Topic: Preimplantation Genetic Screening

Title: COMPARISON OF THE RESULTS OF PREIMPLANTATION GENETIC SCREENING: a-CGH and NGS

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Introduction: chromosomal aneuploidies are known for being the main cause of abnormal development of embryos with normal morphology. During the last decade, new technologies for aneuploidy screening have been introduced in practice. They allow carrying out the screening of all 24 chromosomes in human embryos. Among them microarray comparative genomic hybridization (a-CGH), single nucleotide polymorphism microarrays (SNP arrays) and quantitative polymerase chain reaction (qPCR) have been validated and successfully applied clinically.

The aid of this investigation was to evaluate the possibilities of embryo investigation results by a new method using semi-conductive NGS technology and to compare these with results obtained with array-CGH based on Agilent platform**.**

Materials and Methods: 38 patients undergoing PGS were enrolled in the study. All embryos were cultured to blastocyst stage; trophectoderm biopsy was performed on Day 5 of developmentor. The samples were analyzed using both comparative genomic hybridization and semiconductor high-throughput sequencing (Ion torrent). Whole genome amplification was carried out by PCR-based method for 28 samples and by MDA for 10 samples.

Results: according to a-CGH investigation 25 (65,7%) out of 38 embryos were euploid and 13 (34,3%) were aneuploid. In 36 samples we obtained concordant results for both NGS and a-CGH analysis. Discordant results were obtained only for 2 (5,2%) of all investigated samples. In order to compare Whole Genome Amplification technique, four samples were biopsied 2 times and amplified using WGA-PCR and MDA. In all cases the results were concordant.

The results achieved in this study demonstrate the reliability of the NGS-based protocol for detection of whole chromosome aneuploidies and segmental changes in embryos. NGS-based PGS represents a valuable alternative to other currently available a-CGH techniques.

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