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Incidence of mosaicism in chromosomally abnormal human embryos detected by array-CGH.

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Introduction

Human embryo has many chromosome abnormalities which are aneuploidy such as trisomy and monosomy. Chromosomal abnormalities are took place during not only gametogenesis but also embryonic development. Mosaicism takes place when chromosome abnormalities occurred in embryonic development. The objective of the present study was to determine how much percentage of chromosomally abnormal embryos indicated by array-based comparative genomic hybridization (array-CGH) is mosaicism assessed by Fluorescence in situ hybridization (FISH) in order to offer precise diagnosis for PGD.

Materials and methods

Six good quality surplus chromosomally abnormal embryos vitrified on Day-3 were used after informed consent. Every blastomere from each embryo was analyzed by FISH to reveal mosaicism.

Result

Two of the six embryos were mosaic while four were not. Monosomy 5 embryo by a-CGH consisted of eight blastomeres. Seven of the eight had single signal and the other one had double signals by FISH. Trisomy 13 embryo by a-CGH consisted of nine blastomeres. Eight had triple signals, but the other one had double signals by FISH.

Conclusions

Mosaicism takes place one third (33%) of chromosomally abnormal Day 3 embryos. The data of the present study was lower than the other data reported. PGD of Day3 embryos should be reliable only if a-CGH is used from the present study.