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Abstracts

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DISCUSSION: In this study, lymphocytes were evaluated in 25 sporadic AD cases (9 female and 16 males) and 25 healthy elderly subjects (10 female and 15 males). Multifactorial analyses of variance was used to assess the frequency of PCD in our samples in correlation to age. A statistically significant difference was found in the number of metaphase with PCD, X chromosome comparing AD female and female controls (P = 0.02) and in the number of total PCD, X chromosome in female sample (P = 0.04). Our results showed no positive correlation with age concerning the X chromosome. These results point to a fact that the X chromosome is preferentially affected in AD cases of women and that the amplification of instability of the X chromosome in women are not precisely connected to age.

P02.178
Congenital Lower Lid Entropion with pericentric inversion 9p13-q12 and deletion in Chromosome 10q23-qter
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Entropion is an inversion of the eyelid toward the globe. The lower eyelid is more frequently affected and depending on the underlying disorder. Entropion may be either unilateral or bilateral. Congenital entropion is an extremely rare disorder, usually involves the lower eyelids. It is often familial and seen more frequently in Asians. The possible causes for this condition include structural tarsal plate defects (horizontal tarsal kink syndrome) and shortened posterior lamella (tarsus, conjunctiva, eyelid retractors). It has been reported that congenital entropion is a part of a syndrome involving multiple systemic anomalies. A case of primary congenital upper eyelid entropion with cardiovascular, musculoskeletal, and central nervous system abnormalities and another with congenital heart defect has been reported. But to the best of our knowledge there is no report describing the genetic background of the disease. We report a patient of congenital lower lid entropion and corneal opacity who was referred to us for cytogenetic analysis. GTG-banding of 50 well spreaded metaphases revealed an interstitial deletion in chromosome 10 and pericentric inversion of chromosome 9. Chromosomal analysis showed 46, XX (57%)/46,XX.del(10q23-qter)(10%)/46,XX.inv(9p13-q12)(33%) karyotype. Most publications suggest that pericentric inversion of chromosome 9 is a polymorphic variation and its clinical significance is uncertain. Thus our finding raises the possibility that the congenital lower lid entropion locus may be located on chromosome 10. This represents a severe manifestation of the disease. Finally, a workup of this finding is suggested and more cases of congenital lower lid entropion needs to be screened using cytogenetics.

P02.179
Chromosome segregation in blastomeres from translocation carriers.
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Balanced carriers of reciprocal and Robertsonian translocations suffer from reduced fertility and are at increased risk of recurrent spontaneous abortions or chromosomally unbalanced offspring. During meiosis I, segregation of the translocated chromosomes and their normal homologues produces a variety of unbalanced gametes. Preimplantation genetic diagnosis (PGD) offers to the translocation carriers an alternative to prenatal diagnosis and a possible pregnancy termination. PGD allows not only selection of normal embryos but also provides unique information about the chromosome segregation. Interphase FISH combining subtelomeric and centromeric probes was used to examine the blastomeres of 126 embryos from 6 Robertsonian (2 female/4 male carriers) and 12 reciprocal translocation carriers (6 female/6 male carriers). In Robertsonian translocation carriers, 39.3% normal and 53.6% aneuploid (25% trisomic and 28.6% monosomic) embryos were found. Each reciprocal translocation had a different segregation mode, but at least one balanced blastomere was found in each translocation. The lengths of translocated segments varied from 7.8Mb to 140.4Mb (4% to 73% of the chromosome involved). Alternate segregation (chromosome balanced) was found in 31% blastomeres. In unbalanced blastomeres, 39.4% resulted from adjacent segregation, 15.5% from segregation 3:1 and 14.1% of blastomeres were haploid, polyploid or chaotic. No statistical difference between male and female rates of chromosome abnormalities in both Robertsonian and reciprocal translocation carriers was observed. Comparison of the length of translocated chromosome segment and the type of segregation revealed a trend for higher proportion of normal blastomeres when shorter chromosome segments were involved.

P02.180
Array CGH analysis (two novel deletions) in pigment dispersion syndrome
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Pigment dispersion syndrome (PDS) is an ocular disorder in which melanin granules from the iris pigment epithelium are distributed throughout the anterior segment on various ocular structures. The prevalence of PDS is 2.5% in whites, and 1.6% in black population. In half of cases, PDS progresses to pigmentary glaucoma. The etiology of PDS is not known. Only one locus for PDS has been identified at 7q35-q36. We report the first, a 34-year old, male with PDS and a balanced 10;15 translocation, revealed using GTG banding. Molecular cytogenetic breakpoints were located using fluorescence in situ hybridization (FISH) analysis at chromosome 10cen and on 15q between D15S134 and SNP503 (DNA probes from Abbott-Vysis and Oncore). The proband’s karyotype was interpreted as 46,XY,t(10;15)(p11.11;q11.1). Array CGH analysis using the DNA sample of the patient was genotyped in duplicate with the HumanHap300-Duo Beadchip (Illumina). Array CGH analysis did not show altered DNA sequences in the breakpoints of the translocation, but revealed two novel deletions in 2q22.1 and 18q22.1. These two CNV regions are not previously described in the Database of Genomic Variants. We suppose that the coexistence of t(10;15) and PDS in our patient is a coincidence. However, the deletion in 2q22.1, where the gene LRP1B has been located, may play a major role in the dysembryogenesis of the eye and cause the disorder. As array CGH shows a number of the chromosomal alterations, it is important to use this molecular karyotyping in diagnostic laboratories.

P02.181
Polyomorphic variants and phenotype correlation. Findings in a Brazilian reference center
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Objective: To describe the frequency of polyomorphic variants such as inv(9), 9q+, 16q+ and satellite increase and associated phenotype in a clinical cytogenetics core in a Women’s University Hospital.
Methods: A retrospective review of the laboratory databases identified all cases of polyomorphic variants from January 2003 to December 2007. Clinical records were also evaluated.
Results: The total number of cases analyzed in 5 years was 797. Cytogenetic evaluation of prenatal diagnosis cases with abnormal ultrasound findings were the most frequent (n=602[75.5%]) and showed rates of 73.5% (n=443) of normal karyotypes, 21.8% (n=131) of abnormal karyotypes and 4.7% (n=28) of polyomorphic variants. Out of 162 karyotypes in women who were evaluated for sex-related disorders, 67.3% (n=109) presented a normal karyotype, 23.5% (n=36) presented an abnormal karyotype and 9.2% (n=15) presented polyomorphic variants. Individuals who were investigated for recurrent abortions showed a 9.1% rate (n=5) of polyomorphic variants among 72.7% (n=24) of patients with normal karyotype and 18.2% (n=6) with abnormal karyotypes. Abnormal karyotypes included both structural/numerical and autosomal/sexual abnormalities. Phenotypes associated with polyomorphic variants at prenatal diagnosis were cardiac and facial