Abstract

Preimplantation genetic testing is one of the major clinical procedures that takes place in centres of assisted reproduction all around the world. It is sought out by couples for many reasons discussed in this paper. The causes of couples' arrival to the centres might vary from inherited diseases that future parents do not wish to transfer to their offspring to the risk of transfer of chromosomal structural rearrangements. What's more, infertility might be caused by the greater risk of aneuploidy in embryos which correlates with higher maternal age and other factors. This paper also focuses on new approaches in cytogenetic examinations for reproductive medicine. Furthermore, it discusses clinical methods used to improve diagnosis and treatment of infertile couples and experimental methods that could become the base of new diagnostic tools. These are for example single nucleotide polymorphism methods, array-based methods, new generation sequencing and whole genome amplifications. It also looks back at methods that became old-fashioned, just like for example fluorescent *in situ* hybridisation, and states its pros and cons.

Key words: preimplantation genetic testing, aneuploidy, structural rearrangements, monogenic diseases, biopsy