Mosaicism: Factors in Determination to Implant



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PGT-A is very accurate using Next Generation Sequencing (NGS) to distinguish euploid from aneuploid. Predictive Value Studies transferring embryos while physician is unaware at transfer of chromosomal status (but learning at 13 weeks) reveal 65% success for euploid transfers but almost zero for aneuploid. Transfer of a single euploid embryo in three successive cycles also results in over 90% women achieving pregnancy. Accuracy of NGS is accompanied by 10-15% mosaicism. Several types of mosaicism exist, most favorable being segmental in which cell cycle replication has not been completed. Mosaic embryos can be transferred if no euploid embryo exists. If amount of aneuploid DNA in an embryo is 20-50%, outcome is almost as good as transferring an embryo with less than 20% aneuploid DNA. However, implantation rate is less and miscarriage rate more.

Collaborative longitudinal studies are in progress to determine outcome of transferring a mosaic embryo. The majority results in full term pregnancy with neonatal anomalies no different from the general population. Mosaicism can persist during ongoing pregnancy but only rarely. Counseling couples electing to transfer a mosaic embryo is now a pivotal component of IVF.