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P1–9 | 47,XY,+del (X)(q21.31)/46,XY mosaicism in prenatal diagnosis—Case report of a rare event

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Objectives: Aneuploidies involving the sex chromosomes are the most common anomalies in humans. In many cases, these anomalies are present in mosaic and may involve either the whole chromosome or just part of it. These anomalies constitute a challenge in prenatal diagnosis because it is generally very difficult to establish a reliable genotype-phenotype correlation. Here, we report a rare event of a mosaic in which one cell line carries an additional abnormal X chromosome, with a terminal deletion at q21.31 region, and a normal XY constitution in the majority of the cells.

Methods: A healthy 36-year-old G1P1 woman was referred for prenatal diagnosis at 11 + 5 weeks of gestation for increased nuchal translucency. Chorionic villus biopsy was performed, and molecular rapid aneuploidy result indicated an anomalous situation for the X chromosome in a male fetus. As the material was not sufficient to establish a culture, an amniocentesis was performed at 17 + 3 weeks and karyotyping and microarray were performed in order to characterize the anomalous result.

Results: The results obtained indicated the presence of a mosaic involving an extra X chromosome with a terminal deletion, [47,XY,+del (Xq)/46,XY, arr [GRCh37] Xp22.33q21.31(169921_89283237)x1–2], which is compatible with a Klinefelter syndrome variant.

Conclusions: Pregnancies affected by X chromosome aneuploidies diagnosed prenatally are at an increased risk of adverse fetal and neonatal outcomes. High quality information is critical for informed decision-making in pregnancy following a prenatal diagnosis of sex chromosome aneuploidy.

P1–11 | High resolution chromosomal microarrays in the general prenatal population: Pathogenic CNVs and VOUS in a 2016 state-wide cohort

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Objectives: Since the publication of a landmark study by Wapner et al. showing the advantages of a targeted chromosomal microarray (CMA) over karyotype for prenatal diagnosis, CMA utilization has expanded rapidly. The yield of CMA and rates of variants of uncertain/unknown significance (VOUS) vary according to the CMA platform and indications for testing. This study aimed to analyse the diagnostic yield of whole genome high resolution SNP CMA in a state-wide cohort and compare this to the yield reported in the 2012 study.

P1–12 | ATAD3A deletions: A challenge in prenatal diagnosis

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Objectives: The ATAD3 gene cluster is part of the ATPase family AAA-domain containing proteins consisting of three paralogs, ATAD3A, ATAD3B, and ATAD3C located in tandem on chromosome 1p36.33. The ATAD3 genes encode mitochondrial membrane proteins that contribute to the stabilization of large-mitochondrial protein complexes. Recently, deletions in the ATAD3 gene cluster...