

Topic: Preimplantation Genetic Screening

Title: TROPHECTODERM REBIOPSY RESULTS FROM EMBRYOS WITH INITIAL INCONCLUSIVE RESULTS USING 24-CHROMOSOME SINGLE NUCLEOTIDE POLYMORPHISM (SNP) MICROARRAY

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OBJECTIVE: Report 24-chromosome preimplantation genetic screening (PGS) results on Day 5/6 trophectoderm (TE) rebiopsy samples that had an initial inconclusive result due to uninformative SNP data.

DESIGN: Retrospective analysis.

MATERIALS AND METHODS: Patients were referred for PGS for reasons including prior failed in vitro fertilization (IVF) cycle, recurrent pregnancy loss, and/or advanced maternal age. Following IVF, embryos underwent blastomere or TE biopsy according to each clinic's standard procedures. Biopsy samples, along with parental blood or buccal samples, were shipped from IVF clinics to a single reference laboratory for analysis. Genotyping was performed using Illumina Cyto12 SNP microarray and informatics technique (Parental Support™). Embryo samples that returned results were classified as 'euploid' if no chromosome abnormalities were detected, or 'aneuploid' if monosomy, tri/polysomy, haploidy, triploidy, large deletions/duplications, and/or uniparental disomy was detected.

RESULTS: Of the initial 80,111 embryo samples submitted for PGS, 218 (0.3%), including two blastomere samples and 216 TE samples, had a No Call due to uninformative SNP data. Fifty-nine (27.1%) of these embryos underwent TE rebiopsy for reanalysis. Average maternal age for this patient cohort was 36 years (range 24–44). Rebiopsy results revealed that 34 (57.6%) samples were euploid, 15 (25.4%) were aneuploid, 6 (10.2%) were inconclusive due to insufficient DNA for analysis, and 4 (6.8%) were discordant, meaning the rebiopsied samples were not a genetic match to the original embryo sample.

CONCLUSIONS: Fifty-eight percent of Day 5/6 embryos that underwent rebiopsy of TE with an initial No Call due to uninformative SNP data were euploid. Although the causes of uninformative SNP data are not clear and may result from various factors (e.g., DNA degradation, unusual biology of the sample, sample contamination), these findings suggest that rebiopsy of embryos that received an initial No Call during PGS due to uninformative SNP data should be considered.