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PREVALENCE OF SPECIFIC CHROMOSOME-LEVEL ANOMALIES IN MOSAIC AND ANEUPLOID EMBRYOS

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OBJECTIVE:

We have previously demonstrated that the frequency of individual chromosomal copy number variations identified in human embryos cultured in vitro mirrors those found following early pregnancy loss.¹ It is unclear whether the same chromosomal abnormalities are identified in mosaic embryos. The aim of this study is to determine the frequency of individual chromosomal anomalies in aneuploid versus mosaic embryos.

MATERIALS AND METHODS:

This study included patients who underwent IVF with preimplantation genetic testing for aneuploidy (PGT-A) at a single academic center from January 2020 through March 2021. Trophectoderm biopsies were analyzed using a modified FAST-SeqS NGS-based PGT method and bioinformatics pipeline, which detects whole chromosome and segmental aneuploidies ($\geq 10\text{Mb}$), most types of polyploidy, and many instances of single chromosome uniparental isodisomy through amplification of L1 sites. The reported chromosome complement for each embryo was analyzed. The prevalence of abnormalities for each chromosome among aneuploid and mosaic embryos was determined and compared using chi-square. Bonferroni correction was used to adjust for multiple comparisons with a corrected p value of .002.

RESULTS:

A total of 7,872 embryos from 1,371 patients were included in the study with a mean age of 35.9 years, including 2,763 aneuploid embryos (35.1%) and 1,035 mosaic embryos (13.1%). The chromosomes with the greatest prevalence of abnormalities among aneuploid embryos were 21 (20.6%) and 16 (17.4%) followed by 21 (12.5%) and 15 (11.0%). The lowest prevalences were seen in chromosomes 17 (2.9%) and 12 (2.8%). In mosaic embryos, in contrast, the variation in prevalence of anomalies per chromosome was lower, ranging from 2.4% in both chromosomes 19 and 20 to 7.1% in chromosome 6 and 8.2% in chromosome 2. When comparing the prevalence of abnormalities for each chromosome among aneuploid and mosaic embryos, significant differences were seen in chromosomes 1 (3.7% vs. 6.0%, $p=.0015$), 2 (4.3% vs. 8.2%, $p<.0001$), 5 (3.9% vs. 6.6%, $p=.0004$), 6 (3.9% vs. 7.1%, $p<.0001$), 15 (11.0% vs. 3.4%, $p<.0001$), 16 (17.4% vs. 3.9%, $p<.0001$), 19 (6.0% vs. 2.4%, $p<.0001$), 20 (5.2% vs. 2.4%, $p=.0002$), 21 (12.5% vs. 3.6%, $p<.0001$), and 22 (20.6% vs. 5.7%, $p<.0001$).

CONCLUSIONS:

Specific chromosomes appear to have greater susceptibility to meiotic versus mitotic errors in the process of oocyte maturation and embryo development. These findings suggest that the differential mechanisms governing chromosome segregation during meiosis versus mitosis result in distinct differences in per chromosome copy number reports from embryo biopsies.

IMPACT STATEMENT:



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This study enriches our understanding of the frequencies and mechanisms of individual chromosomal anomalies, and enhances our ability to interpret pre-implantation genetic testing results for our patients.

REFERENCES:

Rodriguez-Purata J et al. Embryo selection versus natural selection: how do outcomes of comprehensive chromosome screening of blastocysts compare with the analysis of products of conception from early pregnancy loss (dilation and curettage) among an assisted reproductive technology population? *Fertil Steril*. 2015 Dec;104(6):1460-66.e1-12.