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Chromosomal polymorphic variants increase the embryo aneuploidy rate in IVF cycles

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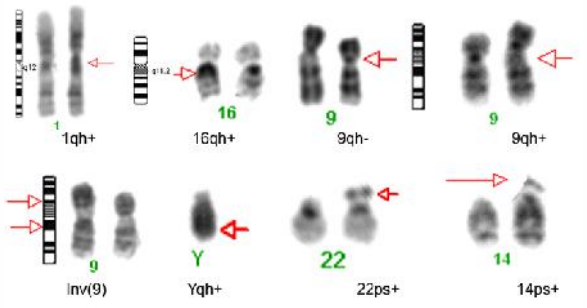
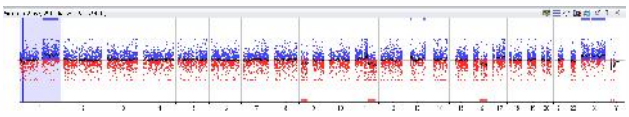
Introduction:

Chromosomal polymorphic variants are considered as normal but previous studies have reported that they are associated with infertility and recurrent abortions although the way is unknown. On the other hand there is a high incidence of chromosome abnormalities in human gametes and embryos that leads to failure of IVF cycles, including oocyte donation cycles. This could be due to aggressive stimulation, male factor or other issues, but there are causes that are yet to be defined. The aim of this study was to analyze if chromosomal polymorphic variations could increase the embryo aneuploidy rate in blastocysts obtained after an IVF cycle.



Examples of chromosomal polymorphic variants (in chromosome 1, 16, 9, Y , 22 and 14) by banding G



Embryo XX (dup 1q, del9p, del11p, del16q)

Materials and methods:

A retrospective study was performed. We included the array-CGH results of 524 embryos from 231 comprehensive chromosome screening (CCS) cycles performed between 2013 and 2014 at Instituto Bernabeu, Alicante, Spain (301 embryos from oocyte donation cycles and 223 from IVF cycles with own oocytes). Whole chromosome imbalances by array-CGH in trophectoderm cells from D5 embryos were detected. Array-CGH analysis was performed using Agilent SurePrint G3 8x60K CGH microarrays previous whole genome amplification (WGA) of genomic DNA. The main outcome measures were embryo aneuploidy rate and implantation rate.

Results:

Significant differences were reported in the embryo aneuploidy rate between carriers and not carriers of chromosomal polymorphisms (46.8% vs. 34.9%; OR=1.638, 95% CI=1.005-2.668). We show that these differences occur in oocyte donation cycles (50.0% vs. 27.7%; OR=2.747, 95% IC=1.039-7.264) but not in cycles with own oocytes, perhaps because the oocytes donors are a homogeneous group without confusion factors like woman age (mean age 25.8 years). The presence of a chromosomal polymorphism in the oocyte donor (and not in the male partner) has a higher risk for embryonic aneuploidy. Moreover, we observed that the implantation rate from euploid embryos transferred were lower in polymorphisms carriers group (42.9% vs. 57.3%) although the difference in this case was not significant.

Discussion:

This study reveals a higher aneuploidy rate in blastocysts from chromosomal polymorphisms carriers than in embryos from individuals with normal karyotype, showing a relationship between these both phenomena and suggesting that chromosomal polymorphisms could be responsible for a higher percentage of aneuploidies in embryos from IVF cycles, mainly in young woman. Karyotyping of individuals, patients and donors, before an IVF cycle is important because the incidence of chromosomal abnormalities, including polymorphic variants, is quite high and application of array-CGH in these cases could improve the IVF results selecting euploid embryos.