of rare fetal cells in the maternal circulation. The fetal erythroblasts are the target cells of choice. However, variable numbers of maternal F cells, containing fetal hemoglobin (HbF), are counted within the population of fetal cells. The determination of fetal cells in maternal blood by fluorescence-activated cell sorting (FACS) based only on the detection of HbF is complicated due to the considerable background of F cells. The aim of our study was to evaluate a new flow cytometric method based on the combination of antibodies directed against HbF and carbonic anhydrase (CA)—a marker in red blood cells fully expressed only after birth—to discriminate fetal erythroblasts from maternal F cells. Whole maternal blood samples obtained from 12 pregnant women with male fetuses (16–22 weeks of pregnancy) were centrifuged in a density gradient of Percoll solution (1.077 g/ml). Anti-HBF-PE, anti-CA-FITC, anti-CD45-PE-Cy5 and Hoechst 33342 staining were used for the following FACS procedure. The Fl 1/Fl 2 histogram showed distinction between fetal erythroblasts (Hb^+, CA^+ low, $CD45^-$), F cells (HbF^+, CA^+, $CD45^-$) and adult red blood cells (HdF^-, CA^+, $CD45^-$). The number of Hb^+, CA^+ low, $CD45^-$, $Ho33342^+$ cells varied from 0.8×10^{-4} to 2.0×10^{-4} (mean 1.3×10^{-4}). The number of cells with the same immunophenotype after FACS sorted onto microscope slides was 3–140 (mean 30). Dual-color FISH using centromeric probes for the X and Y chromosomes indicated that only on average 7.8% of sorted cells were XY positive, while 60% were of maternal origin with XX signals. The remaining erythroblasts had aberrant or no FISH signals because of small and compact nuclei. Thus, FACS with combination of anti-HbF and anti-CA antibodies is not optimal to enrich fetal erythroblasts.

Keywords: Noninvasive prenatal diagnosis, Fetal erythroblasts, Flow cell sorting, FISH

10.P7

Prenatal diagnosis of a partial dup (16p) due to a rare recombinant resulting from a paternal intrachromosomal insertion

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Chromosomal rearrangements involving three breakpoints are relatively rare, about 1/5,000 live births. When a chromosomal segment is moved from one part of a chromosome into another part of the same chromosome, it is considered an intrachromosomal insertion; the orientation of the inserted material in relation to the centromere may remain the same, resulting in a direct insertion, or reversed, resulting in an inverted insertion. A single crossover in the gametogenesis between any of the three breakpoints may result in unbalanced recombinants, leading to phenotypic consequences in the offspring.

Partial trisomy 16p is a rare chromosomal imbalance characterized by mental retardation, prenatal and postnatal growth deficiency, facial anomalies, cleft palate, congenital heart defects, and urogenital anomalies. Previous studies have established that the phenotype of this condition is not related to the extension of the duplicated segment and that the region 16p13.1–p13.3 is critical in determining this disorder.

We report on a prenatal diagnosis performed at 14 weeks. The fetus presented with an increased fetal nuchal translucency and thus was referred for conventional cytogenetic studies.

The chromosomal analysis of the amniotic fluid cells revealed a structurally abnormal chromosome 16, with additional material on 16q. The maternal karyotype was normal, but the father carried an intrachromosomal insertion in chromosome 16: a between-arm insertion of a small segment of the short arm into the distal region of the long arm. To characterize the extension of the imbalance in the fetus, chromosome comparative genomic hybridization (cCGH) analysis was performed.

Fetus karyotype: 46,XY,rec(16)dup(16p)ins(16)(q24p13.2p13.3)pat.ish cgh dup(16)(p13.2p13.3).

The authors emphasize the rarity of this case, explain its possible formation mechanism and compare the fetal phenotype (available after autopsy) with similar cases described in the literature.

Keywords: Prenatal diagnosis, dup (16p), Recombinant chromosome

10.P8

Unusual triploidy presentation in a spontaneous miscarriage

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Triploidy is commonly found in spontaneous abortions, representing about 20% of the chromosomal aberrations in miscarriages. Usually, it is of paternal origin by fertilization of an ovocyte by two spermatozoa (dispermy). Here, we present a case of triploidy found in abortive rests with a 68,XXY,−21 karyotype. The second gestation of a healthy couple with a previous abortion due to mosaicism 45,X/47,XXX ended in a miscarriage at 10 weeks of gestation. Placental abortive rests were analyzed by QF-PCR and karyotype.

QF-PCR analysis revealed a triple dosage for chromosomes 13 and 18, XXY but only two copies of chromosome 21. Cytogenetic analysis of two independent chorionic cultures (mesoderm) confirmed a 68,XXY,−21 karyotype, which is a triploidy with only two chromosomes 21. DNA analysis from both parents (buccal wash) could ascertain a paternal origin of the triploidy (dispermy) and an absence of chromosome 21 from the mother.

In conclusion, two different abnormalities occurred in this gestation: an anomalous 21 nullisomic ovocyte, due to a meiotic nondisjunction, was abnormally fertilized by two spermatozoa. The result would have always been a miscarriage either by the abnormal ovocyte or the triploidy caused by the double fertilization. However, genetic counselling for this couple may vary as, although the risk of triploidy is very low, the risk of aneuploidy should be considered. The previous mosaicism gestation (45,X/47,XXX) due to a post-zygotic error had no significant influence and was not taken into account.

Triploidy may occur with aneuploidies, resulting in complex karyotypes, such as 70,XXY,+21 or, in the reverse, the present one 68,XXY,-21. It is important in these cases to find out the parental origin in order to provide accurate risk of recurrence for future gestations.

Keywords: Triploidy, Miscarriage

10.P9

Array-CGH identification of cryptic submicroscopic imbalances in handicapped children when their mothers are seeking subsequent pregnancies

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Microarray-based comparative genomic hybridization (array-CGH) has revolutionized clinical cytogenetics as it provides a relatively quick method to scan the genome for gains and losses of chromosomal material with significantly higher resolution and greater clinical yield than was previously possible.

In the past few years, these new methodologies have led to the identification of novel genomic disorders in patients with developmental delay/mental retardation and/or multiple congenital anomalies (DD/MR/MCA) as well as the discovery that each individual carries inherited copy number variations whose contributions to genetic variation and complex disease are not yet well understood. This study reported that array comparative genomic hybridization (aCGH) has successfully identified the molecular basis in two handicapped children with developmental delay without definite diagnosis. Furthermore, the normal result of aCGH in subsequent pregnancies ensured the normal children. Our study provided new evidence to support the use of aCGH in women with handicapped