25th European Congress of Obstetrics and Gynaecology

in conjunction with

15th Congress of Turkish Society of Obstetrics and Gynaecology

17 - 21 May 2017
Titanic Deluxe Hotel, Antalya / Turkey

ABSTRACT BOOK

www.ebcog2017.org

P 576 PRENATAL DIAGNOSIS OF MOSAIC TRISOMY 9

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Introduction: Trisomy 9 is an uncommon lethal chromosomal disorder with characteristic features such as growth and mental retardation, dysmorphic faces, low-set malformed ears, microphthalmia, small mouth, bulbous nose, high arched palate, congenital heart defects, genitourinary, skeletal and central nervous system abnormalities. Genetic counselling is difficult because all fetuses do not exhibit these typical phenotypic characteristic, and some can be found normal. Long term survivors are only possible in case of mosaicism. In this case we present mosaic trisomy 9 case with subtle phenotypic clues.

Case: A 41 year-old, gravida 2, abortus 1 patient with a history of previous cesarean section, and with a twenty weeks and two days of pregnancy according to last menstrual period and previous ultrasonographic calculations. She was referred to our clinic because of right fetal renal hypertrophy. Age risk was 1/84 and first trimester combined fetal aneuploidy test results were 1/1440. Fetal biometric measurements were three weeks retarded and right fetal renal pelvis longitudinal diameter was 18.2 mm and transverse diameter was 8.8mm. Amniocentesis was performed because of advanced maternal age, fetal growth restriction, and renal pyelectasis. Mosaic trisomy 9 was detected. Family rejected pregnancy termination. However, oligohydramnios and fetal demise was encountered three weeks later in the course of pregnancy.

Conclusion: Multiple abnormalities and phenotypical characteristics can be detected is trisomy 9, whereas normal phenotype can also be encountered. In this case only observed structural abnormalities were fetal growth restriction and renal pyelectasis.

Keywords: trisomy, prenatal diagnosis, mosaicism