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
- ❑ Sex-linked dominant / recessive disease
- ❑ Combined with HLA typing

3. Procedure

In addition to IVF procedure, 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 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 embryo cycles.
- Carrier blastocysts with minimal risk of developing the disease will not be discarded.

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

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: _____**
- ❑ **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: _____