Prenatal diagnosis of familial pericentric inv(16) in a ultrasonographically normal fetus
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Objective
Pericentric inversions are structural balanced chromosomal rearrangements that rarely cause problems in carriers unless if one of the breakpoints involve disruption a gene. When balanced carriers transmit the rearrangement, it can result in a significant chromosomal imbalance in the offspring. Pairing and crossover during meiotic division occurs by loop formation between the homologous chromosomes to form recombinant unbalanced gametes. Prenatally, inversions are rarely detected. In this report, we present a familial pericentric inversion 16 in a fetus which was detected prenatally.

Methods
A 28 year-old woman at 17th gestational week referred for prenatal diagnosis because of increased Trisomy 21 risk (1/180) in first trimaster screening test. Amniocentesis were performed for fetal karyotyping, 20 cell were analyzed with GTG banding at 550 band levels.

Results
Fetal karyotype was reported as 46, XX, inv(16)(p12;q13). For familial transmission, parental karyotype analysis was performed and father had 46, XY, inv(16)(p12;q13), mother had normal karyotype. Fetal USG at 20th gestational week was normal. The couple had no history of spontaneous abortion or fetal anomaly in previous pregnancies.

Conclusion
For familial transmission of apparently balanced inversion, fetal anomaly risk is low but especially in the presence of ultrasonographic findings advanced molecular investigation such as “SNP array” for detection of submicroscopic deletion or duplication must be planned. To the best our knowledge, this is the first report about familial inversion 16.