 瑪麗醫院 QUEEN MARY HOSPITAL	Department of Obstetrics and Gynaecology	Document No.	OGRM220
		Issue Date	May 2024
	<u>Subject</u> Information on PGT-SR English	Next review date	April 2026
		Approved by	HKU-QMH-KWH CARE
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Information on Preimplantation Genetic Testing (PGT) for Structural Rearrangements

1. Nature

This method aims at determining the chromosomal number of embryos before they are transferred to the womb. It allows selection and transfer of embryos with normal number of chromosomes.

2. Indications

- ☐ Translocation
- ☐ Inversion
- ☐ Others, specify: _____

3. Procedure

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


- Fertilization by intracytoplasmic sperm injection
- Embryo biopsy: removal of 5-10 trophectoderm cells from blastocysts
- Each blastocyst is individually frozen after embryo biopsy
- Genetic study of the cells by next generation sequencing (NGS)
- Transfer of a blastocyst with normal chromosomal number to the womb in frozen transfer cycles

4. Risks

The biopsy of cells from blastocysts may reduce the pregnancy rate (~5%), when compared with no biopsy.

5. Test accuracy and limitations

- PGT has <5% misdiagnosis.
- NGS **CANNOT** detect point mutations, triplet repeat expansions, uniparental disomy and certain form of haploidy and polyploidies (e.g. 69,XXX). NGS may not effectively detect small chromosomal imbalance (<20 Mb).
- NGS **MAY NOT** distinguish balanced translocation carrier from normal blastocyst.
- False result can be attributed to mosaicism of blastocysts.

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6. Follow up

After successful conception, conventional prenatal diagnosis is still needed if clinically indicated.

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:

- ☐ We will proceed to the embryo biopsy even if there is only one blastocyst.
- ☐ There may NOT be any transfer after PGT if all are genetically abnormal or the blastocysts cannot survive after thawing.
- ☐ We CANNOT disclose the sex of the blastocysts unless medically indicated.
- ☐ Only one blastocyst will be replaced in each transfer. Please use contraception during the transfer cycle.

(Affix label)
 Wife's name &
 ID number

(Affix label)
 Husband's name &
 ID number

Signature of Wife: _____

Signature of Husband: _____

Date: _____