Polar body diagnosis by array comparative genomic hybridization reveals a distinct aneuploidy pattern in women of advanced maternal age

Advanced maternal age (AMA) is positively correlated with implantation failure or pregnancy loss, due to higher aneuploidy rate in the oocyte. Array comparative genomic hybridization (aCGH) based polar bodies diagnosis (PBD) offers a more comprehensive view on the oocyte’s chromosomal constitution. Do polar bodies (PB) from women with AMA show a discriminatory aneuploidy signature after aCGH based analysis? Do complex chromosomal aneuploidies occur more often in this cohort? Are the segregation errors distributed equally between meiosis I and meiosis II?

In two IVF-centers, both PBs were simultaneously biopsied and sent to the diagnostic laboratory. After whole genome amplification (WGA) of the PB DNA, the aCGH analysis was performed with 24sure arrays in 24 hours. Test results were analysed in terms of segregation error-frequency, -kind and meiotic origin.

Patients: In a retrospective cohort study PBs from 35 AMA patients (>=38 years, 39 IVF-cycles) were biopsied. The average maternal age of the patient cohort was 41.2 years. Of 434 first and second PBs, 143 (33%) were found to be euploid. We observed segregation errors of all chromosomes with chromosomes 15, 16, 19, 21 and 22 showing the highest error frequency. The frequency of errors occurring at meiosis I didn’t differ significantly from meiosis II errors except for chromosomes 16, 18 and 22: mainly losses of these chromosomes were detected in second polar bodies. Complex aneuploidy patterns were observed in both PBs with a weak correlation to maternal age.

aCGH based PBD identifies a variety of aneuploidies. The similar incidences of meiosis I and II errors imply that both PBs should be tested. PB results of this particular cohort showed: 1) aneuploidies of almost all chromosomes, 2) no numerical difference in the meiotic origin of the errors and 3) a trend towards more complex aneuploidies (aneuploidies>=3).