

Topic: Preimplantation Genetic Screening

**ASSESSMENT OF MOSAICISM CONCORDANCE IN THE WHOLE EMBRYO USING HR-NEXT GENERATION SEQUENCING AND THE CLINICAL IMPACT OF CURRENT CALLING PRACTICES**

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Increased detection and quantification due to advancements in preimplantation genetic screening (PGS), namely high resolution next generation sequencing (hr-NGS), have rendered scientists and clinicians alike uncertain how to proceed when considering the transfer of mosaic embryos. Here hr-NGS by VeriSeq PGS (Illumina) was used to determine concordance rates between either the whole embryo or trophectoderm of embryos previously determined to contain 1 or 2 mitotic aneuploidies. Additionally, clinical outcomes from elective single embryo transfers (eSETs) were obtained from a single center and the associated profiles were blindly classified for mosaicism and correlated to their respective outcomes. In embryos where a second trophectoderm biopsy was obtained, 38% exhibited concordance of mosaic chromosomes in both biopsies, while 46% exhibited no mosaicism in one biopsy when compared to the other. At the chromosome level, 72% (52/72) of chromosomes determined to be mosaic in one biopsy exhibited euploid CNVs in the other. Furthermore, assessment of whole embryos revealed 51% (55/107) of embryos with trophectoderm mosaicism exhibited euploid CNVs. At the chromosome level, 66.4% (95/143) of mosaic chromosomes in the trophectoderm exhibited euploid CNV's in the whole embryo. Euploid CNVs in the whole embryo were observed for 92% (33/36) of chromosomes exhibiting 40% mosaicism as compared to just 27% for chromosomes exhibiting 70% mosaicism. Interestingly, When classifying profiles to clinical outcomes, little difference was observed in implantation (75% v 71.2%), loss (22% v 16%) and ongoing pregnancy rates (58.3% v 59.9%), in transfers of embryos with (n = 24) and without (n = 177) mosaicism, respectively. Overall, this work provides valuable insight into the understanding of trophectoderm mosaicism identified during routine PGS using hr-NGS. Furthermore, these data provide the ground work for adapting mosaicism calling practices to improve upon the selection of embryos that are and are not suitable for transfer.