

## CURRENT METHODS OF PREIMPLANTATION GENETIC DIAGNOSIS IN THE REPUBLIC OF UZBEKISTAN

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**Relevance:** Preimplantation genetic diagnosis has existed for more than 20 years, during which time the range of possibilities of assisted reproductive technologies and the potential of molecular genetic diagnosis of single cells have significantly expanded.

**The study aimed to:** To date, preimplantation diagnosis has evolved from an experimental procedure to a valid and earliest form of prenatal diagnosis, while expanding the range of indications.

**Materials and Methods:** Women of older reproductive age - 35 years and older. Couples with a history of more than three spontaneous early terminations of pregnancy (habitual miscarriage). Men with severe disorders of spermatogenesis - oligoasthenoteratozoospermia, severe oligozoospermia, azoospermia. Couples with repeated unsuccessful IVF attempts - more than three unsuccessful attempts

**Results:** Mitochondrial diseases caused by nuclear DNA mutations are transmitted according to Mendelian laws, hence the pattern of PGD should be the same as for monogenic diseases. The inheritance of mitochondrial diseases caused by mtDNA mutations is complicated by heteroplasmy (genetic heterogeneity of the mitochondrial population), different levels of heteroplasmy in different oocytes, random distribution of mtDNA molecules between blastocyst cells, and the fact that the proportion of mutant mtDNA molecules may vary differently in tissues during the development and life of the organism. This is especially true when the diagnosis is carried out according to the "transport scheme" - that is, different stages of IVF with PGD are performed in different medical institutions located in different cities or even countries. The "transport scheme" is widely used in the Americas and Europe. This is due to the fact that it is not yet economically viable to maintain expensive equipment for genetic research and a staff of highly qualified employees in small and medium-sized ART clinics. Clinical and laboratory departments are connected using special couriers and standard means of communication (Internet, telephone).

**Conclusion:** This has transformed PGD from an experimental procedure to a valid and earliest form of PD, while expanding the range of indications.

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