Objectives

Next generation sequencing (NGS) provides evidence of mosaicism in the blastocyst-stage embryo. Mosaic profiles are often graded as low or high to denote levels of risk. Mosaicism, as it pertains to specific chromosomes, was assessed to determine rates of high- and low-level mosaicism for individual chromosomes.

Design

Retrospective analysis of PGT-A derived mosaicism data in terms of rates per chromosome and levels of mosaicism.

Methods

A total of 6,525 samples were assessed during the time period of this study. Mosaicism rates were determined as a percentage of total samples, total mosaicism event and per chromosome. Additionally, the frequency of high- and low-level mosaics for each chromosome was evaluated.

Results

Of the 6,525 samples that underwent PGT-A testing, 931 (14%) displayed whole aneuploid mosaicism. High- and low-level mosaicism was observed in 47% and 53% of the samples respectively. Male and female samples showed autosomal mosaicism disproportionately at 44% and 56% respectively. Mosaicism in general, and specifically, high-level mosaicism in chromosome 22, occurred at a higher rate than other chromosomes, while mosaicism rates were lowest in chromosomes 12 and 17. Data are summarized in Table 1.

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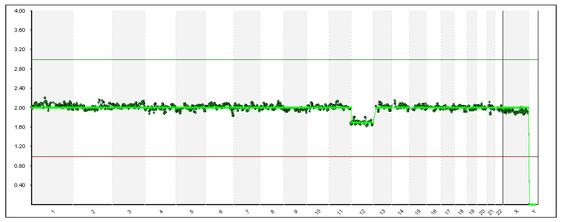
Individual Chromosome Mosaicism Rates After Preimplantation Genetic Testing for Aneuploidy: Implications for Mechanisms Related to the Early Stages of Embryo Development



Example of a Low Mosaic Result

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ASRM 2019
P-318

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Table 1: Mosaicism Rates

Ch #	Total Mosaic	%	High*	%	Low**	%	Female	%	Male	%
1	37	4%	15	41%	22	60%	19	51%	18	49%
2	40	4%	20	50%	20	50%	22	55%	18	45%
3	35	4%	18	51%	17	49%	22	63%	13	37%
4	59	6%	32	54%	27	46%	34	58%	25	42%
5	39	4%	21	54%	18	46%	21	54%	18	46%
6	40	4%	21	53%	19	48%	22	55%	18	45%
7	33	4%	7	21%	26	79%	19	58%	14	42%
8	49	5%	20	41%	29	59%	27	55%	22	45%
9	33	4%	8	24%	25	76%	20	61%	13	39%
10	39	4%	20	51%	19	49%	25	64%	14	36%
11	34	4%	15	44%	19	56%	23	68%	11	32%
12	21	2%	8	38%	13	62%	20	95%	1	5%
13	48	5%	20	42%	28	58%	25	52%	23	48%
14	36	4%	15	42%	21	58%	18	50%	18	50%
15	39	4%	19	49%	20	51%	23	59%	16	41%
16	37	4%	17	46%	20	54%	18	49%	19	51%
17	21	2%	10	48%	11	52%	6	29%	15	71%
18	47	5%	25	53%	21	45%	26	55%	21	45%
19	56	6%	33	59%	23	41%	25	45%	31	55%
20	30	3%	11	37%	19	63%	14	47%	16	53%
21	51	6%	26	51%	25	49%	29	57%	22	43%
22	68	7%	45	66%	23	34%	42	62%	26	38%
X	33	4%	10	30%	23	70%	30	91%	3	9%
Y	6	1%	1	17%	5	83%	0	0%	6	100%

*High Mosaic: A threshold of >50% but =<70%
**Low Mosaic: A threshold of >30% but =<50%

Conclusion

Bridging the gap between preimplantation genetics and prenatal cytogenetics has the potential to be a powerful tool for clinicians treating infertile couples. Currently, many couples view their IVF journeys' end in terms of being pregnant or not pregnant, while often the circumstances are much more complicated. The literature have reported that mosaicism is clinically relevant and clearly contributes to long-term pregnancy outcomes. This report evaluates the rates of mosaicism for individual chromosomes, providing a basis on which to correlate the incidence of preimplantation mosaicism in specific chromosomes, with mosaicism observed in prenatal samples. Additionally, the data highlight the putative uneven distribution of mosaicism in male and female samples.