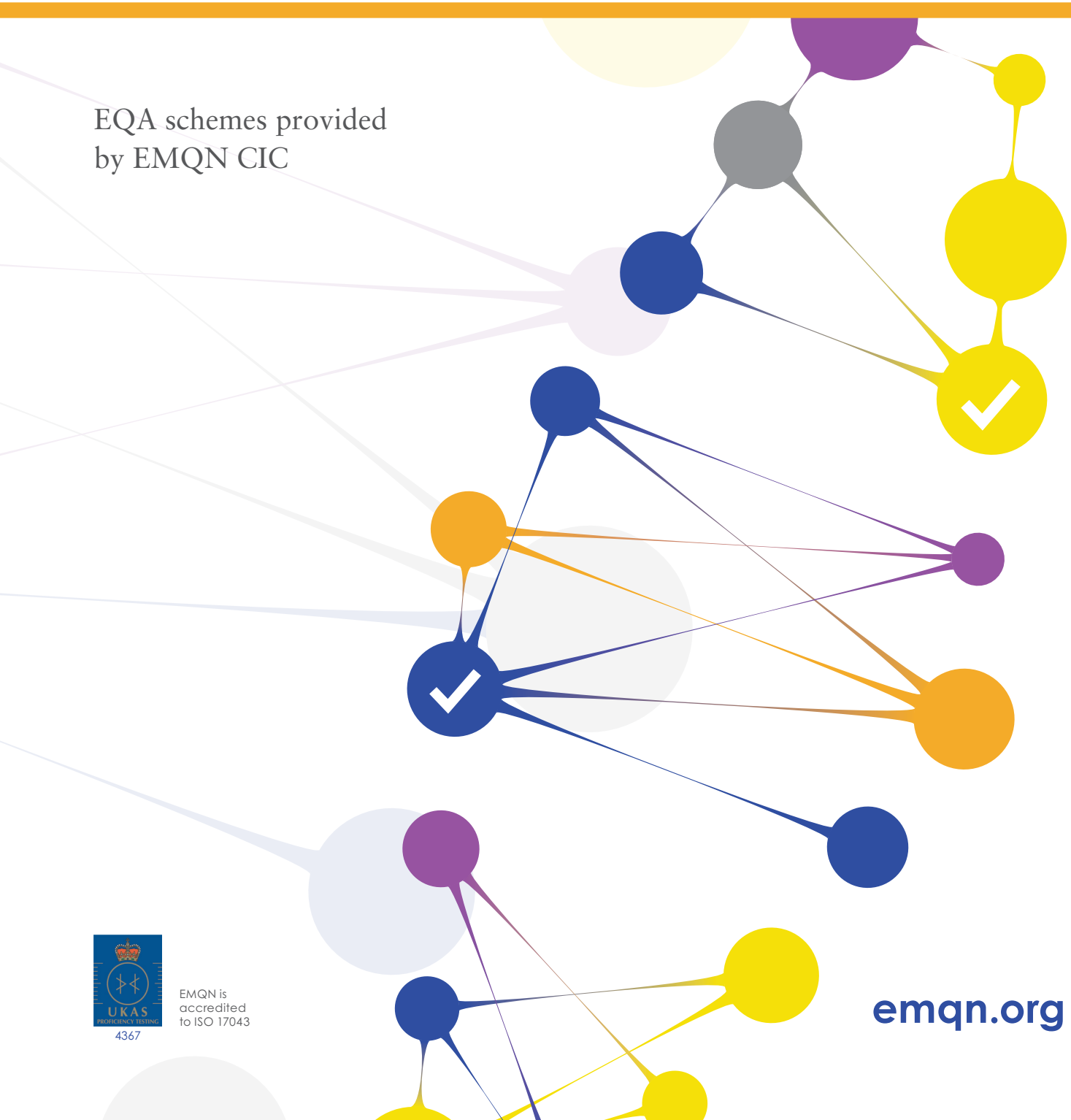
EQA scheme catalogue & participant guide 2020

EQA schemes provided by EMQN CIC



UKAS PROFICIENCY TESTING
4367

EMQN is accredited to ISO 17043

emqn.org

Table of contents

TEL: +44 161 276 6741 FAX: +44 161 276 6606 | WEB: www.emqn.org EMAIL: office@emqn.org

Introduction from EMQN Managing Director

Welcome to our 2020 EQA scheme catalogue and participant guide. The new EQA scheme year promises to be an exciting time for us and we have lots of new developments that we want to share with you, including two new EQA's and changes to our membership structures (see page 4 for more information).

EMQN is a scientific organisation which helps laboratories provide accurate and reliable test results by promoting professional quality in diagnostic genomic testing and appropriate analytical and interpretative performance. We started our life in 1998 with initial grant funding from the European Commission and went on to develop into the successful organisation that we are today, as a hosted service within a large UK public sector hospital (Manchester University NHS Foundation Trust).

We have seen tremendous growth since our humble start in 1999 and now have members in 79 countries; our services are used by approximately 1600 laboratories around the globe. We are very proud of our achievements. However this growth, coupled with regulatory changes within the UK public health system, means that we need to restructure our organization.

The restructure means that we are now operating independently from Manchester University NHS Foundation Trust, as a new legal entity called EMQN CIC. This means that EMQN now operates as a Community Interest Company (CIC). This is a legal model that obliges us to spend at least half of the profits on community benefit. This will enable us to continue to reinvest our profits into research and development, best practice, and training and education in genomics for the benefit of patients. The remainder can be spent on operational costs. There are no shareholders, so nobody personally profits out of EMQN doing well.

Over the coming months we will continue to work closely with all our partners to ensure a smooth transition.

Our work has always been supported by our members and collaborators and we value the partnership and working relationship that we share with them. We envision that this will continue alongside our restructure and that our members will be part of this exciting development and see the benefits that we can offer both to patients and to you.

We will keep in touch regularly with any relevant updates. For any enquiries please contact Michelle Plaiter at communication@plaiter.org

Simon Patton (Managing Director)

New for 2020

We are launching 2 new pilot EQA schemes:

- **Severe Combined Immunodeficiency (SCID)** for variants in different genes relevant to SCID, for example *RAG1/2, ADA*, and *DCLRE1C*. The genes included in the scheme will vary each year.
- **Microsatellite Instability testing (MSI)** is an addition to our molecular pathology schemes.

We have three **Interlaboratory Comparison (ILC's) / Sample Exchange** schemes running:

- **Achondroplasia** (*FGFR3*)
- **Congenital Hyperventilation Syndrome** (*PHOX2B*)
- **Hereditary Cancer Panel Testing**

If you are struggling to find an EQA scheme that meets your needs, for example for a specific rare disease, then please submit an application to our ILC programme. Our threshold for providing this activity is based on a minium of 3 laboratories[1]. Please go to page 67 for more information.

A number of existing EQA schemes have been reviewed and small changes made:

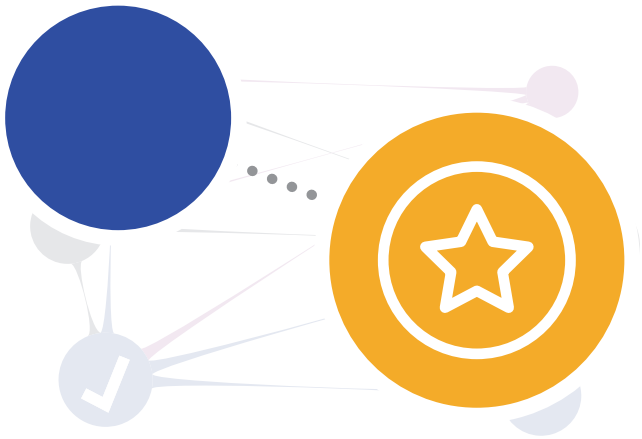
- The CMT scheme name has been amended to **CMT/HNPP** to reflect the fact that the potential gene targets now include *PMP22* (deletion /duplication), *MPZ, GJB1* and other associated genes.
- The FAP scheme has been renamed **Polyposis Syndromes** (FAP, MAP) because *MUTYH* is included.
- Both NIPT schemes have been renamed in recognition that the clinical testing pathway has evolved (see pages 65-66). The renamed schemes are:

[1] Minimum of 3, maximum of 7.

- **Non-invasive prenatal testing (NIPT) for common aneuploidies (including sex chromosomes).**
- **Non-invasive prenatal testing (NIPT) for fetal sexing (X-linked disorders).**

We are working with our colleagues at IQNPath to deliver an EQA for **Tumour Mutation Burden (TMB).** This is still at an early stage but we hope to be part of the IQNPath project to deliver a pilot for TMB in 2020. Watch this space!

We have made a number of changes to our membership structure. From September 2019, the annual EMQN membership fee for 2020 will rise to £150 but laboratories will be able to register up to 10 staff. Additional staff members can be registered in quotas of 5 for a fee of £50 per quota[2]. The change in membership structure will apply to all members and laboratories will be notified when they can add additional laboratory staff members to their account.



[2] For information, the previous membership fee was £55 for one staff member, and £50 for each additional account (up to a maximum of 5 extra). The total equivalent cost was £305.

TEL: +44 161 276 6741 FAX: +44 161 276 6606 WEB: www.emqn.org EMAIL: office@emqn.org

EMQN Membership

EMQN membership makes you a partner in the largest External Quality Assessment (EQA, sometimes called Proficiency Testing) network for molecular testing in the world. Membership offers important benefits for a modest outlay and is open to public and private testing laboratories, commercial manufacturers of relevant instruments, kits and reagents, and to pharmaceutical, veterinary and other laboratories.

Gain Recognition For Your Laboratory

The independent assessment of your laboratory's performance brings a focus to your quality management programme and helps you to gain international recognition for your results.

National accrediting bodies, such as UKAS in the UK, are members of the International Laboratory Accreditation Cooperation (ILAC). Membership of EMQN enables you to satisfy the EQA participation requirements of these bodies.

EMQN includes over 1600 member labs worldwide (including over 80% of the genetic testing laboratories in Europe). Membership of these high performing laboratories extends beyond Europe with members in Australia, Asia and the Americas. These laboratories have chosen EMQN as their EQA provider. By choosing EMQN you join this elite group. By participating in our EQA schemes you set high standards and will gain recognition and respect for a commitment to the highest standards of patient care.

Drive Your Quality Improvement and Innovation

Participation in EMQN's schemes helps to drive your laboratory quality higher and contributes to your quality improvement model. The results of scheme participation contribute towards best practice guideline development and enables the dissemination of innovation. Our members' combined contributions ultimately benefit each individual member laboratory.

EMQN has adopted a continuous improvement model for its EQA process. Each laboratory receives feedback on their performance on each scheme. Schemes and participants are assessed with reference to best practice on individual diseases and the feedback provided includes guidance for laboratory management in genetics. A report is provided from the scheme as a whole, as is a summary report on the EQA schemes in a given year. Based on an independent assessment of results these reports provide a variety of levels of feedback to support the overall improvement model.

EMQN organises and facilitates best practice meetings. Members can contribute to best practice development through these meetings which benefits the testing community and improves patient care. Following a best practice meeting, draft best practice guidelines are produced and published on this and related websites. Access to and use of this information and participation in EQA schemes are key to the diffusion of innovation and the continuous drive for quality.

TEL: +44 161 276 6741
FAX: +44 161 276 6606
WEB: www.emqn.org
EMAIL: office@emqn.org

Access Help and Advice

As a member laboratory requiring help and advice you are invited to contact the EMQN office where, on a case by case basis, we will use our extensive network to access international expertise in molecular genetic testing.

EMQN is best placed to know the high performers and leaders in specific areas. We can help with general performance problems or with specific issues highlighted by scheme participation. Through our quality system these interactions are recorded and followed up to ensure the effective use of the advice provided. These valuable experiences are selectively captured in anonymised case studies and shared with the membership.

EMQN is continually seeking to improve its offerings to member laboratories. We are active in the development of new benefits including our case studies library and a new initiative to enable experience sharing through staff interchange. These new benefits will be published in our news sections and detailed further on this website.

Accelerate Your Adoption of New Technology

EMQN responds to innovative testing technologies with appropriate new EQA schemes to drive quality and accelerate adoption. Where your laboratory is a leader in new technology adoption, EMQN compares and links you to other progressive laboratories in the field.

EMQN closely tracks developments in diagnostic testing as new technologies are adopted and applied. EMQN responds to these developments with new EQA schemes, providing support to progressive laboratories. Participating laboratories benefit from an independent measure of quality early in the adoption process.

Examples of Developments

An example of EMQN leadership is in the development of DNA sequencing schemes. Other schemes currently being piloted, or under consideration include the development of EQA methodologies for:

1. Circulating free DNA (liquid biopsy)
2. Non-invasive prenatal testing
3. Pharmacogentics method to predict patient response to new specialized drugs
4. Next Generation DNA Sequencing
5. New schemes for rare disease such as Severe Combined Immunodeficiencies (SCIDs)

JOIN EMQN TODAY
www.emqn.org

TEL: +44 161 276 6741
FAX: +44 161 276 6606
WEB: www.emqn.org
EMAIL: office@emqn.org

EQA Schemes

Overview

We provide EQA schemes for 4 core areas of genomic medicine:

- Inherited genetic disorders (Germline mutation testing)
- Cancer (Somatic mutation testing)
- Technological approaches used in testing
- Pharmacogentic testing related to drug intolerance

Our EQA schemes aim to mimic real clinical testing as closely as possible with laboratories testing samples accompanied by an appropriate referral, and subsequently submitting fully interpreted clinical laboratory reports to our website. Most of our schemes are covered under our UKAS accreditation. Where possible we offer multi-language support to allow laboratories to submit their reports in their native language. Each scheme has a set of core requirements which are defined in the following pages. Any differences or exceptions are clearly shown.

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.

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.

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 producsts.

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.

TEL: +44 161 276 6741
FAX: +44 161 276 6606
WEB: www.emqn.org
EMAIL: office@emqn.org

Germline mutation testing EQA schemes

Thirty seven EQA schemes are provided in 2020. These EQAs 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. This is clearly shown for each EQA scheme. Please see our website for more information.

TEL: +44 161 276 6741
FAX: +44 161 276 6606
WEB: www.emqn.org
EMAIL: office@emqn.org

Autosomal Dominant Polycystic Kidney disease

KIDNEY DISEASE | INHERITED | GERMLINE

Scheme code	ADPKD-20
Target	Mutations in the *PKD1* and *PKD2* genes.
Sample Material	DNA (in TE Buffer)
Scheme Format	Assessment of genotyping, and biological and clinical interpretation. For each case, participants are expected to return a clinical report which includes a complete interpretation of the results. Full EQA scheme no restrictions on number of participants. Open to laboratories from ALL countries. Three mock clinical cases with matching samples. (3)
Reporting Language	Reports accepted in English or German.
Additional Information	Suitable for sequence based analysis (e.g. NGS / Sanger Sequencing) and copy number variation (e.g. MLPA, NGS based CNV analysis). Mutation screening in any *PKD1* and *PKD2* exons. **This EQA is NOT suitable for labs testing recurrent mutations only.**
Performance criteria	Performance criteria **DO** apply.
Accreditation	This scheme is covered by the scope of EMQN's accreditation.
Collaborator	None

TEL: +44 161 276 6741 FAX: +44 161 276 6606 | WEB: www.emqn.org EMAIL: office@emqn.org

Beckwith-Wiedemann & Silver-Russell syndromes

IMPRINTING | DYSMORPHOLOGY | CANCER | INHERITED | GERMLINE

Scheme code	BWS/SRS -20
Target	BWS: Maternal hypomethylation at ICR2 *(KCNQ1OT1)*, hypermethylation at maternal ICR1 *(H19)*, copy number variants, segmental mosaic UPD11pat, and maternally-inherited mutations of *CDKN1C*. SRS: Paternal methylation at ICR1, CNVs simulating maternalisation of ICR1, UPD7mat, other rare imprinting anomalies, and diverse CNVs.
Sample Material	DNA (in TE Buffer)
Scheme Format	Assessment of genotyping, and biological and clinical interpretation. For each case, participants are expected to return a clinical report which includes a complete interpretation of the results. Full EQA scheme NO restrictions on number of participants. Open to laboratories from ALL countries. Three mock clinical cases with matching samples. (3)
Reporting Language	Reports accepted in English or German.
Additional Information	Suitable for methylation and copy number analysis (e.g. MS-MLPA).
Performance criteria	Performance criteria **DO** apply.
Accreditation	This scheme is covered by the scope of EMQN's accreditation.
Collaborator	None

TEL: +44 161 276 6741
FAX: +44 161 276 6606
WEB: www.emqn.org
EMAIL: office@emqn.org

Cardiac Arrhythmias

CARDIAC DISEASE | INHERITED | GERMLINE

PANEL

Scheme Code	CARDIO(ARR)-20
Target	**Panel testing:** as testing approaches are still not standardized and vary between laboratories, the exact list of genes to be tested is not specified.
Sample Material	DNA (in TE Buffer)
Scheme Format	Assessment of genotyping, and biological and clinical interpretation. For each case, participants are expected to return a clinical report which includes a complete interpretation of the results. No restrictions on number of participants. Open to laboratories from ALL countries. Three mock clinical cases with matching samples. (3) **Aimed at laboratories using a panel based DNA sequencing strategy.** Reports are marked in the context of the panel performed.
Reporting Language	Reports accepted in English.
Additional Information	Suitable for sequence based analysis (e.g. NGS / Sanger Sequencing) and copy number variation (e.g. MLPA, NGS based CNV analysis). Only clinically relevant variants* should be reported. * Only variants that have been validated in the test samples will be assessed.
Performance criteria	✓ Performance criteria **DO** apply..
Accreditation	☆ This scheme is covered by the scope of EMQN's accreditation.
Collaborator	⊖ None

TEL: +44 161 276 6741
FAX: +44 161 276 6606
WEB: www.emqn.org
EMAIL: office@emqn.org

Charcot-Marie-Tooth disease / Hereditary Neuropathy with Pressure Palsies

NEUROLOGICAL DISEASE | INHERITED | GERMLINE

Scheme code	CMT/HNPP-20
Target	Mutation testing in the *PMP22* (deletion /duplication), *MPZ*, *GJB1* and other associated genes.
Sample Material	DNA (in TE Buffer)
Scheme Format	Assessment of genotyping, and biological and clinical interpretation. For each case, participants are expected to return a clinical report which includes a complete interpretation of the results. No restrictions on number of participants. Open to laboratories from ALL countries. Three mock clinical cases with matching samples. (3)
Reporting Language	Reports accepted in English or German.
Additional Information	Suitable for copy number analysis of the *PMP22* gene (e.g. MLPA) and/or sequence analysis (Sanger/ NGS) of *PMP22/MPZ/GJB1* genes.
Performance criteria	Performance criteria **DO** apply.
Accreditation	This scheme is covered by the scope of EMQN's accreditation.
Collaborator	None

TEL: +44 161 276 6741
FAX: +44 161 276 6606
WEB: www.emqn.org
EMAIL: office@emqn.org

Congenital Adrenal Hyperplasia

HORMONE INHERITED GERMLINE

Scheme code	CAH-20
Target	Mutations in the *CYP21A2* gene
Sample Material	DNA (in TE Buffer)
Scheme Format	Assessment of genotyping, and biological and clinical interpretation. For each case, participants are expected to return a clinical report which includes a complete interpretation of the results. No restrictions on number of participants. Open to laboratories from ALL countries. Three mock clinical cases with matching samples. (3)
Reporting Language	Reports accepted in English or German.
Additional Information	Suitable for targeted mutation testing as well as sequence based analysis (e.g. NGS / Sanger Sequencing), copy number analysis (e.g. MLPA, NGS based CNV analysis), and Southern blotting techniques.
Performance criteria	✓ Performance criteria **DO** apply.
Accreditation	☆ This scheme is covered by the scope of EMQN's accreditation.
Collaborator	— None

TEL: +44 161 276 6741
FAX: +44 161 276 6606
WEB: www.emqn.org
EMAIL: office@emqn.org

Duchenne / Becker Muscular Dystrophy

MUSCULAR DYSTROPHY | INHERITED | GERMLINE

Scheme Code	DMD-20
Target	Mutations in the Dystrophin gene
Sample Material	DNA (in TE Buffer)
Scheme Format	Assessment of genotyping, and biological and clinical interpretation. For each case, participants are expected to return a clinical report which includes a complete interpretation of the results. No restrictions on number of participants. Open to laboratories from ALL countries. Three mock clinical cases with matching samples. (3)
Reporting Language	Reports accepted in English, Dutch or German.
Additional Information	Suitable for copy number analysis of the dystrophin gene but we expect labs to refer to point mutation testing if it is used.
Performance criteria	Performance criteria **DO** apply.
Accreditation	This scheme is covered by the scope of EMQN's accreditation.
Collaborator	None

TEL: +44 161 276 6741
FAX: +44 161 276 6606
WEB: www.emqn.org
EMAIL: office@emqn.org

Familial Hypercholesterolemia

CARDIAC DISEARSE | INHERITED | GERMLINE

Scheme code	FH-20
Target	Mutations in the *LDLR*, *APOB* and *PCSK9* genes
Sample Material	DNA (in TE Buffer)
Scheme Format	Assessment of genotyping, and biological and clinical interpretation. For each case, participants are expected to return a clinical report which includes a complete interpretation of the results. Open to laboratories from ALL countries. Three mock clinical cases with matching samples. (3)
Reporting Language	Reports accepted in English, German, French or Spanish.
Additional Information	Suitable for sequence based analysis (e.g. NGS / Sanger Sequencing) and copy number analysis (e.g. MLPA, NGS based CNV analysis). Mutation screening in any *LDLR*, *APOB* and *PCSK9* exons required. **Autosomal Dominant FH only.** **This scheme is NOT suitable for labs testing for recurrent mutations only.**
Performance criteria	Performance criteria **DO** apply.
Accreditation	This scheme is covered by the scope of EMQN's accreditation.
Collaborator	None

TEL: +44 161 276 6741
FAX: +44 161 276 6606
WEB: www.emqn.org
EMAIL: office@emqn.org

Familial SHOX-related disorders

INHERITED GERMLINE

Scheme code	SHOX-20
Target	Mutations in the *SHOX* gene
Sample Material	DNA (in TE Buffer)
Scheme Format	Assessment of genotyping, and biological and clinical interpretation. For each case, participants are expected to return a clinical report which includes a complete interpretation of the results. No restrictions on number of participants. Open to laboratories from ALL countries. Three mock clinical cases with matching samples. (3)
Reporting Language	Reports accepted in English or German.
Additional Information	Suitable for sequence based analysis (e.g. NGS / Sanger Sequencing) and copy number analysis (e.g. MLPA, NGS based CNV analysis). Testing of the short stature homeobox for disorders such as Langer mesomelic dysplasia and Leri-Weill dyschondrosteosis (LWD).
Performance criteria	✓ Performance criteria **DO** apply.
Accreditation	☆ This scheme is covered by the scope of EMQN's accreditation.
Collaborator	⊖ None

TEL: +44 161 276 6741
FAX: +44 161 276 6606
WEB: www.emqn.org
EMAIL: office@emqn.org

Fragile X Syndrome (Full version)

INTERLECTUAL DISABILITY | TRINUCELOTIDE REPEAT DISORDER | INHERITED | GERMLINE

Scheme code	FRAX-20-Full
Target	Triplet repeat expansions (**including the entire range of expansion, +/- methylation, of mutations**) in the *FMR1* gene.
Sample Material	DNA (in TE Buffer)
Scheme Format	**Applicable to labs which are able to perform the FULL diagnosis in each case with a method which detects the ENTIRE range of expansion mutations.** Assessment of genotyping, and biological and clinical interpretation. For each case, participants are expected to return a clinical report which includes a complete interpretation of the results. No restrictions on number of participants. Open to laboratories from ALL countries. Three mock clinical cases with matching samples. (3)
Reporting Language	Reports accepted in English, German or French.
Additional Information	Suitable for PCR-based (e.g. TP-PCR) and Southern blotting techniques. CCG repeat analysis ONLY. **NOTE: this version of the FRAX scheme is applicable only to laboratories that analyse the full range of expansion (+/- methylation patterns).**
Performance criteria	✓ Performance criteria **DO** apply.
Accreditation	☆ This scheme is covered by the scope of EMQN's accreditation.
Collaborator	⊖ None

TEL: +44 161 276 6741
FAX: +44 161 276 6606
WEB: www.emqn.org
EMAIL: office@emqn.org

Fragile X Syndrome (Pre-screening only version)

INTERLECTUAL DISABILITY | TRINUCELOTIDE REPEAT DISORDER | INHERITED | GERMLINE

Scheme code	FRAX-20-Pre screen
Target	Triplet repeat pre-screen for the presence of expansion mutations in the *FMR1* gene.
Sample Material	DNA (in TE Buffer)
Scheme Format	**Applicable to labs which are NOT able to perform the full diagnosis in each case but perform a pre-screen ONLY.** Assessment of genotyping, and biological and clinical interpretation. No restrictions on number of participants. Open to laboratories from ALL countries. Three mock clinical cases with matching samples. (3)
Reporting Language	Reports accepted in English, German or French.
Additional Information	Suitable for PCR-based and Southern blotting techniques. CCG repeat analysis ONLY. **NOTE: This scheme is intended for laboratories that CANNOT test the full range of expansion mutations / methylation patterns, and provide an initial screen for the presence / absence of normal (or small permutation) FRAX repeats.**
Performance criteria	Performance criteria **DO** apply.
Accreditation	This scheme is covered by the scope of EMQN's accreditation.
Collaborator	None

TEL: +44 161 276 6741
FAX: +44 161 276 6606
WEB: www.emqn.org
EMAIL: office@emqn.org

Friedreich Ataxia

NEUROLOGICAL DISEASE | TRINUCELOTIDE REPEAT DISORDER | INHERITED | GERMLINE

Scheme code	FRDA-20
Target	Triplet repeat expansions of mutations in the *FXN* gene
Sample Material	DNA (in TE Buffer)
Scheme Format	Assessment of genotyping, and biological and clinical interpretation. For each case, participants are expected to return a clinical report which includes a complete interpretation of the results. No restrictions on number of participants. Open to laboratories from ALL countries. Three mock clinical cases with matching samples. (3)
Reporting Language	Reports accepted in English, French, German or Dutch.
Additional Information	Suitable for PCR-based and Southern blotting techniques. Point mutation testing not required.
Performance criteria	Performance criteria **DO** apply.
Accreditation	This scheme is covered by the scope of EMQN's accreditation.
Collaborator	None

TEL: +44 161 276 6741
FAX: +44 161 276 6606
WEB: www.emqn.org
EMAIL: office@emqn.org

Hereditary Breast and Ovarian Cancer (BRCA1/2 targeted testing only)

CANCER | INHERITED | GERMLINE

Scheme Code	HBOC-20-BRCA
Target	Mutations in the *BRCA1* and *BRCA2* genes ONLY.
Sample Material	DNA (in TE Buffer)
Scheme Format	**Participants are expected to return a clinical report which includes a complete (biological and clinical) interpretation, in addition to the genotyping results, for each case.** If you DO NOT provide clinical interpretation, then a supporting document MUST be submitted explaining why in order to be excluded from assessment of interpretation. No restrictions on number of participants. Open to laboratories from ALL countries. Three mock clinical cases with matching samples. (3) **NOTE: Laboratories which test the BRCA genes in the context of targeted therapies (e.g., PARP inhibitors) for Ovarian cancer should register for the separate schemes (somatic/germline) related to "Molecular testing of BRCA genes in Ovarian Cancer".**
Reporting Language	Reports accepted in English, French, German, Spanish or Italian.

Continued on next page

TEL: +44 161 276 6741
FAX: +44 161 276 6606
WEB: www.emqn.org
EMAIL: office@emqn.org

Hereditary Breast and Ovarian Cancer (BRCA1/2 targeted testing only)

CANCER | INHERITED | GERMLINE

Additional Information	Suitable for sequence based analysis (e.g. NGS / Sanger Sequencing) and copy number analysis (e.g. MLPA, NGS based CNV analysis). Mutation screening in any BRCA exons required. Some cases may require screening of the full BRCA1 or BRCA2 gene. **NOT suitable for labs testing for recurrent mutations only.** **Choose this scheme if you only test the BRCA1/2 genes.** If you test other genes then register for the HBOC panel testing scheme.
Performance criteria	Performance criteria **DO** apply.
Accreditation	This scheme is covered by the scope of EMQN's accreditation.
Collaborator	None

TEL: +44 161 276 6741
FAX: +44 161 276 6606
WEB: www.emqn.org
EMAIL: office@emqn.org

Hereditary Breast and Ovarian Cancer (Panel testing version)

CANCER | INHERITED | GERMLINE

PANEL

Scheme code	HBOC-20-Panel
Target	**Panel testing of genes (not limited to BRCA1/2) associated with HBOC.** As testing approaches are still not standardised and vary between laboratories, the exact list of genes to be tested is not specified.
Sample Material	DNA (in TE Buffer)
Scheme Format	**Participants are expected to return a clinical report which includes a complete (biological and clinical) interpretation, in addition to the genotyping results, for each case.** If you DO NOT provide clinical interpretation, then a supporting document MUST be submitted explaining why in order to be excluded from assessment of interpretation. No restrictions on number of participants. Open to laboratories from ALL countries. Three mock clinical cases with matching samples. (3) Reports will be marked in the context of the panel performed. **NOTE: Laboratories which test the BRCA genes in the context of targeted therapies (e.g., PARP inhibitors) for Ovarian cancer should register for the separate schemes (somatic/germline) related to "Molecular testing of BRCA genes in Ovarian Cancer".**
Reporting Language	Reports accepted in English, French, German, Spanish or Italian.

Continued on next page

TEL: +44 161 276 6741
FAX: +44 161 276 6606
WEB: www.emqn.org
EMAIL: office@emqn.org

Hereditary Breast and Ovarian Cancer (Panel testing version)

CANCER | INHERITED | GERMLINE

PANEL

Additional Information	Suitable for sequence based panel analysis (e.g. NGS / Sanger Sequencing) and copy number analysis (e.g. MLPA, NGS based CNV analysis). Mutation screening in any HBOC gene panel – to account for differing panel compositions, variants outside the stated scope of an individual laboratory's panel will be excluded from their assessment. Only clinically relevant variants* should be reported. *Only variants that have been validated in the test samples will be assessed. **NOT suitable for labs testing for recurrent mutations only.** **Do not choose this scheme if you only test the BRCA 1/2 genes.** Please register for the HBOC-20-BRCA scheme instead.
Performance criteria	Performance criteria **DO** apply.
Accreditation	This scheme is covered by the scope of EMQN's accreditation.
Collaborator	None

TEL: +44 161 276 6741
FAX: +44 161 276 6606
WEB: www.emqn.org
EMAIL: office@emqn.org

Hereditary Deafness

DEAFNESS | INHERITED | GERMLINE

PANEL

Scheme code	DFNB1-20
Target	Mutations in the *GJB2* and *GJB6* genes (DFNB1). **An optional 4th sample will be included for laboratories that perform panel testing for other genes (for example, *TRIOBP*).** Labs that do not perform panel testing are not required to analyse this sample
Sample Material	DNA (in TE Buffer)
Scheme Format	Assessment of genotyping, and biological and clinical interpretation. For each case, participants are expected to return a clinical report which includes a complete interpretation of the results. No restriction on the number of participants. Open to laboratories from ALL countries. Four mock clinical cases with matching samples will be supplied. (4)
Reporting Language	Reports accepted in English, German and French ONLY.
Additional Information	Suitable for sequence based analysis (e.g. NGS / Sanger Sequencing) and copy number analysis (e.g. MLPA, NGS based CNV analysis).
Performance criteria	✓ Performance criteria **DO** apply.
Accreditation	☆ This scheme is covered by the scope of EMQN's accreditation.
Collaborator	⊖ None

TEL: +44 161 276 6741
FAX: +44 161 276 6606
WEB: www.emqn.org
EMAIL: office@emqn.org

Hereditary Haemochromatosis

INHERITED GERMLINE

Scheme code	HFE-20
Target	Mutations in the *HFE* gene
Sample Material	DNA (in TE Buffer)
Scheme Format	Assessment of genotyping, and biological and clinical interpretation. For each case, participants are expected to return a clinical report which includes a complete interpretation of the results. No restrictions on number of participants. Open to laboratories from ALL countries. Three mock clinical cases with matching samples. (3)
Reporting Language	Reports accepted in English and German ONLY.
Additional Information	Suitable for targeted mutations testing as well as sequence based analysis (e.g. NGS / Sanger Sequencing) and copy number analysis (e.g. MLPA, NGS based CNV analysis).
Performance criteria	Performance criteria **DO** apply.
Accreditation	This scheme is covered by the scope of EMQN's accreditation.
Collaborator	None

TEL: +44 161 276 6741
FAX: +44 161 276 6606
WEB: www.emqn.org
EMAIL: office@emqn.org

Hereditary Recurrent Fevers

INFLAMATORY DISEASE | INHERITED | GERMLINE

Scheme code	HRF-20
Target	Mutations in the *MEFV, MVK, TNFRSF1A* and *NLRP3* genes.
Sample Material	DNA (in TE Buffer)
Scheme Format	Assessment of genotyping, and biological and clinical interpretation. For each case, participants are expected to return a clinical report which includes a complete interpretation of the results. No restrictions on number of participants. Open to laboratories from ALL countries. Four mock clinical cases with matching samples. (4) Laboratories may opt out of cases where the gene specific is not tested in their laboratory.
Reporting Language	Reports accepted in English or German.
Additional Information	Suitable for sequence based analysis (e.g. NGS / Sanger Sequencing) and copy number analysis (e.g. MLPA, NGS based CNV analysis).
Performance criteria	Performance criteria **DO** apply.
Accreditation	This scheme is covered by the scope of EMQN's accreditation.
Collaborator	None

TEL: +44 161 276 6741
FAX: +44 161 276 6606
WEB: www.emqn.org
EMAIL: office@emqn.org

Huntington Disease

NEUROLOGICAL DISEASE | TRIPLET REPEAT DISORDER | INHERITED | GERMLINE

Scheme code	HD-20
Target	Triplet repeat analysis of mutations in the *HTT* gene
Sample Material	DNA (in TE Buffer)
Scheme Format	Assessment of genotyping, and biological and clinical interpretation. For each case, participants are expected to return a clinical report which includes a complete interpretation of the results. No restrictions on number of participants. Open to laboratories from ALL countries. Three mock clinical cases with matching samples. (3)
Reporting Language	Reports accepted in English, Dutch or German.
Additional Information	Suitable for PCR-based analysis techniques ONLY. CAG repeat analysis ONLY.
Performance criteria	✓ Performance criteria **DO** apply.
Accreditation	☆ This scheme is covered by the scope of EMQN's accreditation.
Collaborator	⊖ None

TEL: +44 161 276 6741
FAX: +44 161 276 6606
WEB: www.emqn.org
EMAIL: office@emqn.org

Hypertrophic Cardiomyopathies

CARDIAC DISEARSE | INHERITED | GERMLINE

PANEL

Scheme code	CARDIO(HCM)-20
Target	**Panel testing:** aimed at laboratories using a panel based DNA sequencing strategy. As testing approaches are still not standardized and vary between laboratories, the exact list of genes to be tested is not specified.
Sample Material	DNA (in TE Buffer)
Scheme Format	Assessment of genotyping, and biological and clinical interpretation. For each case, participants are expected to return a clinical report which includes a complete interpretation of the results. No restrictions on number of participants. Open to laboratories from ALL countries. Three mock clinical cases with matching samples. (3) Reports will be marked in the context of the panel performed.
Reporting Language	Reports accepted in English.
Additional Information	Suitable for sequence based analysis (e.g. NGS / Sanger Sequencing) and copy number analysis (e.g. MLPA, NGS based CNV analysis). Only clinically relevant variants* should be reported. *Only variants that have been validated in the test samples will be assessed.
Performance criteria	✓ Performance criteria **DO** apply.
Accreditation	☆ This scheme is covered by the scope of EMQN's accreditation.
Collaborator	⊖ None

TEL: +44 161 276 6741
FAX: +44 161 276 6606
WEB: www.emqn.org
EMAIL: office@emqn.org

Lynch Syndrome

CANCER | INHERITED | GERMLINE

Scheme code	HNPCC-20
Target	Mutations in the *MSH2*, *MLH1*, *MSH6* and *PMS2* genes.
Sample Material	DNA (in TE Buffer)
Scheme Format	Assessment of genotyping, and biological and clinical interpretation. For each case, participants are expected to return a clinical report which includes a complete interpretation of the results. No restrictions on number of participants. Open to laboratories from ALL countries. Three mock clinical cases with matching samples. (3)
Reporting Language	Reports accepted in English, French or German.
Additional Information	Suitable for sequence based analysis (e.g. NGS / Sanger Sequencing) and copy number analysis (e.g. MLPA, NGS based CNV analysis).
Performance criteria	Performance criteria DO apply.
Accreditation	This scheme is covered by the scope of EMQN's accreditation.
Collaborator	None

TEL: +44 161 276 6741
FAX: +44 161 276 6606
WEB: www.emqn.org
EMAIL: office@emqn.org

Mitochondrial DNA (mtDNA) Metabolic Disorders

METABOLIC | INHERITED | GERMLINE

Scheme code	mtDNA-20
Target	Mutations in mtDNA (mitochondrial genome).
Sample Material	DNA (in TE Buffer)
Scheme Format	Assessment of genotyping, and biological and clinical interpretation. For each case, participants are expected to return a clinical report which includes a complete interpretation of the results. No restriction on the number of participants. Open to laboratories from ALL countries. Three mock clinical cases with matching samples will be supplied. (3)
Reporting Language	Reports accepted in English.
Additional Information	Suitable for sequence based analysis (e.g. NGS / Sanger Sequencing) and copy number analysis (e.g. MLPA, NGS based CNV analysis). Metabolic disorders which MAY be covered by the EQA scheme include MELAS, NARP, LHON, MERRF, Leigh syndrome, and Pearson syndrome. Levels of homo/heteroplasmy will be assessed.
Performance criteria	✓ Performance criteria DO apply.
Accreditation	☆ This scheme is covered by the scope of EMQN's accreditation.
Collaborator	Royal College of Pathologists of Australasia Quality Assurance Programs – www.rcpaqap.com.au/.

TEL: +44 161 276 6741
FAX: +44 161 276 6606
WEB: www.emqn.org
EMAIL: office@emqn.org

Monogenic Diabetes

METABOLIC | INHERITED | GERMLINE

Scheme code	MONODIAB-20
Target	Mutations in the *GCK*, *HNF1A*, *HNF1B* and *HNF4A* genes
Sample Material	DNA (in TE Buffer)
Scheme Format	Assessment of genotyping, and biological and clinical interpretation. For each case, participants are expected to return a clinical report which includes a complete interpretation of the results. No restrictions on number of participants. Open to laboratories from ALL countries. Three mock clinical cases with matching samples. (3)
Reporting Language	Reports accepted in English, German or French.
Additional Information	Suitable for sequence based analysis (e.g. NGS / Sanger Sequencing) and copy number analysis (e.g. MLPA, NGS based CNV analysis).
Performance criteria	Performance criteria **DO** apply.
Accreditation	This scheme is covered by the scope of EMQN's accreditation.
Collaborator	None

TEL: +44 161 276 6741
FAX: +44 161 276 6606
WEB: www.emqn.org
EMAIL: office@emqn.org

Multiple Endocrine Neoplasia Type 2

CANCER | INHERITED | GERMLINE

Scheme code	MEN2-20
Target	Mutations in the *RET* proto-oncogene
Sample Material	DNA (in TE Buffer)
Scheme Format	Assessment of genotyping, and biological and clinical interpretation. For each case, participants are expected to return a clinical report which includes a complete interpretation of the results. No restrictions on number of participants. Open to laboratories from ALL countries. Three mock clinical cases with matching samples. (3)
Reporting Language	Reports accepted in English or German.
Additional Information	Suitable for sequence based analysis (e.g. NGS / Sanger Sequencing) and copy number analysis (e.g. MLPA, NGS based CNV analysis).
Performance criteria	Performance criteria **DO** apply.
Accreditation	This scheme is covered by the scope of EMQN's accreditation.
Collaborator	None

TEL: +44 161 276 6741
FAX: +44 161 276 6606
WEB: www.emqn.org
EMAIL: office@emqn.org

Myotonic Dystrophy (Types 1 & 2)

MUSCULAR DYSTROPHY | TRINUCELOTIDE REPEAT DISORDER | INHERITED | GERMLINE

Scheme code	DM-20
Target	**The scope of this scheme has been updated to included triplet repeat analysis of mutations in the *CNBP* gene (DM2)** (optional fourth sample), in addition to the *DMPK* gene (DM1).
Sample Material	DNA (in TE Buffer)
Scheme Format	Assessment of genotyping, and biological and clinical interpretation. For each case, participants are expected to return a clinical report which includes a complete interpretation of the results. No restrictions on number of participants. Open to laboratories from ALL countries. Four mock clinical cases with matching samples. (4)
Reporting Language	Reports accepted in English, Danish or German.
Additional Information	Suitable for PCR-based and Southern blotting techniques. CTG repeat analysis ONLY. An optional DM2 sample will be included. Labs that do not perform DM2 testing are not required to analyse this sample.
Performance criteria	✓ Performance criteria **DO** apply.
Accreditation	☆ This scheme is covered by the scope of EMQN's accreditation.
Collaborator	⊖ None

TEL: +44 161 276 6741
FAX: +44 161 276 6606
WEB: www.emqn.org
EMAIL: office@emqn.org

Osteogenesis imperfecta

CONNECTIVE TISSUE DISORDER | INHERITED | GERMLINE

Scheme code	OI-20
Target	Mutations in the *COL1A1* and *COL1A2* genes.
Sample Material	DNA (in TE Buffer)
Scheme Format	Assessment of genotyping, and biological and clinical interpretation. For each case, participants are expected to return a clinical report which includes a complete interpretation of the results. Open to laboratories from ALL countries. Three mock clinical cases with matching samples. (3)
Reporting Language	Reports accepted in English or Dutch.
Additional Information	Suitable for sequence based analysis (e.g. NGS / Sanger Sequencing) and copy number analysis (e.g. MLPA, NGS based CNV analysis). Mutation screening in any *COL1A1* and *COL1A2* exons required. **This scheme is NOT suitable for labs testing for recurrent mutations only.**
Performance criteria	Performance criteria **DO** apply.
Accreditation	This scheme is covered by the scope of EMQN's accreditation.
Collaborator	None

TEL: +44 161 276 6741
FAX: +44 161 276 6606
WEB: www.emqn.org
EMAIL: office@emqn.org

Phenylketonuria

METABOLIC | INHERITED | GERMLINE

Scheme code	PKU-20
Target	Mutations in the *PAH* gene
Sample Material	DNA (in TE Buffer)
Scheme Format	Assessment of genotyping, and biological and clinical interpretation. For each case, participants are expected to return a clinical report which includes a complete interpretation of the results. No restrictions on number of participants. Open to laboratories from ALL countries. Three mock clinical cases with matching samples. (3)
Reporting Language	Reports accepted in English or German.
Additional Information	Suitable for sequence based analysis (e.g. NGS / Sanger Sequencing) and copy number analysis (e.g. MLPA, NGS based CNV analysis).
Performance criteria	Performance criteria **DO** apply.
Accreditation	This scheme is covered by the scope of EMQN's accreditation.
Collaborator	None

TEL: +44 161 276 6741
FAX: +44 161 276 6606
WEB: www.emqn.org
EMAIL: office@emqn.org

Polyposis Syndromes

CANCER | INHERITED | GERMLINE

Scheme code	FAP-20
Target	Mutations in the *APC* and *MUTYH* genes.
Sample Material	DNA (in TE Buffer)
Scheme Format	Assessment of genotyping, and biological and clinical interpretation. For each case, participants are expected to return a clinical report which includes a complete interpretation of the results. No restrictions on number of participants. Open to laboratories from ALL countries. Four mock clinical cases with matching samples. (4)
Reporting Language	Reports accepted in English or German.
Additional Information	Suitable for sequence based analysis (e.g. NGS / Sanger Sequencing) and copy number analysis (e.g. MLPA, NGS based CNV analysis). **Molecular testing for disorders associated with polyposis - Familial adenomatous polyposis (FAP) and MUTYH-associated polyposis (MAP)**
Performance criteria	Performance criteria **DO** apply.
Accreditation	This scheme is covered by the scope of EMQN's accreditation.
Collaborator	None

TEL: +44 161 276 6741 | FAX: +44 161 276 6606 | WEB: www.emqn.org | EMAIL: office@emqn.org

Porphyrias

METABOLIC | INHERITED | GERMLINE

Scheme code	POR-20
Target	Mutations in the most frequently analysed porphyria genes (for example *PPOX, UROS, HMBS*).
Sample Material	DNA (in TE Buffer)
Scheme Format	Assessment of genotyping, and biological and clinical interpretation. For each case, participants are expected to return a clinical report which includes a complete interpretation of the results. No restrictions on number of participants. Open to laboratories from ALL countries. Three mock clinical cases with matching samples. (3)
Reporting Language	Reports accepted in English.
Additional Information	Suitable for sequence based analysis (e.g. NGS / Sanger Sequencing) and copy number analysis (e.g. MLPA, NGS based CNV analysis).
Performance criteria	Performance criteria **DO** apply.
Accreditation	This scheme is covered by the scope of EMQN's accreditation.
Collaborator	None

TEL: +44 161 276 6741
FAX: +44 161 276 6606
WEB: www.emqn.org
EMAIL: office@emqn.org

Prader-Willi and Angelman Syndromes

IMPRINTING INHERITED GERMLINE

Scheme code	PWAS-20
Target	15q11-q13 studies in order to diagnose Prader-Willi or Angelman syndromes
Sample Material	DNA (in TE Buffer)
Scheme Format	Assessment of genotyping, and biological and clinical interpretation. For each case, participants are expected to return a clinical report which includes a complete interpretation of the results. No restrictions on number of participants. Open to laboratories from ALL countries. Three mock clinical cases with matching samples. (3)
Reporting Language	Reports accepted in English or German.
Additional Information	Suitable for MS-MLPA, methylation-specific PCR and Southern blotting techniques. Methylation analysis of PWS / AS critical region; uniparental disomy / deletion analysis.
Performance criteria	Performance criteria **DO** apply.
Accreditation	This scheme is covered by the scope of EMQN's accreditation.
Collaborator	None

TEL: +44 161 276 6741
FAX: +44 161 276 6606
WEB: www.emqn.org
EMAIL: office@emqn.org

Retinoblastoma

CANCER | INHERITED | GERMLINE

Scheme code	RB-20
Target	Mutations in the *RB1* gene
Sample Material	DNA (in TE Buffer)
Scheme Format	Assessment of genotyping, and biological and clinical interpretation. For each case, participants are expected to return a clinical report which includes a complete interpretation of the results. No restrictions on number of participants. Open to laboratories from ALL countries. Three mock clinical cases with matching samples. (3)
Reporting Language	Reports accepted in English or German.
Additional Information	Suitable for sequence based analysis (e.g. NGS / Sanger Sequencing) and copy number analysis (e.g. MLPA, NGS based CNV analysis).
Performance criteria	Performance criteria **DO** apply.
Accreditation	This scheme is covered by the scope of EMQN's accreditation.
Collaborator	None

TEL: +44 161 276 6741
FAX: +44 161 276 6606
WEB: www.emqn.org
EMAIL: office@emqn.org

RYR1 related Myopathies and Malignant Hyperthermia susceptibility

INHERITED GERMLINE

Scheme code	RYR1-20
Target	Mutations in the *RYR1* gene
Sample Material	DNA (in TE Buffer)
Scheme Format	Assessment of genotyping, and biological and clinical interpretation. For each case, participants are expected to return a clinical report which includes a complete interpretation of the results. No restrictions on number of participants. Open to laboratories from ALL countries. Three mock clinical cases with matching samples. (3)
Reporting Language	Reports accepted in English or German.
Additional Information	Suitable for sequence based analysis (e.g. NGS / Sanger Sequencing) and copy number analysis (e.g. MLPA, NGS based CNV analysis).
Performance criteria	Performance criteria **DO** apply.
Accreditation	This scheme is covered by the scope of EMQN's accreditation.
Collaborator	None

TEL: +44 161 276 6741
FAX: +44 161 276 6606
WEB: www.emqn.org
EMAIL: office@emqn.org

Severe Combined Immunodeficiency (pilot)

PANEL NEW

INHERITED GERMLINE

Scheme code	SCID-20
Target	Variants in different genes relevant to SCID, for example *RAG1/2*, *ADA*, and *DCLRE1C*. The genes included in the scheme will vary each year.
Sample Material	DNA (in TE Buffer)
Scheme Format	Assessment of genotyping, and biological and clinical interpretation. For each case, participants are expected to return a clinical report which includes a complete interpretation of the results. Restrictions on number of participants. (30) Open to laboratories from ALL countries. Three mock clinical cases with matching samples. (3) Reports will be assessed in the context of the panel performed.
Reporting Language	Reports accepted in English.
Additional Information	Suitable for sequence based analysis (e.g. NGS / Sanger Sequencing). Only clinically relevant variants* should be reported. *Only variants that have been validated in the test samples will be assessed
Performance criteria	⊗ Performance criteria **DO** NOT apply.
Accreditation	⊗ This scheme is NOT covered by the scope of EMQN's accreditation.
Collaborator	European Reference Network - Rare Immunodeficiency, Autoinflammatory and Autoimmune diseases ERN-RITA (http://rita.ern-net.eu/)

TEL: +44 161 276 6741
FAX: +44 161 276 6606
WEB: www.emqn.org
EMAIL: office@emqn.org

Spinal Muscular Atrophy

NEUROLOGICAL DISEASE | INHERITED | GERMLINE

Scheme code	SMA-20
Target	Mutations in the *SMN1* gene
Sample Material	DNA (in TE Buffer)
Scheme Format	Assessment of genotyping, and biological and clinical interpretation. For each case, participants are expected to return a clinical report which includes a complete interpretation of the results. No restrictions on number of participants. Open to laboratories from ALL countries. Three mock clinical cases with matching samples. (3)
Reporting Language	Reports accepted in English, Dutch or German.
Additional Information	Suitable for copy number analysis of the *SMN1*/(*SMN2*) gene(s) but if labs do point mutation of testing then we expect them to report it.
Performance criteria	Performance criteria **DO** apply.
Accreditation	This scheme is covered by the scope of EMQN's accreditation.
Collaborator	None

TEL: +44 161 276 6741
FAX: +44 161 276 6606
WEB: www.emqn.org
EMAIL: office@emqn.org

Spinocerebellar Ataxia's

NEUROLOGICAL DISEASE | TRINUCELOTIDE REPEAT DISORDER | INHERITED | GERMLINE

Scheme code	SCA-20
Target	Triplet repeat analysis of mutations in the *ATXN1*, *ATXN2*, *ATXN3*, *CACNA1A* and *ATXN7* genes.
Sample Material	DNA (in TE Buffer)
Scheme Format	Assessment of genotyping, and biological and clinical interpretation. For each case, participants are expected to return a clinical report which includes a complete interpretation of the results. No restrictions on number of participants. Open to laboratories from ALL countries. Three mock clinical cases with matching samples. (3)
Reporting Language	Reports accepted in English, French, German, or Dutch.
Additional Information	Suitable for PCR-based techniques ONLY. CAG repeat analysis ONLY.
Performance criteria	✓ Performance criteria **DO** apply.
Accreditation	☆ This scheme is covered by the scope of EMQN's accreditation.
Collaborator	— None

TEL: +44 161 276 6741
FAX: +44 161 276 6606
WEB: www.emqn.org
EMAIL: office@emqn.org

Stickler Syndrome

DYSMORPHOLOGY | INHERITED | GERMLINE

PANEL

Scheme Code	STICKLER-20
Target	Mutations in the collagen genes *COL2A1*, *COL11A1*, *COL11A2*, *COL9A1*, *COL9A2*.
Sample Material	DNA (in TE Buffer)
Scheme Format	Assessment of genotyping, and biological and clinical interpretation. For each case, participants are expected to return a clinical report which includes a complete interpretation of the results. No restrictions on number of participants. Open to laboratories from ALL countries. Three mock clinical cases with matching samples. (3) Reports will be marked in the context of the panel performed
Reporting Language	Reports accepted in English.
Additional Information	Suitable for sequence based analysis (e.g. NGS / Sanger Sequencing) and copy number variation (e.g. MLPA, NGS based CNV analysis). Only clinically relevant variants* should be reported. *Only variants that have been validated in the test samples will be assessed.
Performance criteria	✓ Performance criteria **DO** apply.
Accreditation	☆ This scheme is covered by the scope of EMQN's accreditation.
Collaborator	⊖ None

TEL: +44 161 276 6741
FAX: +44 161 276 6606
WEB: www.emqn.org
EMAIL: office@emqn.org

Von Hippel Lindau Syndrome

CANCER | INHERITED | GERMLINE

Scheme code	VHL-20
Target	Mutations in the *VHL* gene
Sample Material	DNA (in TE Buffer)
Scheme Format	Assessment of genotyping, and biological and clinical interpretation. For each case, participants are expected to return a clinical report which includes a complete interpretation of the results. No restrictions on number of participants. Open to laboratories from ALL countries. Three mock clinical cases with matching samples. (3)
Reporting Language	Reports accepted in English or German.
Additional Information	Suitable for sequence based analysis (e.g. NGS / Sanger Sequencing) and copy number analysis (e.g. MLPA, NGS based CNV analysis).
Performance criteria	Performance criteria **DO** apply.
Accreditation	This scheme is covered by the scope of EMQN's accreditation.
Collaborator	None

TEL: +44 161 276 6741
FAX: +44 161 276 6606
WEB: www.emqn.org
EMAIL: office@emqn.org

Wilson Disease

CANCER | INHERITED | GERMLINE

Scheme code	WIL-20
Target	Mutations in the *ATP7B* gene
Sample Material	DNA (in TE Buffer)
Scheme Format	Assessment of genotyping, and biological and clinical interpretation. For each case, participants are expected to return a clinical report which includes a complete interpretation of the results. No restrictions on number of participants. Open to laboratories from ALL countries. Three mock clinical cases with matching samples. (3)
Reporting Language	Reports accepted in English.
Additional Information	Suitable for sequence based analysis (e.g. NGS / Sanger Sequencing) and copy number analysis (e.g. MLPA, NGS based CNV analysis).
Performance criteria	Performance criteria **DO** apply.
Accreditation	This scheme is covered by the scope of EMQN's accreditation.
Collaborator	None

TEL: +44 161 276 6741
FAX: +44 161 276 6606
WEB: www.emqn.org
EMAIL: office@emqn.org

Y-Chromosome Microdeletions

ANDROLOGY | INHERITED | GERMLINE

Scheme code	AZF-20
Target	Microdeletions of the Y-Chromosome
Sample Material	DNA (in TE Buffer)
Scheme Format	Assessment of genotyping, and biological and clinical interpretation. For each case, participants are expected to return a clinical report which includes a complete interpretation of the results. No restrictions on number of participants. Open to laboratories from ALL countries. Three mock clinical cases with matching samples. (3)
Reporting Language	Reports accepted in English, French, German or Italian.
Additional Information	Suitable for copy number analysis (e.g. STS analysis).
Performance criteria	Performance criteria **DO** apply.
Accreditation	This scheme is covered by the scope of EMQN's accreditation.
Collaborator	European Academy of Andrology (http://www.andrologyacademy.net/)

TEL: +44 161 276 6741
FAX: +44 161 276 6606
WEB: www.emqn.org
EMAIL: office@emqn.org

Somatic mutation testing EQA schemes

Nine EQA schemes are provided in 2020. These EQAs require FFPE or Plasma samples to be genotyped and full interpretative reports to be submitted (exceptions may apply – see each scheme for details).

We collaborate with other organizations to provide some of these EQA schemes. This is clearly shown for each EQA scheme. Please see our website for more information.

TEL: +44 161 276 6741
FAX: +44 161 276 6606
WEB: www.emqn.org
EMAIL: office@emqn.org

Molecular testing for EGFR gene mutations in cell free DNA (cfDNA)

CANCER | ACQUIRED | SOMATIC | PLASMA

Scheme code	LIQUIDBIOPSY(EGFR)-20
Target	Mutations in the *EGFR* gene (see information below on scheme format).
Sample Material	Artificial plasma containing cfDNA
Scheme Format	Assessment of genotyping, and biological and clinical interpretation. For each case, participants are expected to return a clinical report which includes a complete interpretation of the results. No restrictions on number of participants. Open to laboratories from ALL countries. 3 mock clinical cases with matching samples. (3)
Reporting Language	Reports accepted in English.
Additional Information	**NOTE: This scheme has not been scheduled (at the time of going to press) but EMQN is working with its collaborators and IQNPath to provide it in 2020. EMQN member laboratories will be informed of developments, timeframes and costs once this has been established. If you have any questions, then please contact the EMQN office.**
Performance criteria	✓ Performance criteria **DO** apply..
Accreditation	⊗ This scheme is NOT covered by the scope of EMQN's accreditation.
Collaborator	IQNPath (www.iqnpath.org) Liquid Biopsy working group

TEL: +44 161 276 6741
FAX: +44 161 276 6606
WEB: www.emqn.org
EMAIL: office@emqn.org

Microsatellite Instability testing (pilot)

CANCER | ACQUIRED | SOMATIC

NEW

Scheme code	MSI-20
Target	Microsatellite Instability testing.
Sample Material	Artificial paraffin embedded (FFPE) materials designed to simulate realpatient samples. Matched normal materials are **not** provided. NOTE: Samples provided as cut sections (rolled scrolls) only - we cannot provide slide mounted materials.
Scheme Format	Assessment of genotyping only. For each case, however, participants are expected to return a clinical report which includes a complete biological and clinical interpretation of the genotype (for information only, interpretation is not assessed). Restricted to 30 laboratories. (30) Open to laboratories from ALL countries. 3 mock clinical cases with matching samples. (3)
Reporting Language	Reports accepted in English.
Additional Information	One distribution per year in June. Ten weeks are given for testing and reporting. EMQN will select participants from the list of applicants via an online survey (applicants will be contacted by the EMQN office).
Performance criteria	⊗ Performance criteria **DO NOT** apply.
Accreditation	⊗ This scheme is **NOT** covered by the scope of EMQN's accreditation.
Collaborator	⊖ None

TEL: +44 161 276 6741
FAX: +44 161 276 6606
WEB: www.emqn.org
EMAIL: office@emqn.org

Molecular testing in Lung Cancer (NSCLC)

CANCER | ACQUIRED | SOMATIC

Scheme code	NSCLC-20
Target	Mutations in the *EGFR*, *KRAS* and *BRAF* genes.
Sample Material	Mix of real tumour tissue and artificial paraffin embedded (FFPE) materials designed to simulate real patient samples. **NOTE:** Samples provided as cut sections (rolled scrolls) only - we cannot provide slide mounted materials.
Scheme Format	Assessment of genotyping, and biological and clinical interpretation. For cases 1 and 2, participants are expected to return a clinical report which includes a complete interpretation of the results. The remaining 8 cases require genotyping-only. **The minimum requirement is *EGFR*. If you provide a clinical service for *BRAF* (p.V600E only) gene testing or *KRAS* genes, these should be included in the interpretation of results. The testing of other genes is optional. Only variants that have been validated in the test samples will be assessed.** For *EGFR* testing, labs should expect mutations in all 4 Tyrosine Kinase (TK) domain exons (for example, but not exclusively p.Gly719Ser, p.Thr790Met, deletions in exon 19, and p.Leu858Arg). We only require clinically relevant pathogenic (disease-causing) mutations to be reported, and not common SNPs / variants. No restrictions on number of participants. Open to laboratories from ALL countries. 10 mock clinical cases with matching samples. (10)

TEL: +44 161 276 6741
FAX: +44 161 276 6606
WEB: www.emqn.org
EMAIL: office@emqn.org

Molecular testing in Lung Cancer (NSCLC)

CANCER | ACQUIRED | SOMATIC

Reporting Language	Reports accepted in English.
Additional Information	Mutation allelic frequencies of the artificial FFPE materials have been validated using ddPCR. **We would like to thank AstraZeneca for the educational grant to deliver this EQA.** Consequently, the cost of this scheme has been discounted (from £445) and shows as £0.00 when users purchase it via their EMQN website account.
Performance criteria	Performance criteria **DO** apply.
Accreditation	This scheme is covered by the scope of EMQN's accreditation.
Collaborator	None

TEL: +44 161 276 6741
FAX: +44 161 276 6606
WEB: www.emqn.org
EMAIL: office@emqn.org

Molecular testing in Melanoma

CANCER | ACQUIRED | SOMATIC | SKIN

Scheme code	MELANOMA-20
Target	Mutations in the *BRAF, NRAS, KIT* genes.
Sample Material	Mix of real tumour tissue and artificial paraffin embedded (FFPE) materials designed to simulate real patient samples. **NOTE:** Samples provided as cut sections (rolled scrolls) only - we cannot provide slide mounted materials.
Scheme Format	Assessment of genotyping, and biological and clinical interpretation. For cases 1 and 2, participants are expected to return a clinical report which includes a complete interpretation of the results. The remaining 8 cases require genotyping only. **The minimum requirement is *BRAF* gene testing. If you provide a clinical service for the *NRAS* and *KIT* gene, these should be included in the interpretation of results. The testing of other genes is optional.** We only require clinically relevant pathogenic (disease-causing) mutations to be reported, and not common SNPs / variants. No restrictions on number of participants. Open to laboratories from ALL countries. 10 mock clinical cases with matching samples. (10)
Reporting Language	Reports accepted in English.
Additional Information	Mutation allelic frequencies of the artificial FFPE materials have been validated using ddPCR.
Performance criteria	✓ Performance criteria **DO** apply.
Accreditation	☆ This scheme is covered by the scope of EMQN's accreditation.
Collaborator	⊖ None

TEL: +44 161 276 6741
FAX: +44 161 276 6606
WEB: www.emqn.org
EMAIL: office@emqn.org

Molecular testing in sporadic Colorectal Cancer

CANCER | ACQUIRED | SOMATIC

Scheme code	COLOREC-20
Target	Mutations in the *KRAS, BRAF, NRAS* and *PIK3CA* genes.
Sample Material	Mix of real tumour tissue and artificial paraffin embedded (FFPE) materials designed to simulate real patient samples. NOTE: Samples provided as cut sections (rolled scrolls) only - we cannot provide slide mounted materials.
Scheme Format	Assessment of genotyping, and biological and clinical interpretation. For cases 1 and 2, participants are expected to return a clinical report which includes a complete interpretation of the results. The remaining 8 cases require genotyping only. **The minimum requirement is *KRAS, NRAS* and *BRAF* gene testing. If you provide a clinical service for the *PIK3CA* gene, this should be included. The testing of other genes is optional.** We only require clinically relevant pathogenic (disease-causing) mutations to be reported, and not common SNPs / variants. No restrictions on number of participants. Open to laboratories from ALL countries. 10 mock clinical cases with matching samples. (10)
Reporting Language	Reports accepted in English.
Additional Information	Mutation allelic frequencies of the artificial FFPE materials have been validated using ddPCR.
Performance criteria	✓ Performance criteria **DO** apply.
Accreditation	☆ This scheme is covered by the scope of EMQN's accreditation.
Collaborator	⊖ None

TEL: +44 161 276 6741
FAX: +44 161 276 6606
WEB: www.emqn.org
EMAIL: office@emqn.org

Molecular testing of BRCA genes in Ovarian Cancer (vGermline)

CANCER | INHERITED | GERMLINE

Scheme code	OVARIAN-20 (G)
Target	Mutations in the *BRCA1* and *BRCA2* genes
Sample Material	DNA (in TE buffer).
Scheme Format	Assessment of genotyping, and biological and clinical interpretation (***BRCA1* and *BRCA2* mutation testing within the context of targeted PARP inhibitor treatment**). For each case, participants are expected to return a clinical report which includes a complete interpretation of the results. No restrictions on number of participants. Open to laboratories from ALL countries. 3 mock clinical cases with matching samples. (3)
Reporting Language	Reports accepted in English.
Additional Information	**We would like to thank AstraZeneca for their educational grant to deliver this EQA.** Consequently, the cost of this scheme has been discounted (from £300) and shows as £0.00 when users purchase it via their EMQN website account.
Performance criteria	Performance criteria **DO** apply.
Accreditation	This scheme is covered by the scope of EMQN's accreditation.
Collaborator	Genomics Quality Assessment (GenQA)

TEL: +44 161 276 6741
FAX: +44 161 276 6606
WEB: www.emqn.org
EMAIL: office@emqn.org

Molecular testing of BRCA genes in Ovarian Cancer (vSomatic)

CANCER | ACQUIRED | SOMATIC

Scheme code	OVARIAN-20 (S)
Target	Mutations in the *BRCA1* and *BRCA2* genes (see information below on scheme format).
Sample Material	Artificial paraffin embedded (FFPE) materials designed to simulate a real patient sample. **NOTE:** Samples provided a as cut sections (rolled scrolls) ONLY - we cannot provide slide mounted materials.
Scheme Format	Assessment of genotyping, and biological and clinical interpretation (***BRCA1* and *BRCA2* mutation testing within the context of targeted PARP inhibitor treatment**). For each case, participants are expected to return a clinical report which includes a complete interpretation of the results. No restrictions on number of participants. Open to laboratories from ALL countries. 3 mock clinical cases with matching samples. (3)
Reporting Language	Reports accepted in English.
Additional Information	**We would like to thank AstraZeneca for their educational grant to deliver this EQA.** Consequently, the cost of this scheme has been discounted (from £300) and shows as £0.00 when users purchase it via their EMQN website account.
Performance criteria	Performance criteria **DO** apply.
Accreditation	This scheme is covered by the scope of EMQN's accreditation.
Collaborator	Genomics Quality Assessment (GenQA)

TEL: +44 161 276 6741 | FAX: +44 161 276 6606 | WEB: www.emqn.org | EMAIL: office@emqn.org

Oncogene panel testing

CANCER | AQUIRED | SOMATIC | NGS

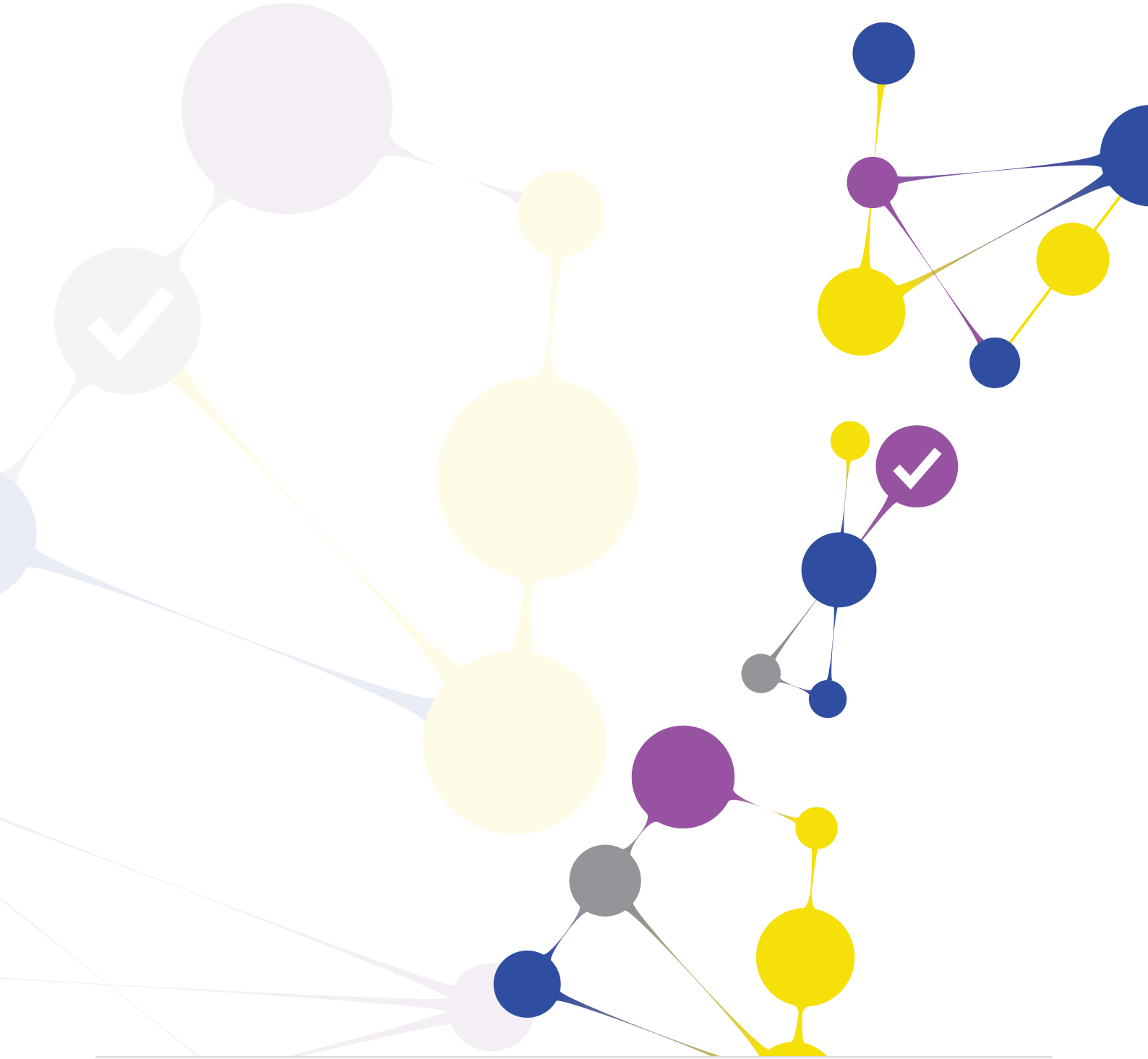
Scheme code	ONCOPANEL-20
Target	Mutations in the *EGFR*, *PIK3CA*, *KRAS*, *HRAS*, *NRAS*, *KIT*, *TP53* and *BRAF* genes.
Sample Material	Rolled sections of artificial formalin fixed paraffin embedded (FFPE) materials designed to simulate real patient samples. **NOTE:** Samples provided a as cut sections (rolled scrolls) only - we cannot provide slide mounted materials.
Scheme Format	Assessment of genotyping ONLY. Labs will be requested to provide information on which genes and mutations the samples were tested for. Therefore, testing for all of the genes shown above is NOT required. We only require clinically relevant pathogenic (disease-causing) mutations to be reported, and not common SNPs / variants. Justification for the inclusion of reported variants will be requested when reporting results to EMQN. No restrictions on number of participants. Open to laboratories from ALL countries. 3 mock clinical cases with matching samples (3)
Reporting Language	Not applicable - this is a genotyping only scheme
Additional Information	This scheme is being offered to help labs using high through put technologies (e.g., NGS, MassArray etc) to accurately validate assay sensitivity and specificity. For specific tumour types, please register for the relevant Lung, Melanoma or Colorectal scheme. High quality reference materials are provided covering a range of genes with ddPCR quantified allelic frequencies.
Performance criteria	✓ Performance criteria **DO** apply.
Accreditation	☆ This scheme is covered by the scope of EMQN's accreditation.
Collaborator	⊖ None

TEL: +44 161 276 6741
FAX: +44 161 276 6606
WEB: www.emqn.org
EMAIL: office@emqn.org

Pharmacogenetics EQA Schemes

One EQA scheme is provided in 2020. This EQA requires DNA samples to be genotyped and full interpretative reports to be submitted (exceptions may apply – see scheme for details).

We sometimes collaborate with other organizations to provide some of these EQA schemes. This is clearly shown for each EQA scheme. Please see our website for more information.



TEL: +44 161 276 6741
FAX: +44 161 276 6606
WEB: www.emqn.org
EMAIL: office@emqn.org

Pharmacogenetics (drug intolerance and effectivity)

INHERITED | GERMLINE | PHARMACOGENETIC

PANEL

Scheme Code	PHARMACO-20
Target	A panel of 45 clinically relevant variants in the *CYP2B6, CYP2C9, CYP2C19, CYP2D6, CYP3A5, DPYD, fVl, HLA-B, SLCO1B1, TPMT, UGT1A1* and *VKORC1* genes.
Sample Material	DNA (in TE Buffer)
Scheme Format	Assessment of genotyping, and biological and clinical interpretation. For each case, participants are expected to return a clinical report which includes a complete interpretation of the results. No restrictions on number of participants. Open to laboratories from ALL countries. Three mock clinical cases with matching samples. (3)
Reporting Language	Reports accepted in English or German.
Additional Information	Suitable for sequence based analysis (e.g. NGS / Sanger Sequencing) and copy number variation (e.g. MLPA, NGS based CNV analysis).
Performance criteria	⊗ Performance criteria **DO** NOT apply.
Accreditation	⊗ This scheme is NOT covered by the scope of EMQN's accreditation.
Collaborator	⊖ None

TEL: +44 161 276 6741
FAX: +44 161 276 6606
WEB: www.emqn.org
EMAIL: office@emqn.org

Technique-specific EQA Schemes

Six EQA schemes are provided in 2020. These EQAs require FFPE, Plasma or DNA samples to be genotyped and full interpretative reports to be submitted (exceptions may apply – see each scheme for details).

We collaborate with other organizations to provide some of these EQA schemes. This is clearly shown for each EQA scheme. Please see our website for more information.

TEL: +44 161 276 6741
FAX: +44 161 276 6606
WEB: www.emqn.org
EMAIL: office@emqn.org

DNA Sequencing - Sanger

SEQUENCING TECHNICAL

Scheme code	SEQ-20-SANGER
Target	Sanger DNA sequencing (gene independent)
Sample Material	DNA (in TE Buffer)
Scheme Format	**Assessment of genotyping, diagnostic interpretation, mutation nomenclature and quality of raw data.** Laboratories which do not genotype can opt out of submitting this data at the results submission stage. Wet lab based exercise distributing g.DNA samples for testing. No restrictions on number of participants. Open to laboratories from ALL countries. 4 cases with matching samples. (4)
Reporting Language	Not applicable - genotyping only EQA.
Additional Information	Suitable for Sanger sequencing technology ONLY.
Performance criteria	Performance criteria **DO** apply.
Accreditation	This scheme is covered by the scope of EMQN's accreditation.
Collaborator	None

TEL: +44 161 276 6741
FAX: +44 161 276 6606
WEB: www.emqn.org
EMAIL: office@emqn.org

DNA Sequencing NGS (vGermline) (pilot)

SEQUENCING | TECHNICAL | INHERITED | GERMLINE | NGS

Scheme code	NEXTGEN (G) -20
Target	Any NGS strategy can be used (single gene, panel testing, exome or genome sequencing) - the laboratory can choose. **The scheme will assess all variants 50bp or less in size.** Larger (>50bp in size) changes including copy number and strutural variants (CNV's, SV's) are covered in a pilot EQA currently in development (please contact office@emqn.org for more information).
Sample Material	g.DNA (in TE Buffer)
Scheme Format	Assessment of genotyping and quality of raw data. **Designed specifically for labs doing NGS based GERMLINE mutation testing ONLY.** Labs doing somatic mutation testing should register for the separate SOMATIC version of the scheme. Wet lab based exercise distributing g.DNA samples for testing. No restrictions on number of participants. Open to laboratories from ALL countries. 1 mock clinical cases with matching samples. (1)
Reporting Language	Not applicable - genotyping only EQA
Additional Information	**EQA scheme allows upto 3 independent analyses to be submitted** (for example a panel, exome and genome test). Laboratories receive comprehensive feedback on variant calling and benchmarking quality of NGS raw data (17 different metrics). Analysis of VCF, BED, BAM and FASTQ files
Performance criteria	⊗ Performance criteria **DO NOT** apply.
Accreditation	⊗ This scheme is **NOT** covered by the scope of EMQN's accreditation.
Collaborator	Genomics Quality Assessment (GenQA)

TEL: +44 161 276 6741
FAX: +44 161 276 6606
WEB: www.emqn.org
EMAIL: office@emqn.org

DNA Sequencing NGS (vSomatic) (pilot)

SEQUENCING | TECHNICAL | AQUIRED | SOMATIC | NGS

Scheme code	NEXTGEN (S) -20
Target	Any NGS strategy can be used (single gene, panel testing, exome or genome sequencing). The laboratory can choose.
Sample Material	g.DNA sample derived from FFPE material. Matching control g.DNA sample (from FFPE) also included.
Scheme Format	Assessment of genotyping and quality of raw data. **Designed specifically for labs doing NGS based SOMATIC mutation testing ONLY.** Labs doing germline mutation testing should register for the separate GERMLINE version of the scheme. Wet lab based exercise distributing DNA samples for testing No restrictions on number of participants. Open to laboratories from ALL countries. 1 mock clinical cases with matching samples. (1)
Reporting Language	Not applicable – genotyping only EQA
Additional Information	**EQA scheme allows upto 3 independent analyses to be submitted** (for example a panel, exome and genome test). Laboratories receive comprehensive feedback on variant calling and benchmarking quality of NGS raw data (17 different metrics). Analysis of VCF, BED, BAM and FASTQ files
Performance criteria	⊗ Performance criteria **DO NOT** apply.
Accreditation	⊗ This scheme is **NOT** covered by the scope of EMQN's accreditation.
Collaborator	Genomics Quality Assessment (GenQA)

TEL: +44 161 276 6741
FAX: +44 161 276 6606
WEB: www.emqn.org
EMAIL: office@emqn.org

Postnatal Constitutional CNV Detection [array/NGS]

INTERLECTUAL DISABILITY | TECHNICAL | INHERITED | GERMLINE

Scheme code	aCGH-20
Target	Genomic deletions and duplications
Sample Material	DNA samples (in TE Buffer)
Scheme Format	Assessment of genotyping, and biological and clinical interpretation. For each case, participants are expected to return a clinical report which includes a complete interpretation of the results. No restrictions on number of participants. Open to laboratories from ALL countries. 2 mock clinical cases with matching samples. (2)
Reporting Language	Reports accepted in English ONLY.
Additional Information	Platform independent - participants use their normal methodology. **Applicable to labs using either array or NGS based technologies to detect large scale genomic / structural changes and CNV.**
Performance criteria	Performance criteria **DO** apply.
Accreditation	This scheme is covered by the scope of EMQN's accreditation.
Collaborator	Genomics Quality Assessment (GenQA)

TEL: +44 161 276 6741
FAX: +44 161 276 6606
WEB: www.emqn.org
EMAIL: office@emqn.org

Non-invasive prenatal testing (NIPT) for common aneuploidies (including sex chromosomes)

SCREENING | PLASMA | NGS | TECHNICAL | ACQUIRED

Scheme code	NIPT(ANEUPLOIDY)-20
Target	NIPT for the 3 most common aneuploidies (Chr 13, 18 and 21). Will include an optional element for the testing and reporting of fetal sex and sex chromosome aneuploidies (in recognition that this extension to testing is offered by many laboratories as part of their clinical service).
Sample Material	Plasma sample(s) containing cffDNA
Scheme Format	Assessment of genotyping, and biological and clinical interpretation. For each case, participants are expected to return a clinical report which includes a complete interpretation of the results. No restrictions on number of participants. Open to laboratories from ALL countries. 3 mock clinical cases with matching samples. (3)
Reporting Language	Reports accepted in English ONLY
Additional Information	None
Performance criteria	✓ Performance criteria **DO** apply.
Accreditation	✗ This scheme is **NOT** covered by the scope of EMQN's accreditation.
Collaborator	Genomics Quality Assessment (GenQA)

TEL: +44 161 276 6741
FAX: +44 161 276 6606
WEB: www.emqn.org
EMAIL: office@emqn.org

Non-invasive prenatal testing (NIPT) for fetal sexing (X-linked disorders)

INHERITED | NGS | PLASMA | SEQUENCING | TECHNICAL

Scheme code	NIPT(SEXING)-20
Target	NIPT for the fetal sex determination in the context of X-linked disorders, which often require testing at an early gestational age.
Sample Material	Plasma sample(s) containing cffDNA
Scheme Format	Assessment of genotyping, and biological and clinical interpretation. For each case, participants are expected to return a clinical report which includes a complete interpretation of the results. No restrictions on number of participants. Open to laboratories from ALL countries. 3 mock clinical cases with matching samples. (3)
Reporting Language	Reports accepted in English ONLY.
Additional Information	None.
Performance criteria	✓ Performance criteria **DO** apply.
Accreditation	✗ This scheme is **NOT** covered by the scope of EMQN's accreditation.
Collaborator	Genomics Quality Assessment (GenQA)

TEL: +44 161 276 6741
FAX: +44 161 276 6606
WEB: www.emqn.org
EMAIL: office@emqn.org

Interlaboratory Comparison (Sample Exchanges)

Three ILC programmes are currently offered (see pages 69-71). However, we will facilitate additional programmes when there is sufficient demand. If you are interested, please complete our "Expression of interest form".

Introduction

Laboratory accreditation standards (for example ISO 17025, ISO15189) mandate that laboratories should participate in EQA schemes (where they exist). If no EQA scheme is available, then the standards require laboratories to participate in interlaboratory comparisons or sample exchanges.

Interlaboratory comparisons (or sample exchanges) between laboratories are a method of monitoring laboratory performance which are suitable for accreditation purposes in certain defined circumstances, for example tests on very rare diseases where there are a small number of labs perform testing, and there are no EQA schemes available. EMQN will provide support for Interlaboratory Comparisons (ILCs) in accordance with EA-4/21 INF: 2018 Guidelines for the assessment of the appropriateness of small interlaboratory comparisons (ILC) within the process of laboratory accreditation. EMQN will facilitate the exchange of materials between laboratories for the ILC, will assess the genotyping results, and will provide a report summarising the results for the ILC.

ILC Proposals and registration

Please submit an Expression of interest form if you would like to propose an interlaboratory comparison study. EMQN will accept a minimum of 3, and a maximum of 7 laboratories for an ILC study. Once sufficient participants register interest, EMQN will contact you to arrange registration. **All ILC studies will be genotyping-only – no assessment of clinical interpretation will be made.**

Cost of particpation

There is an annual fee of £200 to participate per ILC study, and EMQN membership is required. Participating laboratories must commit for a minimum of 3 years (total cost of £600).

TEL: +44 161 276 6741
FAX: +44 161 276 6606
WEB: www.emqn.org
EMAIL: office@emqn.org

Samples

Participating laboratories will contribute DNA samples from clinical cases to EMQN for the ILC study, along with information related to your laboratories' accreditation and the methods used to test the samples.

EMQN will perform basic QC of the DNA sample (to assess the amount of dsDNA and DNA quality), anonymise the materials, and co-ordinate distribution to participating laboratories with matching clinical referral information.

All participanting laboratories will be asked to send between 2 or 3 samples for testing over the 3 year period. EMQN will request sufficient DNA to provide approximately 2 µg per participant (8-16µg per sample; see table on page 3 for estimations of the amount of DNA to be provided). The actual amount of DNA required will be specific for each particular disease; if there is sufficient material, the same samples will be sent to all participants in the interlaboratory comparison study. If this is not possible, sample exchanges will be co-ordinated between smaller groups of laboratories. Please note that if samples are distributed to all participants, you will test at least one of your own samples (anonymised) during the 3 year period.

Number participating labs	Minimum number samples to be provided by each participant over 3 years	Minimum quantity of DNA (ug) per sample to provide DNA to all participants (2 ug per particpant[1])
3	3	8 µg
4	3	10 µg
5	2	12 µg
6	2	14 µg
7	2	16 µg

[1] An additional 2ug of DNA is requested for EMQN quality control of each material, and in the event of need for independent sample validation (if required).

Timelines and instructions

EMQN will provide a handbook for participants with more detailed guidance about our ILC studies.

We will notify laboratories of the timelines for the ILC study; each round will be scheduled to take place within a calendar year.

Results submission

Results will be submitted to EMQN via an online form. Participants will be asked to include the mock patient identifiers, the sample genotype with basic classification of pathogenicity (eg. class 5 pathogenic, class 4 likely pathogenic, class 3 UV), and the relevant reference sequences.

Assessment and certificates

There will be NO poor performance criteria applied to ILC studies. The results will be evaluated by EMQN for accuracy of patient identifiers, as well as the genotype and classification of pathogenicity, which will be assessed in comparison to the consensus result amongst the participating laboratories.

If the consensus results are discordant, EMQN will mediate discussions to resolve the discrepant results. If necessary, EMQN can send a sample for validation by NGS, and an additional fee will be charged to each participating labs to cover the costs of independent sample validation.

A summary report on the results of the ILC study, as well as your individual scores will be issued after the completion of the assessment period and these documents will be available via your EMQN website account.

A certificate of participation will be available following completion of the appeals process, shortly after the final results have been released to the laboratories.

Further information

If you have any questions, and would like to discuss ILC studies further, please contact the EMQN team at office@emqn.org.

TEL: +44 161 276 6741
FAX: +44 161 276 6606
WEB: www.emqn.org
EMAIL: office@emqn.org

Achondroplasia (FGFR3) - ILC

INHERITED | ILC | SAMPLE EXCHANGE

Scheme code	ACH(ILC)-20
Target	*FGFR3* testing for Achondroplasia
Scheme Format	Assessment of genotyping only. Restrictions on number of participants. (7) Open to laboratories from ALL countries. Variable - maximum of 3 samples.
Reporting Language	Reports not required.
Additional Information	None.
Performance criteria	⊗ Performance criteria **DO** NOT apply.
Accreditation	⊗ This scheme is **NOT** covered by the scope of EMQN's accreditation.
Collaborator	⊖ None

TEL: +44 161 276 6741 | FAX: +44 161 276 6606 | WEB: www.emqn.org | EMAIL: office@emqn.org

Congenital Hyperventilation Syndrome (PHOX2B) - ILC

INHERITED | ILC | SAMPLE EXCHANGE

Scheme code	PHOX2B(ILC)-20
Target	*PHOX2B* testing for Congenital Hyperventilation Syndrome
Scheme Format	Assessment of genotyping only. Restrictions on number of participants. (7) Open to laboratories from ALL countries. Variable - maximum of 3 samples.
Reporting Language	Reports not required.
Additional Information	None.
Performance criteria	⊗ Performance criteria **DO** NOT apply.
Accreditation	⊗ This scheme is **NOT** covered by the scope of EMQN's accreditation.
Collaborator	⊖ None

TEL: +44 161 276 6741 | FAX: +44 161 276 6606 | WEB: www.emqn.org | EMAIL: office@emqn.org

Hereditary Cancer Panel Testing - ILC

INHERITED | ILC | SAMPLE EXCHANGE

Scheme code	HCANCER(ILC)-20
Target	Hereditary cancer panel testing.
Scheme Format	Assessment of genotyping only. Restrictions on number of participants. (7) Open to laboratories from ALL countries. Variable - maximum of 3 samples.
Reporting Language	Reports not required.
Additional Information	None.
Performance criteria	⊗ Performance criteria **DO** NOT apply.
Accreditation	⊗ This scheme is **NOT** covered by the scope of EMQN's accreditation.
Collaborator	⊖ None

TEL: +44 161 276 6741
FAX: +44 161 276 6606
WEB: www.emqn.org
EMAIL: office@emqn.org

How to participate

The website plays an important part in the operation of EMQN. The web address is www.emqn.org

To participate in our EQA schemes you need to be a registered member of EMQN. There is a fee for this which is payable EVERY year.

Registered members of EMQN get an account for their laboratory on the EMQN website. The account allows users to:

- Register for, and participate in, our EQA schemes,
- Manage your EQA schemes, return EQA results, view EQA scheme status, review EQA performance from previous years,
- View and download your EQA reports (and past EQA reports),
- Check all scheme purchases,
- Download copies of certificates of participation,
- Add/delete additional staff members,
- Update contact information.

To register to participate in our EQA schemes:

- Log into your account (www.emqn.org).
- Click the "Purchase" button to select the scheme(s) in which you wish to participate.
- Please check that your laboratory details are up to date to ensure the smooth running of the scheme.
- Each laboratory will be charged an annual membership fee of £150 (invoiced automatically). A laboratory user account can have upto 10 registered users. Options are available to add more users if required.
- If you register to participate in a scheme, we assume that your laboratory participates in that scheme. Thus a registration fee has to be paid, independently whether you submit results or not.

TEL: +44 161 276 6741
FAX: +44 161 276 6606
WEB: www.emqn.org
EMAIL: office@emqn.org

EQA 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. We therefore offer a number of options to help laboratories manage the costs of EQA participation and EMQN membership. Examples include:

- Discounted participation in our EQA activities for laboratories in developing / evolving economies (see https://www.emqn.org/participating-in-eqa/discounted-costs-participation/ for more information).
- A single membership fee for a laboratory allowing up to 10 users to be registered with EMQN, with the option to buy additional quota of membership. **Note:** this facility will go live at the end of October 2019.

Where distributor arrangements exist, then their 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/).

Membership	ANNUAL Fee (GBP, £)
All laboratories (up to 10 users per laboratory account)	150
Additional 5 users (multiples of 5, payable annually)	50

Germline mutation testing EQA schemes	ANNUAL Fee (GBP, £)	
Autosomal Dominant Polycystic Kidney disease	300	✪
Beckwith-Wiedemann and Silver-Russell syndromes	300	✪
Cardiac Arrhythmias	300	✪
Charcot-Marie-Tooth disease / Hereditary Neuropathy Pressure Palsies	300	✪
Congenital Adrenal Hyperplasia	300	✪
Duchenne / Becker Muscular Dystrophies	300	✪
Familial Autosomal Dominant Hypercholesterolemia	300	✪
Fragile X Syndrome (Full scheme)	300	✪
Fragile X Syndrome (Pre-screen scheme)	300	✪
Friedreich Ataxia	300	✪
Hereditary Breast and Ovarian Cancer (BRCA1/2 targeted testing)	300	✪
Hereditary Breast and Ovarian Cancer (Panel testing)	300	✪
Hereditary Deafness	300	✪
Hereditary Haemochromatosis	300	✪
Hereditary Recurrent Fevers	300	✪
Huntington Disease	300	✪

✪ Accredited ⊗ Not accredited

TEL: +44 161 276 6741 FAX: +44 161 276 6606 WEB: www.emqn.org EMAIL: office@emqn.org

Germline mutation testing EQA schemes (cont.)	ANNUAL Fee (GBP, £)	
Hypertrophic Cardiomyopathies	300	✪
Lynch Syndrome	300	✪
Mitochondrial DNA (mtDNA) Metabolic Disorders	300	✪
Monogenic Diabetes	300	✪
Multiple Endocrine Neoplasia (Type 2)	300	✪
Myotonic Dystrophy (Types 1 & 2)	300	✪
Osteogenesis Imperfecta	300	✪
Phenylketonuria	300	✪
Polyposis Syndromes (FAP, MAP)	300	✪
Porphyrias	300	✪
Prader-Willi and Angelman syndromes	300	✪
Retinoblastoma	300	✪
RYR1 related Myopathies and Maligant Hyperthermia	300	✪
Severe Combined Immunodeficiency (pilot) NEW	100	⊗
Short Stature Homeobox Gene Testing	300	✪
Spinal Muscular Atrophy	300	✪
Spinocerebellar Ataxias	300	✪
Stickler Syndrome	300	✪
Von Hippel Lindau Syndrome	300	✪
Wilson Disease	300	✪
Y-Chromosome Microdeletion testing	300	✪

Pharmacogenetic EQA schemes	ANNUAL Fee (GBP, £)	
Pharmacogenetics (drug intolerance and effectivity)	**300**	✪

Somatic mutation testing EQA schemes	ANNUAL Fee (GBP, £)	
Microsatellite Instability testing (MSI) (pilot) NEW	100	⊗
Molecular testing in Melanoma	445	✪
Molecular testing in Lung cancer	0	✪
Molecular testing in Colorectal cancer	445	✪
Molecular testing for Oncogenes (panel testing)	445	✪
Molecular testing (germline) of BRCA genes in Ovarian cancer	0	⊗
Molecular testing (somatic) of BRCA genes in Ovarian cancer	0	⊗
Molecular testing for EGFR gene mutations in cfDNA	TBC[1]	⊗

[1] This scheme is co-ordinated by IQNPath in conjunction with EMQN and other EQA providers. No cost has been agreed at the time of going to press - please see EMQN website for regular updates.

✪ Accredited ⊗ Not accredited

TEL: +44 161 276 6741
FAX: +44 161 276 6606
WEB: www.emqn.org
EMAIL: office@emqn.org

Technique specific EQA schemes	ANNUAL Fee (GBP, £)	
DNA Sequencing (Sanger)	300	✪
DNA Sequencing (NGS v Germline)	610	⊗
DNA Sequencing (NGS v Somatic)	610	⊗
Postnatal Constitutional CNV Detection [array/NGS]	375	✪
NIPT for common aneuploidies (including sex chromosomes)	375	⊗
NIPT for fetal sex determination (X-linked disorders)	375	⊗

Interlaboratory Comparisons (Sample Exchanges)	ANNUAL Fee (GBP, £)	
Achondroplasia (*FGFR3*)	200	⊗
Congenital Central Hypoventilation Syndrome (*PHOX2B* expansions)	200	⊗
Hereditary Cancer Panel	200	⊗

✪ Accredited ⊗ Not accredited

TEL: +44 161 276 6741
FAX: +44 161 276 6606
WEB: www.emqn.org
EMAIL: office@emqn.org

Extra 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, a 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 EXTRA materials from our catalogue whilst the schemes are open for registration (see pages 78-81). These materials are priced as follows:

Somatic mutation testing EQA schemes	Fee (GBP, £) (extra sets 1, 2, or 3)
Lung Cancer	224, 269, 312
Colorectal Cancer	224, 269, 312
Melanoma	224, 269, 312
Oncogene panel testing	224, 269, 312

Technical EQA schemes	Fee (GBP, £) (extra set)
DNA Sequencing (NGS v Germline)	104
DNA Sequencing (NGS v Somatic)	104

Other EQA schemes in the catalogue	Fee (GBP, £)
EXTRA samples are not available for purchase (except those listed above)	N/A

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. Testing of replacments 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.

Schemes		Fee (GBP, £)
Lung, Colorectal and Melanoma	(1-4 samples)	85
	(5-7 samples)	155
	(8-10 samples / full set)	225
Oncogene panel	(1-3 samples / full set)	225
All other schemes		50

TEL: +44 161 276 6741 FAX: +44 161 276 6606 WEB: www.emqn.org EMAIL: office@emqn.org

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.

Purchase orders

If you require a purchase order quote to register for the EQA scheme then please contact us (office@emqn.org). **Please ensure the annual £150 registration fee is included in the total amount stated on the purchase order.**

Other information

EMQN is a community interest company (CIC) registered in England (Number: 12020789. VAT/Tax Number: 329563282).

For more information please contact us (finance@emqn.org) or see our website (https://www.emqn.org/participating-in-eqa/terms-conditions/).

Terms and Conditions

EMQN 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 (https://www.emqn.org/participating-in-eqa/terms-conditions/).

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 https://www.emqn.org/privacy-policy/).

EQA scheme timetable

R = registration period, S = survey period

Scheme	Page	September (19)	October (19)	November (19)	December (19)	January (20)	February (20)	March (20)	April (20)	May (20)	June (20)	July (20)	August (20)	September (20)	October (20)	November (20)	December (20)
GERMLINE MUTATION TESTING EQA'S																	
Autosomal Dominant Polycystic Kidney disease	09	R	R	R		S	S	S									
Beckwith-Wiedemann and Silver-Russell syndromes	10	R	R	R		S	S	S									
Cardiac Arrhythmias	11	R	R	R		S	S	S									
Charcot-Marie-Tooth disease / Hereditary Neuropathy with Pressure Palsies	12	R	R	R		S	S	S									
Congenital Adrenal Hyperplasia	13	R	R	R		S	S	S									
Duchenne / Becker Muscular Dystrophies	14	R	R	R		S	S	S									
Familial Autosomal Dominant Hypercholesterolemia	15	R	R	R		S	S	S									
Familial SHOX related disorders	16	R	R	R		S	S	S									
Fragile X Syndrome	17	R	R	R		S	S	S									
Friedreich Ataxia	19	R	R	R		S	S	S									
Hereditary Breast and Ovarian Cancer (BRCA1/2 targeted testing)	20	R	R	R		S	S	S									
Hereditary Breast and Ovarian Cancer (Panel testing)	22	R	R	R		S	S	S									
Hereditary Deafness	24	R	R	R		S	S	S									
Hereditary Haemochromatosis	25	R	R	R		S	S	S									
Hereditary Recurrent Fevers	26	R	R	R		S	S	S									
Huntington Disease	27	R	R	R		S	S	S									

TEL: +44 161 276 6741
FAX: +44 161 276 6606
WEB: www.emqn.org
EMAIL: office@emqn.org

R = registration period, S = survey period

Scheme	Page	September (19)	October (19)	November (19)	December (19)	January (20)	February (20)	March (20)	April (20)	May (20)	June (20)	July (20)	August (20)	September (20)	October (20)	November (20)	December (20)
Hypertrophic Cardiomyopathies	28	R	R	R		S	S	S									
Lynch Syndrome	29	R	R	R		S	S	S									
Mitochondrial DNA	30	R	R	R		S	S	S									
Monogenic Diabetes	31	R	R	R		S	S	S									
Multiple Endocrine Neoplasia (Type 2)	32	R	R	R		S	S	S									
Myotonic Dystrophy (Types 1 & 2)	33	R	R	R		S	S	S									
Osteogenesis Imperfecta	34	R	R	R		S	S	S									
Phenylketonuria	35	R	R	R		S	S	S									
Polyposis Syndromes (FAP, MAP)	36	R	R	R		S	S	S									
Porphyrias	37	R	R	R		S	S	S									
Prader-Willi and Angelman syndromes	38	R	R	R		S	S	S									
Retinoblastoma	39	R	R	R		S	S	S									
RYR1 related Myopathies	40	R	R	R		S	S	S									
Severe Combined Immunodeficiency (pilot)	41	R	R	R		S	S	S									
Spinal Muscular Atrophy	42	R	R	R		S	S	S									
Spinocerebellar Ataxias	43	R	R	R		S	S	S									
Stickler Syndrome	44	R	R	R		S	S	S									

TEL: +44 161 276 6741
FAX: +44 161 276 6606
WEB: www.emqn.org
EMAIL: office@emqn.org

R = registration period, S = survey period

Scheme	Page	September (19)	October (19)	November (19)	December (19)	January (20)	February (20)	March (20)	April (20)	May (20)	June (20)	July (20)	August (20)	September (20)	October (20)	November (20)	December (20)
Von Hippel Lindau Syndrome	45	R	R	R		S	S	S									
Wilson Disease	46	R	R	R		S	S	S									
Y-Chromosome Microdeletion testing	47	R	R	R		S	S	S									
PHARMACOGENETICS EQA'S																	
Pharmacogenetics (drug intolerance and effectivity)	59	R	R	R	R	R	R				S	S	S				
SOMATIC MUTATION TESTING EQA'S																	
Molecular testing for EGFR gene mutations	49	To be confirmed - see EMQN website for updates															
Microsatellite Instability Testing (MSI) (pilot)	50	R	R	R	R	R	R				S	S	S				
Molecular testing in Lung cancer	51	R	R	R	R	R	R				S	S	S				
Molecular testing in Melanoma	53	R	R	R	R	R	R				S	S	S				
Molecular testing in Colorectal cancer	54	R	R	R	R	R	R				S	S	S				
Molecular testing (germline) of BRCA genes in Ovarian cancer	55	R	R	R	R	R	R							S	S	S	
Molecular testing (somatic) of BRCA genes in Ovarian cancer	56	R	R	R	R	R	R							S	S	S	
Molecular testing for Oncogenes (panel testing).	57	R	R	R	R	R	R				S	S	S				
TECHNICAL EQA'S																	
DNA Sequencing (Sanger)	61	R	R	R	R	R	R							S	S	S	
DNA Sequencing (NGS v Germline)	62	R	R	R	R	R	R							S	S	S	

TEL: +44 161 276 6741
FAX: +44 161 276 6606
WEB: www.emqn.org
EMAIL: office@emqn.org

R = registration period, S = survey period

Scheme	Page	September (19)	October (19)	November (19)	December (19)	January (20)	February (20)	March (20)	April (20)	May (20)	June (20)	July (20)	August (20)	September (20)	October (20)	November (20)	December (20)
DNA Sequencing (NGS v Somatic)	63	R	R	R	R	R	R							S	S	S	
Postnatal Constitutional CNV Detection (array/NGS)	64	R	R	R	R	R	R			S	S	S	S				
NIPT for common aneuploidies (including sex chromosomes)	65	R	R	R	R	R	R							S	S	S	
NIPT for fetal sexing (X-linked disorders)	66	R	R	R	R	R	R							S	S	S	
INTERLABORATORY COMPARISON'S (SAMPLE EXCHANGES)																	
Achondroplasia	69	R	R	R	R	R	R			S	S	S	S				
Congenital Central Hypoventilation Syndrome	70	R	R	R	R	R	R			S	S	S	S				
Hereditary Cancer Panel	71	R	R	R	R	R	R			S	S	S	S				

Accreditation of EMQN

EMQN is accredited by the United Kingdom Accreditation Service (UKAS) to ISO17043. The scope of our accreditation can be found at https://www.emqn.org/participating-in-eqa/ukas-accreditation/. The accreditation status of all our EQA schemes is clearly shown within each of the schemes listed in this catalogue.

UKAS PROFICIENCY TESTING 4367

TEL: +44 161 276 6741 FAX: +44 161 276 6606 | WEB: www.emqn.org EMAIL: office@emqn.org

EQA Participant user guide

Participation

EMQN EQA scheme participation is on a voluntary, confidential basis. As a member, you will be actively encouraged to participate in all relevant schemes. Participation is anonymous; laboratories are identified in the EMQN database by a unique reference number known only to the EMQN office staff.

Registration

Prior to registration we advise that laboratories ensure they can receive samples of the type sent by EMQN as part of the EQA scheme. Charges will still apply if the lab cannot receive samples due to customs or import licence issues. Please see Terms and Condition, section 4.11.

Assessment

EMQN aim to publish the genotypes of the scheme samples 7 days after the closing date for result submission.

Marks are allocated according to fixed criteria agreed in advance by the scheme organiser(s) and assessors prior to receiving the returns. Marking is divided into two categories: genotyping and interpretation (except for pilot schemes, for which interpretation is not marked). Reports are marked by two independent expert assessors from different countries, at least one having experience of EQA schemes. All marking is moderated and differences resolved at an assessment meeting. You are advised to check your results as soon as the scheme genotypes are published. This is important for patient care, as the EQA may identify a systematic problem with your testing proceedures.

The aim of each EMQN EQA schemes is primarily educational; it is an opportunity for your laboratory to critically review its performance. Where possible, the standards expected follow best practice guidelines. Schemes test the ability of your lab to provide accurate genotyping and interpret the results; they also assess clerical accuracy.

Each category is marked out of 2.0, where marking is subtractive with deductions for errors, missing information or erroneous conclusions.

All reports are independently assessed by at least 2 assessors.

Laboratory testing

Your laboratory is asked to perform its normal range of tests on the samples. Assessors take note of best practice guidelines, so you are advised to check available guidelines before beginning your EQA tests. Before testing, refer to the scheme documents, which are available online. If you cannot complete your tests for any reason then contact EMQN. If the testing process fails you can request replacement samples.

Reporting

Most of our EQA schemes require you to provide a clinical report. You are asked to prepare it in your normal laboratory format and style and submit it in an approved file format. Example reports are available to EMQN members. Include your interpretation of the genotype in the context of the mock clinical information supplied with each sample. If you do not offer clinical interpretation of the genotype result, then you must supply a document clarifying why no clinical interpretation is given.

Genetic test reports may be transmitted to other nongenetics 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 'standalone report' following relevant best practice guidance where available.

TEL: +44 161 276 6741
FAX: +44 161 276 6606
WEB: www.emqn.org
EMAIL: office@emqn.org

For general guidance on reporting, please see European Journal of Human Genetics 2013:10.1038/ ejhg.2013.125. It is strongly recommended that you include the analytical limitations of your test in the report.

Your returns must be anonymous and identified only by your individual EMQN laboratory reference number. Reports are submitted via the EMQN website.

Do not upload password-protected files. Also, make sure that the report is in a language accepted by the scheme (as detailed in the scheme instructions); use of any other language may affect EMQN's ability to assess your submission.

The deadline for return of reports is usually 8 weeks from receipt of samples. Specific EQA scheme documentation will provide more information, so please check them carefully.

Sample dispatch

For practical EQA exercises sufficient sample material (for example DNA, tissue, plasma etc.) is supplied to perform all necessary analyses. You will be notified in advance, through the website, of the planned date of sample dispatch for each scheme.

Materials for the schemes are obtained from either lymphoblastoid cell lines, reference material providers, normal volunteers or patients through special arrangements with collaborating physicians, or participating laboratories. In all cases, informed consent has been given for the use of the sample in EQA.

The materials distributed are provided as specimens for the sole purpose of enabling external quality assessment at the recipients laboratory they are not controls for patient tests and must NOT be used as such.

EQA samples are validated by the scheme organiser(s) and at least one other independent, accredited expert centre. Genotypes are generally checked by more than one analytical method. Each sample dispatch will be accompanied with a dispatch note and scheme checklist. Details scheme instructions can be downloaded from your the scheme page in your EMQN user account.

You will receive advanced warning of dispatches sent to you. Please check that the delivery address on the system is up to date. A tracking number will sent to you in an email from the courier (e.g., DHL, FEDEX etc). If you do not receive your sample within 5 days of shipping you must contact the EMQN office.

Please see the following documents for the relevant terms and privacy policy:

Terms and Conditions

Privacy Policy

Sample receipt

When you receive the samples, please check the dispatch note carefully, and check the samples for damage. Notify EMQN immediately of any discrepancies or problems. Any relevant storage information will be clearly described in the scheme instructions which are available to download from the scheme page (accessed via your EMQN website account).

The samples themselves will be marked in the usual way, including patient name. You will not be provided with patient referral details: these will be a part of the scheme instructions, which you will need to download from the EMQN website.

Confidentiality

Laboratory information is confidential between you and the EMQN office (and in exceptional circumstances the Scheme Organiser(s) and the Scientific and Strategic Advisory Board (SSAB)). Only your laboratory's allocated

TEL: +44 161 276 6741
FAX: +44 161 276 6606
WEB: www.emqn.org
EMAIL: office@emqn.org

unique EMQN reference number will identify its scores within distributed summary reports. The fact that your laboratory participates in EMQN schemes is not confidential.

Replacement Samples

From time to time, laboratories may require additional samples (for example due to technical error, sample mix ups etc). Replacement samples cannot be guaranteed, but will be sent if available. There is a charge to cover reasonable costs associated with sending replacement samples (see page 76).

Individual Laboratory Reports

You will receive an individual laboratory report (ILR) for each scheme in which you participate. The report will give a breakdown of your overall score in the form of a simple grid and includes any agreed comments from the assessors. If any laboratory is considered to have performed poorly in an EQA scheme, direct contact will be made by the EMQN office staff with the laboratory's primary contact person.

Final Scheme Reports

For each scheme, a final report summarising the correct genotypes, expected interpretative points, average scores and any recurring problems / mistakes is made available via the EMQN website to all participating laboratories. The final scheme report allows you to review your own performance, and make changes to your laboratory's practices if considered necessary. Final scheme reports form an important part of the EMQN continuous improvement model, and all participants are urged to study them carefully.

Appeals

If you do not agree with the marks you have received for an EQA scheme, you are given a period of time (up to 21 days after the release of the provisional results) to submit a written appeal to the EMQN Office. An appeal must be submitted online, using a form available from the EMQN website. The appeal will be considered by the scheme assessment team and a response communicated back to the laboratory via the final ILR.

In cases where the scheme organiser(s) and scheme assessment team cannot come to a conclusion, the appeal is forwarded to the EMQN Scientific and Strategic Advisory Board (SSAB) for a decision. The **EMQN cannot consider any appeal submitted after the set deadline**; in these cases the originally assigned marks will stand.

There is NO appeals process for pilot EQA schemes.

Feedback to EMQN

EMQN welcomes written comments about feedback. Confidential communications about an EQA scheme should be made to the EMQN Office. A customer satisfaction survey may be sent out to participants shortly following each final EQA scheme report.

Complaints procedure

Most complaints received by EMQN consist of minor misunderstandings or problems with specimens. These can usually be resolved over the telephone with EMQN office staff. If a complaint is received it will be logged along with the action taken. EMQN Office Staff will attempt to address the complaint as soon as possible by letter, or email. If the participant is not satisfied with the response by EMQN office staff then the matter will be brought to the Scientific and Strategic Advisory Board (SSAB) at their next meeting. A response will be made in light of the advice given by the SSAB. Participants who feel that their cause for complaint requires a

TEL: +44 161 276 6741 | FAX: +44 161 276 6606 | WEB: www.emqn.org | EMAIL: office@emqn.org

more formal response are invited to contact, in writing, the Chairman of the EMQN Board.

Collusion

EQA scheme participants are reminded that it is the duty of the scheme provider to try to prevent collusion between participating laboratories. You must keep your EQA results private and not collude with other laboratories. Participants suspected of colluding in their EQA scheme returns will be contacted and may be excluded from participation in future scheme(s).

Subcontracted activities

EMQN is responsible for all design, planning, review and oversight of EQA schemes. Some activities such as the manufacture of materials or peer review by scheme assessors are sub contracted. However, EMQN remains responsible for the oversight of all subcontracted activities.

Working with EMQN

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

Help and Advice

Our website has a comprehensive Frequently Asked Questions (FAQ) section, including user guides and short help video's. If you cannot find an answer to your questions, then please do not hesitate to contact us (office@emqn.org). Our staff are always happy to help you.

TEL: +44 161 276 6741 | FAX: +44 161 276 6606 | WEB: www.emqn.org | EMAIL: office@emqn.org

EMQN

Registered address:
EMQN, c/o Trustech, Citylabs 1.0
Nelson Street
Manchester
M13 9NQ, United Kingdom

TEL: +44 161 276 6741
FAX: +44 161 276 6606
WEB: www.emqn.org
EMAIL: office@emqn.org

@EMQNOffice https://linkedin/company/emqn