"The newest approach for aneuploidy screening is next-generation sequencing (NGS). This method not only allows us to detect all 24 chromosome abnormities, but also determines the percentage of cells carrying the detected aneuploidy"

Preimplantation genetic screening is strongly advisable for the following couples:

- advanced maternal age couples
- couples with repeated IVF failure and unsuccessful embryo transfers
- recurrent pregnancy losses and impaired child births (caused by chromosomal abnormalities)
- men with an abnormal sperm analysis (presence of pathological sperm forms) and low sperm concentrations
- couples that are carriers of chromosomal translocations and abnormalities
- use of TESE, MESA samples for fertilisation
- couples after a chemotherapy or actinotherapy for oncological diseases

Because preimplantation genetic screening increases the success of each IVF cycle, it is recommendable not only to couples with a genetic risk.

The newest approach for aneuploidy screening is next-generation sequencing (NGS). This method not only allows us to detect all 24 chromosome abnormities, but also determines the percentage of cells carrying the detected aneuploidy. Embryos can contain several cell lines with different chromosome complements. This phenomenon, when cells from one embryo carry a different chromosomal number, is called mosaicism. Mosaic embryo transfer, although sometimes resulting in healthy childbirths also carries with it many risks – such as a lower implantation rate and a higher abortion rate (in comparison to transfers of euploid embryos).

Mosaicism detection is therefore one of the main advantages of next-generation sequencing, in comparison to chip based comparative genomic hybridisation (aCGH),which is limited by its resolution.

Currently an obsolete method of aneuploidy detection is fluorescent in situ hybridisation (FISH), allowing detection of certain chromosomes only, making it nowadays unsuitable for PGS.

Preimplantation genetic diagnostics is, unlike genetic screening, a set of methods specifically targeting the transmission of a certain genetic risk or condition from one or both parents to their offspring. This may be, for example, a structural chromosomal abnormality, frequently a chromosomal translocation. Chromosomal translocations are detected in a manner similar to PGS, but with a higher resolution potential, allowing us not only to distinguish full chromosomal abnormalities, but also small chromosomal section alterations. Each couple prior to IVF treatment undergoes karyotyping to diagnose potential chromosomal abnormities or rearrangements e.g. translocations.

Another big group of disorders targeted by preimplantation genetic diagnosis are rare inherited disorders. Rare inherited disorders (also called monogenic diseases) are a very variable group of severe conditions, oncogenic predispositions and syndromes caused by genetic factors. Although these diseases vary significantly, they share the same origin: a single-gene mutation. It is possible to perform PGD for known mutations (either point mutations, repetitions, deletions or inversions), and therefore select unaffected embryos.

The most up-to-date method of PGD for rare inherited disorders is karyomapping, based on a whole genome detection of single-nucleotide polymorphisms. It is the whole genome coverage that allows us to detect various human single-gene mutations in embryos in the course of a normal IVF cycle. This represents a big advantage to the previously used PCR (polymerase chain reaction) based methods for single-gene mutation detection.

In view of the fact that only DNA of the treated couple and a single reference of known disease status (either from an offspring or from any other affected relative) is necessary, karyomapping significantly eases the stress, which the treated family is exposed to. In some cases, only DNA of the treated couple is sufficient for obtaining a clear result. By evaluating a single reference sample with DNA samples of the treated couple, we are able to mark the set of genetic information characteristic for the specific mutation (a haplotype linked with the mutation) and therefore exclude affected embryos from transfer. Hence, karyomapping represents an exquisite tool for termination of transmission of rare inherited disorders in the affected family.

In addition to rare inherited diseases, this method is also suitable for detection of some oncological predispositions, such as BRCA1 and BRCA2 gene mutations.

Karyomapping is also the only known tool, allowing us to detect balanced translocations in embryos. Balanced translocations are inherited from parent to offspring and their carriers often do not show any visible symptoms. Nevertheless, balanced translocations are a frequent cause of aneuploidies in embryos. Through karyomapping it is possible to distinguish between embryos carrying these types of translocations and between completely unaffected embryos.

Last, but not least, karyomapping also allows us to perform chromosomal screening for aneuploidies. We are now able to select embryos not only without inherited single-gene defects,