

Monash IVF

Fact Sheet

Preimplantation Genetic Diagnosis (PGD) for Aneuploidy Screening



Key points:

- Error/s in the early development of the sperm, egg or embryo can lead to an abnormal number of chromosomes in the developing embryo (ie: missing or extra chromosomes).
- An abnormal chromosome number can cause implantation failure, miscarriage, or the birth of a child with a chromosome abnormality (eg: Down syndrome).
- Some couples have an increased risk of producing embryos with an abnormal chromosome number.
- Preimplantation Genetic Diagnosis (PGD) with aneuploidy screening can be used to screen embryos for abnormalities in chromosome number. Only embryos which are found to be “normal” for the tested chromosomes are considered suitable for transfer to the uterus.
- PGD is NOT 100% accurate. Confirmatory prenatal diagnosis is highly recommended if a pregnancy is achieved following PGD.

What is Aneuploidy Screening?

An individual's genetic information is packaged into strings of DNA called chromosomes. Normal embryos contain 46 chromosomes, or 23 chromosome pairs. These chromosome pairs are labelled 1 to 22 (the autosomes) and X and Y (the sex chromosomes). Some embryos can have an abnormal number of chromosomes (ie: missing or extra chromosome/s) due to errors in cell division in the developing egg, sperm or embryo. This is known as chromosomal aneuploidy. An aneuploid embryo will fail to implant, miscarry, or result in the birth of an affected child. PGD testing can be used to screen for the most commonly observed chromosomal aneuploidies (ie: those involving chromosomes 13, 15, 16, 17, 18, 21, 22, X and Y; see Figure 1A). This testing may be appropriate for:

- Couples with advanced maternal age (>36 years)
- Couples who have experienced repeated implantation failure
- Couples who have experienced repeated miscarriage
- Couples who have experienced repeated IVF failure
- Couples who have previously had a pregnancy with a chromosomal abnormality
- Couples where one partner has an altered sex chromosome complement (eg: XXY)

How is this test done?

Embryo biopsy is performed on Day 3 after egg collection (please refer to the “Preimplantation Genetic Diagnosis” fact sheet for further information relating to the embryo biopsy procedure). The biopsied cells are tested using a technique called Fluorescence In Situ Hybridisation (FISH). This technique involves the use of fluorescent dyes which are used to tag specific chromosomes. These tags show as coloured dots which can be used to indicate the number of chromosomes present in the biopsied cell. Testing is performed in a two round process and takes approximately 8 hours to complete. The first round of the test involves analysis of chromosomes 15, 17, X and Y, while the second round of the test involves analysis of chromosomes 13, 16, 18, 21 and 22. Figure 1B provides an example of a “normal” male and “normal” female FISH result, as well as an example of an “abnormal” male with trisomy 21 (ie: three copies of chromosome 21 which results in Down syndrome) and an “abnormal” female with trisomy 18 (ie: three copies of chromosome 18 which results in Edward's syndrome).

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Figure 1A: Chromosomes tested during Aneuploidy Screening, and the location of the FISH probes

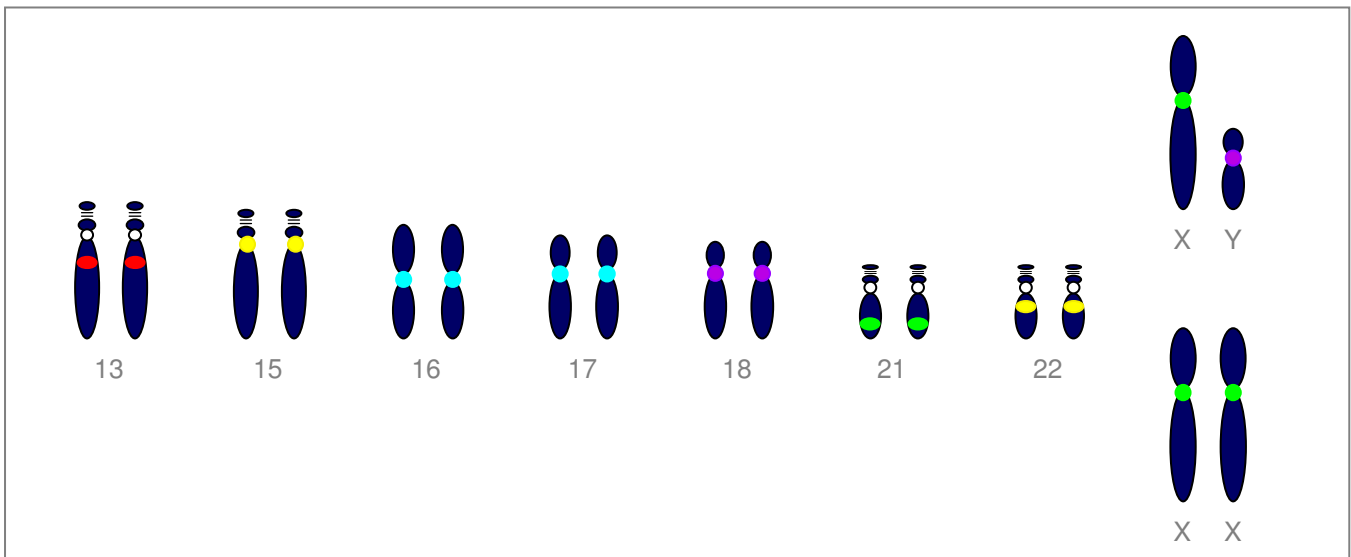
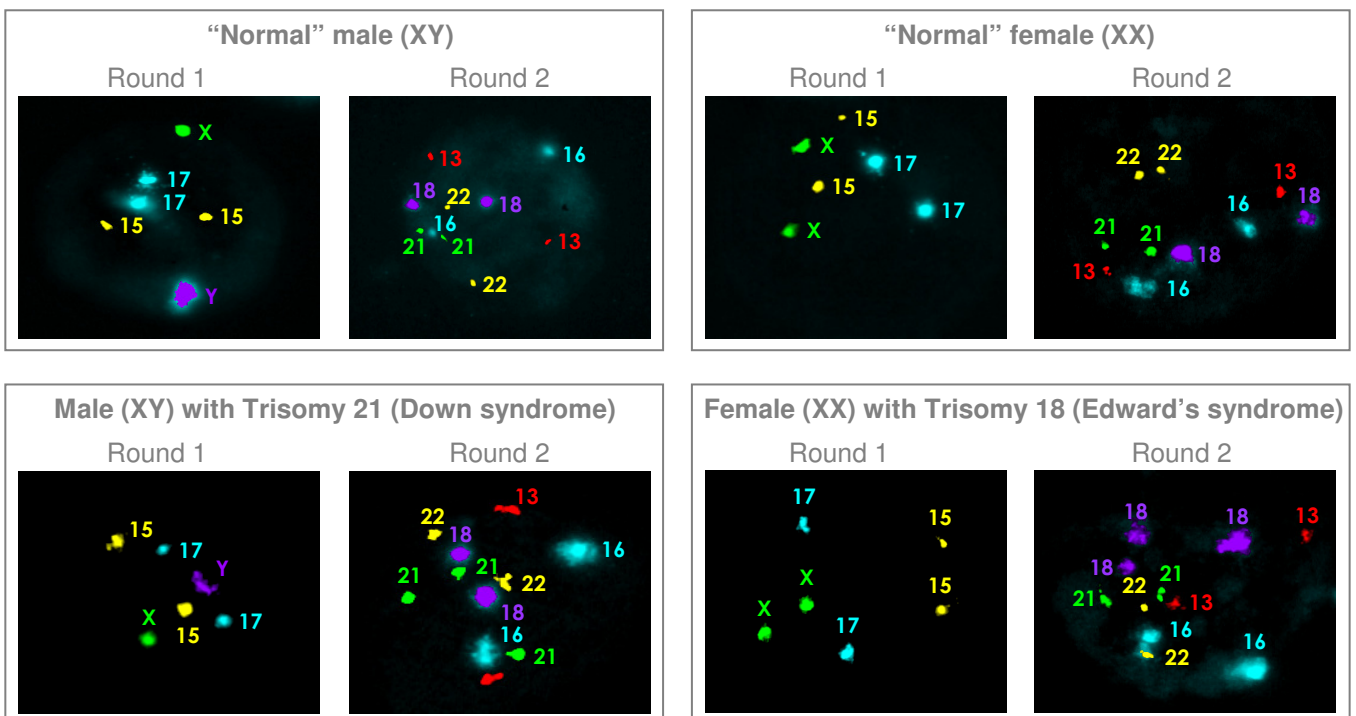


Figure 1B: FISH analysis of biopsied cell/s



The embryo is kept in culture while testing of the biopsied cell/s proceeds. Embryos identified as being "normal" for the chromosomes tested can be transferred on Day 4 or Day 5. When a number of "normal" embryos are identified, morphological criteria are also used to determine those best for transfer. Surplus "normal" embryos which are not transferred but which continue to develop satisfactorily to the blastocyst stage may be frozen. These embryos may be used in a subsequent IVF cycle if the couple does not achieve a pregnancy with the fresh embryos. Chromosomally abnormal embryos are discarded or donated to research/training with the couple's consent.

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Other important information

- PGD for Aneuploidy Screening is appropriate as an embryo selection process if more than two 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 one or two embryos on Day 3, it may be considered more beneficial to transfer these embryos without biopsy.
- Due to the complexity of FISH testing it may not be possible to obtain a conclusive result for some or all embryos (or for some chromosomes). In this case, the embryos can either be transferred without a genetic result or frozen if they reach an appropriate stage of development (ie: form a blastocyst).
- PGD for Aneuploidy Screening involves the analysis of nine chromosomes (ie: 13, 15, 16, 17, 18, 21, 22, X and Y). The test does not give any information relating to other chromosomes, other genetic conditions or other abnormalities.
- This test is only a screening test and therefore cannot provide an absolute guarantee of the chromosome status of the embryo. In some embryos, the biopsied cell/s may not be representative of the whole embryo. Given this, an error rate of approximately 10% is associated with this test and **prenatal diagnosis is highly recommended in an ensuing pregnancy.**

What are the costs?

Information relating to the cost of PGD is available from Monash IVF. Please note that additional costs will be incurred for IVF.

Quality systems

Monash IVF employs a very high standard of quality assurance. Through the application of quality systems the laboratory provides standards of excellence in quality service, care and advice.