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Validation of microarray comparative genomic hybridization for comprehensive chromosome analysis of embryos.

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Source

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Abstract

OBJECTIVE:

To validate and determine the best array-comparative genomic hybridization (aCGH; array-CGH) protocols for preimplantation genetic screening (PGS).

DESIGN:

Embryos had one cell removed as a biopsy specimen and analyzed by one of two array-CGH protocols. Abnormal embryos were reanalyzed by fluorescence in situ hybridization (FISH).

SETTING:

Reference laboratory.

PATIENT(S):

Patients donating embryos or undergoing PGS.

INTERVENTION(S):

Embryo biopsy, array-CGH, FISH reanalysis.

MAIN OUTCOME MEASURE(S):

Diagnosis, no result rate and error rate.

RESULT(S):

Method one produced 11.2% of embryos with no results and a 9.1% error rate compared with 3% and 1.9% for method two, respectively. Thereafter, only method two was used clinically. The aneuploidy rate for cleavage-stage embryos was 63.2%, significantly

increasing with maternal age. The chromosomes most involved in aneuploidy were 16, 22, 21, and 15. We report the first live births after array-CGH combined with single blastomere biopsy.

CONCLUSION(S):

Array-CGH is proved to be highly robust (2.9% no results) and specific (1.9% error rate) when applied to rapid (24-hour) analysis of single cells biopsied from cleavage-stage embryos. This comprehensive chromosome analysis technique is the first to be validated by reanalyzing the same embryos with another technique (e.g., FISH). Unlike some alternative techniques for comprehensive chromosome screening, array-CGH does not require prior testing of parental DNA and thus advance planning and careful scheduling are unnecessary.