

be used throughout pregnancy, at relatively low costs. As many of the chromosomal syndromes have similar sonographic findings, a definitive diagnosis cannot be determined based on ultrasound alone, usually requiring follow-up genetic investigations.

Prenatal diagnosis of chromosome abnormalities through the analysis of amniocytes or chorionic villus samples is the standard of prenatal care. The application of chromosomal microarray analysis in routine chromosomal analysis has rapidly and substantially increased the diagnostic yield in clinical cytogenetics.

**Materials and Methods:** We selected three prenatal cases with minor ultrasound findings and major structural chromosomal abnormalities. Molecular karyotyping analysis was performed with either oligo or SNP-based microarrays developed for the detection of copy-number alterations: microdeletions, microduplications, aneuploidy and unbalanced translocations.

**Results:** Chromosomal microarray analysis revealed abnormal results for each of the three mentioned cases that are summarized in the table.

Case	Sonographic findings	Genomic microarray result
1	Unilateral multicystic dysplastic kidney	arr[GRCh37]17q12 (34817422_36168104)x1
2	Unilateral club foot	arr[GRCh37]17p11.2 (16657318_20433723)x3
3	Ventriculomegaly	arr[GRCh37]1q42.2q44 (233141951_249205158)x3,13q33.2q34 (106642740_115107733)x1

**Conclusions:** Identification of any fetal malformation should alert the sonographer for further investigation. Ultrasonographic markers may hint to genetic imbalances associated with severe consequences after birth. Our cases highlight the need of further invasive tests and genetic analysis, as that many rare microdeletion/microduplication syndromes can be present even with a normal sonographic exam. Therefore, the invasive testing remains the gold standard for prenatal diagnosis of chromosomal syndromes.

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#### E-P01.26

##### MicroRNA-200 regulated trophoblast invasion by targeting EG-VEGF in preeclampsia

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Preeclampsia is a severe gestational complication characterized by new onset of high blood pressure and proteinuria after 20 weeks of gestation. It is one of the leading hypertensive disorders in pregnant women, affecting 2 to 8% of pregnancies worldwide. Recently, endocrine gland-derived vascular endothelial growth factor (EG-VEGF) was regarded as a critical factor for embryo implantation and placental development. Micro-RNA 200 family, including miR-200a, -200b, -200c, -141- and -429, were highly expressed in human placenta and maternal circulation during pregnancy. Both EG-VEGF and miR-200 family have been shown to be associated with several pregnancy complications, including abnormal embryo implantation, intrauterine fetal growth restriction, preterm birth and preeclampsia. Besides, miR-200 family was predicted to target on 5'UTR of EG-VEGF. In order to investigate the roles of miR-200 family and EG-VEGF in preeclampsia, we analyzed these miRNAs and EG-VEGF expression in the pregnant women of healthy control (n=55) and preeclampsia (n=33). The expression level of miR-141 and miR-200a in preeclamptic women was significantly higher in maternal blood (p <0.05), while EG-VEGF was significantly lower in the maternal blood and placenta of preeclamptic women (p <0.05). Further, we verified miR-141 and -200a were target on 5'UTR region of EG-VEGF and affect the migration and invasion ability of trophoblast (HTR-8/SVneo). The results suggest miR-141 and -200a target on EG-VEGF and inhibit trophoblast migration and invasion, and may therefore predispose to develop preeclampsia in human early pregnancy.

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#### E-P01.27

##### Association of oxidative stress-related genes with miscarriage

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**Introduction:** 15% of all human pregnancies end in miscarriage before 12 weeks of gestation. Oxidative stress may play a very important role in the miscarriage with unknown etiology. This study was conducted to investigate the association of polymorphisms in oxidative stress-related genes with miscarriage.

**Materials and Methods:** A total of 127 women with miscarriage and 138 controls were genotyped for SOD1 rs4998557, SOD2 rs4880, CAT rs1001179, GPX4 (rs713041), EDN1 rs5370 and NOS3 rs2070744.

**Results:** A protective effect of SOD1 rs4998557-G allele on spontaneous abortion was shown in individual SNP

analysis:  $P=0.03$ ,  $OR = 0.49$ , 95% CI 0.26-0.93. It was established that the genotype AsnAsn EDN1 rs5370 was associated with an increased risk of missed abortion in the first trimester ( $OR 6.7$ , 95% CI 1.3-34.2). The multi-factor dimensionality reduction approach revealed gene-gene interactions for NOS3, GPX4 and EDN1 genes on spontaneous abortion. Cumulative gene risk score analysis demonstrated that genotype LysLys198 EDN1 /-786TT NOS3 / 718CC GPX4 was associated with spontaneous abortion ( $P=0.003$ ,  $OR = 4.28$ , 95% CI 1.63-11.2). The missed abortion was associated with interaction of GPX4, NOS3 and SOD2 genes. Cumulative gene risk score analysis demonstrated that more than three risk alleles in the genes GPX4 (rs713041-T), NOS3 (rs2070744-C), SOD2 (rs4880-Val) were associated with missed abortion ( $P = 0.038$ ,  $OR = 4.23$ , 95% CI 1.2-15.1).

**Conclusion:** Gene-gene interactions of oxidative stress related genes are able of modulating of miscarriage risk. This study was supported by the federal assignment № 6.6762.2017 from Russian Ministry of Science and Education.

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#### E-P01.28

**The incidence of particular types of chromosomal numerical aberrations in miscarriages is associated with maternal age**

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It is commonly known that the risk of giving birth to a child with trisomy 13, 18 and 21 increases rapidly with the maternal age. The relationship between advanced maternal age and incidence of Down syndrome was initially reported more than 75 years ago. On the other hand it has been proven that maternal age has no impact on the incidence of monosomy X in liveborns.

In this study 579 unselected products of conceptions were analysed using commercially available kits for chromosomes 13, 15, 16, 18, 21, 22, X and Y (QF-PCR) and for all chromosomes (MLPA). In contrast to QF-PCR, MLPA is unable to detect 69,XXX triploidy. Overall 170 different trisomies, 38 monosomies X, 40 triploidies and 8 double

aberrations were detected using MLPA or QF-PCR. Using MLPA, trisomies of all chromosomes except of chromosome 1 and 19 were diagnosed and the most common trisomies were of chromosome 16, while for QF-PCR the most common aberration was monosomy X. Similarly as in previously described studies, average age of women with trisomic pregnancy was significantly higher than in women with euploid pregnancy (35.73 vs 33.63  $p<0.005$ ) and the difference in maternal age for euploid foetuses and foetuses with X monosomy was insignificant (32.73 vs 33.63,  $p = 0.31$ ). Interestingly, frequency of triploidy in aborted foetuses seemed to decrease with maternal age (average age 31.23 vs 33.63,  $p<0.05$ ). Meiotic error in egg is a leading genetic cause of trisomy, however still little is known about the molecular basis of aneuploidies related to maternal age.

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#### E-P01.31

**Results of prospective study of utilisation of Trisomy test in noninvasive prenatal testing for trisomies of chromosomes 21, 18 and 13**

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**Introduction:** Noninvasive prenatal testing of most common trisomies, based on analysis of circulating DNA from blood of pregnant women, becomes an important part of prenatal screening.

**Aim:** Aim of the work was prospective study of utilisation of Trisomy test for noninvasive prenatal testing (NIPT) of the most common trisomies of chromosomes 21, 18 and 13.

**Materials and Methods:** From September 2015 till April 2017, 4109 samples of pregnant women were analyzed using Trisomy test. For high risk samples detection whole genome low coverage scan was used in association with home-made bioinformatic pipeline and our own biostatistical approach.

**Results:** Of 4109 analysed samples 3847 were reported as euploid and 76 as trisomic. After analysis of the first blood sample, 184 cases were found to be nonreportable, after second blood sample analysis only 42 samples were still unreportable, so no call rate of the test was 1%. Among