Re: Isolated ventricular septal defects in the era of advanced fetal echocardiography: risk of chromosomal anomalies and spontaneous closure rate from diagnosis to age of 1 year. O. Gómez, J. M. Martínez, A. Olivella, M. Bennasar, F. Crispi, N. Masoller, J. Bartrons, B. Puerto, E. Gratacós. *Ultrasound Obstet Gynecol* 2014; **43**: 65–71.

Gómez *et al.* have examined the characteristics and outcome in a group of 248 fetuses diagnosed with isolated ventricular septal defects (iVSD) over a 6-year period in a tertiary referral center. They focused on the association with extracardiac defects and the rate of spontaneous closure by the age of 1 year, with complete data on 211 of them.

They found that cases of iVSD represented 25% of their total volume of cardiac pathology, a higher proportion than reported in other fetal studies. The mean gestational age at diagnosis of iVSD was 30 (range, 17–41) weeks. Crucial to their diagnosis was the observation of bidirectional flow across the defect on color flow mapping in the fetus, thus excluding a false-positive diagnosis in an artifact.

For the sake of simplicity, and because of limited numbers, they divided their cases for analysis into just two groups: perimembranous and muscular defects. Muscular defects were more common than were perimembranous defects, by a factor of seven. There was only one clinically significant chromosomal defect, found in association with a perimembranous defect.

The authors measured the size of the defect and compared it with the size of the aortic root as a ratio. This ratio is used as a rough guide for the pediatric cardiologist: a defect larger than half the size of the aorta is likely to be symptomatic. The well-recognized tendency for most VSDs is to close spontaneously and this was confirmed by the authors in that they found that over 80% of the defects they detected did so, some even before birth. Spontaneous closure was more common in the muscular defects than in the perimembranous region and was related to size, the larger defects being less likely to close. Both of these observations are consistent with postnatal experience.

Why did the authors find so many, mainly muscular, iVSDs in their series? Detailed echocardiography in asymptomatic neonates shows small muscular defects on color flow mapping in up to 20% of cases, almost all of which close spontaneously. In contrast, echocardiography in infants presenting with a cardiac murmur or breathlessness usually around 2 months of age, will show mostly perimembranous defects, hence the contradictions in reported findings depending on the timing and method of examination. Detailed echocardiography in the last 10 weeks of pregnancy might be expected, therefore, to show a similar high incidence of small muscular defects, as found here. There were no iVSDs diagnosed before 17 weeks and over 70% were diagnosed in the last 10 weeks of pregnancy, an unusual time for fetal echocardiography in most experience. The proportion of muscular to perimembranous iVSDs found in this study may not, therefore, be applicable to other centers, in which most fetal echocardiograms are performed around 20 weeks' gestation.

The rate of spontaneous closure might have been different had the authors been able to further classify the perimembranous defects into inlet, outlet or doubly committed, as this can affect the tendency towards closure.

Measurement of a defect, particularly in the muscular septum, is notoriously inaccurate, as it is a one-dimensional measurement of a defect which is often elliptical in shape. The authors acknowledge this and describe their attempts to minimize measurement error. Three-dimensional imaging of a muscular defect using spatiotemporal image correlation may be used to overcome this potential for inaccuracy in the future.

The low (3%) rate of need for surgical closure again reflects the particular population in this study, but it is of interest that it was not necessary in any defect, regardless of its position, that measured smaller than 50% of the aortic size. However, it is important to realize that the criteria for surgical closure do vary somewhat in different surgical centers.

In contrast to previous series, the authors found a low rate of chromosomal anomalies in association with iVSD, but this may be because they were examining mainly what proved to be otherwise normal pregnancies late in gestation and they excluded those with known extracardiac anomalies or those found subsequently to have extracardiac malformations, all of which are likely to have biased their data towards chromosomally normal fetuses. Even so, their conclusion, that karyotyping is not necessary in the setting of a fetus with a muscular iVSD, a low risk from the nuchal scan and no other abnormal findings, is probably correct. In contrast, the detection of a significant perimembranous defect probably is an indication for the offer of karyotyping, although the precise risk would be difficult to quantify accurately.

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DOI: 10.1002/uog.13237