

Original Article

Diverse Applications of Preimplantation Genetic Testing in Assisted Conception at a Nigerian Fertility Centre: A Case Series

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ABSTRACT

Background: Preimplantation genetic testing (PGT) enables genetic assessment of embryos generated through in vitro fertilization before embryo transfer. Its applications include testing for chromosomal aneuploidy and monogenic disorders, with potential utility in reducing transmission of serious inherited conditions. Experience with PGT remains relatively underreported in sub-Saharan Africa.

Objective: To describe three successful applications of PGT for differing reproductive indications at a Nigerian fertility centre.

Methods: This descriptive case series reports three couples who underwent in vitro fertilization with PGT at a Kingswill Specialist Hospital, Lagos. Clinical and reproductive information was obtained from medical records. The indications comprised family balancing, a family history of oculocutaneous albinism, and prevention of sickle cell disease. Relevant PGT approaches were incorporated into assisted conception treatment before embryo transfer. Pregnancy and neonatal outcomes were documented.

Results: The first case involved a 39-year-old para 4 woman with four daughters who sought assisted conception with PGT and subsequently delivered twin boys following embryo transfer. The second couple had significant family histories of oculocutaneous albinism and underwent PGT for a monogenic disorder, resulting in the birth of a phenotypically unaffected female infant. In the third case, both partners had HbAS genotypes and had previously had two children with sickle cell disease (HbSS). Following PGT for a monogenic disorder, an unaffected embryo was transferred, resulting in the birth of a male infant with an HbAA genotype.

Conclusion: These cases illustrate the diverse potential applications of PGT in an African assisted-reproduction setting.

Keywords: Assisted Reproductive Technology; In Vitro Fertilization; Preimplantation Genetic Testing; PGT-A; PGT-M; Sickle Cell Disease; Oculocutaneous Albinism; Nigeria

Introduction

Preimplantation genetic testing (PGT) represents an important intersection between assisted reproductive technology and genomic medicine. [1] It enables genetic assessment of embryos generated through in vitro fertilization (IVF) before selection for embryo transfer, thereby providing couples with information that may influence reproductive decision-making. [1, 2] Contemporary PGT is broadly classified according to its intended purpose into preimplantation genetic testing for aneuploidy (PGT-A), preimplantation genetic testing for monogenic disorders (PGT-M), and preimplantation genetic testing for structural chromosomal rearrangements (PGT-SR). [1-3]

PGT-A evaluates embryos for numerical chromosomal abnormalities and is principally intended to facilitate the selection of chromosomally suitable embryos for transfer. Aneuploidy is an important contributor to implantation failure, miscarriage and adverse reproductive outcomes, particularly with advancing maternal age. [1,3] However, the precise clinical benefits and appropriate indications for routine PGT-A remain subjects of continuing scientific debate. [4-7] PGT-M, in contrast, is designed to identify embryos at risk of inheriting a defined monogenic disorder where the familial pathogenic variant or molecular diagnosis has been established. [1,3,5]

The potential importance of PGT-M is particularly evident in settings with a substantial burden of inherited diseases. [5, 8] Sickle cell disease is a major public health problem in sub-Saharan Africa and is inherited in an autosomal recessive pattern. Couples in whom both partners carry the haemoglobin S allele have a 25% probability of an affected HbSS offspring with each conception. PGT-M provides such couples with a reproductive option that can reduce the likelihood of transferring an embryo affected by sickle cell disease. [3,4] Successful live birth following PGT for sickle cell disease has previously been reported in a resource-limited African setting. [8]

Oculocutaneous albinism (OCA) represents another group of predominantly autosomal recessive inherited disorders for which PGT-M may be considered following appropriate molecular characterization. OCA is genetically heterogeneous, and pathogenic variants involving genes including OCA2 and TYRP1, among others, have been described in populations of African ancestry. [8] Consequently, establishing the specific familial pathogenic variant is important before developing a targeted PGT-M strategy.

Beyond disease prevention, PGT technologies may intersect with parental preferences regarding

offspring sex. The ethical and regulatory acceptability of non-medical sex selection varies considerably between jurisdictions. [3,9-11] In some settings, family balancing has emerged as an important reason why couples seek assisted reproductive treatment involving embryo genetic testing. A recent Nigerian study reported that elective sex selection or family balancing represented the predominant indication among couples undergoing PGT at a fertility centre, whereas PGT-M was used less frequently for inherited conditions including haemoglobin disorders and oculocutaneous albinism. [3]

Despite increasing availability of advanced assisted reproductive technologies in Africa, detailed reports describing the practical application and successful reproductive outcomes of PGT remain limited. This case series describes three distinct applications of PGT at a Nigerian fertility centre: assisted reproduction involving embryo sex information for family balancing; PGT-M in a couple with a strong family history of oculocutaneous albinism; and PGT-M to prevent sickle cell disease in a couple who had previously had two affected children.

Patients and Methods

Ethics Approval

Ethical approval for the study was obtained from the **Ethics and Research committee of 68 Nigerian Army Reference Hospital, Lagos Nigeria. [68 NARHY/ADM/ObGyn/ART/28.** The study was conducted in accordance with applicable institutional ethical standards and the principles of the Declaration of Helsinki.

Study Design and Setting

This was a descriptive case series of three couples who underwent IVF with PGT at Kingswill Specialist Hospital in Nigeria. The cases were selected to illustrate distinct clinical and reproductive applications of PGT encountered in assisted reproductive practice.

Clinical information was obtained retrospectively from patients' medical records, including demographic characteristics, obstetric and family histories, indication for PGT, type of genetic testing performed, assisted reproductive treatment and pregnancy outcomes.

The couples underwent clinical assessment and counselling before assisted conception treatment. IVF treatment involved controlled ovarian stimulation, oocyte retrieval, fertilization and embryo culture according to the center's established clinical protocols. Embryos considered appropriate for genetic assessment underwent embryo biopsy followed by genetic analysis relevant to the indication for testing.

Embryo selection and subsequent transfer were undertaken following review of the genetic testing results and counselling of the couples.

PGT-A was used where chromosomal assessment of embryos was undertaken, whereas PGT-

M was applied where testing was directed towards a defined or suspected monogenic disorder. Pregnancy outcomes following embryo transfer were obtained from clinical records and follow-up information.

Methods

Embryos selected for preimplantation genetic testing underwent biopsy following embryo culture, with genetic analysis performed using **next-generation sequencing (NGS), polymerase chain reaction (PCR)-based testing**. For PGT-A, embryo samples were analysed for chromosomal copy-number abnormalities and sex-chromosome constitution. For PGT-M, targeted molecular analysis was performed according to the specific genetic indication. In the case involving oculocutaneous albinism, testing was directed towards the previously identified familial pathogenic variant in **OCA2/TYRP1**, while in the sickle cell disease case, embryo genotyping targeted the pathogenic HBB variant responsible for haemoglobin S, permitting classification of embryos as HbAA, HbAS or HbSS. Genetic results were reviewed before embryo selection, and transfer decisions were made following appropriate genetic and reproductive counselling.

Case 1

A 39-year-old para 4 woman with four living female children presented to the fertility centre seeking assisted reproductive treatment and PGT. Her principal reproductive preference was to have male offspring following four previous female births. Following counselling, the patient underwent IVF with PGT-A and embryo transfer. Embryo genetic testing provided information regarding chromosomal status and embryo sex before selection for transfer. The treatment resulted in a successful clinical pregnancy. The pregnancy progressed to delivery, and the patient subsequently delivered live twin male infants. This case illustrates the application of embryo genetic testing in the context of assisted reproduction where family balancing was an important component of the couple's reproductive decision-making.

Case 2

A couple presented for reproductive genetic assessment because of a strong family history of oculocutaneous albinism. The female partner reported two siblings affected by oculocutaneous albinism, while the male partner reported one affected sibling.

Prior to PGT-M, molecular genetic evaluation was undertaken to establish the familial genetic basis of oculocutaneous albinism. Testing identified a pathogenic variant in the **OCA2/TYRP1** gene, which was considered responsible for the familial disorder. Because of this molecular diagnosis, a patient-specific PGT-M strategy was developed to identify embryos at

risk of inheriting the familial pathogenic variant. Embryos were biopsied and analyzed for the identified familial variant, and an embryo classified as **unaffected** was selected for transfer following genetic counselling. The transfer resulted in the birth of a female infant with no clinical evidence of oculocutaneous albinism.

This case demonstrates the potential role of PGT-M in reproductive planning for couples with a strong family history of an inherited disorder. The case also highlights the importance of establishing the causative familial genetic variant before PGT-M, given the genetic heterogeneity of oculocutaneous albinism.

Case 3

A couple in whom both partners had HbAS genotypes presented for reproductive counselling. The couple had previously had two children with sickle cell disease, both with an HbSS genotype. Given their reproductive history and the autosomal recessive inheritance of sickle cell disease, the couple sought assisted conception to reduce the likelihood of having another child affected by the condition.

PGT-M was undertaken specifically to determine the haemoglobin genotype of the embryos. Following embryo biopsy, genetic analysis targeting the β -globin (HBB) gene variant responsible for haemoglobin S was performed. Of the 7 embryos successfully genotyped, 2 were HbAA, 3 were HbAS and 2 were HbSS. Embryos with the HbSS genotype were excluded from consideration for transfer. Although HbAS embryos were unaffected by sickle cell disease and were available for transfer, an HbAA embryo was preferentially selected following counselling of the couple. Transfer of the HbAA embryo resulted in a successful pregnancy and the birth of a male infant, whose HbAA genotype was subsequently confirmed by postnatal haemoglobin testing. Three HBAS and one HBAA embryos were cryopreserved for the couple for future use.

This case illustrates the clinical application of PGT-M for preventing transmission of sickle cell disease in an at-risk couple with a history of two previously affected children.

Table 1: Comparative Case Table

Characteristic	Case 1	Case 2	Case 3
Principal indication	Family balancing/male offspring preference	Prevention of oculocutaneous albinism	Prevention of sickle cell disease
Relevant history	39 years; para 4; four female children	Female partner had two affected siblings; male partner had one affected sibling	Both partners HbAS; two previous children HbSS
PGT approach	IVF with PGT-A*	PGT-M	PGT-M
Genetic condition targeted	None	Oculocutaneous albinism	Sickle cell disease
Embryo selection objective	Chromosomal assessment with embryo sex information	Selection against the familial OCA disorder	Selection of an embryo not affected by HbSS
Pregnancy outcome	Successful twin pregnancy	Successful pregnancy	Successful pregnancy
Neonatal outcome	Twin live-born males	Live-born female without clinical albinism	Live-born male
Postnatal genetic/phenotypic outcome	Male phenotype	No clinical evidence of OCA	HbAA genotype

*PGT-A is primarily a test for embryonic aneuploidy rather than a diagnostic test developed specifically for non-medical sex selection. Where comprehensive chromosome testing reveals sex-chromosome constitution, this information may permit embryo sex identification subject to applicable ethical and regulatory frameworks.

Discussion

This case series illustrates three distinct applications of preimplantation genetic testing within assisted reproductive practice in Nigeria. Although all three couples underwent IVF incorporating embryo genetic assessment, their motivations and clinical indications differed substantially. These agree with the study of *Adegbola* and colleagues who found in their study that couples have different motivations for seeking fertility treatment. [12]

PGT has undergone considerable technological evolution. Earlier approaches to embryo chromosome screening were limited by the number of chromosomes that could be evaluated, technical limitations of single-cell analysis and the possibility of diagnostic error. Advances in blastocyst culture, trophectoderm biopsy, vitrification and molecular genetic techniques have substantially changed contemporary PGT practice. Nevertheless, PGT-A and PGT-M have fundamentally different objectives and should not be considered interchangeable. [2, 4]

In Nigeria, the utilization of PGT for family balancing appears to reflect broader sociocultural influences on reproductive preferences. A recent retrospective study from a Nigerian fertility centre reported that elective sex selection was the most frequent indication among couples undergoing PGT, substantially exceeding its utilization for monogenic disease prevention. [3, 13] This suggests that the pattern of PGT utilization in some African settings may differ

from settings where regulatory frameworks restrict embryo sex selection to avoidance of sex-linked disorders.

The second and third cases illustrate the disease-prevention potential of PGT-M. Unlike PGT-A, PGT-M involves targeted testing for a specific monogenic disorder and usually requires identification and characterization of the relevant pathogenic variant within the affected family. [1,3, 4] This distinction is particularly important when counselling couples with inherited conditions.

Our cohort also reveals successful treatment that resulted in the birth of an HbAA male infant. The outcome is consistent with previous reports demonstrating that PGT can be successfully applied to prevent sickle cell disease in resource-constrained settings. [8] Considering the substantial burden of sickle cell disease in sub-Saharan Africa, the increasing availability of PGT-M represents a significant milestone in reproductive care for couples at risk of transmitting the condition to their offspring. By enabling the identification and preferential transfer of embryos unaffected by sickle cell disease before pregnancy is established, PGT-M offers affected couples an important reproductive option and may alleviate the considerable psychological, emotional, social, and financial burden associated with recurrent births of affected children. Wider access to PGT-M, supported by appropriate genetic counselling and robust ethical

frameworks, could therefore contribute meaningfully to reducing the burden of sickle cell disease among high-risk families in the region.[14]

Taken together, these cases demonstrate a spectrum of motivations for PGT, ranging from parental reproductive preferences to prevention of significant inherited disease. They also highlight important differences in the ethical justification and clinical counselling associated with these indications. PGT-M for serious monogenic disease provides a potential alternative to natural conception followed by prenatal diagnosis and possible pregnancy termination.

Advanced reproductive genetic technologies.

The clinical significance of these cases extends beyond the individual reproductive outcomes and is particularly relevant to assisted reproductive practice in Nigeria and other resource-constrained settings. They demonstrate that PGT can be incorporated into

IVF care for both reproductive preference and prevention of inherited monogenic disorders where appropriate laboratory and genetic expertise are available. In settings such as Nigeria, where conditions such as sickle cell disease remain highly prevalent and access to prenatal diagnosis, genetic counselling and advanced reproductive technologies may be limited, PGT-M may provide an additional reproductive option for selected high-risk couples.

Strengths and Limitations

A strength of this case series is that it demonstrates diverse real-world applications of PGT within an African assisted-reproduction setting.

However, it should be acknowledged. First, the report includes only three cases and therefore cannot provide estimates of treatment effectiveness, implantation rates, live-birth rates or the overall success of PGT within the study population.

Conclusion

Preimplantation genetic testing has diverse applications within assisted reproductive practice, ranging from embryo chromosomal assessment and family balancing to prevention of inherited monogenic disorders. This case series demonstrates successful live births following PGT in three distinct reproductive

scenarios at a Nigerian fertility centre. PGT-M may have relevance in populations with a substantial burden of inherited disorders such as sickle cell disease and may also provide reproductive options for families affected by other monogenic conditions, including oculocutaneous albinism.

Acknowledgements

Ethics Approval: Ethical approval for the study was obtained from the **Ethics and Research committee of 68 Nigerian Army Reference Hospital, Lagos Nigeria.** [68 NARHY/ADM/ObGyn/ART/28. The study was conducted in accordance with applicable institutional ethical standards and the principles of the Declaration of Helsinki.

Individual Patient Consent Statement: Written informed consent was obtained from all patients/couples for the use of their anonymized clinical information for scientific publication. All potentially identifying information was removed from the manuscript to preserve patient confidentiality.

Data Availability: The data supporting the findings of this case series are available from the corresponding author upon reasonable request, subject to ethical and confidentiality restrictions. The complete individual patient records are not publicly available because they contain confidential clinical and genetic information.

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