 瑪麗醫院 QUEEN MARY HOSPITAL	<b>Department of Obstetrics and Gynaecology</b>	<b>Document No.</b>	<b>OGRM225</b>
	<b>Subject</b> <b>Information on PGT-SR English</b>	<b>Issue Date</b>	<b>April 2020</b>
		<b>Next review date</b>	<b>July 2022</b>
		<b>Approved by</b>	<b>HKU-QMH-KWH CARE</b>
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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 (wife / husband)
- Inversion (wife / husband)

### **3. Procedure**

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


- Fertilization by intracytoplasmic sperm injection
- Removal of 3-5 trophoblast cells from blastocysts and each blastocyst individually frozen (or occasionally 1 cell from a day 3 embryo)
- Genetic study of the cells by next generation sequencing (NGS)
- Transfer of a blastocyst with normal chromosomal number to the womb in subsequent frozen-thawed transfer cycles.

### **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.
- NGS **CANNOT** detect point mutations nor polyploidies (e.g. 69,XXX). NGS may not effectively detect small chromosomal imbalance (<20 Mb).
- NGS **CANNOT** 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:

- 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 blastocyst transfer after PGT if all blastocysts have abnormal chromosomal number.**
- We CANNOT disclose the sex of the blastocyst, 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: \_\_\_\_\_