**Objective**

Atrioventricular septal defect (AVSD) constitutes an indication for fetal karyotyping, despite the existing risk of pregnancy loss related to invasive procedures. The aim of this study was to assess the rate of trisomy 21 in fetuses diagnosed with an AVSD in the era of first trimester screening and to ascertain if the rate differs according to first trimester risk for trisomy 21.

**Methods**

Fetuses diagnosed with an AVSD from 2002 to 2014 were retrospectively identified. The overall rates of trisomy 21 and other aneuploidies were calculated among cases with normal situs. The prevalence of trisomy 21 and other aneuploidies was also assessed in the subgroups of women with low and high first trimester risk for trisomy 21, using a cut-off risk value of 1:150.

**Results**

A total of 116 fetuses were identified. AVSD was diagnosed at median gestation of 21 weeks (IQR: 17.6-22.4). Among 103 fetuses with normal situs, 72 fetuses had an isolated AVSD, and 31 fetuses had an AVSD associated with other abnormalities. The prevalence of trisomy 21 among fetuses with normal situs was 45% (95%CI: 35-54%). Within the low risk group the rate was 41% (95%CI: 27-57%) while in the high risk group the rate was 67% (95% CI: 50-80%), significantly higher than in the low risk group (p= 0.036).

**Conclusion**

Despite first trimester combined screening, the rate of trisomy 21 among fetuses diagnosed with an AVSD in the second trimester remains high. Thus, advising for fetal karyotyping by invasive procedure is still a reasonable choice irrespective of pregnancy being identified as high risk or low risk group in the first trimester.