

# Preimplantation Genetic Testing - Monogenic disorders (PGTM) for sickle cell anaemia prevention



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## OBJECTIVES

The breakthrough birth of an unaffected offspring via PGT-M <sup>1</sup> has brought hope to carriers of sickle cell anaemia (SCA). The wider implications of this technology in a low-resource setting need assessment. The objective of this study was to assess the prevalence of SCA in embryos of carrier couples, pregnancy outcomes and technical complications.

## DESIGN

This was a retrospective cross-sectional study of 201 embryos from 40 couples, tested for the presence of the HBB c.20A>T p.Glu6Val haemoglobin gene variant (HbS) using PGT-M from 2014-2021.

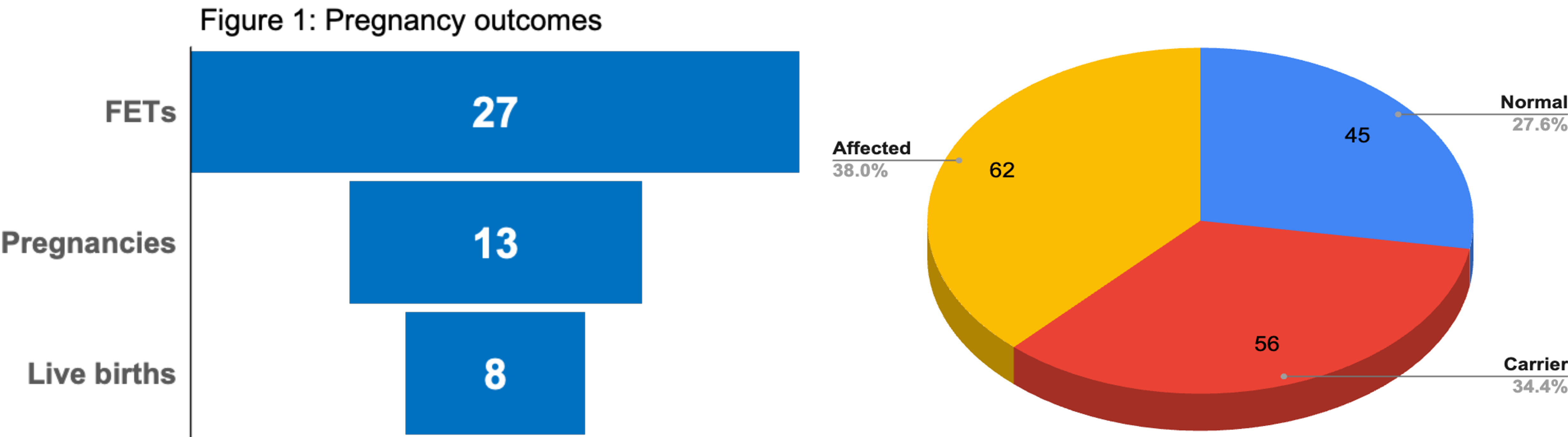
## METHOD

All embryos tested within the stated time frame were included in the study. From 2018, analysis was carried out at Cell Genomics Laboratory in Abuja and prior to this at Reprogenetics Laboratory in the UK. Cells were processed using the Illumina DNA extraction kit and the PGT-M protocol of Applied Biosystems, then analyzed using the Genetic Analyzer 3500 Series which operates with Sanger sequencing methodology. Genotype report was interpreted as unaffected [normal (HbAA) or carrier (HbAS)] and affected (HbSS).

## RESULTS

Mean maternal age was **31.48** (23-41). **101** embryos (50.2%) were **unaffected** (45 normal (22.39%), 56 carriers (27.86%)) and **62** were **affected** (30.85%). For 38 embryos, no result was obtained. Between 2018 and 2022, **27** patients had **embryo transfers** with 4 awaiting frozen embryo transfer (FET). There were **13 pregnancies** (48.1%) with 3 ongoing and **8 live births**, all of whom are confirmed to be unaffected.

Figure 2: Outcomes of PGT-M analysis



## CONCLUSION

PGT-M is a viable option for couples who are both carriers of HbS to prevent transmission of sickle cell disease to their offspring.

## REFERENCES

<sup>1</sup> Ibrahim W, Christopher D, Mohamed C. Live birth following preimplantation genetic testing to prevent sickle cell disease in a low resource setting: A case report. Afr J Reprod Health. 2020 Dec;24(4):218-220. doi: 10.29063/ajrh2020/v24i4.22. PMID: 34077086.

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