Journal of Fertilization: In Vitro - IVF-Worldwide, Reproductive Medicine, Genetics & Stem Cell Biology

OPEN ACCESS Freely available online

Perspective

Chromosomal Mosaicism Patterns in Trophectoderm Biopsies and their Association with Blastocyst Expansion Rates

Helena Novak*

Department of Human Cytogenetics, Central European Medical University, Prague, Czech Republic

DESCRIPTION

Preimplantation genetic testing has become an important component of assisted reproductive treatment for couples affected by recurrent pregnancy loss, repeated implantation failure, advanced maternal age, or inherited genetic disorders. During this procedure, a small number of cells are removed from the trophectoderm layer of blastocysts and analyzed for chromosomal abnormalities before embryo transfer. Advances in next-generation sequencing technology have improved detection sensitivity, leading to increased identification of chromosomal mosaicism within human embryos. Mosaic embryos contain both euploid and aneuploid cell populations, creating significant discussion regarding embryo viability and transfer decisions in fertility practice.

Blastocyst development involves rapid cellular division and differentiation during the first several days after fertilization. The trophectoderm eventually contributes to placental formation, while the inner cell mass develops into fetal tissue. During these early mitotic divisions, errors in chromosome segregation may occur, resulting in mosaic cell populations. Unlike meiotic errors arising during gamete formation, mitotic abnormalities develop after fertilization and can affect varying proportions of embryonic cells. The biological significance of these abnormalities remains incompletely understood because some embryos appear capable of self-correction during later development.

Embryologists frequently evaluate blastocyst expansion as an indicator of developmental progression before biopsy and cryopreservation. Expansion reflects fluid accumulation within the blastocoel cavity and coordinated cellular activity during trophoblast differentiation. Fully expanded blastocysts often demonstrate higher implantation potential than slowly developing embryos. Researchers have therefore examined whether blastocyst expansion rates correlate with chromosomal stability and mosaicism frequency.

Several investigations have reported increased rates of chromosomal abnormalities among delayed or poorly expanded blastocysts. Embryos reaching full expansion on day five after fertilization generally display higher euploidy rates than embryos requiring day six or day seven culture. However, developmental timing alone does not reliably predict chromosomal status because many slower embryos still produce healthy pregnancies after transfer. Mosaic embryos in particular present a challenge because their developmental potential varies according to the proportion and type of abnormal cells present.

Next-generation sequencing has enabled more sensitive detection of intermediate chromosomal copy number patterns interpreted as mosaicism. Earlier genetic screening methods often classified embryos simply as normal or abnormal, potentially overlooking mixed cellular populations. Current sequencing platforms can identify partial chromosomal gains or losses involving twenty to eighty percent of sampled cells. While this increased sensitivity provides additional information, it has also complicated clinical interpretation because mosaic results do not always correspond with poor reproductive outcome.

The relationship between mosaicism and blastocyst expansion may reflect underlying cellular stress during embryogenesis. Chromosomal imbalance can disrupt cell cycle regulation, metabolic activity, and apoptosis pathways. Embryos containing high proportions of aneuploid cells may therefore display slower cavitation and impaired trophoblast growth. Conversely, embryos with limited mosaic involvement may continue developing relatively normally if euploid cell populations remain dominant.

Maternal age strongly influences chromosomal abnormality rates in preimplantation embryos. Oocytes from older women exhibit increased susceptibility to meiotic nondisjunction, spindle instability, and mitochondrial dysfunction. Although meiotic errors differ biologically from postzygotic mosaicism, age-related cellular stress may contribute indirectly to mitotic instability during cleavage-stage development. Studies have reported higher overall mosaicism frequencies among embryos derived from advanced maternal age cycles, though findings remain inconsistent across laboratories.

Laboratory culture conditions may also influence mitotic stability during embryogenesis. Temperature fluctuations,

Correspondence to: Helena Novak, Department of Human Cytogenetics, Central European Medical University, Prague, Czech Republic, E-mail: giulia. helena.novak.cemu@genetixmail.org

Received: 01-Sep-2025, Manuscript No. JFIV-25-41836; **Editor assigned:** 03-Sep-2025, PreQC No. JFIV-25-41836 (PQ); **Reviewed:** 17-Sep-2025, QC No. JFIV-25-41836; **Revised:** 24-Sep-2025, Manuscript No. JFIV-25-41836 (R); **Published:** 01-Oct-2025, DOI: 10.35841/2375-4508.25.13.424

Citation: Novak H (2025). Chromosomal Mosaicism Patterns in Trophectoderm Biopsies and their Association with Blastocyst Expansion Rates. J Fertil *In Vitro* IVF World w Reprod Med Gent Stem Cell Biol. 13:424.

oxidative stress, pH imbalance, and culture media composition can affect spindle formation and chromosomal segregation during early cleavage stages. Time-lapse incubation systems have allowed embryologists to monitor developmental kinetics continuously without repeated embryo handling. Some investigations suggest that abnormal cleavage patterns, multinucleation, or delayed cytokinesis correlate with higher mosaicism risk in resulting blastocysts.

Interpretation of trophectoderm biopsy findings remains limited by sampling constraints. Only a small number of cells are removed from the outer embryonic layer, meaning biopsy results may not fully represent the chromosomal composition of the entire embryo. Mosaicism may vary substantially between different embryonic regions. Some embryos classified as mosaic through trophectoderm biopsy may contain predominantly euploid inner cell mass tissue capable of normal fetal development. Conversely, apparently euploid biopsies may occasionally overlook localized abnormal cell populations elsewhere in the embryo.

CONCLUSION

Current evidence suggests that chromosomal mosaicism represents a relatively common feature of human preimplantation embryos, particularly during blastocyst development in assisted reproductive cycles. Blastocyst expansion rates appear associated with chromosomal stability to some extent, though the relationship remains biologically complex and clinically variable. Continued research involving embryology, genetics, developmental biology, and bioinformatics will remain important for refining interpretation of trophectoderm biopsy findings and improving reproductive decision-making in assisted conception programs.