

EQA scheme catalogue 2025

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### Overview

#### We provide External Quality Assessment (EQA) for 10 core areas of genomics:

- Genomic and inherited disorders (rare diseases)
- Molecular pathology (tissue, plasma)
- Pharmacogenetic testing (drug intolerance)
- Point of Care Testing NEW
- Pre-analytical (sample handling) NEW
- Prenatal testing (including non-invasive testing)
- Postnatal testing
- Technology (including Next Generation Sequencing)
- Interlaboratory comparison (sample exchange)
- Variant classification and interpretation



#### Our EOA schemes:

- We aim to mimic real clinical testing as closely as possible. Laboratories are sent samples accompanied by a referral and are asked to perform testing using normal procedures.
- We provide materials that cover the range of common test requests relevant for the indication e.g., molecular pathology, diagnostic, predictive, family follow up/cascade screening and point of care testing.
- Laboratories submit fully interpreted clinical reports (for the majority of our schemes) or upload results to our website. A subset of technical schemes focus on genotyping and/or benchmarking the analytical process.
- We assess interpretation and advice for follow up by measuring compliance against best practice. Specific educational feedback is provided to laboratories, including information on genotyping and interpretation errors and advice to ensure clerical inaccuracy does not lead to patient harm.
- Scheme reports are provided which summarise data across the scheme such as different assays used, common errors and learning points.
- Some technical schemes provide data for benchmarking against other laboratories or over time.
- Where possible we offer multi-language support to allow laboratories to submit their reports in their native language.
- Most of our schemes are covered under our UKAS accreditation (ISO 17043:2010)

### New for 2025

You spoke, we listened! We continue to expand the scope of our EQA schemes to meet the needs of our users and address the increasing use of NGS panel testing and clinically focused genomic methods. We have updated some of our EQA schemes based on your feedback, helping to save costs and time:

- 1. Reducing the maximum number of molecular pathology cases from 10 to 5.
- 2. Using virtual cases to cover rare genotypes, whilst still providing cases with DNA for all EQA schemes to test the full analysis workflow.
- 3. Piloting flexible EQA options, allowing you to participate in schemes that better align with your laboratory practices and diagnostic service scope. For example, new in 2025, the hereditary hearing loss scheme has options for either targeted locus or gene panel testing.

If successful, we will roll out these strategies more widely in 2026 to better meet the needs of laboratories. To view detailed information on this year's EQA schemes, please go to <a href="https://www.emqn.org/our-eqa-schemes/">https://www.emqn.org/our-eqa-schemes/</a>).

	Scheme	New	Extension to scope	Description of change
**	Inherited Retinal Disorders (IRD)		<b>~</b>	Scope updated to offer cases involving genes that are known to cause either syndromic or non-syndromic IRD. Format modified to better assess the analytical pipelines of laboratories offering comprehensive testing services.
	Homologous Recombination Deficiency (HRD)	<b>~</b>		Pilot EQA scheme. Supported by MSD
	Pan-Tumour Fusions (FUSION)	<b>~</b>		Pilot EQA scheme. Suitable for laboratories testing for clinically relevant gene fusions in solid tumours.
	Breast cancer (ESR1 cfDNA testing)		<b>✓</b>	Offered as a full EQA scheme.
	Urothelial / bladder cancer (FGFR SNV/fusion testing)		<b>✓</b>	Offered as a full EQA scheme. Supported by Johnson & Johnson.
Ā	cfDNA extraction from plasma	<b>✓</b>		In partnership with SensID GmbH and supported by AstraZeneca. Two surveys in 2025; three artificial plasma samples for cell free DNA (cfDNA) extraction for each survey. Use it to monitor the quality of your cfDNA extraction procedures and to benchmark results against other laboratories.
	DNA Sequencing - NGS (v Germline) copy number variants (CNV)		<b>~</b>	Scheme has been improved: you can now upload two sets of data for this EQA. Test two different techniques, platforms, gene panels, or kits for the price of one submission. Collaboration with GenQA.
	Point of Care Testing (POCT) for MT-RNR1	<b>~</b>		Designed for the Genedrive® MT-RNR1 assay. Six sample distributions per year.
	Device verification package for MT-RNR1 (POCT)	<b>✓</b>		Patient-like swab-based materials designed to help support laboratories implement and verify POCT devices for <i>MT-RNR1</i> testing.

## **Pricing/Fees**

EMQN is a community interest company (CIC) registered in England (Number: 12020789, VAT / Tax Number: 329563282). As a CIC, we recognise the financial constraints being imposed upon many laboratories and therefore we keep our participation fees as low as possible. Consequently, we offer a number of options to help laboratories manage the costs of EQA participation and EMQN membership.

#### **Examples include**

- Membership of EMQN is free. You must be a member of EMQN to purchase our EQAs.
- We offer a **7.5% discount for 6 or more** EQA schemes purchased, with this increasing to a **15% discount when 12 or more** EQA schemes have been purchased. **To qualify for theses discounts you must purchase all schemes in one order.** Note: ILC activities do not qualify for a discount (see page 12).
- **Discounted participation** in our EQA activities for laboratories in developing / evolving economies (see <a href="https://www.emqn.org/our-eqa-schemes/">https://www.emqn.org/our-eqa-schemes/</a> for more information).
- Some EQA schemes are free, or participation costs are partially subsidised by sponsorship from corporate sponsors.

EMQN partners with several distributor organisations worldwide to make shipping, billing and invoicing easier in certain regions. Where distributor arrangements exist, then there may be differences in the pricing structure due to VAT/Tax, logistics and handling costs. For more information, please contact us (office@ emqn.org) or see our website (https://www.emqn.org/participating-in-eqa/terms-conditions/).

# Guidance for genotyping-only laboratories

Genetic test reports may be transmitted to other non-genetics health professionals and may also cross national boundaries. Therefore, whilst we recognise the different legislative requirements in various parts of the world, it is EMQN policy to encourage a comprehensive 'stand-alone report' following relevant best practice guidance, where available. Most of our EQA schemes therefore require interpretation of the genotype in the context of the clinical information provided.

**However, genotyping-only laboratories can still take part in our EQA schemes** by submitting a supporting document to the relevant scheme explaining why they do not provide clinical interpretation. Please note laboratories submitting genotyping reports are required to classify the variants they have detected and include those that are of clinical significance in their reports.

### Guidance for commercial kit manufacturers

Participation in EQA schemes is a valuable activity for commercial kit manufacturers and can be used for both validation of new test methodologies, as well as post market surveillance of existing products to ensure compliance with the requirements of the In-vitro Diagnostics Regulation (IVD-R).

We welcome and encourage the participation of commercial kit manufacturers with EMQN - we usually require a declaration of reason for use of the EQA and there is normally no requirement to provide clinical interpretation of test results. If you would like to discuss this further, then please contact us.

web: www.emqn.org

mail: office@emqn.org tel: +44 (0) 161 757 1591

## Registering to participate

- 1. Go to the EMQN website: <a href="https://www.emqn.org">https://www.emqn.org</a>
- 2. Click the "LOGIN" button, enter your username and password, and go to the "Purchase" page. **Please note** that both the main contact person AND additional staff members registered to the account can register for schemes. **Note:** only staff with permission to purchase schemes will see the Purchase page.
- 3. If you do not remember your password, click 'Forgotten your password' on the login page.
- 4. Select the schemes you wish to participate in. **Important:** click the "Check out and Complete" button to finalise the registration.
- 5. The registration period is open **from 16th September 2024**. The closing date is dependent on the scheme please see the website for details.
- 6. All participating laboratories are required to pay the appropriate fees to register. When a registration form is submitted, we assume that your laboratory will participate in the scheme, and materials are prepared accordingly. Therefore, a registration fee has to be paid, regardless of whether you submit results or not. If your laboratory circumstances change, you can request to be removed from a scheme by contacting office@emqn.org before samples are dispatched. However, if samples have been dispatched the fees will be charged.
- 7. An invoice will be prepared (including those schemes that have not been dispatched yet) and sent by email in April 2025. Laboratories with distribution agents will receive an invoice directly from them.
- 8. EQA materials will be shipped to the laboratories and should be analysed by your laboratory's routine protocol. See the online EQA scheme catalogue for the month samples will be shipped. Point of Care Testing (POCT) schemes are shipped bimonthly.
- 9. The laboratory is required to submit results to the EMQN website before the reporting deadline.
- 10. Validated genotypes of EQA materials will be published shortly after the reporting deadline and detailed results will be available 3-4 months afterwards.
- 11. For more details of all the schemes, please see the online EQA scheme catalogue that is available from https://www.emgn.org

web: www.emqn.org

mail: office@emgn.org

# Genomic and Inherited disorders EQA schemes



These EQA schemes require genomic DNA samples to be genotyped and full interpretative reports to be submitted. Genotyping, Interpretation and Clerical Accuracy are assessed by EMQN. We collaborate with other organizations to provide some of these EQA schemes. Please see <a href="https://www.emqn.org/our-eqa-schemes/">https://www.emqn.org/our-eqa-schemes/</a> for more information.

Scheme	Registration period	Distribution month	Results deadline	ISO 17043 status	Fee (GBP)
Autosomal dominant polycystic kidney disease (optional gene panel testing for hereditary cystic kidney disease)	16.09.2024 - 30.11.2024	January	March	Accredited	445
Beckwith-Wiedemann and Silver-Russell syndromes	16.09.2024 - 30.11.2024	January	March	Accredited	445
Cardiac genetics (arrhythmias)	16.09.2024 - 30.11.2024	January	March	Accredited	445
Cardiac genetics (hypertrophic cardiomyopathies)	16.09.2024 - 30.11.2024	January	March	Accredited	445
Charcot-Marie-Tooth disease / Hereditary Neuropathy with liability for pressure palsies (optional gene panel testing for other CMT- associated genes)	16.09.2024 - 30.11.2024	January	March	Accredited	445
Congenital Adrenal Hyperplasia	16.09.2024 - 30.11.2024	January	March	Accredited	445
Duchenne / Becker Muscular Dystrophy (optional gene panel testing for muscular dystrophies / hereditary myopathies)	16.09.2024 - 30.11.2024	January	March	Accredited	445
Inherited Retinal Disorders (IRD)	16.09.2024 - 30.11.2024	September	November	Pending	445
Familial autosomal dominant hypercholesterolemia	16.09.2024 - 30.11.2024	January	March	Accredited	445
Familial SHOX gene related disorders	16.09.2024 - 30.11.2024	January	March	Accredited	445
Fragile X Syndrome	16.09.2024 - 30.11.2024	January	March	Accredited	445
Friedreich Ataxia	16.09.2024 - 30.11.2024	January	March	Accredited	445

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mail: office@emgn.org

Scheme	Registration period	Distribution month	Results deadline	ISO 17043 status	Fee (GBP)
Hereditary amyloidosis	16.09.2024 - 30.11.2024	January	March	Not accredited	445
Hereditary breast / ovarian cancer	16.09.2024 - 30.11.2024	January	March	Accredited	445
Hereditary hearing loss	16.09.2024 - 30.11.2024	January	March	Accredited	445
Hereditary Haemochromatosis	16.09.2024 - 30.11.2024	January	March	Accredited	445
Huntington disease	16.09.2024 - 30.11.2024	January	March	Accredited	445
Lynch syndrome (hereditary non-polyposis colon cancer)	16.09.2024 - 30.11.2024	January	March	Accredited	445
Mitochondrial DNA metabolic disorders	16.09.2024 - 30.11.2024	January	March	Accredited	445
Monogenic Diabetes	16.09.2024 - 30.11.2024	January	March	Accredited	445
Multiple Endocrine Neoplasia Type 2	16.09.2024 - 30.11.2024	January	March	Accredited	445
Myotonic dystrophy (Types 1 and/or 2)	16.09.2024 - 30.11.2024	January	March	Accredited	445
Osteogenesis imperfecta	16.09.2024 - 30.11.2024	January	March	Accredited	445
Phenylketonuria	16.09.2024 - 30.11.2024	January	March	Accredited	445
Polyposis Syndromes	16.09.2024 - 30.11.2024	January	March	Accredited	445
Porphyrias	16.09.2024 - 30.11.2024	January	March	Accredited	445
Prader-Willi and Angelman Syndromes	16.09.2024 - 30.11.2024	January	March	Accredited	445
Rare neurological disease gene panel	16.09.2024 - 30.11.2024	January	March	Not accredited	445
Rare neuromuscular disease gene panel	16.09.2024 - 30.11.2024	January	March	Accredited	445

Scheme	Registration period	Distribution month	Results deadline	ISO 17043 status	Fee (GBP)
Retinoblastoma	16.09.2024 - 30.11.2024	January	March	Accredited	445
RYR1 related Myopathies and Malignant Hyperthermia susceptibility	16.09.2024 - 30.11.2024	January	March	Accredited	445
Severe Combined Immunodeficiencies gene panel	16.09.2024 - 30.11.2024	January	March	Accredited	445
Spinal Muscular Atrophy	16.09.2024 - 30.11.2024	January	March	Accredited	445
Spinocerebellar Ataxias	16.09.2024 - 30.11.2024	January	March	Accredited	445
Stickler syndrome	16.09.2024 - 30.11.2024	January	March	Accredited	445
Systemic Autoinflammatory Diseases	16.09.2024 - 30.11.2024	January	March	Accredited	445
Von Hippel Lindau Syndrome	16.09.2024 - 30.11.2024	January	March	Accredited	445
Wilson Disease	16.09.2024 - 30.11.2024	January	March	Accredited	445
Y-Chromosome Microdeletion testing	16.09.2024 - 30.11.2024	January	March	Accredited	445

## Molecular Pathology EQA schemes



These EQAs require FFPE, plasma, or germline DNA samples to be genotyped and fully interpretative reports are to be submitted (exceptions may apply – see website for comprehensive details on each scheme). We collaborate with other organizations to provide some of these EQA schemes. This is clearly shown for each EQA scheme. Please see <a href="https://www.emqn.org/our-eqa-schemes/">https://www.emqn.org/our-eqa-schemes/</a> for more information.

#### Tissue (FFPE)

Scheme	Registration period	Distribution month	Results deadline	ISO 17043 status	Fee (GBP)
Breast cancer (AKT Pathway gene testing - PIK3CA, AKT1, PTEN) <sup>1</sup>	16.09.2024 – 28.02.2025	June	August	Accredited	285
Colorectal cancer	16.09.2024 – 28.02.2025	June	August	Accredited	630
Lung (NSCLC) cancer – COMMON BIOMARKERS <sup>2</sup>	16.09.2024 – 28.02.2025	June	August	Accredited	285
Lung (NSCLC) cancer-NEW and EMERGING BIOMARKERS 3	16.09.2024 – 28.02.2025	June	August	Accredited	115
Melanoma	16.09.2024 – 28.02.2025	June	August	Accredited	630
Microsatellite Instability testing	16.09.2024 – 28.02.2025	June	August	Accredited	455
Oncogene Panel Testing	16.09.2024 – 28.02.2025	June	August	Accredited	630
Ovarian, breast, prostate and pancreatic cancers (Somatic testing of <i>BRCA1</i> , <i>BRCA2</i> and HRR genes) (PARPi) <sup>4</sup>	16.09.2024 – 28.02.2025	September	November	Accredited	285
Pan-Tumour Fusions	16.09.2024 – 28.02.2025	September	November	Not accredited	735
Urothelial / bladder cancer (FGFR (FGFR SNV/fusion testing) 5	16.09.2024 – 28.02.2025	September	November	Not accredited	285

<sup>&</sup>lt;sup>1</sup>Limited to 100 places. Partial subsidised by sponsorship from AstraZeneca.

<sup>&</sup>lt;sup>2</sup>Limited to 450 places. Partial subsidised by sponsorship from AstraZeneca.

<sup>&</sup>lt;sup>3</sup> Limited to 450 places. Partial subsidised by sponsorship from AstraZeneca. You must purchase the Lung cancer – COMMON BIOMARKERS scheme to qualify for this scheme.

<sup>&</sup>lt;sup>4</sup>Delivered in conjunction with GenQA Limited to 350 places. Partial subsidised by sponsorship from MSD, and AstraZeneca.

 $<sup>^{\</sup>rm 5}\text{Limited}$  to 300 places. Partial subsidised by sponsorship from Johnson & Johnson.

#### Plasma (cfDNA)

Scheme	Registration period	Distribution month	Results deadline	ISO 17043 status	Fee (GBP)
Breast cancer (ESR1)	16.09.2024 – 28.02.2025	September	November	Not accredited	285
Lung cancer (EGFR, KRAS) <sup>1</sup>	16.09.2024 – 28.02.2025	September	November	Accredited	285
Prostate cancer (BRCA1, BRCA2) (PARPi) <sup>2</sup>	16.09.2024 – 28.02.2025	September	November	Not accredited	285

<sup>&</sup>lt;sup>1</sup> Delivered in conjunction with GenQA. Limited to 300 places. Partial subsidised by sponsorship from AstraZeneca.

#### **Germline DNA**

Scheme	Registration period	Distribution month	Results deadline	ISO 17043 status	Fee (GBP)
Ovarian, breast, prostate and pancreatic cancers (germline testing of <i>BRCA1</i> and <i>BRCA2</i> (PARPi) <sup>3</sup>	16.09.2024 – 28.02.2025	September	November	Accredited	285 <sup>7</sup>

<sup>&</sup>lt;sup>3</sup> Delivered in conjunction with GenQA·Limited to 250 places. Partial subsidised by sponsorship from MSD, and AstraZeneca

<sup>&</sup>lt;sup>2</sup> Delivered in conjunction with GenQA Limited to 120 places. Partial subsidised by sponsorship from MSD, AstraZeneca and Johnson & Johnson

# Pharmacogenetics EQA Schemes



Our EQA schemes require DNA samples to be genotyped and full interpretative reports to be submitted. We may collaborate with other organizations to provide some of these EQA schemes. This is clearly shown-please see <a href="https://www.emqn.org/our-eqa-schemes/">https://www.emqn.org/our-eqa-schemes/</a> for more information.

Scheme	Registration period	Distribution month	Results deadline	ISO 17043 status	Fee (GBP)
Pharmacogenetics (panel testing)	16.09.2024 – 28.02.2025	June	August	Accredited	455
Pharmacogenetics (DPYD / UGT1A1 testing only)	16.09.2024 – 28.02.2025	June	August	Pending	455

# Point of Care Testing (POCT) EQA Schemes



Our EQA schemes are designed to specific POCT test technologies and include:

- Swab-based EQA materials
- Bimonthly (six distributions per year)
- Three samples per distribution (18 samples per year)
- Suitable for use with Genedrive® POCT devices.

Please see <a href="https://www.emqn.org/our-eqa-schemes/">https://www.emqn.org/our-eqa-schemes/</a> for more information.

Scheme	Registration period	Distribution month	Results deadline	ISO 17043 status	Fee (GBP)
POCT testing for MT-RNR1 NEW	Not restricted – join anytime from October 2024	February, April, June, August, October, December	14 days after distribution	Not accredited	1300

## **Device Verification Kits**

Leverage EMQN's package of EQA materials to help you implement and verify new devices.

Our Device Verification Kits contain:

- 4x positive controls (High, medium, low and <LOD cell counts)</li>
- 4x negative controls (High, medium, low and <LOD cell counts

Please see <a href="https://www.emgn.org/our-ega-schemes/">https://www.emgn.org/our-ega-schemes/</a> for more information.

Scheme		No. kits <sup>1</sup>	Fee (GBP)
Device Verification Kit (Suitable for GeneDrive® MT-RNR1 assay)		1	890
		2	1155
	NEW	3	1422
	WEN	4	1690
		5	1955

<sup>&</sup>lt;sup>1</sup> If you wish to purchase more than 5 kits, please contact the EMQN office. The unit cost increase is £268 / kit.

## Managed POCT service

We can provide different levels of service to cater for the unique needs of our customers in Point of Care settings. We offer a **Fully Managed Service** for sites that do not have the support of a local laboratory Point of Care Department for performance oversight. EMQN will perform the tasks that are generally the remit of a Point of Care co-ordinator e.g. Registration of instruments and maintenance of the database on EMQN's platforms, result entry, report issue and interpretation, poor performance management. This also includes a dedicated helpline service to provide customers with direct access to experienced personnel who can offer advice on method performance, interpretation and troubleshooting. **Contact the EMQN office to discuss your needs** (office@emqn.org; +44.161.757.1591).

# Pre-analytical (Sample handling) EQA Schemes



These EQAs are designed to test the pre-analytical processes that are routinely used by a laboratory as part of their testing strategy.

Please see <a href="https://www.emqn.org/our-eqa-schemes/">https://www.emqn.org/our-eqa-schemes/</a> for more information.

web: www.emqn.org

mail: office@emgn.org

Scheme	Registration period	Distribution month	Results deadline	ISO 17043 status	Fee (GBP)
cfDNA extraction from plasma <sup>1</sup> NEW	16.09.2024 – 28.02.2025	March, September	May, November	Not Accredited	02

SensID GmbH (https://www.sens-id.com) manufactures the materials used for this scheme. Separate nucleosome ctDNA controls are available from SensID GmbH directly. Inclusion herein does not represent an endorsement of their product or service by EMQN CIC.

<sup>&</sup>lt;sup>2</sup>Costs fully subsidised by sponsorship from AstraZeneca

# Technology / Technique-specific EQA Schemes



These EQAs require DNA, FFPE, or fresh frozen samples to be tested and genotyped only. We collaborate with other organisations to provide some of these EQA schemes. This is clearly shown for each EQA scheme.

Please see <a href="https://www.emqn.org/our-eqa-schemes/">https://www.emqn.org/our-eqa-schemes/</a> for more information.

Scheme	Registration period	Distribution month	Results deadline	ISO 17043 status	Fee (GBP)
DNA Sequencing (Sanger)	16.09.2024 – 28.02.2025	June	August	Accredited	455
DNA Sequencing – NGS (Germline SNVs and indels) <sup>1</sup>	16.09.2024 – 28.02.2025	September	November	Accredited	715
DNA Sequencing – NGS (Germline CNVs) 1	16.09.2024 – 28.02.2025	September	November	Not accredited	505
DNA Sequencing - (Somatic SNVs and indels + matched germline sample) 1	16.09.2024 – 28.02.2025	September	November	Not accredited	715
DNA Sequencing -(Somatic SNVs and Indels - no matched germline sample) 1	16.09.2024 – 28.02.2025	September	November	Not accredited	1115

<sup>&</sup>lt;sup>1</sup> Delivered in conjunction with GenQA.

# Prenatal testing EQA Schemes



These EQAs require plasma samples to be genotyped and full interpretative reports to be submitted. We collaborate with other organizations to provide some of these EQA schemes. This is clearly shown for each EQA scheme. Please see <a href="https://www.emqn.org/our-eqa-schemes/">https://www.emqn.org/our-eqa-schemes/</a> for more information.

Scheme	Registration period	Distribution month	Results deadline	ISO 17043 status	Fee (GBP)
NIPT for common aneuploidies (including sex chromosomes) <sup>1</sup>	16.09.2024 – 28.02.2025	October	November	Accredited	471
NIPT for fetal sexing (X-linked disorders) <sup>1</sup>	16.09.2024 – 28.02.2025	October	November	Accredited	471

<sup>&</sup>lt;sup>1</sup> Delivered in conjunction with GenQA.

## Postnatal testing EQA Schemes



This EQA requires DNA samples to be genotyped and full interpretative reports to be submitted. We collaborate with another organization to provide this EQA scheme. This is clearly shown - please see <a href="https://www.emqn.org/our-eqa-schemes/">https://www.emqn.org/our-eqa-schemes/</a> for more information.

web: www.emqn.org

mail: office@emqn.org tel: +44 (0) 161 757 1591

Scheme	Registration period	Distribution month	Results deadline	ISO 17043 status	Fee (GBP)
Postnatal Constitutional CNV detection [array / NGS] 1	16.09.2024 – 28.02.2025	May	July	Accredited	471

<sup>&</sup>lt;sup>1</sup> Delivered in conjunction with GenQA.

# Interlaboratory Comparison (ILC) Sample Exchanges



EMQN provides these EQA activities where there are a small number of laboratories performing testing, and/or there are no EQA schemes available. The sample types vary (normally DNA) and EMQN facilitates the exchange of materials between laboratories for the ILC, plus assessment of the genotyping results, and provision of a report summarizing the results. To participate, laboratories need to contribute samples. Please see <a href="https://www.emqn.org/our-eqa-schemes/">https://www.emqn.org/our-eqa-schemes/</a> for more information.

ILC	Registration period	Distribution month	Results deadline	ISO 17043 status	Fee (GBP)
Achondroplasia (FGFR3)	16.09.2024 – 28.02.2025	July	September	Not accredited	305
Congenital central hypoventilation syndrome (CCHS) (PHOX2B)	16.09.2024 – 28.02.2025	July	September	Not accredited	305
Fascioscapulohumeral Muscular Dystrophy (Type 1)	16.09.2024 – 28.02.2025	July	September	Not accredited	305
Fascioscapulohumeral Muscular Dystrophy (Type 2)	16.09.2024 – 28.02.2025	July	September	Not accredited	305

# Variant Classification and Interpretation



EMQN provides an online educational individual competence assessment programme designed to assess the accuracy of variant classification. We collaborate with another organization to provide this activity. Registration includes access to educational webinars by international experts. Please see <a href="https://www.emqn.org/our-eqa-schemes/">https://www.emqn.org/our-eqa-schemes/</a> or more information.

Scheme	Registration period	Online Assessmen	Results t deadline	ISO 17043 status	Fee (GBP)
BRCA and HRR gene variant classification 1	TBC <sup>9</sup>	May/ October	June/ November	Not accredited	0

<sup>&</sup>lt;sup>1</sup>Delivered in collaboration with GenQA. Funded by an unrestricted grant from MSD and AstraZeneca

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# Additional samples

The amount of EQA scheme material we ship for each EQA scheme is based upon the average requirements for routine laboratory testing. In some instances, the amount we ship may not be sufficient (for example, when a laboratory is using a technology which requires higher/larger amounts of input DNA, or which only tests for one gene at a time). In these circumstances, laboratories have the option to purchase ADDITIONAL samples from our catalogue whilst the schemes are open for registration. Additional samples are chargeable and will be invoiced - they are not covered underany of our corporate sponsorship agreements. These samples are priced as follows:

EQA schemes 1	Fee (GBP, £)
Germline Schemes (DM, FRAX, and FRDA)	138 (1set)
Molecular Pathology Schemes (Lung, Melanoma, Colorectal and Oncogene Panel)	285 (1 set), 351 (2 sets), 388 (3 sets)
DNA Sequencing (NGS v Germline SNVs and Indels)	143 (1 set), 186 (2 sets)
DNA Sequencing (NGS v Germline CNVs)	143 (1 set), 186 (2 sets)
cfDNA Extraction	285 (1 set)

<sup>&</sup>lt;sup>1</sup>Availability is restricted to a limited number of schemes. Please consult the online EQA catalogue for up-to-date information.

## Replacement samples

From time to time, laboratories may require replacement samples (for example, due to technical error, sample mix ups etc.). Replacement samples cannot be guaranteed but will be sent if available. Refer to the scheme instructions for information on how to request a replacement sample. Testing of replacements samples must be within the same timeframe as the originals. There is a charge to cover reasonable costs associated with sending replacement samples and these charges apply per request. This charge is a FLAT RATE independent of geographical location (see below) and will be applied to the requesting laboratory's EMQN invoice. Please note: If your organisation has previously raised a PO number then you may need to adjust this to take account of costs associated with replacement samples.

EQA schemes <sup>1</sup>	Fee (GBP, £)
All Germline schemes	138
All Molecular Pathology schemes <sup>2</sup>	285
Fusion gene testing scheme	358
cfDNA Extraction scheme	285
All Pharmacogenetics schemes	143
DNA Sequencing (NGS v Germline SNVs and Indels)	143
DNA Sequencing (NGS v Germline CNVs)	143
DNA Sequencing (NGS v Somatic only)	358
DNA Sequencing (NGS v Somatic + matched normal)	412
DNA Sequencing (Sanger)	143
All NIPT schemes	285

<sup>&</sup>lt;sup>1</sup>Except ILCs where we CANNOT provide any replacement samples.

<sup>&</sup>lt;sup>2</sup>Except Fusion gene testing EQA scheme

#### Invoices

#### VAT/TAX

The pricing of all products sold by EMQN (EQA schemes and memberships) is exclusive of VAT/Tax which will be charged on our invoices as follows:

- Customers from the United Kingdom (UK): Our products are liable to VAT and this will be added to all
  invoices.
- Customers from the European Union (EU): Our products are liable to UK VAT/Tax (20%) unless the customer can supply their VAT/Tax number; those customers will be required to account for VAT/Tax in their own region under the reverse charge scheme. The VAT/Tax number can be updated in the users EMQN website account.
- Customers outside of the EU or UK: VAT/Tax will not be charged on invoices.

#### **Payment terms**

Our payment terms are 30 days. There are penalties for late payment and interest (Bank of England base rate plus 1%), charged per month, will be applied to outstanding balances after the invoice payment date. Non-payment of invoices will mean **access to your laboratory account will be suspended until payment is received** (see section 4.3 of our terms and conditions: (<a href="https://www.emqn.org/terms-conditions/">https://www.emqn.org/terms-conditions/</a>).

#### Purchase orders

If you require a purchase order quote to register for the EQA scheme, then please contact us at <a href="mailto:office@emgn.org">office@emgn.org</a>.

#### Other information

EMQN is a community interest company (CIC) registered in England (Number: 12020789. VAT/Tax Number: 329563282). For more information please contact us (<a href="mailto:finance@emqn.org">finance@emqn.org</a>) or see our website (<a href="https://www.emqn.org/terms-conditions/">https://www.emqn.org/terms-conditions/</a>).

web: www.emqn.org

mail: office@emgn.org

### Terms and conditions

EMQN CIC is supported financially by subscription fees. By joining us, and/or registering for an EQA scheme, you are agreeing to abide by our terms and conditions. A copy of our full terms and conditions can be downloaded from our website - please go to the following page (<a href="https://www.emqn.org/terms-conditions/">https://www.emqn.org/terms-conditions/</a>).

## Privacy and data protection

EMQN is committed to ensuring that your personal information is protected and never misused. We've read lots of Privacy policies and understand that they can be complicated. We've tried to make ours as clear as possible and summarised how EMQN handles your personal information. To read our Privacy Policy, please go to our website - (see <a href="https://www.emqn.org/privacy-policy">https://www.emqn.org/privacy-policy</a>).

### Accreditation of EMQN

EMQN is accredited by the United Kingdom Accreditation Service (UKAS) to ISO17043:2010. The scope of our accreditation can be found at <a href="https://www.emqn.org/about-emqn/#accreditation">https://www.emqn.org/about-emqn/#accreditation</a>
The accreditation status of all our EQA schemes is clearly shown within each of the schemes listed in this catalogue.



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## Working with EMQN

If you would like to join one of our assessment teams, please go to our <u>"Become an EQA Assessor"</u> section of the website for more information

## Help and advice

Our website has a comprehensive <u>Help and Frequently Asked Questions (FAQ) section</u>, including user guides and short help videos. If you cannot find an answer to your questions, then please do not hesitate to contact us (<u>office@emgn.org</u>). Our staff are always happy to help you.

#### Visit our website for more information

www.emqn.org

#### Contact us

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