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Ethical and Clinical Considerations in the Transfer of High-Level Mosaic Embryos: A Case Study and Framework for Informed Consent

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Abstract

Objective: The transfer of high-level mosaic embryos remains controversial due to concerns regarding embryo viability, pregnancy outcomes, and potential fetal abnormalities. However, emerging evidence suggests that selected mosaic embryos may result in healthy live births, creating ethical and clinical challenges for fertility clinics and patients.

Case presentation: A 43-year-old woman undergoing publicly funded In Vitro Fertilization (IVF) with Intracytoplasmic Sperm Injection (ICSI) produced three blastocysts. Preimplantation Genetic Testing for Aneuploidy (PGT-A) identified two aneuploid embryos and one high-level mosaic embryo demonstrating mosaic trisomy 2 and partial trisomy 20pter-p11.23. Despite institutional policy against the transfer of high-level mosaic embryos, the patient and her partner requested transfer after receiving comprehensive counselling. A multidisciplinary process involving genetic counselling, ethics review, legal consultation, psychosocial support, and development of a customized informed consent document was undertaken.

Results: Following completion of the informed consent process, the patient underwent Frozen Embryo Transfer (FET) and achieved pregnancy. Prenatal evaluation, including Non-Invasive Prenatal Testing (NIPT), detailed ultrasound assessment, and amniocentesis, demonstrated a chromosomally normal female fetus. The pregnancy progressed without major complications, resulting in the birth of a healthy infant who demonstrated normal development at three months of age.

Conclusion: This case highlights the importance of individualized decision-making and multidisciplinary informed consent when considering the transfer of high-level mosaic embryos. As evidence regarding mosaic embryo outcomes continues to evolve, fertility clinics may benefit from flexible frameworks that balance patient autonomy with clinical responsibility and ethical oversight.

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Introduction

Preimplantation Genetic Testing for Aneuploidy (PGT-A) has significantly enhanced embryo selection during in vitro fertilization by identifying chromosomal abnormalities before embryo transfer. While PGT-A has improved embryo selection strategies, the identification of embryos classified as mosaic—containing both euploid and aneuploid cell populations—has introduced substantial clinical and ethical uncertainty.

Historically, many fertility clinics have discouraged or prohibited the transfer of high-level mosaic embryos because of concerns regarding implantation failure, miscarriage, congenital anomalies, and uncertain developmental outcomes. However, accumulating evidence suggests that some mosaic embryos retain reproductive potential and can result in healthy live births. Furthermore, limitations in current PGT-A technology may contribute to false-positive diagnoses and imperfect prediction of embryonic developmental potential.

As the scientific understanding of embryo mosaicism continues to evolve, clinicians must navigate the complex balance between patient autonomy, institutional policies, and ethical responsibility. This case report describes the transfer of a high-level mosaic embryo following an extensive multidisciplinary informed consent process and proposes a framework for clinical decision-making in similar cases.

Case presentation

A 43-year-old woman undergoing publicly funded IVF with ICSI produced three blastocysts suitable for PGT-A analysis. Next-generation sequencing identified two aneuploid embryos and one high-level mosaic embryo demonstrating mosaic trisomy 2 and partial trisomy 20pter-p11.23.

Because the clinic maintained a policy against transferring high-level mosaic embryos, the case required individualized review. Despite counselling regarding potential risks and uncertainties, the patient and her partner expressed a strong desire to proceed with the transfer of the mosaic embryo.

The case was reviewed by the clinic's ethics committee and subsequently approved for consideration under exceptional circumstances. Written informed consent was obtained from the patient for both treatment and publication of this case report.

Multidisciplinary informed consent process

Genetic counseling

The patient underwent extensive counselling with a certified genetic counsellor. Discussions focused on the biological basis of embryo mosaicism, limitations of PGT-A testing, potential diagnostic inaccuracies, and the range of possible reproductive outcomes. Potential outcomes reviewed included failed implantation, miscarriage, congenital abnormalities, chromosomal disorders, and healthy live birth.

Particular emphasis was placed on the uncertainty associated with mosaic embryo transfer and the inability of current testing technologies to predict fetal outcomes with complete accuracy.

Ethics review

Given the proposed deviation from institutional policy, the case underwent formal ethics review. The committee evaluated patient autonomy, reproductive rights, potential risks to the future child, and the broader implications of permitting transfer of high-level mosaic embryos.

The committee concluded that proceeding with treatment was ethically permissible provided that comprehensive counselling was documented and the patient demonstrated a clear understanding of the associated risks and uncertainties.

Legal consultation

A reproductive law specialist reviewed the case to ensure compliance with applicable regulations and standards of care. Legal consultation focused on documentation of informed decision-making, provider responsibilities, and liability considerations.

Recommendations from the legal review informed the development of a customized informed consent document specific to the transfer of a high-level mosaic embryo.

Customized informed consent

A detailed consent document was developed outlining:

- The embryo's specific genetic findings.
- Current evidence regarding outcomes associated with mosaic embryo transfer.
- Limitations of PGT-A technology.
- Potential maternal, fetal, and neonatal risks.
- Available alternatives include additional IVF cycles, donor gametes, donor embryos, adoption, or discontinuation of fertility treatment.

The consent document explicitly stated that the clinic does not routinely recommend transfer of abnormal embryos because of increased risks, including miscarriage, stillbirth, congenital abnormalities, severe lifelong disability, maternal complications, and neonatal mortality.

Limitations of prenatal screening and diagnostic procedures, including amniocentesis, were also reviewed. The patient and her partner acknowledged understanding these risks and confirmed that their decision to proceed was voluntary and informed. The final document was reviewed and signed by the patient, her partner, and the attending physician.

Psychosocial support

Recognizing the emotional complexity of the decision, psychological counselling was offered to assist the patient in processing uncertainty, managing expectations, and ensuring that the decision was made free from coercion or misunderstanding.

Clinical outcome

Following completion of the multidisciplinary review and consent process, the patient underwent frozen embryo transfer. Pregnancy was achieved following the transfer of the high-level mosaic embryo. Prenatal evaluation included NIPT, serial ultrasonography, and diagnostic amniocentesis. All investigations demonstrated normal fetal development and no evidence of chromosomal abnormalities.

The pregnancy progressed without major complications, resulting in the delivery of a healthy female infant. Developmental assessment at three months of age was reported as normal.

Discussion

This case highlights the complex ethical and clinical considerations surrounding the transfer of high-level mosaic embryos. Although institutional policies are designed to promote patient safety and standardize care, rigid exclusion of all mosaic embryos may not fully reflect the current scientific literature.

Several studies have demonstrated that mosaic embryos retain developmental potential and may result in successful pregnancies and healthy live births. Greco and colleagues reported successful live births following transfer of mosaic blastocysts, challenging the assumption that all mosaic embryos are nonviable. Additionally, limitations in PGT-A technology may contribute to false-positive mosaic diagnoses, further complicating clinical decision-making.

The successful outcome observed in this case does not eliminate the risks associated with mosaic embryo transfer; however, it demonstrates that favourable outcomes remain possible when patients are appropriately selected and thoroughly counselled.

The informed consent process served as the cornerstone of ethical decision-making in this case. Through collaboration among genetics, ethics, legal, medical, and psychological professionals, the patient was empowered to make an informed decision while ensuring that clinicians fulfilled their ethical obligations.

Proposed framework for informed consent

Based on this case and existing professional guidance, the following framework may assist clinics considering the transfer of mosaic embryos:

1. Comprehensive education regarding mosaicism, PGT-A limitations, and available evidence.
2. Shared decision-making that respects patient values and reproductive goals.
3. Multidisciplinary review involving genetics, ethics, legal, and psychosocial experts when appropriate.
4. Detailed documentation of risks, uncertainties, alternatives, and patient understanding.
5. Ongoing prenatal and postnatal follow-up when pregnancy is achieved.

Conclusion

As assisted reproductive technologies continue to evolve, clinical and ethical frameworks must evolve alongside them. This case demonstrates that transfer of a high-level mosaic embryo can be ethically considered within a structured multidisciplinary framework that prioritizes informed consent, patient autonomy, and careful clinical oversight.

Future research should focus on improving the interpretation of mosaic PGT-A findings, refining embryo selection algorithms, and establishing evidence-based guidelines that support individualized patient care while maintaining the highest standards of safety and ethical practice.

Author declarations

Ethics approval and patient consent

The case was reviewed and approved by the clinic's ethics committee. Written informed consent was obtained from the patient for treatment and publication of this case report.

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Conflict of interest

The authors declare no conflicts of interest.

Author contributions

All authors contributed to the conception, preparation, review, and approval of the final manuscript.

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