MITOCHONDRIAL DNA COPY NUMBER, ASSESSED DURING PREIMPLANTATION GENETIC DIAGNOSIS WITH NEXT GENERATION SEQUENCING - SELECTION CRITERIA FOR EMBRYO TRANSFER PRIORITIZATION

K. Lukaszuk[1,2], S. Pukszta[1], W. Kuczynski[3,4], C. Cybulska[1], J. Liss[1]
[1]*Reproduction and Fertility Clinic, INVICTA, Poland*
[2]*Department of Obstetrics and Gynecological Nursing, Faculty of Health Sciences, Medical University of Gdansk, Poland*
[3]*Centre for Reproductive Medicine, KRIOBANK, Poland*
[4]*Department of Gynecology and Oncological Gynecology, Medical University of Bialystok, Poland*

Preimplantation Genetic Diagnosis with Next Generation Sequencing allows simultaneous testing of chromosomal imbalances and mitochondrial DNA (mtDNA) copy number. The amount of mtDNA could be an indirect measure of the mitochondrial functions and the quality of the mtDNA.
We set out to observe the amounts of mtDNA among 359 investigated embryos and compare that with the amount of genomic DNA. MtDNA relative amount was assessed as number of reads for mtDNA / number of reads for genome DNA ratio x 1000. Our results showed a significant difference in mtDNA amount depending on embryo biopsy day. Results from day 3 biopsy had much higher mtDNA to genome DNA ratio than those from day 5 material (5.54 vs. 1.2, p0.001, respectively). Additionally, we wanted to determine whether there were differences in the quantity of mtDNA in aneuploid and normal embryos, embryos which implanted and those that did not, embryos of good and poor morphology, and embryos from older and younger women. We found important difference in mtDNA amount in day 3 biopsy samples from normal and aneuploid embryos (4.6 vs 6.0 p=0.01, respectively). No correlation between mtDNA copy number and remaining investigated parameters was found for day 3 samples. For day 5 samples, besides implantation rate, no correlation was established. Obtained results suggest a link between mitochondrial and chromosomal nondisjunction. Higher amount of mtDNA copy number was found in embryos that were aneuploid, thus indicating that mtDNA copy number assession has a potential of becoming a criteria for embryo selection.