Aspiro 2022

The 11th Virtual Congress of the Asia Pacific Initiative on Reproduction

Addressing the Challenges of Human Reproduction

Thursday, 28 April - Sunday, 1 May, 2022

COMPARISON OF NEXT-GENERATION SEQUENCING LIBRARY PREPARATION METHODS FOR CHROMOSOME ANEUPLOIDY SCREENING IN PREIMPLANTATION EMBRYOS

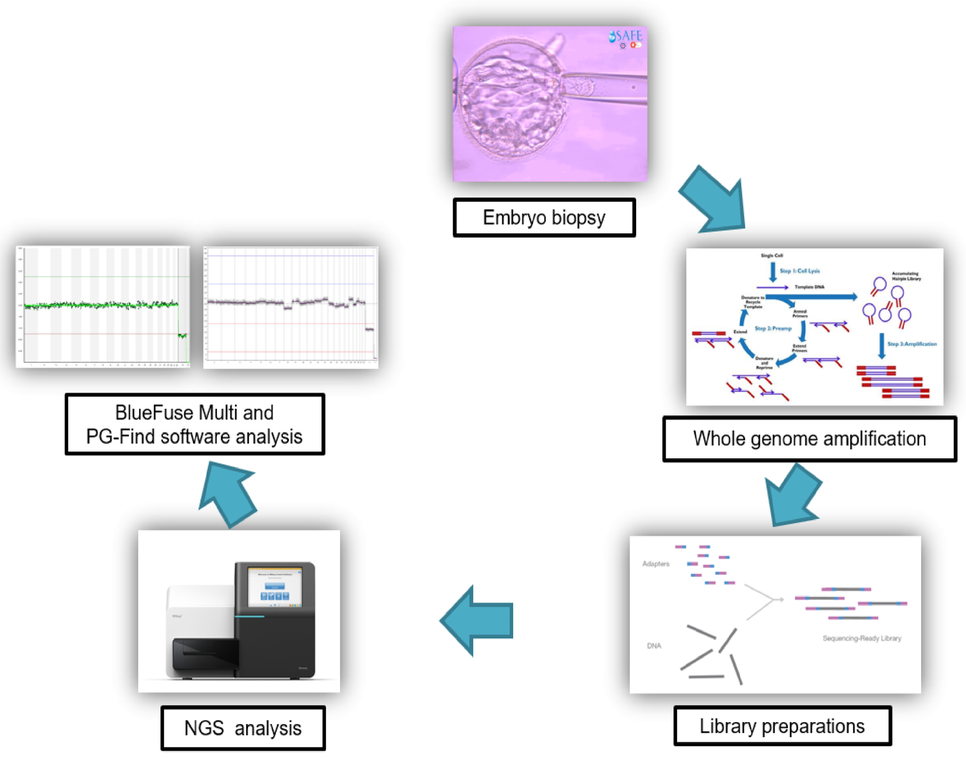
Ms. Saripaporn Suwannasing[1], Ms. Wanwisa Suksalak[1], Ms. Chanakarn Jiramongkolrat[1], Ms. Sirikul Kulanuwat[1], Ms. Wipavee Tangsee[1], Ms. Sunanta Nabu[1], Ms. Sujin Chanchamroen[1,2], and Dr. Wiwat Quangkananurug[1,2]

[1]Next Generation Genomic Co. Ltd, Bangkok, Thailand. [2]Safe Fertility Center Co. Ltd, Bangkok, Thailand.

Background and Aims

To analyze and compare the performance of two next-generation sequencing (NGS) library preparation kits, VeriSeq PGS (Illumina, USA) and PG-Seq kits (PerkinElmer, USA) for the detection of whole, segmental, and mosaic aneuploidies in preimplantation genetic testing for aneuploidy (PGT-A).

Methods



124 trophectoderm biopsied

Whole genome amplification
SurePlex WGA kit (Illumina, USA)

Library preparations were performed using
- VeriSeq PGS (Illumina, USA)
- PG-Seq preparation reagent kits (PerkinElmer, USA)

All libraries were sequenced on the Illumina MiSeq platform (Illumina, USA)

The sequencing results were analyzed by
- BlueFuse Multi analysis software for Veriseq PGS
- PG-Find analysis software for PG-Seq

Results

Using VeriSeq preparation reagent kit, embryos were classified as euploid 14.5% (18/124), aneuploid 74.2% (92/124) and mosaicism 10.5% (13/124), respectively. The PG-Seq based NGS results were identified 19.3% (23/124) euploid embryos, 70.2% (87/124) aneuploid and 9.7% (12/124) mosaic embryos.

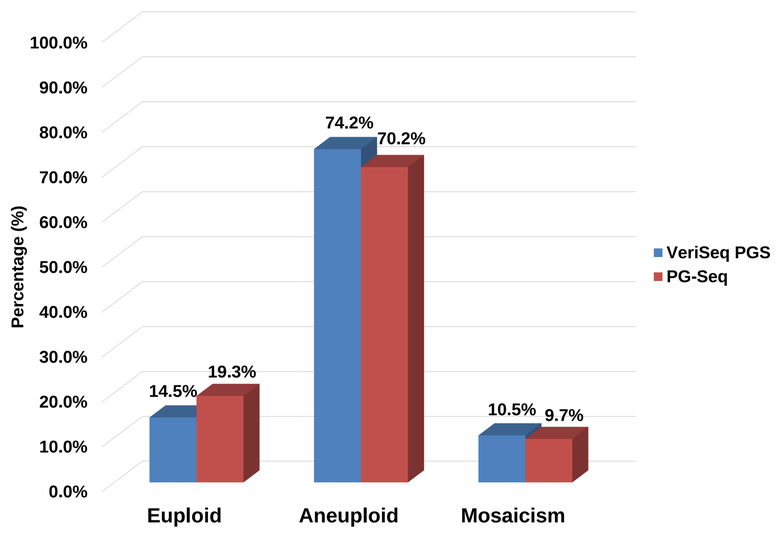


Fig 1. The comparison of NGS based VeriSeq PGS and PG-Seq results.

The concordance rate of two library preparation kits was 93.5% (116/124, kappa = 0.81) with fully chromosomal concordance 82.3% (102/124) and partially chromosomal concordance 11.3% (14/124). The chromosomal discordance rate per analyzed chromosome between two methods was 1.65% (49 of 2,976 analyzed chromosomes).

Table 1. The concordance rate of VeriSeq PGS and PG-Seq library preparation kits

Performance characteristic	N = 124
Fully chromosomal concordance	82.3% (102/124)
Partially chromosomal concordance	11.3% (14/124)
Chromosomal discordance	6.4% (8/124)

Conclusions

Our findings demonstrate that the genomic DNA amplified by SurePlex WGA can be applied and analyzed by PG-Seq technology. The SurePlex WGA combined with PG-Seq kit is efficient of detecting chromosome aneuploidy in PGT-A. Our study highlights the importance of the evaluation of library preparation prior to clinical use in order to know the limitation of each method as well as to improve the interpretation of NGS results.

References

1. Rostislav Navratil, Jakub Horak, Miroslav Hornak, David Kubicek, Maria Balcova, Gabriela Tauwinklova, Pavel Travnik, Katerina Vesela, Concordance of various chromosomal errors among different parts of the embryo and the value of re-biopsy in embryos with segmental aneuploidies, *Molecular Human Reproduction*, Volume 26, Issue 4, April 2020, Pages 269–276

Acknowledgements

Next Generation Genomic Co. Ltd and SAFE Fertility Center, Bangkok, Thailand.