

O-250 Wednesday, October 10, 2018 11:30 AM

SEGMENTAL ANEUPLOIDY IN BLASTOCYSTS: WHEN THE CHROMOSOMES BREAK.

M. Insua,^a M. Escriba,^a X. Vendrell,^b V. Peinado,^c T. Viloria.^d ^aIVI Valencia, Valencia, Spain; ^bReproductive Genetics Unit, Sistemas Genomicos, Paterna, Spain; ^cPGD Molecular Cytogenetics, IGENOMIX, Valencia, Spain; ^dIVF-Laboratory, IVIRMAValencia, Valencia, Spain.

OBJECTIVE: Our objective is to describe quantitatively and qualitatively segmental aneuploidies (SA) in trophoectoderm samples, defined as a loss or gain of a chromosomal fragment and its relationship with clinical and embryological parameters.

DESIGN: Clinical retrospective historical study

MATERIALS AND METHODS: 3628 blastocysts were studied of 844 cycles of PGT-A. Trophoectoderms were analyzed by NGS (next generation sequencing). SA was considered, if the lost / gained fragment measured was higher than 5 Mb. The diagnosed aneuploidies were classified as: complete chromosome, single segmentals (only a segmented chromosome, with or without complete chromosome aneuploidy) and pure segmentals (PSA, segmented chromosome unique without additional aneuploidy). We defined: prevalence, type, size, distribution and chromosomal topology (arm p or q) and its relation to: clinical indication, blastocyst stage and quality of the MCI and of the trophoectoderm.

RESULTS: 8.6% (314/3628) of blastocysts showed SA associated or not with complete chromosome aneuploidy; 7.9% (288/3628) exhibited unique SA, and 4.4% (161/3628) PSA. The incidence of PSA was not related to clinical or embryological parameters, except for the quality of the trophoectoderm. Chromosomes 19, 22, and Y did not exhibit PSA. PSAs were more frequent in the q arm of the metacentric and submetacentric chromosomes. Its size was greater in q than in p. The PSA/ chromosome ratio was constant. The PSA in q was greater than in p. The ratio PSA / arm was lower in arm q. The description of the PSAs only relates to intrachromosome topographic parameters

CONCLUSIONS: PSA is chromosome-dependent with clear topographic effect. In addition, it does not vary with maternal age, but it does vary with the morphology of the blastocyst, as a possible indicator of chromosomal instability in the trophoectoderm. Supported by: IVIRMA Private Grant.