PRENATAL DIAGNOSIS OF AURICULO-CONDYLAR SYNDROME (CASE REPORT)

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OBJECTIVE
The auriculo-condylar syndrome (ACS) is caused by abnormalities of the first and second pharyngeal arches during embryonic development. It follows the autosomal dominant pattern of inheritance. The syndrome is characterized by wide phenotypic variation, with affected individuals expressing clinical signs of variable severity due to variable expressivity of the responsible genes. Clinical signs of the syndrome include auricular malformation, hypoplasia of the mandibular condyles, anomalies of the temporomandibular joints, malocclusion and in more severe cases cleft palate, glossoptosis, facial asymmetry and respiratory problems.

METHODS
Case report:
- 35 years old primigravida
- pregnancy after intrauterine insemination (IUI)
- 1st trimester ultrasound scan at 13th week with normal sonoanatomy
- combined test negative, RT21 1/3250
- integrated screening negative, RT21 1/3300, RNTD 1/5000
- 20th week ultrasound scan with normal biometry, atypically flat facial profile „face plate”, NB 4.6mm, hypertelorism, otherwise normal fetus sonoanatomy
- based on the US findings suspected for the Binder’s syndrome, it was indicated AMC
- AMC was performed at 21st week with a result of a microdeletion 20p12.3p12.2, this region of the chromosome contains the PLCB4 gene, which is associated with the auriculo-condylar syndrome

RESULTS
The amniocentesis revealed the auriculocondylar syndrome of the fetus. The parents opted for termination of pregnancy. We recommended DNA testing of both parents. The comparable microdeletion 20p12.3p12.2 was found in the father of the fetus by examination SNP array. We suggest a possible way of fertilization by IVF with preimplantation genetic screening (PGS) by the method array CGH for next pregnancy.

CONCLUSION
Auriculo-condylar syndrome (ACS) is a rare syndrome with an autosomal dominant pattern of inheritance. ACS can be caused by mutations in either the PLCB4 or GNAI3 gene. A disease locus for family with ACS has been mapped to 1p21.1-23.3. Some people who have one altered copy of the PLCB4 or GNAI3 have no features related to auriculo-condylar syndrome. This situation is now as reduced penetrance. PGS (preimplantation genetic screening) with array CGH (comparative genomic hybridization) enables all genome screening of unbalanced chromosomal abnormalities with high resolution. But the principle and the most important is the detail 20th week ultrasound scan which can induce the suspicion for a rare syndromes.