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Evaluation of Cardiac Defect in a Fetus with 8p Interstitial Deletion

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ABSTRACT About 30% of all malformations are of genetic origin, and congenital heart defect (CHD) is one of them. Here we report a case at 17 weeks of gestation with a small ventricular septal defect (VSD) revealed an abnormal karyotype of 46,XY,del (8) (p11.2-p21) in an amniotic fluid sample. The fetus at 21 weeks showed a progress of VSD to double outlet right ventricle, and in addition showed cleft lip, Intra Uterine Growth Retardation (IUGR) and oligohydramnios. Apart from being the first prenatal case to report a deletion spanning the region 8(p11.2-p21), this case also highlights the importance of prenatal diagnosis for cases with even a small VSD in the early scan and special attention should be paid to chromosome 8p and a proactive follow-up should be carried out to see if there is progression in the heart defect and other multiple congenital/phenotypic anomalies.