

<sup>1</sup>Hunan Normal University School of Medicine- Changsha- 410013- China  
<sup>2</sup>National Engineering and Research Center of Human Stem Cell- Changsha-410078- China  
<sup>3</sup>Clinical Research Center for Reproduction and Genetics in Hunan Province- Reproductive and Genetic Hospital of CITIC-Xiangya- Changsha 410078- China  
<sup>4</sup>Laboratory of , ,

**Study question:** To analyze factors that could influence meiotic segregation patterns for reciprocal translocation carriers.

**Summary answer:** Involvement of an Acr-ch, female gender, and lower TARI (ratio of translocated segment I over the chromosome arm) were independent risk factors for alternate segregation.

**What is known already:** Reciprocal translocation is one of the more common structural rearrangements of chromosomes, which is associated with reproductive risks, such as infertility, spontaneous abortion and the delivery of babies with mental retardation or developmental delay. Extensive studies on meiotic segregation patterns of sperm, blastomere, and blastocysts have identified several factors that may influence the generation of unbalanced rearrangement of reciprocal translocations, including carrier's gender and age, location of breakpoints, chromosome type, and the quadrivalent structure. However, some results are controversial.

**Study design, size, duration:** A retrospective study from October 2013 to December 2019, a total of 10846 blastocysts originating from 2871 oocyte retrieval cycles from 2253 couples with one of the partners carrying reciprocal were investigated. The mean maternal age was 29.97±4 years (20–47years).

**Participants/materials, setting, methods:** Trophectoderm biopsy of blastocysts was performed on the 5th or 6th day of development. Whole genome amplification (WGA) was performed on all samples, and the WGA was analyzed with SNP array or NGS. Segregation patterns of quadrivalent in 10846 blastocysts were analyzed. Risk factors for segregation patterns were explored through analyzing carriers' demographic and cytogenetic characteristics using multivariate generalized linear mixed models (GLMMs).

**Main results and the role of chance:** The percentage of normal/balanced blastocysts was 34.3%, and 2:2 segregation was observed in 90.0% of blastocysts. Increased TARI (the ratio of translocated segment I over the chromosome arm) was noted as an independent protective factor for the proportion of alternate segregation ( $P=0.004$ ). The female gender and involvement of an Acr-ch were found independent risk factors for alternate segregation ( $P<0.001$ ). A higher TARI reduced the risk of adjacent-1 segregation; longer translocated segment and female gender increased the risk of adjacent-2 segregation ( $P=0.009$  and  $P<0.001$ , respectively). Female gender and involvement of an Acr-ch enhanced the risk of 3:1 segregation ( $P<0.001$  and  $P=0.012$ , respectively).

**Limitations, reasons for caution:** About 1400 blastocysts were not diagnosed in the 2871 cycles, which might cause bias in the results. Secondly, the interchromosomal effect of reciprocal translocations was not analyzed in this study.

**Wider implications of the findings:** In conclusion, a carrier's gender, involvement of an Acr-ch, and location of breakpoints may influence the segregation patterns. Besides, involvement of an Acr-ch, female gender, and lower TARI are independent risk factors for alternate segregation. These results may provide more appropriate genetic counseling for couples with balanced translocation.

**Trial registration number:** no

### O-206 Meiotic segregation analysis for reciprocal translocation carriers: Assessment of factors influencing meiotic segregation patterns

P. Xie<sup>1</sup>, H. Liang<sup>2</sup>, P. Yangqin<sup>3</sup>, T. Yueqiu<sup>2</sup>, L. Ge<sup>4</sup>

<sup>1</sup> Hunan Normal University School of Medicine- Changsha- 410013- China

<sup>2</sup>National Engineering and Research Center of Human Stem Cell- Changsha-410078- China, basic medicine, changsha, China ;

<sup>2</sup>National Engineering and Research Center of Human Stem Cell- Changsha-410078- China  
<sup>3</sup>Clinical Research Center for Reproduction and Genetics in Hunan Province- Reproductive and Genetic Hospital of CITIC-Xiangya- Changsha 410078- China  
<sup>4</sup>Laboratory of , , ;

<sup>3</sup>Clinical Research Center for Reproduction and Genetics in Hunan Province- Reproductive and Genetic Hospital of CITIC-Xiangya- Changsha 410078- China, genetic department, changsha, China ;