Retrospective Cohort Analysis on Couples Seeking Pre-implantation Genetic Testing over a 5-year Period

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Introduction

Preimplantation genetic testing (PGT) is a method that allows screening of embryos for genetic diseases or chromosomal disorders during assisted reproduction cycles. It serves as an alternative to prenatal diagnosis for at-risk couples by offering early detection and selection of unaffected embryos prior to embryo transfer into the uterus. PGT is classified into three types: PGT for aneuploidy screening (PGT-A), PGT for monogenic diseases (PGT-M) and PGT for structural rearrangements (PGT-SR). With increasing acceptance and utility of PGT in recent years, this study aims to review the usage of PGT in Queen Mary Hospital, Hong Kong.

Methods

This is a single-centred, retrospective review on the usage of PGT in Hong Kong over the past 5 years. PGT cases performed in HKU-QMH Centre of Assisted Reproduction and Embryology (HKU-QMH CARE) from January 2015 to September 2020 were collected. The indication for PGT, type of PGT requested, anticipated reproductive risks, and demographics are collected for analysis.

Results

A total of 411 couples requested PGT during the study period. The mean age of women was 34.3 years old (range from 21 to 43).

- 192 couples (47%) requested PGT-M for monogenic disorders
- 89 couples (22%) requested PGT-SR for balanced chromosomal changes
- 10 couples (2%) requested PGT-A for (mosaic) sex aneuploidy in the couple
- 120 couples (29%) requested PGT-A because of recurrent pregnancy loss, repeated implantation failure, advanced maternal age and previous abnormal pregnancy

Among the couples requesting PGT-M:

- 130 couples were at-risk of passing on an autosomal recessive (AR) condition
- 44 couples were at-risk of passing on an autosomal (AD) condition
- 18 couples were at-risk of passing on an X-linked (XL) condition

Conclusion

This study reviewed the PGT usage of Hong Kong couples at a single centre over a 5-year period. Most couples requested PGT for a known genetic disease risk. With the advancement in genomic medicine, PGT will continue to play a critical role in assisting reproduction for at-risk couples.