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LETTER TO THE EDITOR

Re-analysis of aneuploidy blastocysts with an inner cell mass and different regional trophectoderm cells

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Dear Editor

We read with interest the study by Huang et al., aiming to explore which part of the trophectoderm best represents the inner cell mass after array comparative genomic hybridization (aCGH) analysis [1]. Abnormal blastocysts diagnosed by aCGH were re-biopsied in four regions: in the inner cell mass (ICM) and in three trophectoderm (TE) sites. The biopsied pieces were processed through multiple annealing and looping-based amplification cycle sequenced for 24-chromosome aneuploidy screening.

Fifty of 51 (98.04%) ICM samples were concordant with at least one of the TE biopsies derived from the same embryos. There were 43 blastocysts in which ICM and the other three TE pieces were consistent. Discordance among the four pieces occurred in eight out of the 51 blastocysts (15.7%). One blastocyst was discordant between the ICM and the other three TE pieces, and seven blastocysts were discordant between one of TE and the other three biopsied pieces.

Previous studies of multiple TE biopsies have reported significant higher levels of false-positive diagnoses, up to 50% divergence between biopsies of the same embryos in the same laboratories and up to approximately 80% divergence between multiple biopsies in different laboratories [2–4]. Moreover, surprisingly, our recently published study, which used the same design as that of Huang et al., went unnoticed. We evaluated in eight embryos concordance of multiple TEBs and in four embryos concordance of TE and ICM biopsies. Discordant results (i.e., mosaicism) were observed in three out of eight embryos [5].

TE mosaicism may be present in at least half of all embryos. In addition, laboratory platforms used in assessing TE biopsies may offer different diagnostic sensitivities and specificities in detecting chromosomally abnormal cell lines. That is why the recently released practice guidelines for preimplantation genetic screening (PGS) 2.0 by the Preimplantation Genetic Diagnosis International Society (PGDIS) [6] have recommended to PGS laboratories, to use Next-Generation Sequencing (NGS), which is capable of measuring chromosomal copy numbers, as the only diagnostic platform used in assessing TE mosaicism. While in our study we used NGS [5], Huang et al. [1] did not. These different platforms may explain the rather low mosaicism rate in the study by Huang et al., as compared to that of others [2–5].

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