

Using Single Nucleotide Polymorphism (SNP) Analysis to Determine Embryo Ploidy Status

Claire Murphy Jones¹, Amy Jordan¹, Eugene Toh¹, Rebecca Brohammer², Yuri Wagner², Juergen Liebermann², Cengiz Cinnioglu¹

¹Luminary Genetics, Santa Clara, California, USA; ²Fertility Centers of Illinois, Chicago, IL, USA

Objective

- To determine the rate of haploidy and triploidy from embryos with normal fertilization check
- To better characterize how fertilization check and NGS-only PGT-A can miss haploid and triploid embryos

Materials and Methods

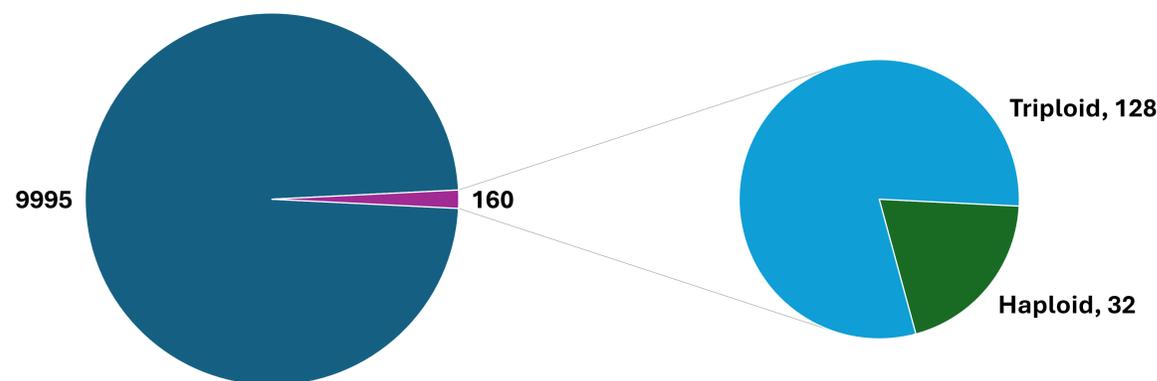
- PGT-A with SNP-NGS results were evaluated from a single IVF clinic from January 2024 through January 2026
- All embryo biopsies sent from this clinic were reported as 2PN via fertilization check
- Haploidy and triploidy rates via SNP-NGS were obtained
- Additional data from embryo samples labeled haploid or triploid XXX was collected:
 - Number of euploid embryos in the same cycle
 - Grading and day of biopsy of the embryos from that cycle

Results

Haploidy and Triploidy Rates

- 10,155 embryo samples originally identified as 2PN via fertilization check underwent PGT-A with SNP-NGS analysis
- 160 (1.58%) embryo samples were identified to have haploidy or triploidy
 - 32 (0.32%) haploidy
 - 128 (1.26%) triploidy
- Of these 160 embryo samples, 86 (53.75%) were haploid or triploid XXX
 - Of these 86 samples:
 - 26 (30.23%) were part of a cycle with no euploid embryos
 - 22 (25.58%) had euploid embryos in the same cycle, but had the same or better embryo grade and day of biopsy as the euploid embryos in the cycle

Ploidy Status of Embryos Identified as 2PN via Fertilization Check



Conclusions

- 1.58% (160/10,155) of embryo samples found to be haploid or triploid via SNP-NGS were 2PN at fertilization check
- 0.85% (86/10,155) of embryo samples would have been miscalled as euploid and options for transfer via NGS-only PGT-A; the use of SNP analysis identified them as haploid or triploid XXX
- 30.23% (26/86) of embryos would have been the only option for transfer from that patient's cycle and an additional 25.58% (22/86) would have been transferred first if compared by embryo grade and day of biopsy alone
- Therefore, 0.85% (86/10,155) of haploid and triploid embryos were at risk of being transferred, with 55.81% (48/86) of this subset at risk of being transferred first

Embryo Grade	Biopsy Day	Embryo Summary	Sex	Interpretation	Link™ (Cohort QC)
Blastocyst : XBBB	6	Aneuploid	XXX	Triploid	Match
Blastocyst : XBBB	7	Aneuploid	XX	Monosomy 16	Match
Blastocyst t:XBBB	7	Euploid	XX	Euploid	Match