection click, and is more audible in aortic focus; on the other hand, the murmur of aortic regurgitation is a decreasing diastolic murmur of high frequencies, and is more audible in accessory aortic focus.

VSD can be classified into four subtypes: membranous/perimembranous, muscular/trabecular, inlet/inflow or subaortic/outflow; the latest being the case of the patient [8]. In addition, the septal defects can also be classified according to the magnitude of the shunt, determined by the size of the VSD in the echocardiographic evaluation: small (diameter <5 mm/m^{2}SC), moderate (5 - 10mm/m^{2}SC) or large (>10mm/m^{2}SC). The patient has a shunt of 5.5 mm/m^{2}SC, thus classifying her as a moderate VSD [8].

The magnitude of VSD directly interferes with the patient's clinical presentation: (i) small VSD: asymptomatic child, adequate growth, normal thorax, systolic tactil fremitus that may be present and more localized. Presence of systolic murmur in the left sternal border since the 1st week of life and usually more localized (may be protosystolic); (ii) moderate VSD: heart failure (HF) may not exist or be later. Holosystolic murmur predominates accompanied by fremitus. Moderate precordial bulging and discrete precordial impulses are common physical signs. Harrison groove is not visible; (iii) large VSD: systolic murmur appears at the end of the 1st or 2nd month together with dyspnea and frequent interruption of breastfeeding, deficient weight gain and recurring pneumonia. Chest with precordial bulging, visible systolic thrusts, left-sided palpable ichthyus, visible Harrison's groove, absent fremitus, and increased anteroposterior diameter are some other findings. In auscultation there is discrete systolic murmur in left sternal lip, mitral diastolic murmur and hyperphonetic S2 [8].

The diagnosis can be suspected by the pediatrician or general practitioner and confirmed by a pediatric cardiologist, who, in addition to the clinical history and physical examination, uses complementary tests such as: Chest X-ray, which will be normal in cases of small VSD; in cases of moderate VSD, mild cardiomegaly presents with enlargement of the left atrium and left ventricle, increased pulmonary vascular markings, and bulging of the pulmonary arch can be found. In severe VSD, important cardiomegaly appears with enlarged left ventricle, left atrium and right ventricle may be present; in VSD with pulmonary hypertension, the cardiac area is unchanged, the pulmonary hilum is prominent with bulging of the pulmonary arch. Electrocardiogram evidences alterations from moderate VSD, which will have left ventricular overload. In large VSD, the overload will be biventricular, and in the VSD with pulmonary hypertension the finding will be right ventricular overload. The echocardiogram is able to demonstrate the defect, classify the type and size, evaluate the associated lesions and cardiac repercussions and estimate pulmonary pressure and the pulmonary flow/systemic flow ratio (QP/QS) [8].

The treatment in cases of VSD varies with its classification, because it is known that in some situations the defect closes spontaneously before the patient completes 2 years of life. However, some patients will need drug therapy, which may be necessary in cases with signs of heart failure or pulmonary hypertension. Surgical treatment is indicated in moderate VSD after the first year of life in children with good growth, with QP/QS > 2:1 and pulmonary vascular resistance > 4U/m^2; in large VSD, in infants who do not present good response and inadequate weight gain, surgery should be performed before the patient completes 6 months. Surgery is also indicated in all cases where associated aortic insufficiency exists, regardless the size of the VSD.

However, in patients with subaortic membrane, there are 5 specific criteria to assess the need for surgical intervention, which are: Systolic gradient in left ventricular outflow tract greater than 30 mmHg, left ventricular hypertrophy; valvular area (there is no evidence of from what value the procedure should be performed); presence of symptoms such as dyspnea, angina and syncope and aortic valve insufficiency. The presence of only one of these criteria is enough to consider surgical treatment [6]. Our patient presented 2/5 criteria ratifying surgical indication.

There is, in addition to the conventional method of subaortic membrane resection, the possibility of percutaneous dilation of the stenosis by balloon catheter. This method proved to be safe and effective, but it is only indicated after an echo doppler cardiographic study, by means of evidence of subaortic membrane of thin thickness and distant from the aortic valves, absence of associated muscle component or important aortic insufficiency, which was not the case in our patient [6].

Various types of sub-aortic stenosis have been described; namely, membranous, as a thin fibrous diaphragm below the aortic valve; fibromuscular, as a fibromuscular ring is located in the left ventricular outflow tract; tunnel stenosis; and hypertrophic obstructive cardiomyopathy. The term discrete sub-aortic stenosis is applicable to the first two [9]. The exact incidence of this association as VSD with discrete sub-aortic stenosis is not well known.

To what extent the VSD and subaortic fibrous ridge are associated to septal malalignment? There has been previously published the foregoing association as consistent as 100% ($p< 0.00001$) in a study by Zilelensky at al. [10]. In addition, it has been proposed that the turbulent flow coming from the septal malalignment could be the responsible to enhance the fibrous subaortic membrane. Thus, it may also damage the aortic valve leaflets and produce aortic valve regurgitation [11].

The surgical closure of a VSD can stimulate the progression of a previously silent subaortic ring. According to this, the progression of a fibrous subaortic membrane could be present after VSD closure if misdiagnosed or overlooked the subaortic membrane [12,13].

As a conclusion, VSD is, among congenital heart defects, the most frequent pathology, and may progress with development of subaortic membrane and generate stenosis, and in rare cases aortic insufficiency. Thus, it is extremely important to actively search for congenital anomalies in early life in or-

der to early diagnose and treat these patients as soon as possible, according to the current guideline's parameters [14].

Therefore, what is worthy to emphasize in this article is the importance of being aware about this association, given the possibility of worsening if the subaortic membrane is overlooked at the moment of the VSD closure. It carries a high risk of left ventricular outflow tract obstruction in the mid or long run after operation. Thus, this association should be routinely searched in the presence of VSD, and once detected, it always be totally excised.

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