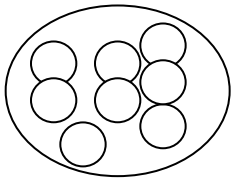
Information on Mosaic Embryos after PGT for Aneuploidy

1. What is meant by mosaic embryos?

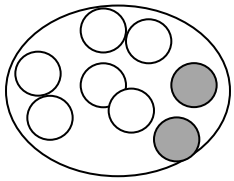
Preimplantation genetic testing (PGT) for aneuploidy aims at determining the chromosomal number of embryos before they are transferred to the womb. It allows selection and transfer of embryos with normal number of chromosomes.

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:

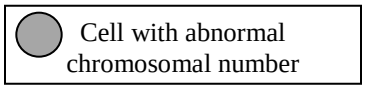
a. normal (<20% of cells biopsied are abnormal)
b. low level mosaic abnormal (20 - < 50 % of cells biopsied are abnormal)
c. high level mosaic abnormal (50% - 80% of cells biopsied are abnormal)
d. abnormal (> 80% of cells are abnormal)



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 or blastocysts. According to the data in our laboratory, 10% of blastocysts are suspected to be 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. low level mosaic segmental
b. low level mosaic monosomy (but not 45,X)
c. low level mosaic trisomy (those involving 2, 7, 13, 14, 15, 16, 18 and 21 in a lower priority)*
d. high level mosaic segmental
e. high level mosaic monsomy (but not 45,X)
f. do not replace high level mosaic trisomies involving chromosomes 13, 14, 16, 18 and 21*
g. do not replace complex mosaic i.e. 3 or more chromosomes

*For mosaicism involving trisomies, the following order shall be considered for replacement:

i. Trisomies involving chromosome 1, 3, 10, 12, 19 have the highest priority for transfer
ii. Trisomies 4, 5 and 47, XYY
iii. Trisomy 2, 17, 22
iv. Trisomies 6, 9 and 15
v. Trisomies 8, 20, 47,XXX and 47, XXY

5. Follow up

After successful conception, conventional prenatal diagnosis by amniocentesis is needed. After delivery, it is advisable to follow up the development of the baby.

6. Alternative

You can start another IVF cycle with PGT to increase the chance of identifying a normal blastocyst with normal chromosome number for transfer. 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: _____