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Editorial

Atrioventricular septal defect—anatomic characteristics in patients with and without Down's syndrome

Bruno Marino

The recent issue of ***Cardiology in the Young*** (October 1991, Volume 1, Number 4) was concerned exclusively with papers on atrioventricular septal defects. These papers were interesting and accurate. Little attention was paid, nonetheless, by the various authors to the anatomic differences between patients with and without Down's syndrome, and, obviously, no consideration was given to possible implications for clinical and surgical aspects of the lesion. The presence of Down's syndrome cannot be considered occasional in patients with an atrioventricular septal defect since it occurs in one-third to one-half of cases, according to different published series.[1-3] One of the differences between patients with and without Down's syndrome is the higher prevalence of forms with common, as opposed to separate, valvar orifices in the presence of trisomy 21 and, vice versa, of the forms with separate right and left valvar orifices in patients with normal chromosomes.[2-4] Furthermore, when the variant with a common atrioventricular orifice is present in patients without Down's syndrome, Rastelli type A[5] and the so-called intermediate form, prevail.[6]

Another interesting finding is that, in patients with an atrioventricular septal defect without Down's syndrome, the left (or "mitral") valve tends to be attached to the crest of the ventricular septum, and this can be the substrate for obstruction of the left ventricular outflow tract.[7,8]

In 1986, we provided the first statistical documentation of the prevalence of obstructions of the left ventricular outflow tract in patients with atrioventricular septal defect without Down's syndrome,[9] including both patients with a common atrioventricular orifices and also those with separate right and left valvar orifices. We also cited previous anatomic studies[1-10] that supported these data. Thus, in 1978, Freedom, Bini and Rowe[10] reported 12 patients with significant hypoplasia of the left ventricle of whom only one (8.3%) had Down's syndrome. In 1984, Van Praagh and colleagues[1] described 12 patients with stenosis of the subaortic outflow tract of whom three (25%) had Down's syndrome. Penkoske and colleagues[2] reported only six specimens (15%) with left-sided anomalies among 39 cases with Down's syndrome, while left-sided anomalies were found in 28 out of 56 (50%) hearts from patients without Down's syndrome. In a more recent report, de Leon and colleagues[11] described the surgical treatment of 12 patients with subaortic stenosis. Only three of them had Down's syndrome. Interestingly, the subaortic obstruction was recognized before repair of the atrioventricular septal defect in only two cases. One could speculate that increasing awareness of this potential problem in patients without Down's syndrome might allow for an earlier diagnosis, thus avoiding the need of reoperation in such patients.

In contrast, data reported by Gow and colleagues[12] seem to diverge from these concepts. In 30 patients with an atrioventricular septal defect and either aortic coarctation or subaortic stenosis, they did not find significant differences between patients with (13 cases) and without (17 cases) Down's syndrome. In their selection of patients, however, the authors excluded two cases with bicuspid aortic valve: one patient with severe aortic stenosis and all patients with right ventricular dominance. Our studies[3,9,13-16] in patients with atrioventricular septal defect have suggested that the main difference between patients with and without Down's syndrome is probably related to the morphology of the left ventricular inlet. Thus, malalignment or malorientation of the atrioventricular junctions (right ventricular dominance or double outlet right atrium), and malformations of the left atrioventricular valve (double orifice) and of its subvalvar apparatus (parachute valve), occur more often in patients without Down's syndrome. These associated anomalies may cause various degrees of hypoplasia of the left ventricle[3,9,10,13,15,16] and also affect the development of the left ventricular outflow tract and/or the aortic arch.

This increased prevalence of right ventricular dominance, and the association with left-sided obstructions

in the subgroup of patients with usual atrial arrangement, atrioventricular septal defect, and normal chromosomes are similar to those observed in patients with isomerism of the left atrial appendages and the polyspenia syndrome.[17]

Another recently recognized feature[18,19] is the rarity of additional ventricular septal defects in the apical muscular septum of patients with atrioventricular septal defect and Down's syndrome. This new information was recently endorsed by the investigators of the Baltimore-Washington Infant Study who had previously confirmed the rarity of left-sided anomalies in patients with Down's syndrome.[20] While the causes of this anatomical differences are unknown, their important clinical and surgical implications should be recognized. Patients with a degree of left-sided obstruction, the most severe being stenosis of the inlet, may present early in life with profound congestive heart failure.[9,10,13,15] The surgical options vary according to the type and severity of the associated malformations. Recent studies describe a higher surgical mortality in patients without Down's syndrome after repair of cases with both separate[15] and common[22-24] atrioventricular orifices. It is also the case that infants without Down's syndrome carry a significantly higher hazard of need for reoperation to correct severe post-operative regurgitation of the left atrioventricular valve compared to those with trisomy 21.[25-27]

In conclusion, therefore, patients with atrioventricular septal defect without Down's syndrome differ from those with trisomy 21 because of the higher risk of left-sided anomalies. This notion must be considered in the light of anatomical studies, in the diagnostic assessment, and in the surgical treatment of these patients. Furthermore, these children with normal chromosomes may also have serious extracardiac anomalies representing, in a number of cases, recognizable syndromes.[23,28,29]

The increasing interest for the genetic imprinting in congenital heart diseases may now stimulate further studies on morphogenesis and anatomy of atrioventricular septal defect to the better understanding of clinical and surgical findings of all categories of patients with this malformation.

Department of Pediatric Cardiology
Bambino Gesù Hospital
Via San Onofrio 4
Rome, Italy

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