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Genome analyses of single human oocytes

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Abstract:

Single-cell genome analyses of human oocytes are important for meiosis research and preimplantation genomic screening. However, the nonuniformity of single-cell whole-genome amplification hindered its use. Here, we demonstrate genome analyses of single human oocytes using multiple annealing and looping-based amplification cycle (MALBAC)-based sequencing technology. By sequencing the triads of the first and second polar bodies (PB1 and PB2) and the oocyte pronuclei from same female egg donors, we phase the genomes of these donors with detected SNPs and determine the crossover maps of their oocytes. Our data exhibit an expected crossover interference and indicate a weak chromatid interference. Further, the genome of the oocyte pronucleus, including information regarding aneuploidy and SNPs in disease-associated alleles, can be accurately deduced from the genomes of PB1 and PB2. The MALBAC-based preimplantation genomic screening in in vitro fertilization (IVF) enables accurate and cost-effective selection of normal fertilized eggs for embryo transfer.

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