chromosomal rearrangement. CGH and posterior array-CGH detected normal profiles, excluding any imbalances.

In conclusion, our findings confirm that the risk of having an abnormal child for carriers of paracentric inversions is low because the risk of production of unbalanced gametes by a carrier is expected to be small and most of the zygotes resulting from the unbalanced gametes would be so grossly abnormal that the embryos would be lost early, even before implantation.

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Keywords: Prenatal, CGH arrays, MFISH

1.P141

DNA methylation analysis of a de novo balanced X;13 translocation in a girl with abnormal phenotype: evidence for functional duplication of the whole short arm of the X chromosome

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We report on a 13-month-old girl showing dysmorphic features and a delay in psychomotor development. She was diagnosed with a balanced de novo translocation 46,X,t(X;13)(p11.2;p13) and non-random inactivation of the X chromosome. Fluorescence in situ hybridization analysis, using the X chromosome centromere and XIST region-specific probes, showed that the XIST locus was not involved in the translocation. Selective inactivation of paternal X, which was involved in translocation, was revealed by the HUMARA assay. The pattern of methylation of five genes located within Xp, which are normally silenced on an inactive X chromosome, corresponded to an active (unmethylated) X chromosome. These results revealed that in our proband, the X chromosome involved in the translocation (Xt) was preferentially inactivated. However, genes located on the translocated Xp did not include XIST. This resulted in functional Xp disomy, which most probably accounts for the abnormal phenotype in our patient.

Keywords: COBRA, DNA methylation, Duplication, HUMARA locus, X chromosome, Translocation

1.P142

A rare de novo unbalanced complex rearrangement involving chromosomes 12, 18 and 20 in a child with dysmorphic features

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Complex chromosomal rearrangements (CCRs) are rare structural abnormalities that involve three or more breakpoints located on two or more chromosomes and are often associated with developmental delay, mental retardation and congenital anomalies.

Here, we report the case of a rare de novo CCR in a girl who was 9 months old when first reported to us. At 15 months old, her clinical features included marked hypotonia, severe psychomotor delay, progressive postnatal microcephaly, strabismus, depressed nasal root, hands and feet malformations, heart defects, recurrent respiratory infections and bilateral hearing deficit still in study.

Conventional cytogenetic analysis revealed an unbalanced complex rearrangement, involving chromosomes 12, 18 and 20, and an apparent loss of material of chromosome 18 resulting from an interstitial deletion.

Further molecular cytogenetic studies were performed: whole chromosome painting probes for the involved chromosomes and chromosomal comparative genomic hybridization. These studies revealed that apparently no other chromosomes were involved and confirmed a del(18)(q21.1q22) of

approximately 17 Mb on the derivative chromosome 18. The latter chromosome also had material from der (12) to der(20) in its constitution.

As most CCRs involving chromosome 18q show rearrangements in the q21, some authors argue that this region might be a breakpoint "hotspot". On the other hand, cases of single deletions on 18q are predominantly terminal. Interstitial deletions are much rarer, and to our knowledge, this is the first report of a CCR with a del(18)(q21.1q22). The phenotype of patients with deletions within this region, reported so far, seems very similar to the one of our patient, and this may contribute to a better understanding of the genotype–phenotype correlation in this type of structural abnormalities.

1.P143

Translocation t(Y;5) in an azoospermic male

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Balanced translocations of a segment of Yq onto an autosome usually cause azoospermia and/or hypogonadism in male carriers. Yq-autosome translocations are rare and several such cases have been reported in the literature. Translocations involving Yq12 are observed in fertile males whereas in sterile males the breakpoint is located in the Yq11 euchromatic region containing the azoospermia factor (AZF) locus. Here we describe a reciprocal translocation between the long arm of the Y chromosome and the long arm of chromosome 5 in an azoospermic male. The patient was diagnosed with azoospermia during routine infertility examinations including clinical history, spermiogram and blood hormone levels. A testicular biopsy which showed maturation arrest on histology confirmed the diagnosis. Cytogenetic analysis with standard GTG-banding on the peripheral lymphocytes of the patient was performed. The karyotype was 46, X, t(Y;5)(q11,2; q35). We conclude that the balanced translocation that we found in this phenotypically normal male is the apparent cause of the azoospermia resulting in infertility.

Keywords : Translocation t(Y;5); Azoospermia

1.P144

Infant with a duplication of 12p and a deletion of 18q resulting from mother's t(12;18) balanced translocation

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We report a female infant, second live child of healthy non consaguineous parents (mother had three miscarriages, and one stillborn child). Maternal age at birth was 31 and paternal age was 38. Gestation was followed for IUGR, the proband was born at 39 weeks gestation, Apgar 9/10, weight 2230g and length 47 cm. She presented with multiple dysmorphic facial features, short neck, hypothyreosis, dysplastic pulmonal valve with mitral and tricuspidal insuffitienty, dilatative cardiomyopathy and pulmonal hypertension, also lumbosacral hypertrychosis and simian crease on both her palms. She also had postnatal developmental delay. Further investigation did not reveal any other congenital malformations.

Chromosomal analysis was performed using GTG-banding according to standard procedure on peripheral lymphocytes. Cytogenetic evaluation on high resolution GTG banding showed aberrant chromosome 18 in all metaphases: 46,XX,der(18). The karyotype of the mother was: 46,XX,t(12;18)(p13.1;q23). FISH analysis using specific locus probes was performed to indentify the duplication of 12p and deletion of 18q. The karyotype of the patient after FISH was 46,XX,der(18)t(12;18)(p12.3;q23)mat

Clinical characteristics were consistent with the data from the literature, however we can expect various clinical presentations, and therefore we need to follow up the patient's development and the family for genetic counselling.

Keywords : Partial trisomy 12p, Partial monosomy 18q, Balanced translocation