Qamre Alam et al. JBC Genetics. 2025;8(2):092-094
DOI: 10.24911/JBCGenetics.11-2356

SHORT COMMUNICATION

IVF empowered by NGS: redefining reproductive medicine through PGT-A and PGT-M

Qamre Alam[1*], Mohamed Ali Ateyah[1], Mohamed Hani Ali[1], Kadhem Alkhenaizi[1]

Introduction

Medicine has always strived to cure disease. However, when it comes to genetic disorders, the truth is sobering: 95% of these conditions remain without a cure, and only a handful, perhaps 5%, benefit from licensed therapies (1). For families at risk, this means facing heartbreaking choices, limited options, and lifelong burdens. But what if the best way to address these disorders is not to treat them at all, but to prevent them from arising in the first place?

This is precisely what Preimplantation Genetic Testing (PGT) has made possible. By examining embryos before implantation during *in vitro* fertilization (IVF), PGT empowers couples and clinicians to make decisions that were unthinkable a generation ago. With PGT for Aneuploidy (PGT-A), embryos with chromosomal abnormalities can be identified, sparing families the anguish of miscarriage or the birth of a child with severe chromosomal syndromes (2). With PGT for Monogenic Disorders (PGT-M), embryos carrying single-gene diseases, such as sickle cell disease, can be avoided altogether (3).

At the Molecular Genomics and Precision Medicine Department, ExpressMed Diagnostics and Research, Bahrain, we have witnessed this transformation firsthand. To date, we have conducted nearly 700 PGT-A cases and 25 PGT-M cases, the majority of the latter for families affected by sickle cell disease, a significant health burden in our region.

Our own analysis has highlighted the striking impact of maternal age on chromosomal abnormalities. Among women aged 25-30 years, the incidence of embryo aneuploidy was already as high as 60%. Alarmingly, in women over 40 years, this figure rose to nearly 90%. These findings reinforce a truth long known in reproductive medicine: advancing maternal age is the single most important factor driving chromosomal errors, failed implantation, and miscarriage. PGT-A therefore provides a powerful safeguard for older women seeking to conceive, improving the likelihood of selecting a viable embryo.

The advantages are compelling. PGT-A improves implantation rates, reduces pregnancy loss, and accelerates the path to a successful live birth. PGT-M prevents the inheritance of specific genetic diseases, breaking the cycle of transmission within families. These are not abstract outcomes, they are life-changing realities.

The technology itself has advanced remarkably. With Next-Generation Sequencing, we can now analyze embryos with extraordinary accuracy, detecting chromosomal errors and pinpointing single-gene mutations in a single workflow (4). This highly sophisticated platform enables high-throughput, cost-effective, and precise results, making PGT an indispensable tool in modern precision reproductive medicine.

However, it is important to recognize that PGT is not without challenges. The high cost of testing and IVF procedures can limit access for many couples, particularly in low-resource settings. Furthermore, biological complexities such as embryonic mosaicism—where both normal and abnormal cells coexist in the same embryo—can complicate diagnosis and interpretation, occasionally leading to uncertain outcomes. Addressing these limitations through better counseling, standardization, and cost optimization remains a crucial goal for the field.

Another important dimension of PGT is the ability to determine the genetic sex of embryos. While this must always be approached with strict ethical safeguards,

Correspondence to: Qamre Alam
*Molecular Genomics and Precision Medicine Department, EXPRESSMED Diagnostics and Research, Zinj, Bahrain.
Email: alamqa2022@gmail.com
Full list of author information is available at the end of the article.
Received: 04 September 2025 | **Revised (1):** 04 October 2025 | **Accepted:** 08 October 2025

it is essential in cases of sex-linked disorders such as hemophilia or Duchenne muscular dystrophy (5). Here, choosing the "right" gender is not a preference but a prevention.

The implications for public health are profound. In regions like the Middle East, where consanguinity and genetic disorders remain common, PGT offers a preventive strategy at the earliest possible stage, before life begins (6). Instead of managing untreatable disorders across a lifetime, we now have the opportunity to stop them from being passed on at all.

Importance of PGT-A in the GCC and Bahrain

The Gulf Cooperation Council (GCC) countries, including Bahrain, face unique genetic health challenges due to high rates of consanguineous marriages and the prevalence of hereditary conditions such as sickle cell disease, thalassemia, and metabolic disorders. In Bahrain, for example, sickle cell disease continues to be one of the most common inherited disorders, placing significant social and healthcare burdens on families and the medical system. PGT-A and PGT-M provide a direct means to address these challenges by reducing miscarriage rates, preventing the birth of affected children, and lowering the long-term healthcare costs associated with chronic genetic disease. Importantly, these technologies also offer hope to couples who might otherwise feel they have no safe reproductive options. As awareness and accessibility increase, PGT could become a cornerstone of reproductive health strategies in the GCC, complementing existing newborn screening and premarital testing programs.

Conclusions and Future Perspectives

PGT-A and PGT-M represent a paradigm shift in reproductive medicine, moving the focus from treatment to prevention. By enabling couples to avoid the transmission of genetic disorders and reducing the emotional and physical toll of failed pregnancies, PGT offers a vision of healthier families and healthier societies.

Nonetheless, for this promise to be fully realized, the field must confront key limitations such as cost, accessibility, and biological uncertainty. Continued innovation and collaboration will be necessary to make these technologies universally available and ethically sustainable.

Emerging adjunct technologies are poised to further transform this landscape. Artificial intelligence is increasingly being integrated into embryo selection and genetic data interpretation, enhancing accuracy and reducing human bias. Non-invasive embryo testing, analyzing Deoxyribonucleic Acid from spent culture media rather than embryo biopsy, may 1 day minimize risks to embryo integrity. Additionally, advanced imaging and time-lapse systems now allow for real-time assessment of embryo development, complementing genetic insights to improve selection outcomes.

Looking ahead, the future of PGT lies in broader accessibility, integration with personalized medicine, and continued advances in genomic technologies. Cost reduction, streamlined laboratory workflows, and greater awareness will be essential for ensuring equity of access. Ethical oversight will remain critical, particularly as technology expands into areas such as polygenic risk scoring and expanded carrier screening.

For Bahrain and the GCC, embracing PGT as part of a comprehensive public health strategy against genetic disease could dramatically reduce the prevalence of inherited disorders in future generations. By prioritizing prevention before birth, we can ensure that the promise of precision medicine is realized where it matters most: at the very beginning of life.

Acknowledgment
I would like to express my sincere gratitude to the Molecular Genomics and Precision Medicine Department, EXPRESSMED Diagnostics and Research, Bahrain, and the entire team, especially Sara Jaffer and Jannat, for their continuous support and assistance throughout this work.

List of Abbreviations

AI	Artificial intelligence
GCC	Gulf Cooperation Council
IVF	*In vitro* fertilization
NGS	Next generation sequencing
PGT-A	Preimplantation Genetic Testing for Aneuploidy
PGT-M	Preimplantation Genetic Testing for Monogenic Disorder

Conflict of interest
The authors of this article have no affiliations with or involvement in any organization or entity with any financial interest or non-financial interest in the subject matter or materials discussed in this manuscript.

Funding
None.

Consent to participate
Not applicable.

Ethical approval
Not applicable.

Author details
Qamre Alam[1], Mohamed Ali Ateyah[1], Mohamed Hani Ali[1], Kadhem Alkhenaizi[1]
1. Molecular Genomics and Precision Medicine Department, EXPRESSMED Diagnostics and Research, Zinj, Bahrain

References

1. Umair M, Rafeeq M, Alam Q, editors. Rare genetic disorders: advancements in diagnosis and treatment. 1st ed. Singapore: Springer Singapore; 2024. https://doi.org/10.1007/978-981-99-9323-9
2. Alam Q, Rafeeq M, Umair M, editors. Innovations and implications in molecular diagnostics. 1st ed. Boca Raton, FL: CRC Press; 2025. https://doi.org/10.1201/9781003489177
3. Alyafee Y, Alam Q, Tuwaijri AA, Umair M, Haddad S, Alharbi M, et al. Next-generation sequencing-based pre-

implantation genetic testing for aneuploidy (PGT-A): first report from Saudi Arabia. Genes. 2021;12(4):461.

4. World Health Organization. Genes and human diseases. Geneva, Switzerland: WHO; 2023.
5. Practice Committee of the American Society for Reproductive Medicine. The use of preimplantation genetic testing for aneuploidy (PGT-A): a committee opinion. Fertil Steril. 2018;109(3):429–36.
6. Malik SD, Al-Shafai M, Abdallah AM. The special features of prenatal and preimplantation genetic counseling in Arab countries. Genes. 2022;13(2):167. https://doi.org/10.3390/genes13020167