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Perspective

Mosaic Embryos in Preimplantation Genetic Testing: Clinical Interpretation and Laboratory Considerations

Santiago Almeida*

Department of Human Genetics and Embryology, Universidade Federal do Litoral, Recife, Brazil

DESCRIPTION

Preimplantation genetic testing has become an integral component of modern assisted reproduction, allowing embryology laboratories to examine chromosomal constitution before embryo transfer. Among the findings reported through next-generation sequencing and related technologies, mosaicism has emerged as one of the most debated outcomes. Mosaic embryos contain two or more genetically distinct cell lines within the same embryo, typically involving a mixture of euploid and aneuploid cells. The interpretation of such findings presents clinical, biological, and ethical challenges that continue to shape reproductive decision-making.

Embryonic mosaicism arises during mitotic divisions after fertilization rather than from meiotic errors in gametes. A normal zygote may acquire chromosomal errors during early cleavage stages, leading to divergent cell populations. The proportion and distribution of abnormal cells vary widely, influencing developmental potential. Some embryos display low-level mosaicism with limited abnormal cells confined to the trophectoderm, while others demonstrate widespread chromosomal imbalance affecting both trophectoderm and inner cell mass.

The introduction of trophectoderm biopsy has improved survival rates of embryos during genetic testing compared with earlier cleavage-stage biopsy techniques. However, because only a small number of cells are sampled from the outer layer of the blastocyst, the possibility of sampling bias remains. A biopsy showing mosaic results may not fully represent the genetic composition of the entire embryo. This limitation has led to cautious interpretation by geneticists and clinicians when reporting mosaic findings.

Advances in sequencing technology have increased sensitivity for detecting low-level chromosomal variation. While this improvement allows identification of subtle abnormalities, it also raises concerns regarding overdiagnosis. Technical noise, amplification artifacts, and laboratory variability may contribute to classification of embryos as mosaic even when biological significance is uncertain. For this reason, many laboratories categorize mosaic results into low, moderate, and high ranges based on the proportion of abnormal reads, though thresholds vary internationally.

Clinical outcomes following transfer of mosaic embryos have been reported in multiple observational studies. In some cases, embryos classified as low-level mosaic have resulted in healthy live births without detectable chromosomal abnormalities. These outcomes suggest that self-correction mechanisms may exist during early embryonic development. Selective cell death, preferential proliferation of euploid cells, and restricted contribution of abnormal cells to the fetus have all been proposed as possible explanations. Nevertheless, not all mosaic embryos demonstrate developmental capacity, and implantation rates are generally lower compared with euploid embryos.

Different chromosomal abnormalities carry distinct developmental implications. Mosaicism involving chromosomes associated with viable aneuploidies may result in more favorable outcomes than those involving chromosomes critical for early embryonic development. For example, mosaic changes affecting certain autosomes may allow continued development, whereas complex or multiple chromosome abnormalities often correlate with poor implantation potential. However, predicting outcome based solely on chromosomal pattern remains unreliable.

Clinical decision-making regarding mosaic embryo transfer involves careful counseling of patients. Patients often face emotional and ethical difficulty when deciding whether to transfer embryos with uncertain potential. Some fertility programs prioritize euploid embryos exclusively, while others consider transfer of selected mosaic embryos when no euploid embryos are available. Counseling must include discussion of possible implantation failure, miscarriage risk, and the theoretical possibility of chromosomal abnormalities in offspring, even though many reported cases have resulted in healthy newborns. Postnatal follow-up studies of infants born after mosaic embryo transfer remain limited but generally reassuring. Available data indicate that most children demonstrate normal developmental milestones and no detectable chromosomal abnormalities when tested using standard diagnostic methods. However, long-term surveillance

Correspondence to: Santiago Almeida, Department of Human Genetics and Embryology, Universidade Federal do Litoral, Recife, Brazil, E-mail: s.almeida.ivfgenetics@medmailglobal.org

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remains necessary to fully assess health outcomes, particularly because subtle effects may not be immediately apparent at birth.

Laboratory techniques used for embryo biopsy and analysis also influence mosaic detection rates. Variations in whole-genome amplification methods, sequencing depth, and bioinformatic algorithms can produce differences in classification outcomes between laboratories. Standardization efforts have been proposed to reduce variability and improve consistency in reporting. Quality control procedures, including validation of analytical pipelines and external proficiency testing, are increasingly emphasized within reproductive genetics laboratories. Embryo culture conditions may also contribute to the appearance of mosaicism. Some researchers suggest that *in vitro* conditions might influence mitotic error rates during early cleavage stages. Temperature fluctuations, oxidative stress, and suboptimal culture media composition have been considered potential contributors. However, distinguishing between culture-induced artifacts and intrinsic embryonic abnormalities remains difficult.

CONCLUSION

The biological significance of mosaicism also intersects with developmental cell competition. Early embryos possess mechanisms that may eliminate abnormal cells through apoptosis or selective exclusion from the inner cell mass. This phenomenon may explain why some mosaic embryos correct themselves during development. The efficiency of such correction likely depends on timing, degree of mosaicism, and chromosomal involvement. Mosaic embryo research continues to reshape understanding of early human development. Rather than representing a simple abnormality, mosaicism reflects the dynamic nature of embryonic cell division and genetic regulation. Continued investigation into its biological behavior, clinical outcomes, and laboratory detection methods will contribute to more informed reproductive decision-making and improved patient care in assisted reproductive medicine.