



EXPERIENCE WITH A TARGETED NEXT GENERATION SEQUENCING (TNGS) PLATFORM FOR COMPREHENSIVE CHROMOSOME SCREENING (CCS) ON OVER 16,000 EMBRYO BIOPSIES

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OBJECTIVE: There is a broad range in the reported frequencies of mosaic and segmental aneuploidies detected in embryo biopsies by NGS-based platforms. Thus, we sought to evaluate the overall test performance of a proprietary tNGS platform, including the frequency of reported abnormalities.

DESIGN: Retrospective analysis of clinical results.

MATERIALS AND METHODS: Between July 1, 2016 and May 1, 2017, 16,127 embryo biopsies were submitted for CCS testing by NGS from several referring IVF centers. The frequencies of each result category were calculated. Segmental abnormalities are classified as segments that are either mosaic or full range gains or losses that are less than an entire chromosome. Mosaic results are classified as full chromosome aneuploidy where the copy number is in between normal and abnormal. Nonconcurrent results were defined as either samples that didn't pass QC, were suspected of having contamination, or were suspected of being polyploid and a second biopsy was recommended. Unamplified results indicated DNA amplification failure. Biopsies from embryos where a parent carries a chromosome rearrangement were removed from this analysis.

RESULTS: Of the 16,127 embryo biopsies tested, 97.5% produced a conclusive result (15,725/16,127), while 1.5% (242/16,127) were nonconcurrent and 1% (160/16,127) were unamplified. Of the samples that yielded conclusive results, the overall euploid rate was 55.1% (8,666/15,725) and 24.9% (3,921/15,725) of the samples had at least one whole chromosome aneuploidy (without segmental aneuploidy or mosaicism). The frequency of samples with isolated mosaicism or segmental aneuploidy (and no whole chromosome aneuploidy) were 4.7% (733/15,725) and 7.5% (1,175/15,725), respectively. A small portion of samples (0.7%, 103/15,725) were positive for at least one mosaic range chromosome and a segmental abnormality on a separate chromosome, without any other aneuploidy. The remaining samples, 7.2% (1,127/15,725), were diagnosed as abnormal due to at least one whole chromosome abnormality, but also had mosaic range and/or segmental aneuploidies.

CONCLUSIONS: This study is designed to further define the frequency of reported abnormalities on tNGS-based aneuploidy screening within a large sample cohort. These frequencies can be used for counseling patients on their expectation of result outcomes when submitting samples for CCS testing via a tNGS platform