



ONLY SEGMENTAL OR MOSAIC ANEUPLOID EMBRYOS AVAILABLE FOR TRANSFER: A RARE PHENOMENON IN PREIMPLANTATION GENETIC SCREENING (PGS) CYCLES USING TARGETED NEXT GENERATION SEQUENCING (NGS)

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OBJECTIVE: Use of NGS for PGS improves resolution for detecting segmental aneuploidy (SEG) and mosaicism (MOS). However, these additional categories create difficult management decisions when the only available embryo for transfer falls into one of these categories. With this in mind, this study sought to describe: 1) how often the only available embryo carries a SEG or MOS diagnosis, and 2) evaluate whether this risk is modified by age and the number of embryos tested in a cohort.

DESIGN: Retrospective descriptive

MATERIALS AND METHODS: All cycles at a single center utilizing targeted NGS based PGS between July 2016 and March 2017 were included in the analysis. Each embryo's PGS results was placed into 1 of 4 categories: 1) aneuploid (at least 1 whole chromosome aneuploidy present), 2) SEG (no whole chromosome aneuploidy, but deletion or duplication present), 3) MOS (no whole chromosome aneuploidy or segmental present, but mosaic range diagnosis present), and 4) euploid (no abnormalities). Only euploid, SEG, and MOS embryos were considered eligible for transfer. The percentage (%) of cycles in which the only eligible embryo for transfer carried a SEG or MOS diagnosis was then calculated for the entire study population, according to female age, and according to the number of embryos in a given cohort. The relationship between female age and each diagnostic category was also evaluated by logistic regression.

RESULTS: A total of 1364 cohorts resulting in 5876 embryos were evaluated. The overall prevalence of aneuploidy, SEG, and MOS per embryo were 32.5%, 7%, and 4% respectively. Female age was strongly associated with the presence of whole chromosome aneuploidy ($p < 0.001$). However, there was no association between female age and SEG ($p = 0.26$) or MOS ($p = 0.23$) results. The % of cycles in which the only available embryo contained a SEG or MOS was 4.9% across the entire cohort. The risk was similar across all ages and never exceeded 7% in any age group. The % of SEG only or MOS only cycles also decreased as number of embryos in the cohort increased and was $< 1\%$ when at least 4 embryos were available for testing.

CONCLUSIONS: Few PGS cycles analyzed by NGS result in scenario in which the only available embryo for transfer carries a SEG or MOS diagnosis. This low risk is consistent across all age groups as female age is not associated with an increased risk of SEG or MOS diagnosis. These findings are important in guiding pre-cycle PGS counseling and expectations.

Cohort outcome after targeted NGS based PGS according to age					
	Average number of embryos tested per cohort	At least 1 euploid available	Only segmental aneuploidy available	Only mosaic range available	All aneuploid
<35 (n=478)	6.4 (1.9 - 11.1)	94.7%	0.9%	2%	2.4%
35-37 (n=345)	4.5 (1 - 7.9)	88.2%	0.6%	1.4%	9.8%
38-40 (n=316)	3.8 (1.0 - 6.5)	75.7%	2.1%	3.1%	19.1%
41-42 (n=138)	2.5 (0.6 - 4.4)	42%	2.3%	3.7%	52%
>42 (n=87)	2 (0.7 - 3.3)	27.6%	2.3%	3.3%	66.6%