 瑪麗醫院 QUEEN MARY HOSPITAL	Department of Obstetrics and Gynaecology	Document No.	OGRM221
	<b>Subject</b> <b>Information on PGT-M English</b>	Issue Date	April 2020
		Next review date	July 2022
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## Information on Preimplantation Genetic Testing (PGT) for Monogenic Diseases

### 1. Nature

This method aims at determining the genetic make-up of embryos before they are transferred to the womb. It allows selection and transfer of embryos unaffected by the monogenic disorder which the fetus is at risk for.

### 2. Indications

- Autosomal dominant / recessive disease (wife / husband)
- Sex-linked dominant / recessive disease (wife / husband)
- Combined with HLA typing

### 3. Procedure

In addition to IVF procedure, the following steps are involved:


- Fertilization by intracytoplasmic sperm injection
- Removal of 3-5 trophectoderm cells from blastocysts and each blastocyst individually frozen
- Genetic study of the cells by polymerase chain reaction (PCR)
- Checking correct chromosome number can be performed at the same time
- Genetically-transferrable blastocyst will be transferred to the womb in subsequent frozen-thawed cycles.
- Carrier blastocysts with minimal risk of developing the disease will not be discarded

### 4. Risks

The biopsy of cells from blastocysts carried less adverse effect on the pregnancy rate, compared with those from embryos.

### 5. Test accuracy and limitations

- PGT has <5% misdiagnosis.
- We only test for the indicated mutation(s) only.
- False result can be attributed to allelic dropout, aneuploidy, undetected recombination or mosaicism of embryos.

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## 6. Follow up

After successful conception, conventional prenatal diagnosis (ultrasound follow-up, chorionic villus sampling or amniocentesis) will be required to confirm the diagnosis made before implantation.

After delivery, it is advisable to follow up the development of the baby.

## 7. Alternative

The alternative is to try natural conception and undergo prenatal diagnosis when pregnant.

### Note:

- For X-linked recessive disorders, the couple after counselling requested to discard the carrier blastocysts. Reasons: \_\_\_\_\_
- Please use contraception during the treatment cycle.
- We will proceed to the blastocyst biopsy if there is even one blastocyst. Please make sure that you can be contacted by telephone during the treatment cycles.
- There may *NOT* be any transfer after PGT if blastocysts are not cleaving after PGT or all are genetically abnormal.
- We CANNOT disclose the sex of the blastocysts unless medically indicated.
- Only one blastocyst will be replaced in each transfer after PGT.

(Affix label)  
Wife's name &  
ID number

(Affix label)  
Husband's name &  
ID number

Signature of Wife: \_\_\_\_\_

Signature of Husband: \_\_\_\_\_

Date: \_\_\_\_\_