Poster # 17


rosa.morra@emqn.org henry.thomas@emqn.org

Expanding the Boundaries of Point-of-Care Testing: Development of EQA Material for Pharmacogenetic POCT

Rosa Morra, Henry Thomas, Victoria Williams, Simon Patton
EMQN CIC, 3 Exchange Quay, Manchester M5 3ED

Background

Point-of-Care Testing (POCT) is expanding beyond its traditional applications in biochemistry into the field of genomics, particularly pharmacogenetics (PGx)[1,2]. This evolution introduces new challenges in quality assurance, requiring robust systems to ensure accurate and reliable results at the point of care. A leading example of PGx POCT is the Genedrive® MT-RNR1 ID Kit, a NICE-approved device developed by Genedrive PLC. The kit detects the m.1555A>G variant in the *MT-RNR1* gene, which is associated with an increased risk of aminoglycoside-induced hearing loss in neonates.

Aims and Objectives

The EMQN team aimed to support the implementation of the Genedrive® MT-RNR1 POCT by:

- Developing artificial **External Quality Assessment** (EQA) **materials** tailored for this PGx POCT
- Validating the **EQA materials** for **suitability and compatibility** with the Genedrive® MT-RNR1 ID Kit.
- Creating **device verification sample** packages to support clinical implementation.
- Authoring a **clinical verification protocol guideline** for PGx POCT devices, aimed at standardising implementation processes.
- Launching an **EQA scheme for MT-RNR1 POCT**—the first of its kind.

Workflow Overview

The EQA material was designed to closely mimic real patient samples. The MT-RNR1 POCT is a non-invasive test that uses buccal swabs for sample collection.

- **Cell Line Provision and Expansion**

Patient Cell lines with relevant genetic markers are provided by collaborator clinicians and expanded to create sufficient material for testing preparation.

- **Quality Control of Cell Count** to ensure reproducibility and swab coating reliability.
- **Swab Coating with Cells**

FLOQSwabs are coated with precise cell quantities at varying concentrations to simulate clinical sample variability.

- **Validation of Swabs**

Swabs undergo semi-quantitative and qualitative testing ensuring accuracy, homogeneity, low failure rate, and precision

EMQN EQA material validation

Figure 1. Workflow of Genedrive® MT-RNR1 ID kit used to validate the EMQN patient like buccal swab.

Operator	Variant (% conc.)	WT (% conc.)
EMQN 1	100 (N=70)	100 (N=50)
EMQN 2	100 (N=70)	100 (N=50)
External	100 (N=12)	100 (N=12)

Table 1. Concordance (conc.) rates for the EQA material (swab-like) in detecting the correct *MT-RNR1* genotype (variant or wild-type (wt)) across three operators using the Genedrive POCT kit. All operators achieved 100% concordance, demonstrating complete accuracy and precision of the EQA material.

Attribute	Description
AQL Level	Level II applied across batches using Genedrive® MT-RNR1 ID Kit
Post-Reconstitution Stability	24h at 4°C (cell released in assay buffer)
Open Pouch Stability	24h at RT, once pouch is opened

Table 2. Quality and stability attributes of the EQA swabs. Acceptable Quality Level (AQL). Level II refers to a standard inspection level used in quality control, where a statistically determined sample size is tested to ensure batch reliability.

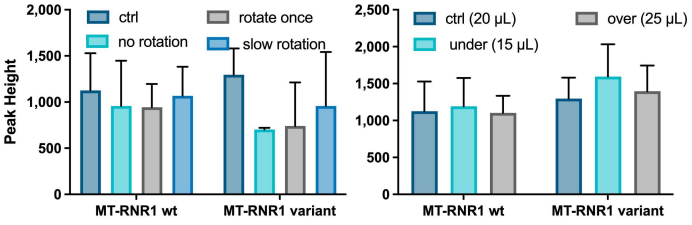
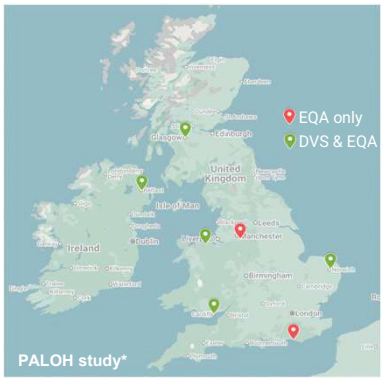


Figure 2. Robustness and stress behaviour of EMQN swab-like material. **Left:** Melting curve peak heights following protocol deviations in cell release (Step 1, Fig. 1). **Right:** Melting curve peak heights with altered assay volumes (Step 2, Fig. 1). Bars represent mean ± SD (N=4). Statistical analysis was performed using one-way ANOVA, *no significant differences* were observed compared to the control condition ($p > 0.05$).

Real world Impact



Map showing NHS Trust centres participating in the MT-RNR1 EQA scheme. Green markers indicate centres that also clinically verified the Genedrive MT-RNR1 POCT devices using EMQN verification material and protocol.

* Pharmacogenetics to Avoid Loss of Hearing (Phase II): Genedrive MT-RNR1 POCT implementation across UK.

EQA overview & outcome

- **Surveys Frequency**: Bi-monthly
- **Sample #**: 3 per survey
- **Result submission window**: 3 weeks
- **Results published**: 2 weeks after the submission deadline.

Survey	Devices enrolled	Full marks	Test failures
01	1	1	0
02	8	7	1
03	16		

Table 3. Results of the MT-RNR1 EQA scheme per survey. Survey 03 results to be published on 6th of Oct 25.

Conclusions

- Developed Artificial materials for **EQA and clinical device verification**
- Verified compatibility with the Genedrive® MT-RNR1 ID Kit
- Published **Device Verification Guidelines** for PGx POCT
- Supported PALOH study for MT-RNR1 POCT implementation (see map)
- Launched the **first genetic POCT scheme** in January 2025

References

1. McDermott JH, et al BMJ Open (2021);11:e044457. doi: 10.1136/bmjopen-2020-044457
2. Burke, Kerry A et al. JMD vol. 27,3 (2025): 209-215. doi:10.1016/j.jmoldx.2024.12.001

Acknowledgements
EMQN team, Genedrive team, Dr. McDermott JH