Mosaicism in human embryos: etiology and pregnancy outcome

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About 15-30% of human preimplantation embryos produced during ART treatments contain two or more cytogenetically different cell lines and are mosaic. Mosaics can involve the presence of different abnormal cells lines in the absence of any euploidy one, or a mixture of euploid and aneuploid cell lines. Mosaicism has been described in cleavage stage embryos over two decades ago with extensinve numbers of embryos fully analyzed by FISH. With the transition from FISH to molecular Comprehensive chromosome analysis (CCA) techniques such as aCGH, SNP arrays and qPCR, that analyzes a blastocyst biopsy as a whole, mosaicism became hard to detect. Recently, high-resolution next-generation sequencing (hr-NGS), have allowed mosaicism to be detected with much greater sensitivity than earlier CCA methods.

Recent studies show that blastocysts classified as mosaic by hr-NGS implant less and miscarry more than euploid embryos, but have more potential to reaching term than fully aneuploid ones, which have close to zero. We recommend that in the absence of euploid embryos, mosaic embryos can be considered for transfer in the absence of fully euploid embryos.

Unlike meiotic aneuploidy, mosaicism occurs post-meiotically and does not increase with maternal age. Instead, ART related procedures seem to affect mosaicism rates per center.