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Oral Presentation

Assessment of aneuploidy concordance between trophectoderm and inner cell mass in 100 blastocysts by next-generation sequencing

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Introduction

A key criticism of pre-implantation genetic testing for aneuploidy (PGT-A) is that a clinical trophectoderm (TE) biopsy might not correctly represent the entire blastocyst. To date, studies addressing the question of concordance between TE and inner cell masse (ICM) biopsies have relied on currently superseded technologies and/or small sample sizes.

Materials & methods

Here we analyze 100 blastocysts that were classified as aneuploid by PGT-A, probing the genetic make-up of their paired ICMs using high-resolution NextGen Sequencing (hr-NGS).

Results

When one or more whole chromosomes were aneuploid in the clinical TE biopsy, the corresponding ICM was aneuploid in 90 out of 93 blastocysts (96.8%). Nonetheless, when the clinical TE biopsy only contained segmental (sub-chromosomal) aneuploidies, the ICM was aneuploid in only 4 out of 7 cases (57.1%). Combined, and when all types of aneuploidy were

considered, an aneuploid TE biopsy correctly predicted aneuploidy in the ICM in 93 out of 100 cases. Of the 7 out of 100 blastocysts that were TE-ICM discordant, the ICM was mosaic in 2 samples and euploid in 5 samples. A second TE biopsy collected from the 7 TE-ICM discordant blastocysts showed concordance with the original clinical TE biopsy in only 2 out of 7 cases (28%).

Conclusions

A clinical TE biopsy containing aneuploidy is highly indicative of an aneuploid ICM, meaning that the blastocyst is indeed extremely unlikely to implant and lead to healthy pregnancy. Concomitantly, the results suggest clinical worth in re-evaluating blastocysts deemed ‘aneuploid’ in select cases by TE re-biopsy, particularly in instances of segmental aneuploidies.