17th World Congress in Fetal Medicine

Prenatal diagnosis of cat eye syndrome – case series

Kazdepka-Zieminska A, Jagielska I, Soponska-Brzoszczyk P
Department of Obstetrics and Gynecology, L. Rydygier Collegium Medicum in Bydgoszcz, Nicolaus Copernicus University, Bydgoszcz, Poland., Bydgoszcz, Poland

Objective

Cat eye syndrom is rare congenital disease caused by trisomic or tetrasomic presence of choromosome 22. Phenotype of cat eye syndrome is variable - most common abnormalities are preauricular tags, anal atresia, congenital heart disease and coloboma of the iris.

Methods

We report two cases of trisomy of chromosome 22 recognized during pregnancy. The family history of both patients revealed no congenital defects.

Results

A 34-year-old gravida 2, para 1, was referred to the clinical hospital at 28 weeks of gestation in order to perform amniocentesis. The mother was treated for hypertension with angiotensin II receptor blocker (sartan) replaced by methyldopa at the first prenatal visit. Due to late date of the first antenatal checkup, first trimester screening was inappropriate. Second trimester ultrasound showed abnormal facial profile, atrioventricular septal defect and suspicion of overalapping fingers. At 28 weeks fetal growth restriction with abnormal cerebroplacental ratio were noticed. Invasive testing revealed trisomy of chromosome 22. A 35-year-old gravida 4, para 3, was identified at the first trimester screening with high risk for Down's syndrome (1: 5) and Edward syndrome (1: 5). Nuchal translucency was increased (3, 71 MoM; 4, 9mm at 12 week of gestation), while bHCG and PAPP-A concentrations were normal. Amniocentesis showed trisomy of chromosome 22. Ultrasound examination in the second trimester revealed abnormal facial profile, two-vessel umbilical cord, persistent left superior vena cava and right-sided aortic arch.

Conclusion

Detailed ultrasound examination in the first and second trimester allows perinatal detection of cat eye syndrome.