Prenatal diagnosis of tetrasomy 13q31.3q34 in fetus with congenital diaphragmatic hernia and hydronephrosis

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Objective
To describe a case of a fetus with increased nuchal translucency in the first trimester and congenital diaphragmatic hernia with urogenital anomalies in the second trimester as nonspecific ultrasound markers for tetrasomy 13q31.3q34.

Methods
The first and the early second trimester ultrasound scans with fetal echocardiography were performed in a 33 years old primigravida (post IVF). Amniocentesis due to increased risk for aneuploidies in the first trimester was performed at 15 weeks of gestation. Origin of the markers was determined using karyotyping and array comparative genomic hybridization (aCGH, Agilent SurePrint G3 Human CGH Microarray Kit 4 x 180K platform).

Results
Abnormalities of the first trimester combined test encompassed increased nuchal translucency (5.5mm at 55.9mm CRL) and left sided pyelectasis. Increased risks of trisomy 21 (1:11) and trisomy 18 (1:207) were revealed. The second trimester fetal echo showed no cardiac anomaly and in anomaly scan left sided diaphragmatic hernia, hydronephrosis with ureter dilatation and increased nuchal fold were detected. Cytogenetic studies of amniocytes showed a karyotype as follows: 48, XX, +mar1, +mar2. Parents’ karyotypes were normal. Tetrasomy of the region 13q31.3 to 13q34 arr[hg19] 13q31.3q34(92507936-115092648) was showed in aCGH. The patient decided to terminate pregnancy at 18 week of gestation.

Conclusion
The increased risk of trisomies in the first trimester combined test is not only related to the common abnormalities. The use of aCGH is a good option determining rare conditions.