

Making sense of PGS: EBM, RCTs, mosaicism and reconciling the two sides

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Mosaicism is defined as the presence of more than one chromosomally distinct cell populations in a tissue or organism. In essence we are all mosaics given that some tissues have haploid or polyploid cells and clinical consequences of aneuploid/diploid mosaics are well documented. In human embryos mosaicism is commonplace and some studies suggest that the majority are mosaic. There are a variety of mechanisms that lead to mosaicism however a broad classification of mosaic embryos is 1. Meiotic; 2. Post-zygotic; 3. Chaotic. Mosaicism provides the backdrop for an ongoing argument about the benefits preimplantation genetic screening (PGS) with opinions ranging from assertions that all IVF cases should include PGS to those who think PGS should not be performed at all. This talk will consider the biological and clinical implications of mosaicism in a field where gathering an evidence base for the efficacy of interventions presents unique challenges.