PGT BY MULTIPLEX PCR IN PATIENTS WITH RISK OF GENETIC AND CHROMOSOMAL ABNORMALITIES

Krasnopolskaya, Ksenia; Sesina, Nina; Voskoboeva, Elena; Badalyan, Gohar; Bocharova, Tatiana

Introduction: PGT is a modern method that allows to increase the probability of a healthy child birth in patients with monogenic genetic diseases (PGT-M) or structural chromosomal rearrangements (PGT-SR) and also without aneuploidies (PGT-A).Aim of study: To evaluate the effectiveness of the transfer of embryos diagnosed after PGT as normal in groups of patients with monogenic diseases, genetic structural rearrangements of chromosomes, and patients, whose embryos were tested for aneuploidy.Materials and methods: For the period 2017 - 2018, 156 cycles with PGT were conducted (15 PGT-SR, 14 PGT-M, 127 PGT-A). Trophectoderm biopsy was performed on blastocyst (day 5-6). A total of 477 embryos were examined. Molecular genetic analysis was performed by Multiplex PCR after the full-genome amplification reaction. The number of embryos per transfer was (PGT-SR - 1.08; PGT-M - 1,36; PGT-A - 1,57).Results: After 129 embryo transfers (15 PGT-SR, 15 PGT-M, 99 PGT-A), 47 clinical pregnancies occurred (7 PGT-SP (pregnancy rate index (PRI)=46.7%), 6 PGT-M (PRI=40%), 34 PGT-A(PRI=38.2%)). Development of 6 pregnancies stopped early on 6-7 weeks and in two cases noted it was a blighted ovum. One missed abortion was noted in the group of PGT-M (16.6%). No pregnancy losses were noted in the group of PGT-SR. In the PGT-A 5 pregnancies stopped in development (14.7%). At the moment, 11 pregnancies have ended with the timely birth of 11 healthy children; one pregnancy has ended with preterm birth at 28 weeks. In 17 patients, the pregnancy is currently progressing. Genetic or chromosomal pathology of the fetus was not observed in any case. The coincidence of chromosomal/genetic status in all born children after diagnosis with a certain PGT is shown.Summary: In patients with risks of chromosomal or genetic abnormalities of the offspring, it is advisable to carry out PGT.