IMPACT OF POLYMORPHIC INVERSIONS IN BLASTOCYST ANEUPLOIDY RATES

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INTRODUCTION There are different types of inversions considered as polymorphic variants. Some of them involved rearrangements in heterochromatin, as inv(9)(p11q12)/(p11q13)/p12q13). Others include euchromatin and the most common is inv(2)(p11.2q13). There is no agreement on their potential impact on reproductive outcome and embryo aneuploidy risk.OBJECTIVETo determine if polymorphic inversions: inv(9)(p11q12)/(p11q13)/p12q13) and inv(2)(p11.2q13) affect aneuploidy rates in patients undergoing Preimplantation Genetic Testing (PGT).MATERIALS AND METHODRetrospective study (January 2016- July 2018) including 218 trophectoderm biopsies in female or male carriers of the polymorphic inversions. Two women age groups were considered: ≤37 years (n=133 embryos) and >37 years (n=85 embryos). And a control group of 6,105 embryos from couples with normal karyotype, subdivided in the same age ranges: ≤37 years (n=2,168 embryos) and >37 years (n=3,937 embryos). Next Generation Sequencing (Thermo Fisher Scientific, USA) was performed for the chromosomal analysis.RESULTSAneuploidy rates were as follows:Carriers with women age ≤37 years: 36.8% (37.5% vs. 35.8% in female and male carriers respectively, without statistical differences according to the gender of the carrier).Carriers with women age >37 years: 71.8% (70.0% vs. 73.3% in female and male carriers respectively, without statistical differences according to the gender of the carrier).Control group with women age ≤37years: 44.0%.Control group with women age >37years: 64.6%.No differences in overall aneuploidy were observed for each carrier subgroup when compared to the age matched control group. However, for carriers of inv(2)(p11.2q13) aneuploidy for chromosome 2 was significantly increased in women >37, compared to the control group (36.4% vs. 6.3; p=0.0006).CONCLUSIONCarriers of these variant inversions do not show an increase in overall aneuploidy rates. However, a specific increase for the chromosome involved in the rearrangement should be considered. Therefore, PGT should be considered if associated with miscarriages and or/implantation failures.