Chromosomal Mosaicism In Embryos From Young ART Patients Determined By Array Comparative Genomic Hybridisation (aCGH)

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Abstract

Aim: Chromosomal mosaicism, where different cells have a different chromosomal complement, reportedly affects approximately 60% of cleavage stage embryos. Of particular concern are diploid mosaic embryos which contain a mixture of diploid and aneuploid cells as they will confound preimplantation genetic screening (PGS) outcomes because the tested cell may not represent the embryo. The majority of this information is derived after analysis of less than 10 chromosomes and cells diagnosed as diploid may be aneuploid for chromosomes that were not tested for. This study reports the extent of mosaicism of all 24 chromosomes in cleavage stage embryos from young, successful ART patients.

Methods: 23 day-3 embryos were allowed to succumb, separated into 115 individual blastomeres (mean 5.0/embryo) and subjected to single cell arrayCGH using Sureplex whole genome amplification and 24sure BAC array analysis (Bluegnome, UK).

Results: Seven (30%) embryos were euploid in every cell, 5 (22%) had a consistent aneuploidy in every cell, which is presumed to be meiotic in origin, and 11 (57%) were mosaic. Of the mosaic embryos, 6 (26%) were aneuploid mosaic where every cell was aneuploid but with different aneuploidies in different cells. Five (22%) embryos were diploid mosaic with a mean of 31% aneuploid and 69% diploid cells per embryo.

Conclusion: This study shows that, even in good quality embryos from young ART patients, i) only a minority of embryos are euploid in every cell, ii) meiotic aneuploidy affects about 1 in 5 embryos, iii) diploid mosaicism affects only about 1 in 5 embryos and a minority of these blastomeres are aneuploid.