



مركز بنون لتقنية مساعدة الإنجاب
Banoon Assisted Reproduction Technology Centre



الخدمات الطبية الملكية - المستشفى العسكري
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Pre-implantation Genetic Diagnosis Aneuploidy Screening (PGD-AS)

Usually, following In-Vitro Fertilization (IVF), the healthiest looking embryos are selected for transfer to the patient, assessed by morphological appearance and development rate (i.e. cell number & quality). However, this gives no information about the genetic makeup of the embryo.

Preimplantation Genetic Diagnosis (PGD) is a test performed on embryos before returning them to mother's womb to detect any abnormalities in them, either at gene level or at chromosome level (PGD-AS).

The chromosomes are the physical structure that contains the DNA and genes necessary for our development. A normal human cell contains 46 chromosomes or 23 pairs of chromosomes. Twenty two pairs of them are called (autosomes) and are identical in both men and women, while the chromosomes in the 23rd pair are the sex chromosomes. Normally, women sex chromosomes are the same (XX), while men sex chromosomes are different (XY).

Sperms and eggs contain half the total number of 46 chromosomes, 23 chromosomes each. A normal fertilized embryo is derived by the fusion of 23 chromosomes from an egg and 23 chromosomes from a sperm. **Any abnormal division in eggs or sperms might result in a greater or fewer chromosomes than the normal 23 pairs**, and any embryos that are derived from these sperms or eggs carry extra or missing chromosomes (Aneuploidy).

Performing PGD-AS

PGD is performed in conjunction with In-Vitro Fertilization (IVF), embryo biopsy is performed on day 2-3 of egg collection where a hole is drilled in the zona (the outer shell) of the embryo using a laser beam and 1-2 cells are gently removed from the embryo and then, the biopsied cells are tested using a technique called FISH.

Fluorescence In-Situ Hybridization (FISH)

It refers to the technique of "painting" the whole chromosome with a genetic probe bound to a colored marker. FISH allows us to count the chromosomes based on color staining. After the test, embryos identified as having no imbalance for the chromosomes tested then can be transferred on day 5 or day 6.

Indications for PGD-AS

Embryos screened using PGD-AS may have higher rate of implantation, lower risk of spontaneous miscarriage and reduced risk of having a trisomic offspring (e.g. Down's syndrome)

PGD-AS is recommended in the following cases:

- Advanced maternal age. (≥ 36 years old)
- Recurrent pregnancy loss. (≥ 3 miscarriages)
- Repeated IVF failure. (≥ 2 implantation failure)
- Families at risk for gender related diseases

PGD-AS for family balancing could be considered in special cases.

Most common chromosome defects

The most common chromosome abnormalities found in miscarriages are:

- Trisomy 18: three copies of chromosome no. 18 (i.e. Edward's Syndrome)
- Trisomy chromosomes 13, 15, 18, 21 or 22: i.e. 3 copies of either the mentioned chromosomes. (e.g. Trisomy 21 is Down's Syndrome)
- Triploidy: i.e. 3 copies of the whole set of chromosomes.
- Defects affecting sex chromosomes: X or Y: e.g. Turner Syndrome (XO) - Klinefelter's Syndrome (XXY).

PGD-AS limitations

- Occasionally, two chromosomes might be lying on top of each other, which would underestimate the number of chromosomes and lead to a false interpretation.
- Some embryos have two separated cell lines, called mosaic; a mosaic embryo can be read falsely as normal or abnormal.
- In total, we believe that false normal rate is about 5% and that false abnormal rate is about the same, 5%.

Other important information

- Embryo biopsy has been applied extensively in major IVF clinics over the world and does not appear to have adverse affect on the embryo's potential to implant or develop into pregnancy.
- It may not be possible to obtain FISH result from some or all of the biopsied embryos. Some embryos may not be of sufficient quality to biopsy. Or in some embryos the test result may not be conclusive.
- Embryo biopsy with chromosome testing is appropriate as an embryo selection process only if at least 5 embryos are suitable for biopsy on day 3. Embryo biopsy is an invasive procedure that may not be considered advisable with only a small number of embryos. If there are only 2 embryos on day 3, it may therefore be considered more beneficial to transfer these embryos without biopsy.
- This test is not diagnostic test because it cannot provide an absolute guarantee of the chromosome status of embryo from analysis of only one cell. In some embryos, one cell may not be representative of the whole embryo.
- Prenatal Diagnosis is advised to confirm the PGD results.