Prenatal diagnosis of WAGR syndrome at 20 gestational week
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- 22 year-old caucasian ethnicity woman in her second pregnancy was referred to our perinatology clinic at 14 gestational week due to increased (4mm) nuchal translucency. The couple were nonconsanguineous. They were counselled regarding the possibility of genetic or chromosomal causes, and they opted for chorion villus sampling. The result of conventional karyotype analysis showed a normal compliment of chromosomes 13, 18, and 21 and an XX (female) chromosome complement at 18 gestational week then sonographic examination was performed. Abnormal examination findings were absent cavum septum pellucidum with suspicion of agenesis of corpus callosum and hypoplasia of vermis with widened cisterna magna. The performing microarray analyse was asked from genetic department with before CVS material. Microarray analysis at 20 gestational week indicated a deletion of approximately 8.6 Mb. Deletion of this region is associated with WAGR 11p13 retardation syndrome. At 20 gestational week examination, fetal heart was structurally normal and cranial findings were same with before examination. There was not any renal and genital pathology. After genetic consultation the pregnancy was terminated. This report is the second report about WAGR syndrome that diagnosed prenatally in English recent literature. In order to diagnose this rare syndrome, although the conventional karyotyping resulted normal, when there is a sonographic abnormalmality, microarray analysis should be performed. The prognostic information related to diagnosis from array analysis may alert other potential co-morbid conditions that cannot be predicted on the basis of sonographic examination alone. This is particularly important for providing the most accurate interpretation to parents that helps make a decision about the option of pregnancy termination.