

IMPROVED MOLECULAR APPROACH FOR THE CHROMOSOMAL ANALYSIS OF PRODUCTS OF CONCEPTION.

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Abstract Body

Introduction: The analysis of products of conception (POC) is essential to determining the cause of a pregnancy loss and counselling couples in future pregnancies.

Objective: To present a molecular approach for chromosomal analysis of POC, including the possibility to detect triploidy and to identify maternal cell contamination (MCC).

Methodology:

Direct DNA extraction was performed from three different sections of each specimen and from maternal blood. Chromosome status was determined by CGH arrays (Illumina, San Diego, USA) or Next Generation sequencing (NGS) (Thermofisher, MA USA). STRs analysis of maternal and specimen DNA was added in the cases with euploid results to discard MCC and to identify triploidy (AmpFISTR Identifier Plus (Applied Biosystems, CA, USA).

Results: We analyzed 605 POC cases, with average maternal age of 36.5±4.3 years, and mean gestational age at miscarriage of 7.8±1.5 weeks. The percentage of POC samples with non-conclusive results after the first dissection was 30.0%, mostly due to MCC. After three dissections, we reduced non-informativity rate to 18.7%. We provided results (either normal or abnormal) in 81.3% of the cases. A statistically significant increase in detection rate was obtained with the multi-sampling strategy (p=0.0010, Fisher's exact test). We found 53.3% of abnormal POCs, with trisomies as the most common abnormality (79.9%).

Deletions/duplications (>5Mb) were found in 4.2% of the cases. Monosomy X was found in 11% of the cases and monosomy 21 in 1% of the cases.

Conclusions: Molecular methods such as aCGH and NGS are more reliable and do not require a previous cell culture, avoiding the possibility of cell culture failure and increasing the rate of informative results. Multisampling and STRs analysis increases the efficiency and reliability of chromosomal analysis in POCs.