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USE OF CGH-ARRAY TO DETECT CHROMOSOME ABNORMALITIES IN FIRST TRIMESTER MISCARRIAGE

ABSTRACT

Background and Objectives

Miscarriage affects 10 to 15% of all pregnancies, with over 80% occurring in the first trimester. Approximately 50% are caused by chromosomal abnormalities. Comparative Genomic Hybridisation-array (CGH-array) is a molecular technology that can be applied to products of conception collected following a miscarriage. Its use in the South African private health sector for investigation of an early pregnancy loss is relatively new.

This study aimed to quantify chromosomal abnormalities underlying first trimester miscarriages in the South African setting using CGH-array, as well as to demonstrate that CGH-array has a low test failure rate.

The objectives were to describe the characteristics of the study population, namely patients seeking infertility treatment in South Africa; to describe the results of the CGH-array analysis performed on products of conception; and to determine the rate of test failure.

Methods

This was a retrospective descriptive study conducted at Vitalab Centre for Assisted Conception, which is a private fertility clinic in Johannesburg, South Africa. The study population consisted of patients who had suffered either a sporadic or recurrent first trimester miscarriage from March 2014 to November 2016. Recurrent miscarriage was defined as the loss of three or more consecutive pregnancies. Chorionic villus samples were obtained under direct vision using a hysteroscope. The samples were analysed by Genesis Genetics, a private laboratory in Midrand, South Africa.

Results

There were 101 samples from 99 patients included in the study. The mean maternal age was 33.9 ± 4.8 years and the patients were predominantly white, employed and in heterosexual partnerships. 79% of the miscarriages were sporadic (the patient had experienced fewer than three consecutive pregnancy losses) and 71% of the pregnancies were conceived using assisted reproductive techniques.

The overall rate of chromosomal abnormalities was 55.4%. Autosomal trisomy accounted for 71.4% of these, followed by structural abnormalities (25%) and monosomy X (7.1%). Chromosomes 16 and 22 were the commonest trisomies, each compromising 19% of the autosomal trisomies. There were three cases of 48 XXY with Trisomy 19 which may represent a test artifact. There were no cases of polyploidy identified. The ratio of female to male euploid results was 2.2:1 and the test failure rate was 0%.

Conclusion

The detection rate of CGH-array in the South African setting is in keeping with international standards. Despite its known limitations and the potential artefacts identified in this study, it is a promising tool to investigate first trimester miscarriages in the local setting.