

ADVANCEMENT OF PGT BY NGS: THE PROBLEM OF MOSAICISM AND ITS INTERPRETATION

Musatova, Elizaveta¹; Kaimonov, Vladimir¹; Mironova, Irina¹; Pomerantseva, Ekaterina¹

¹Center of Genetics and Reproductive Medicine «Genetico»

Abstract Body

Mosaic aneuploidies are the problem of advancement of preimplantation genetic testing (PGT) by next generation sequencing (NGS). The factors to increase the proportion of mosaic embryos need to be studied. The frequency of samples with mosaic aneuploidies varies from clinic to clinic and may amount from 4 to 44% according to published data. This abstract contains anonymized frequency data of mosaic embryos and embryo DNA degradation among some IVF clinics for potential estimate of the embryology stage effect. Another factor affecting the proportion of mosaic embryos may be incorrect interpretation of chromosomal profiles by different specialists (readers) of PGT laboratories.

The materials of study were trophoctoderm samples of embryos, obtained with patients' own oocytes and donor oocytes for PGT for aneuploidies (PGT-A). PGT-A performed by NGS (Veriseq, Illumina) and aCGH (Cytochip, Illumina) and analysis data was performed by BlueFuse Multi software (Illumina). 11 embryologists from different IVF clinics and took part in this study.

Four IVF clinics were selected for comparison of mosaic embryos proportion among embryos from donor oocyte: we observed a statistically significant difference between these IVF clinics. The frequency of mosaic embryos in this experiment varied from 16 to 25%. We estimated the proportion of embryo DNA degradation and mosaic embryos from patients' own oocytes between different embryologists.

The frequency of mosaic embryos in this experiment varied from 11 to 25%, and proportion of samples with DNA degradation – from 0 to 12,5%. In another study we compared the difference in interpretation of the same chromosome profiles by 10 readers from different PGT laboratories and obtained interesting results: interpretation of chromosome profiles by another reader can lead to a change in the clinical fate of the embryo.

Study of mosaicism causes is an important aspect that allows to increase the effectiveness of IVF protocols.