



REBIOPSY AND PREIMPLANTATION GENETIC SCREENING (PGS) REANALYSIS FOR EMBRYOS WITH AN INITIAL NON-DIAGNOSTIC RESULT YIELDS A EUPLOID RESULT IN THE MAJORITY OF CASES

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OBJECTIVE: After PGS, a small percentage of embryo biopsies do not yield a result suitable for a diagnosis. There are limited data regarding the efficacy of rebiopsying these embryos and whether they are more or less likely to be euploid. This study seeks to determine how often a diagnosis is obtained when embryos with previously non-diagnostic PGS results are thawed, re-biopsied and reanalyzed.

DESIGN: Retrospective cohort study of all PGS cases from a large IVF center referred to a single genetics laboratory.

MATERIALS AND METHODS: All embryos that underwent PGS from October 2009 until April 2017 were included. Trophectoderm biopsies were analyzed on one of three genetic screening platforms - SNP microarray, real-time PCR, or next generation sequencing (NGS), depending upon date of submission. Biopsies with non-diagnostic results - either nonconcurrent (results did not pass quality control measures due to noise/uninterpretable results) or unamplified (not enough genetic material available for evaluation) - were noted. Embryos that were subsequently rebiopsied for PGS reanalysis were further examined to classify results as euploid, abnormal or non-diagnostic.

RESULTS: Of the 44,543 embryos submitted for PGS, 1536 (3.4%) yielded nonconcurrent results and 164 (0.4%) yielded unamplified results. Rebiopsy for PGS reanalysis occurred in 364 (23.7%) cases and, after rebiopsies were analyzed, the diagnostic rate was 98.4% with the assignment of 241 (66.2%) euploid diagnoses. The likelihood of a euploid diagnosis following rebiopsy was not significantly different than the euploid rate for embryos that were assigned a diagnosis after initial biopsy (66.2 versus 61.8% respectively, $P=0.08$).

CONCLUSIONS: The rebiopsy and PGS reanalysis of embryos initially designated as non-diagnostic yields an interpretable result in the vast majority of cases and the embryos are just as likely to be euploid. As a result, serious consideration should be given to re-evaluation of undiagnosed embryos.