

Ventricular Septal Defect with Aortic Regurgitation. An Unsolved Problem

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In the varied, complex, and broad spectrum of congenital heart disease, carriers of ventricular septal defect are not uncommon (1.5 to 2 per 1000 live births). Since the prognosis of this defect is good, it could be thought to have scant relevance, especially since we know that 60% of cases remain asymptomatic and 70% close spontaneously with time. The rest, that is to say, the symptomatic defects, require either surgery during infancy or can be managed with medical treatment and slowly pass into the group of asymptomatic ventricular septal defects, due to either a real decrease in size or a relative decrease deriving from changes in the body surface area. Nevertheless, in a small percentage of cases the proximity of the defect to anatomical structures of relevance, such as the atrioventricular or semilunar valves, compromises their function and modifies not only the clinical condition, but also the long-term prognosis of patients.

In this commentary we will discuss only defects in the outlet septum, also known as *subarterial defects*. In these cases, the anatomical position of the defect frequently affects the mobility and correct function of the aortic semilunar cusps, leading to the appearance of aortic incompetence, which is generally mild at first but progressive in nature. In most cases this demands surgical intervention, not only of the defect, but also the aortic valve. These interventions do not always resolve the condition. The percentage of this type of defects is low, ranging from 3% to 4.5% according to series.^{1,2} However, it seems to have a greater incidence in some ethnic groups³ in which conal malformations are more frequent, and in such groups the frequency of this type of defects can reach 8.5% to 10% of all cases of ventricular septal defect.

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It seems to be clear that in no case is incompetence present at birth. Although the series with the longest follow-ups, which are the oldest series, do not have early echocardiographic studies, the color Doppler technique used in the most recent series confirms this. With respect to the severity of the aortic incompetence, its speed of progression, and its relation with the size or location of the defect, the results vary as widely as the anatomical forms and the involvement of the aortic semilunar cusps. This explains the different results obtained and the variety of surgical techniques used to resolve the condition.^{1,4,5} In order to understand the problem, it is sufficient to recall certain anatomical aspects that have been studied in depth by some groups.⁶⁻⁸

The *subarterial defects*, or the defects of the outlet septum, that can cause aortic incompetence are of two basic types. The *supracristal* or *conal* defects affect the development of the conal septum and are located below the anteriormost portion of the right coronary cusp, and also include some forms of outlet septum defects. The *infracristal* defects are located immediately below the conal septum and affect the outermost third of the right coronary semilunar cusp, the commissure, or the non-coronary cusp. The cases that most frequently cause deterioration of the aortic cusps are the supracristal defects, whereas the aortic valve is involved only in larger infracristal defects and after a long evolution. In the *supracristal* defects, the deficient development of the conal septum (*crista supraventricularis*) allows continuity between the aortic and pulmonic cusps, which is why they are also known as doubly committed ventricular septal defects, in which both the aortic and pulmonic cusps form the roof of the defect. The defect in the infundibular septum is not usually very large and the mechanism of development of aortic incompetence proposed would be a lack of support of the semilunar cusp related with the defect, resulting in a gradual prolapse through the defect, which appears in the right ventricular infundibulum. This type of defect includes an anatomic variant that can be difficult to identify without precise echocardiographic studies, in which a supracristal defect coexists

with a small muscular ridge between the aortic and pulmonic cusps (actually intraconal defects) to which the aortic ring would be attached, making the aortic incompetence less significant.⁷

The review of the literature disclosed reports on other factors that favor the development of aortic prolapse. An anomaly of the aortic wall itself^{1,6} is the factor most frequently mentioned. Whereas some groups refer to thinning,⁶ others describe the cusp as thick and rigid.¹ Since no anatomopathological studies of this structure exist, this explanation is used to account for observations as different as the evolutionary changes in the disease, with thinning corresponding to the initial phase and fibrosis and rigidity to the later effects of jet stream injuries. The fact that there are isolated forms of aneurysm of the Valsalva aortic sinuses,⁹ which can occasionally be perforated, obliges us to consider the primary anomaly of the vascular wall as a coadjuvant factor in the development of prolapse. It could also explain the variations in the progression of aortic incompetence in this pathology. The severity of aortic incompetence seems to be clearly related with time of evolution. Since supravalvular defects are generally small, the prolapse is initially non-existent or very mild. For this reason it is interesting to discuss the finding by Hernández Morales et al, published in this issue of the journal,¹⁰ which describes a more intense aortic incompetence in patients with small defects. This observation is difficult to explain without invoking the effect of the «primary vascular lesion,» to which would be added the protrusion with time of the sinus of Valsalva toward the defect as a result of the Venturi effect of the jet stream on the aortic cusp during systole. It is also likely that the size of the defect is related to the degree of aortic incompetence in some series, without considering the location factor. Some series may include patients with infravalvular defects that, despite being larger, are less frequently associated with aortic prolapse.⁷

The natural history of these defects is a progressive deterioration of the semilunar cusp involved, attended by progression of the valvular insufficiency.^{2,11} Sometimes the prolapse is included in the defect, eventually producing occlusion of the defect and modifying the previous clinical condition to the point that it is expressed as severe aortic incompetence. In these advanced forms, diagnostic echo-Doppler may be difficult because it fails to demonstrate a ventricular septal defect that may be found in surgery. It is important to note that we do not know of the existence of any publication citing the presence of pulmonic valve incompetence or cusp prolapse related with ventricular septal defect. Since jet stream injuries also occur in the pulmonic valve, the defective pulmonic ring provides poor support, and both the aortic and pulmonic cusps have a common origin (in the truncal cushions), the lack of involvement of this valve would have to be attributed to the protection provided by the low pulmonary diastolic

pressure. This reasoning suggests that preventive treatment with vasodilators may be appropriate in cases of incipient aortic regurgitation. All these factors, and the knowledge that this type of defect does not close spontaneously,^{1-3,6-8,10} suggest a medical strategy of early surgery, before the lesion progresses.

The second group of defects that can cause aortic regurgitation are *infravalvular* defects. In these defects the infundibular septum is conserved and the contiguity with the aortic valve depends on the type of defect,⁶⁻⁸ which can be located in the intercommissural zone of the right coronary cusp and non-coronary cusp, or under the non-coronary cusp. In the first case, aortic incompetence is established in the commissure due to poor coaptation of the leaflet margins, and the jet stream of aortic regurgitation is channeled to both the left ventricle and right ventricle as a result of the normal anatomic straddling of the aortic annulus on the interventricular septum. These defects are generally larger than the supravalvular defects and usually do not cause prolapse of the aortic cusps. Infravalvular defects in the posteriormost region to the right of the conal septum are usually larger, and often affect the membranous septum.⁶⁻⁸ A small muscular ridge may separate it from the corresponding cusp, which is why aortic prolapse is usually important and involves the non-coronary cusp when it eventually appears. It is likely that in these cases jet stream injuries and the primary anomaly of the vascular wall contribute to the poor evolution.

There are as many surgical techniques,^{1,2,5,11,12} as types of defect and aortic valve involvement. Generally speaking, in patients with a doubly committed supravalvular defect, in which the lack of support of the semilunar cusp seems to be the basic mechanism underlying the prolapse, simple closure of the defect with a patch should solve the problem. This technique would be ideal for cases with a small muscular ridge and conserved aortic ring. Highly evolved cases with a large prolapse require the placement of a double patch, one on the defect and the other on the aneurysm. A small plicature is added to enhance patch support.⁵ The best results are obtained, as can be concluded from a review of the literature, in cases with infravalvular defects in which the double patch is placed immediately below the commissure separating the right coronary and non-coronary cusps. Closure of the defect with a patch, associated with plasty of the commissural zone, suffices to control the evolution of aortic incompetence. Finally, defects that are more removed from the valve, located near the membranous septum, and are associated with prolapse usually involve more important prolapse and are difficult to correct since closing the defect does little or nothing to improve the support of the semilunar cusp. Consequently, major action is needed but it generally fails to resolve the problems of the aortic valve. By applying a variety of techniques, it is possible to reduce the degree of aortic incompetence

and slow its progression in most cases, but reinterventions² for valve replacement or homograft implantations are not infrequent in the intermediate or short term.

In view of these findings, we can assume that a series of factors coincide in this malformation as manifested by a condition with specific characteristics that has been previously identified (Laubry-Pezzi). In such cases, we should suspect the presence of simultaneous anomalies in the development of truncal (semilunar cusps) or conal (conal septum or *crista supraventricularis*) structures. Likewise, it is fairly straightforward to draw certain conclusions of practical utility for the treatment of children with these anomalies: *a)* The clinical diagnosis of a suspected ventricular septal defect, although small, must be followed by a color Doppler echocardiographic study to identify its location. If the defect is located in the outlet septum, its relation with the aortic and pulmonic semilunar cusps and the presence or absence of aortic regurgitation must be clearly defined, as noted by Hernández Morales et al;¹⁰ *b)* once the diagnosis is made, periodic follow-up visits should be scheduled for the purpose of promptly detecting the appearance of aortic incompetence or distortion of the aortic cusps; *c)* we propose the evaluation of protection against hypertension (vasodilators) of the aortic valve in patients in which an evolution towards aortic regurgitation and/or prolapse of a cusp can be anticipated, and *d)* surgery should be scheduled without awaiting the development of incompetence or a prolapse, unless mild, and performed soon in the case of infracristal defects in which surgical treatment is less capable of resolving the problem once prolapse has appeared. Only an intervention of this type can improve the long-term prognosis of these patients, in which the need for eventual implantation of an aortic prosthesis or homograft hangs over their heads like a sword of Damocles.

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