# Evaluation of the Global Standard of NIPT clinical services

Zandra C. Deans[1], Farrah Khawaja[1], Weronika Gutowska-Ding[2], Simon J. Patton[2], Fiona J. Morgan[1]

1. GenQA, Department of Laboratory Medicine, NHS Lothian, Edinburgh, EH45 9DX, United Kingdom, 2. EMQN CIC, Enterprise House, Manchester Science Park, Manchester, M15 5SE, United Kingdom

## Introduction

The delivery of External Quality Assessment (EQA) for non-invasive prenatal testing (NIPT) is a mechanism to independently measure the standard of laboratory testing for these rapid services. Continual participation in EQA is particularly important when the development of technology and the scope of testing is fast moving. EQA is provided for NIPT for common aneuploidies, early fetal sexing and common microdeletions.

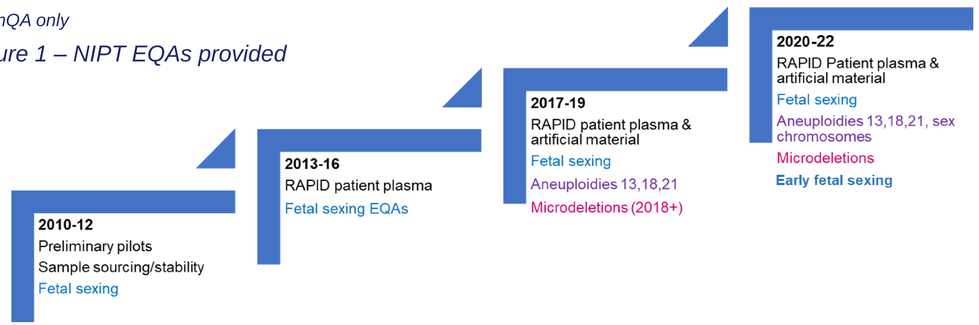
The two EQA Schemes, GenQA and EMQN are global providers of proficiency testing and have delivered assessments for NIPT since 2013. A review of eight years of EQA provision in this field has identified areas for development to improve testing processes and clear reporting of results.

## Material & methods

Fetal sexing assessment has been provided since 2013 and testing for common aneuploidies and microdeletion EQAs* were implemented in 2017 and 2018 respectively (Figure 1).

*GenQA only

*Figure 1 – NIPT EQAs provided*




Two plasma samples with corresponding clinical cases are provided for each EQA run and participants are expected to perform routine testing and submit clinical reports for assessment. Maternal plasma was sourced from RAPID Biobank and artificial material from SeraCare/LGC Group.

Genotyping accuracy and appropriate interpretation and reporting of the result is assessed against peer-ratified marking criteria. Participants are provided with individual tailored scoring and direct feedback comments. Poor performance is assigned when a critical genotyping error is reported, or an erroneous interpretation of the result is included on the report.

## Conclusion

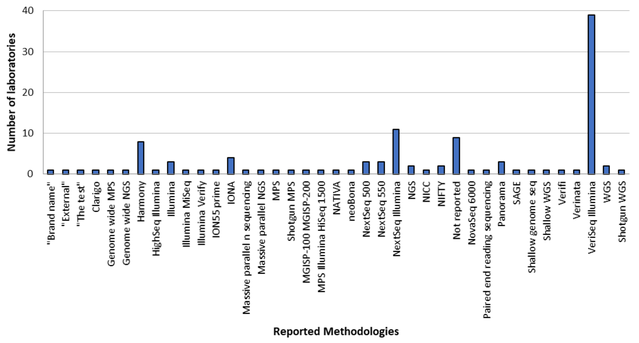
As testing strategies and technology advances, there continues to be areas where good practice and learnings identified by EQA can improve NIPT clinical services. Dissemination of these findings help shape global guidelines for high quality testing, supports laboratories implementing new services and ensure delivery of excellent patient care.

## Results

### NIPT for common aneuploidies

- Increased participation since 2017 with 140 laboratories taking part in the 2021 EQA.
- Poor performance is assigned to approximately 5% of laboratories per EQA run, however the 2020 assessment identified a higher poor performance. One case was high risk for sex chromosome aneuploidy XYY and 8.8% of laboratories reported this as a low risk result; a further two laboratories erroneously reported the presence of trisomy 13. There was no commonly used method by these laboratories.
- A wide range of testing methods are employed which are described in multiple ways (Figure 2).

*Figure 2 – Testing methodologies in 2020 NIPT aneuploidies EQA*



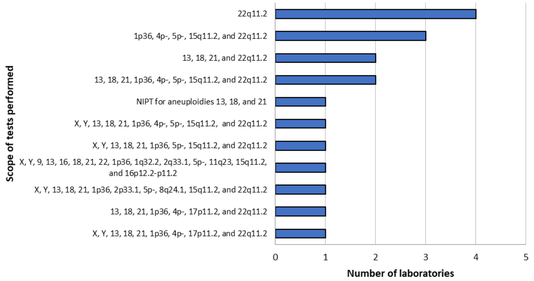

### NIPT for fetal sexing

- Initial fetal sexing NIPT EQAs included 19 participants, and this has doubled with approximately 40 laboratories now participating annually.
- The standard of testing is generally very high with minimal performance issues.
- As fetal sexing has become more commonly performed alongside aneuploidy testing, this EQA has been amended to tailor testing to early fetal gestation testing.

### NIPT for common microdeletions

- Laboratories frequently utilised methods not designed to detect microdeletions and therefore failed to report a high risk result where a microdeletion was present.
- Many reports failed to provide an adequate description of the limitations of the test.
- A wide range of microdeletions were tested for (Figure 3).

*Figure 3 – Scope of testing performed for NIPT microdeletion EQA in 2021*




### Reporting issues

Areas of improvement to reporting NIPT results have been identified and are relevant to all three NIPT EQAs (Figure 4).

*Figure 4 – Requirements and issues identified by NIPT EQAs*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info@genqa.org ● www.GenQA.org ● +44 (0)131 242 6898

**Corresponding author**: Zandra Deans (Sandi.Deans@ed.ac.uk)