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## PP-10 Phenotypic manifestations of copy number variation in chromosome 11p11.2-p11.12

Cagdas Nurettin Emeklioglu<sup>1</sup>, Elif Akkoc Demirel<sup>2</sup>, Mirac Ozalp<sup>3</sup>

Karabuk University Trainning and Research Hospital, Department of Obstetrics and Gynecology, Karabuk, Türkiye <sup>2</sup>Prof. Dr. Cemil Tascioglu City Hospital, Department of Obstetrics and Gynecology, Istanbul,

Türkiye

<sup>3</sup>Prof. Dr. Cemil Tascioglu City Hospital, Department of Obstetrics and Gynecology, Perinatology Clinic, Istanbul, Türkiye

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**Objective:** Advanced maternal age pregnancies are an increasing incidence, especially in developed countries. As the age of conception increases, the risk of chromosomal anomalies and genetic diseases also increases. Detection of chromosomal anomalies such as deletion, microdeletion or duplication-microcopying has become possible with developing technology and increasing clinical applications such as new generation sequencing analysis and chromosomal microarray analysis.<sup>[1]</sup> Chromosome 11 encompasses approximately 135 million DNA building blocks and contains approximately 1300-1400 proteincoding genes.<sup>[2]</sup> These proteins perform various functions in the body. Changes in the structure or copy number of the 11th chromosome lead to different clinical pictures. The common features of previous cases in this extremely rare chromosomal anomaly are autism, mental and motor retardation, growth retardation, polyhydramnios, dysmorphic findings, hypotonia, and macrosomia.<sup>[1,3]</sup>

**Methods:** The computer-based and ultrasonography records of the case, who applied to the Perinatology outpatient clinic of Prof. Dr. Cemil Taşçıoğlu City Hospital at the 13th week of pregnancy, had NIPT due to advanced maternal age and the result was reported as multiple chromosomal anomaly, were analyzed retrospectively from the hospital system and the history of the ultrasonography device. Fetal ultrasonography examination was performed using Mindray Resona 7 device and its 1.2-6 MHz convex abdominal probe. Ultrasonography findings and patient history were noted.

Case: 41-year-old patient with gravida 2, parity 1 applied to our clinic for the first trimester screening test. In the anamnesis taken from the patient, it was learned that she had a history of cranial hemorrhage due to methyl alcohol poisoning 6 years ago, that she had vision loss at a rate of 60%, and that she continued to use alcohol regularly throughout her pregnancy. Non-invasive prenatal testing (NIPT) was recommended to the patient due to advanced maternal age, and a cystic lesion of approximately 5 cm in the left ovary was observed in the left ovary, which was compatible with endometrioma. Amniocentesis was recommended to the patient after NIPT revealed multiple chromosomal anomaly. Amniocentesis was performed on the patient at the 20th week of her pregnancy. In the ultrasonography performed at the 20th gestational week, the fetus was measured as compatible with 22 weeks. Macrosomia, macrocephaly and polyhydramnios were present. TORCH and OGTT test results were normal. Genetic analysis was requested from the parents after the fetus with a normal karyotype result was found to have a 2.65 Mb (303 probe) copy number variation (CNV) increase in the 11p11.2-p11.12 region of 11p11.2p11.12(47814450\_50468342)x3 case in array CGH.

After the parents' genetic tests were reported as normal, de-novo mutation was considered and genetic counseling was given to the patient. During this period, all fetal biometric measurements continued as >97th percentile. Fetocyte and termination procedure was performed at the 32nd week of pregnancy with the decision of the pregnant woman and her husband, after the findings of macrosomia, macrocephaly and polyhydramnios were accompanied by ultrasound, regular alcohol use throughout the pregnancy and the presence of 2.65 Mb CNV in the 11th chromosome. On the 1st day after termination, the patient was discharged without any complaints.

**Results:** In patients with advanced maternal age risk factors, first trimester combined screening test and antenatal screening tests, especially NIPT, are of great importance in determining the risk of fetal chromosomal anomalies. Multidisciplinary management of rare chromosomal anomalies such as the case we have presented, together with Medical Genetics, provides detailed information to families. The increasing complexity of genetic counseling as a result of advanced genetic studies can be overcome by joint studies between branches.

Keywords: Microarray, chromosomal anomaly, NIPT, fetal anomaly, prenatal screening tests.

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# PP-11 Prenatal diagnosis of a placental cyst with intracystic hemorrhage case report

Ceylin Elbistanlı<sup>1</sup>, Melda Kuyucu<sup>2</sup>, Mehmet Serdar Kütük<sup>3</sup> <sup>1</sup>Bezmialem Vakıf University, Faculty of Medicine, İstanbul, Türkiye <sup>2</sup>Bezmialem Vakıf University, Department of Obstetrics and Gynecology, İstanbul, Türkiye <sup>3</sup>İstinye University, Department of Obstetrics and Gynecology, İstanbul, Türkiye DOI:10.59215/prn.23.031supp026

**Objective:** Placental cysts (PC) are usually benign masses on the surface of the placenta and can be detected with ultrasonography in the early stages of pregnancy. Although PCs are clinically insignificant and don't require additional follow-up in most cases, larger cysts may cause intrauterine growth restriction (IUGR), intrauterine asphyxia, and preterm delivery. In this study, we present a case of a placental surface cyst complicated with intracystic hemorrhage.

Case: A 33-year-old, gravida 2 para 1 woman was referred to Bezmialem Vakıf University, Obstetrics and Gynecology clinic at 36 weeks of gestation with the detection of a placental cyst at detailed ultrasound. Besides the rhesus incompatibility and a dermoid cyst, the patient's obstetrics, personal, and family histories were normal. Her detailed ultrasound report showed a thin-walled, anechoic placental surface cyst measuring 2.7x1.8 cm at its widest point, located adjacent to the umbilical cord insertion to the placenta. In our sonographic examination at the 36th week, the fetal development, and the amniotic fluid index were within normal limits. We observed a placental cyst with the dimensions of 6x7.3 cm containing a 5.3x3.8 cm echogenic area suggesting intracystic bleeding. Middle cerebral artery peak systolic velocity was 39 cm/s. A healthy, 3480-g female neonate was born at 38 weeks of gestation via cesarean section. Separation of the placenta resulted in uterine inversion. After the eversion of the uterus, the placenta was sent to pathology. The pathological examination confirmed a placental subchorionic cyst with an undamaged capsule and an area of infarct measuring 8x7x4.5 cm. When the capsule was discharged, brown-colored hemorrhagic fluid was revealed



**Fig 1.** Ultrasound showing the placental cyst with intracystic hemorrhage area



**Fig 2.** The Doppler Ultrasound reveals the umbilical cord insertion site and the placental cyst, which notably exhibits no detectable blood flow.



**Fig 3.** The image depicting the placenta alongside a placental cyst, with the umbilical cord positioned on the right side

**Conclusion:** In this case, we presented the diagnosis and management a subchorionic placental cyst complicated with intracystic hemorrhage. Despite its benign course in most cases, subchorionic placental (surface) cysts are prone to hemorrhage as in our case. The extent of the hemorrhage and the position of the cyst may be predictive for perinatal complication such as IUGR, fetal anemia and fetal distress. Therefore, follow-up US of the cyst with regard to size, echo-texture, MCA-PSV should be implemented in these cases. More cases should be collected and reported to establish antenatal follow-up and delivery protocol for PC cases.

Keywords: Hemorrhage, placental cyct, ultrasound

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