

Monash IVF

Fact Sheet

Preimplantation Genetic Diagnosis (PGD) with Sex Selection for X-linked Disorders



Key points:

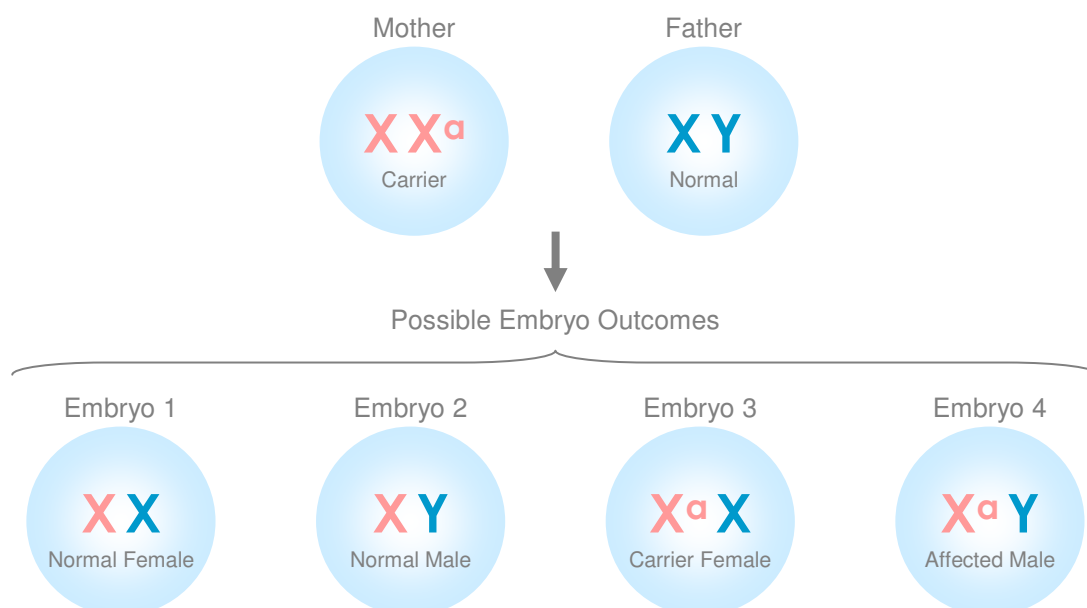
- Individuals with a family history of an X-linked genetic disorder may be at risk for passing the condition on to their children.
- Preimplantation Genetic Diagnosis (PGD) is a viable option for couples at risk of passing on a specific X-linked genetic disorder to their child, and significantly increases the chance of having a healthy baby.
- To undertake PGD one or both partners must have had previous genetic testing to confirm that they are at risk of passing the X-linked genetic disorder on to their children.
- PGD is NOT 100% accurate. Confirmatory prenatal diagnosis is highly recommended if a pregnancy is achieved following PGD.

What is an X-linked disorder?

An individual's genetic information is packaged into strings of DNA called chromosomes. Normal human cells contain 46 chromosomes, or 23 chromosome pairs. These chromosome pairs are labelled 1 to 22 (the autosomes) and X and Y (the sex chromosomes). Females carry two X chromosomes, while males carry an X and a Y chromosome.

X-linked disorders such as Haemophilia or Muscular Dystrophy are caused by different gene changes on the X chromosome. Because these gene changes are carried on the X chromosome, the probability of a particular embryo being affected is dependent upon its sex (Figure 1). The potential risks to offspring will vary according to the parental origin of the gene change.

Figure 1: An example of the possible embryo outcomes following sex selection for an X-linked disorder carried by the mother. The X chromosome carrying the gene change is indicated by X^a.



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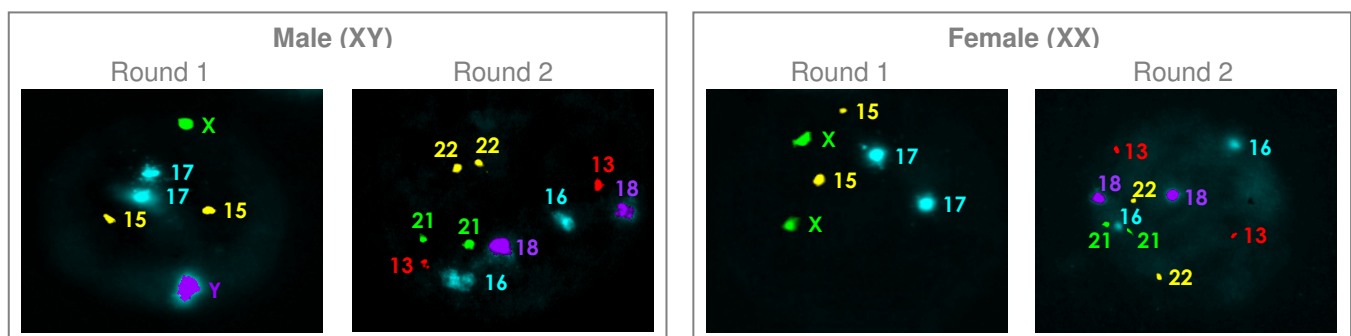
Preimplantation Genetic Diagnosis (PGD) with Sex Selection for X-linked Disorders



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. Using FISH, the sex of the embryo is determined by testing for chromosomes X and Y. Chromosomes 13, 15, 16, 17, 18, 21 and 22 are also analysed as these are the chromosomes most frequently involved in implantation failure, miscarriage or abnormalities at birth (eg: Down syndrome) (Figure 1). Testing takes approximately 8 hours to complete.

Figure 1: FISH analysis of biopsied cell/s



The embryo is kept in culture while the testing of the biopsied cells proceeds. Embryos of the desired sex which are identified as being “normal” for the chromosomes tested can be transferred on Day 4 or Day 5. When a number of “normal” embryos of the desired sex 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 do not achieve a pregnancy with the fresh embryos. Chromosomally abnormal embryos are discarded or donated to research/training.

Other important information

- Due to the complexity of FISH testing it may not be possible to obtain a conclusive result for some or all embryos. 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 with sex selection involves an analysis of the sex chromosomes (ie: X and Y), in addition to seven other chromosomes (ie: 13, 15, 16, 17, 18, 21 and 22) frequently involved in implantation failure, miscarriage or abnormalities at birth. The test does not give any information relating to any 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.