00243 Frequency of Cytogenetic Abnormalities in Products of Conception

Kee Su Keyau, Lim Soon Tiong Alvin, Tien Sim Leng
Singapore General Hospital

Aims: Miscarriages are common during pregnancy. 10-15% end in clinical miscarriages. 50-60% is due to chromosomal abnormalities. The aim of the study is to evaluate the frequency, spectrum of cytogenetic abnormalities in products of conception (POC) and their correlation with maternal age.

Methodology: A retrospective study of 357 POC samples from Jan 2000-March 2018 was performed at the Cytogenetics Laboratory, SGH. Specimens comprising maternal decidua (n=35) were excluded. The abnormality rate and abnormalities were compared between two age groups; <35 years and ⩾35 years. Chromosomal abnormalities were grouped according to autosomal trisomies, sex chromosome aneuploidies, polyploidies, unbalanced structural abnormalities, double abnormalities and mosaicisms.

Result: 88.2% of 322 POC samples were successfully karyotyped. 38 samples failed either because of non-viable tissues or contamination. The abnormality rate was 47.5% (135/284). Women ⩾35 years had a significantly increased abnormality rate (61%; 72/118) compared to women <35 years (38%; 63/166) ($p<0.001$). Autosomal trisomies (58.5%) were the most common aberration where trisomies 16, 21 and 22 were most frequent, followed by trisomies 15 and 13. Monosomy X, polyploidies and mosaicisms accounted for 10.4%, 9.6% and 13.3%, respectively. Women ⩾35 years had increased autosomal trisomy and double abnormality rates compared to women <35 years (70.8% vs. 44.4% and 5.6% vs. 0%, respectively). In contrast, women <35 years had a greater incidence of monosomy X, mosaicisms and polyploidies (17.5% vs. 4.2%, 19% vs 8.3%, and 12.7% vs 6.9%, respectively).

Conclusion: Cytogenetic abnormalities are a major cause of miscarriages so cytogenetic studies are recommended for all POC. Women ⩾35 years had significantly increased abnormalities compared with younger women ($p<0.001$). Fluorescence in situ hybridization (FISH) or cytogenomic microarray (CMA) analysis methods can act as important adjunct methods when there is tissue culture failure.