UNDETECTED CHROMOSOMAL ABNORMALITIES IN COUPLES UNDERGOING IVF

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Abstract Body

Balanced and unbalanced chromosomal abnormalities have been shown to be present in a small, but significant percentage of couples consulting for infertility. Therefore, karyotyping has become an obligatory biological examination in the management of IVF couples.

Here we present two cases, where the parental chromosomal abnormalities were revealed via embryo testing. Preimplantation genetic testing for aneuploidy (PGT-A) was performed by next generation sequencing using VeriSeq (Vitrolife). In both IVF couples a standard kayotyping from peripheral blood has been performed prior to the start of IVF treatment resulting in normal karyotypes in all persons involved. First couple: 6 embryos were examined and in 4 of them the same mosaic 3-8 Mb long deletion on chromosome 8 was revealed. Array CGH (8x60K, Agilent) examination confirmed 4.5 Mb long deletion 8q22.3q23.3 in healthy father. The couple accepted transfer the only euploid embryo with 8q deletion; subsequent prenatal testing in undergoing pregnancy confirmed paternal deletion. Second couple: 5 embryos were examined and in 3 of them the same mosaic 5-10 Mb long duplication on chromosome 10 was revealed. Array CGH examination confirmed 7.2 Mb long duplication 10q22.3q23.2 in healthy oocyte donor. Penetrance of relatively frequent 10q22.3q23.2 duplication is incomplete and euploid embryos with duplication were not accepted for the transfer.

Karyotypes of both chromosomal imbalances carriers have been revised and no error was found in the scope and performance of the examination. In this context the question arises whether the standard karyotype examination is sufficient enough, especially for gamete donors.