



## Integrating innovation and insight in reproductive genetics and genomics: the essential role of advanced counseling

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### INTRODUCTION

The rapid evolution of reproductive genetics and genomics, as outlined in the call for this Special Issue, presents both remarkable opportunities and formidable challenges. From novel sequencing technologies to the ethical dimensions of embryo selection, the field is advancing at a pace that demands parallel progress in how we interpret, communicate, and apply this knowledge in clinical care<sup>[1]</sup>. In this perspective, we argue that the future of reproductive medicine hinges on a synergistic advancement in two domains: genomic technology and genomic counseling. Each subtopic highlighted in this issue not only represents a scientific frontier but also a counseling imperative.

### NOVEL TECHNOLOGIES AND THE INTERPRETATION IMPERATIVE

Long-read sequencing and optical genome mapping are revolutionizing our ability to detect structural variants and complex rearrangements relevant to infertility and prenatal diagnoses. However, these technologies generate data of unprecedented complexity. The role of genetic and genomic counseling must evolve from explaining the reproductive risk of a single-gene result to facilitating understanding of genomic architecture, including a combination of variants of various degrees of reproductive risk<sup>[2]</sup>.

### PREIMPLANTATION GENETIC TESTING: BEYOND THE TEST RESULT

The expansion of preimplantation genetic testing (PGT) for aneuploidy (PGT-A), monogenic disorders (PGT-M), and structural rearrangements (PGT-SR), along with the emerging promise of non-invasive PGT (niPGT), requires nuanced counseling frameworks. Counselors must guide couples through the probabilistic nature of PGT-A, the family-specific logistics of PGT-M, the complexities of translocation risk, and the validation nuances of niPGT. This demands a shift from simple test facilitation to integrated reproductive planning, weighing laboratory, clinical, and



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personal values<sup>[3]</sup>.

### **REPRODUCTIVE CARRIER SCREENING: FROM PANELS TO POPULATIONS**

As carrier screening expands from targeted ethnicity-based panels to comprehensive genomic screening, counseling faces dual challenges: communicating residual risk effectively and navigating incidental findings. The implementation challenge is not merely logistical but deeply educational, requiring counselors to develop strategies for pre-test education and post-test decision-support that align with diverse patient values and family goals<sup>[4]</sup>.

### **INFERTILITY & PREGNANCY LOSS: FROM PHENOTYPE TO MECHANISM**

The genomic dissection of male and female infertility and the etiology of recurrent pregnancy loss (RPL) and stillbirth is transforming idiopathic conditions into multi-omics explainable and predictable diagnoses. For counselors, this means moving beyond syndromic genetics to interpreting oligogenic, epigenetic, and multifactorial contributions. Counseling must provide a bridge between molecular diagnosis and clinical reality, helping patients understand what a genetic finding means for treatment options, prognosis, and future conceptions<sup>[5]</sup>.

### **EMBRYONIC DEVELOPMENT: COUNSELING IN THE EARLIEST STAGES**

Research into the genetic control of early human embryogenesis directly informs PGT and the understanding of early pregnancy failure. Counselors must be prepared to discuss the implications of embryonic mosaicism, developmental potential, and the limits of current testing - conversations that sit at the intersection of cutting-edge science, clinical uncertainty, and profound personal hope.

### **VARIANT INTERPRETATION AND VALIDITY: THE CORE OF TRUST**

The clinical interpretation of genetic variants and gene-disease validity is the bedrock of ethical reproductive genetics. Counselors are frontline interpreters, tasked with explaining variant of uncertain significance (VUS), conflicting classifications, and the evolving nature of evidence. This requires continuous education and close collaboration with clinical laboratory geneticists to ensure counseling accuracy and contextual relevance in fast-moving areas such as haploinsufficiency and imprinting disorders<sup>[6]</sup>.

### **GUIDELINES AND CONSENSUS: TRANSLATING EVIDENCE INTO PRACTICE**

The development and application of expert consensus and professional guidelines are critical for standardizing care. Genetic counselors are both implementers and contributors to these frameworks, ensuring they are practical, patient-centered, and adaptable to real-world clinical diversity. Their role is essential in operationalizing guidelines into actionable counseling protocols.

### **UTILITY OF AI IN REPRODUCTIVE GENETICS AND GENOMICS**

Artificial intelligence (AI) is poised to address two of the most pressing challenges in reproductive genetic and genomic counseling: the need to scale workforce capacity and the imperative to equip counselors with the skills to navigate increasingly complex genomic data. In the realm of education and training, AI-powered platforms offer scalable, interactive learning environments that can simulate diverse reproductive counseling scenarios. These tools can provide trainees with immediate feedback, adapt to individual learning curves, and expose them to a broader range of clinical cases than traditional training alone allows, thereby accelerating competency development and fostering clinical confidence. Beyond training, AI agents - including virtual

assistants and chatbot interfaces - hold promise for expanding access to high-quality reproductive genetic and genomic counseling, particularly in underserved or remote regions. When thoughtfully integrated, these digital tools can handle pre-test education, collect family history, and provide post-test informational support, freeing human counselors to focus on the nuanced psychosocial and decision-making aspects of care. However, AI should be positioned not as a replacement for human expertise but as a force multiplier - enhancing efficiency, extending reach, and enabling counselors to devote their specialized skills to the complex, emotionally nuanced conversations that define the heart of reproductive genetic and genomic counseling.

## **ETHICAL, LEGAL, AND SOCIAL IMPLICATIONS: THE COUNSELING VANGUARD**

Finally, the Ethical, Legal, and Social Implications (ELSI) of emerging reproductive technologies cannot be an afterthought. Counselors are often the first to encounter dilemmas regarding embryo selection, disclosure of adult-onset conditions in PGT, equity in access to technology, and cross-border reproductive care. Consider, for instance, the use of polygenic risk scores for complex conditions in embryo selection - a practice that raises fundamental questions about the goals of reproductive medicine, the definition of serious disease, and the potential for eugenic overtones. Similarly, the expansion of non-invasive prenatal testing has created situations where women receive high-probability results for conditions they did not explicitly consent to be screened for, challenging traditional informed consent frameworks. In the realm of direct-to-consumer genetic testing, individuals increasingly arrive at counseling with preconceived interpretations of their reproductive risks, requiring counselors to address both clinical accuracy and the psychological impact of unsolicited findings.

Yet relying on individual counselors to resolve these dilemmas in real time is neither sustainable nor equitable. What is needed is proactive ELSI infrastructure: multidisciplinary ethics committees that include genetic counselors, clear institutional policies that anticipate technological advances, and professional guidelines that provide consistent ethical guidance. Such infrastructure would not replace counselor judgment but would support it, ensuring that ethical decision-making is systematic, documented, and aligned with best practices. Furthermore, training programs must incorporate ethics education that goes beyond theoretical principles to include practical skills in ethical deliberation, conflict resolution, and policy advocacy. By embedding ELSI considerations into the fabric of clinical practice and professional development, the field can move from reactive problem-solving to proactive stewardship of these powerful technologies.

## **CONCLUSION**

The subtopics of this Special Issue collectively map the expanding universe of reproductive genetics. In each domain, the genetic counselor serves as an indispensable integrator - translating complex data into comprehensible insight, aligning technological possibility with personal values, and advocating for ethical rigor alongside scientific progress. As the field advances, so too must our investment in counseling education, research, and practice, ensuring that every genomic breakthrough is matched by the human wisdom necessary to wield it well. The future of reproductive health depends not only on what we can discover but equally on how we choose to care.

## **DECLARATIONS**

### **Authors' contributions**

Drafted and revised the manuscript: He Y

Conceptualized and revised the manuscript: Shen Y

Both authors approved the final version for submission.

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Not applicable.

### AI and AI-assisted tools statement

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### Conflicts of Interest

Shen Y is the founder of Synergene (Jiangxi) Education Co., Ltd. He Y is employed by Synergene (Jiangxi) Education Co., Ltd. Shen Y serves as a Section Editor of the *Journal of Translational Genetics and Genomics*. He was not involved in any aspect of the editorial process for this manuscript, including reviewer selection, manuscript handling, or editorial decision-making.

### Ethical approval and consent to participate

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### Consent for publication

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## REFERENCES

1. An Y, Shen Y, Ma Y, Wang H. Research needs for birth defect prevention and control in China in the genomic screening era. *BMJ*. 2024;386:e078637. DOI
2. Moustakli E, Christopoulos P, Potiris A, et al. Long-read sequencing and structural variant detection: unlocking the hidden genome in rare genetic disorders. *Diagnostics*. 2025;15:1803. DOI
3. Fernandes SLE, de Carvalho FAG. Preimplantation genetic testing: a narrative review. *Porto Biomed J*. 2024;9(4):262. DOI
4. Clinical Appropriateness Guidelines. Genetic testing. Appropriate use criteria: carrier screening in the reproductive setting. GEN05-0925.2. Available from: <https://guidelines.carelonmedicalbenefitsmanagement.com/wp-content/uploads/2025/05/Carrier-Screening-in-the-Reproductive-Setting-2025-09-20.pdf?utm> [Last accessed on 23 Apr 2026].
5. Kirovov Z, Gyokova EH, Hinkova NH, Konova EI. Recurrent pregnancy loss: etiology, pathophysiology, diagnosis and treatment. *J Biomed Clin Res*. 2025;18:1-10. DOI
6. Han C, Zhang Y, Xue J, Liu Y, An Y, Li H. Pregnancy outcomes and short-term follow-up of fetuses with recurrent microdeletion-microduplication syndromes featuring variable penetrance in prenatal diagnosis. *J Transl Genet Genom*. 2025;9:114-29. DOI

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