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Trophectoderm biopsy and transfer in a subsequent frozen thaw embryo replacement cycle in preimplantation genetic diagnosis cycles

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Introduction

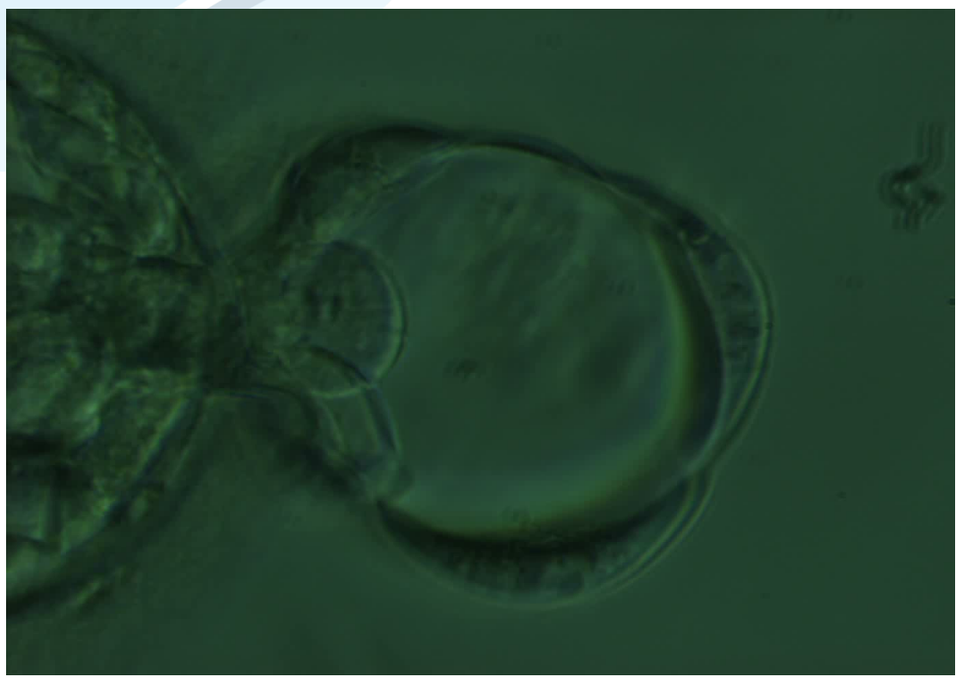
Blastocyst biopsy compared with cleavage stage biopsy has been reported to have a higher rate of genotyping, less amplification failure, less allele drop out and fewer embryos to biopsy. It is also associated with higher implantation and pregnancy rates than cleavage stage biopsy. Furthermore, FER can prevent late onset ovarian hyperstimulation syndrome (OHSS), is associated with improved perinatal outcomes and a higher ongoing pregnancy rate.

Aim

To determine the pregnancy outcome of trophectoderm biopsy and transfer in a subsequent frozen embryo replacement (FER) cycle in couples undergoing preimplantation genetic diagnosis (PGD).

Materials and Methods

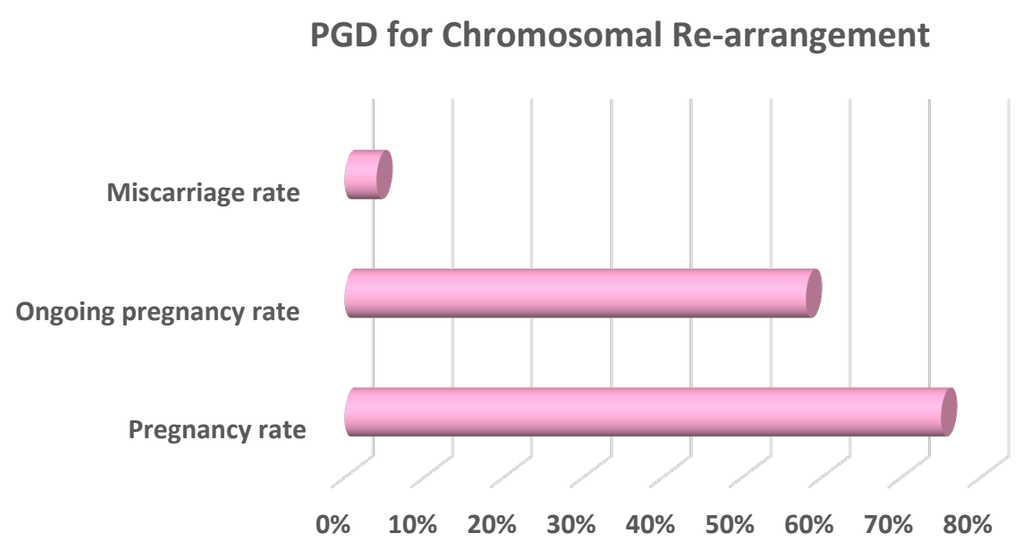
This is a retrospective case series of PGD cycles performed from July 2014 to December 2015.
Couples were either referred from a regional genetic centre, or self-referred. They were counselled and standard IVF protocols were applied.
Eighty five couples had biopsy at blastocyst stage and vitrification. 66/85 couples had a suitable embryo to transfer. Of the remaining 19 couples, 12 are still waiting, whilst 7 couples had blastocysts which were not suitable for transfer.
Embryos were cultured to blastocyst and biopsied as described by Gardner, 2007 and as shown in the following video.

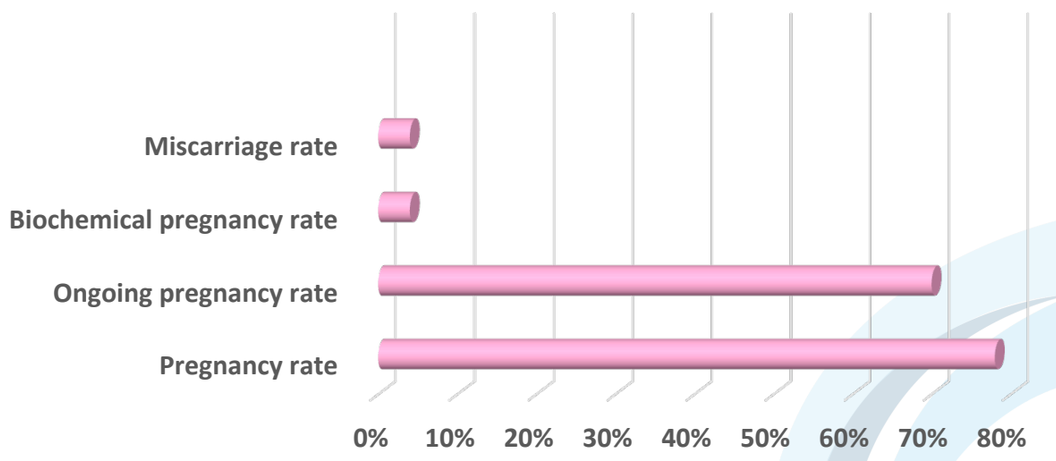


The cells were sent to the reference laboratory for genotyping. Standard procedures for thaw of embryos were performed and blastocysts were transferred in a subsequent medicated FER cycle. Patients carried out their urinary pregnancy test 16 days later.

Results

A total of 85 cycles of trophectoderm biopsy and vitrification were performed. 72 were for single gene disorders and 13 for chromosomal rearrangements. The total number of blastocysts biopsied was 398 and 182 (48%) blastocysts were suitable for transfer.
All couples had elective single embryo transfer.
There were a total of 54 PGD cycles for single gene disorder with known pregnancy outcome, 42 FER cycles resulted in a positive pregnancy test. The ongoing pregnancy rate was 38/54, 2/54 FER cycles resulted in biochemical pregnancies, 2/54 resulted in first trimester miscarriage and 12/54 FER cycles had a negative pregnancy test.
Of 12 PGD cycles for chromosomal rearrangements with known pregnancy outcome, 9 FER cycles resulted in a positive pregnancy test. The ongoing pregnancy rate was 7/12 and 2/12 of such cycles resulted in first trimester miscarriage.





Conclusion and Limitations

Trophectoderm biopsy, vitrification and FER can be successfully applied to PGD cycles for single gene disorders and chromosomal rearrangements. This approach increases the rate of genotyping and moreover, implantation and pregnancy rates exceed those following cleavage stage biopsy.
This is the largest number of pregnancy outcomes so far reported from a trophectoderm biopsy, vitrification and FER dataset for single gene disorders and chromosomal rearrangements. However, results are retrospective and non-randomized.