ABSTRACT

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Study Program	:	Bioinformatics
Title	:	Development and Validation of Aneuploidy Detection Program for Targeted-Next Generation Sequencing Data

Preimplantation Genetic Testing (PGT) is a technique employed in an In Vitro Fertilization procedure to detect any genetic defect in the embryo prior to implantation. Aneuploidy is one of the conditions screened during PGT to ensure every chromosome in the embryo has the right copy number. There are several procedures to perform aneuploid detection but the use of Next-Generation Sequencing allowed whole-genome sequencing of the embryo and aneuploidy assessment for all 23 chromosome pairs. Here we propose a new method for aneuploidy detection method with the use of AmpliSeq for Illumina, a targeted-sequencing chemistry suite that allows targeted sequencing of genomic regions of interest. In addition to aneuploidy detection, we are also interested in microdeletion and microduplication syndrome (MMS) detection. In the bioinformatics side of the study, we aim to develop an easy to use application with a graphical user interface for aneuploidy detection for targeted sequencing. Using a reference file that was constructed from genomic normal male samples without whole genome amplification (WGA), we were able to correctly determine the chromosome copy number of genomic samples without WGA. However, the chromosome copy number of single-cell samples and samples that have been subjected to whole genome amplification was not able to be determined. MMS were unable to be detected because the MMS samples in this study were not genomic samples and have low read counts.