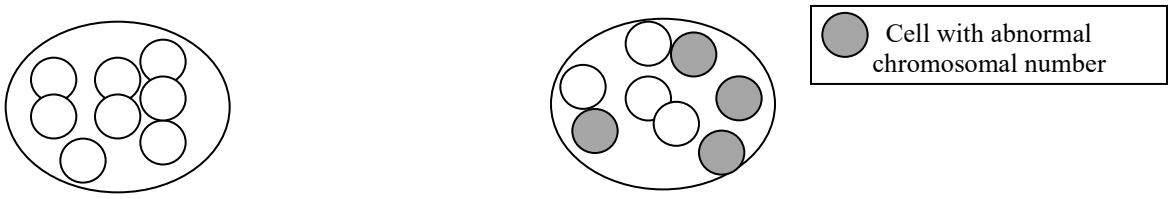
Information on Mosaic Embryos after PGT for Aneuploidy

1. What is meant by mosaic embryos?

Preimplantation genetic testing (PGT) for aneuploidy 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.

Mosaicism of embryos occurs when two or more cell populations with different chromosomal makeup are present within the same embryo. Embryos can be categorized into the following:

a. normal (<30% of cells biopsied are abnormal)
b. low level mosaic abnormal (30 - < 50 % of cells biopsied are abnormal)
c. high level mosaic abnormal (50% - 70% of cells biopsied are abnormal)
d. abnormal (>70% of cells are abnormal)



Normal embryos

Mosaic embryos

2. Why does this occur?

Mosaicism results from mitotic errors occurring after fertilization, occasionally in the first cleavage but more commonly in the second or third cleavage. It is common in embryos. According to the data in our laboratory, 10% of blastocysts are suspected to be mosaic, among which 70% are of low level mosaic.

Biopsy of several cells at the blastocyst stage and use of new diagnostic techniques such as next generation sequencing, have led to increased reporting of mosaicism after PGT.

3. What will happen if these mosaic embryos are replaced? Or what are the risks?

Possible outcomes:

- Not pregnant or reduced implantation rate
- Increased miscarriage rate
- Congenital abnormalities
- Uniparental disomy (UPD, when a person receives two copies of a chromosome from one parent and no copy from the other parent)
- Increased risk of intrauterine growth restriction and other pregnancy complications
- Delivery of healthy babies

4. What is the recommendation when these mosaic embryos are replaced?

Priority of replacing the mosaic embryos

a. mosaic with segmental regions
b. low level mosaic with whole chromosome (1 →2 chromosomes)
c. high low mosaic with whole chromosome (1 →2 chromosomes)

*We do not advise to replace mosaic embryos involving more than 2 whole chromosomes.

5. Follow up

With emergence of mosaic embryos, international data shows there were more than a few thousand mosaic embryos being replaced already. Among livebirths reported, only several livebirths were found to have persistent mosaic genetic make-up.

After successful conception, conventional prenatal diagnosis by amniocentesis is needed. Current technology of non-invasive prenatal DNA testing is unable to confirm mosaic genetic make-up in mosaic embryos.

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

6. **Alternative**

For private patients, you may start another PGT cycle to increase the chance of identifying a blastocyst with normal chromosome number for transfer while the mosaic embryos can remain frozen in the laboratory or be discarded.

(Affix label) Wife's name & ID number	(Affix label) Husband's name & ID number

Signature of Wife: _____ Signature of Husband: _____

Date: _____