

EQA scheme catalogue & participant guide 2018

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Introduction from EMQN Director

Welcome to our 2018 EQA scheme catalogue and participant guide. This is our 21st year of operation and we've come a long way in that time. Our days of EU grant funded start-up finished in 2002 and since then we have been entirely funded by subscription fees paid by you, our customers.

We've grown from 1 EQA scheme and 25 participants (1997), to 50 different EQAs and 1400 regular users in 2017. Our customer base has evolved too over this time and we are now a truly global provider of EQA activities with customers in 68 different countries using our services.

We are hosted by Central Manchester University Hospitals NHS Foundation Trust (CMFT) and based within one of the largest Genomic Diagnostic Laboratories in the UK (the Manchester Centre for Genomic Medicine). This gives us a valuable interface with current and developing clinical practice across the full spectrum of clinical genomic services.

Our activities are supported by a global team of 180 experts in the fields of Genetics, Pathology and Bioinformatics.

Our experts spend countless hours monitoring testing trends in these fields to bring you EQA schemes that:

- Challenge and educate our users
- Keep our offerings contemporary with new programmes to meet new developments in diagnostic practice.

I'd like to thank you for your continued custom and we look forward to working with you in 2018.

Best wishes,

Dr Simon Patton (EMQN Director)

New this year

The clinical diagnostic testing landscape is changing rapidly with the widespread introduction of panel testing and clinically-focused genomic methods.

Therefore in 2018 we will be moving to offering EQA schemes which meet the needs of laboratories which are offering virtual panel analysis (via clinical exomes, whole exomes, or genomes), as well as targeted tests. Consequently this year we are offering two new pilot EQA schemes for cardiac genetics.

The schemes are for:

- Arrhythmias (page 7) **NEW**
- Hypertrophic Cardiomyopathies (page 26) **NEW**



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.

The EMQN includes over 2000 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 diffusion 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 best practice meetings. Members can contribute to best practice development through these meetings which the testing community as a whole. 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.



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

The 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 NGS Sequencing technologies
- 2 Non-invasive prenatal testing
- 3 Pharmacogenetics method to predict patient response to new specialized drugs

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EQA schemes

Overview

We provide EQA schemes for 3 core areas of laboratory medicine:

- Inherited genetic disorders (Germline mutation testing)
- Cancer (Somatic mutation testing)
- Technological approaches used in testing

Our EQA schemes aim to mimic real clinical testing as closely as possible with laboratories. 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.

If you are a lab that is not providing a full clinical interpretation of a test results (e.g., kit manufacturers, commercial sequencing hubs etc), then please contact the EMQN office for advice on the ways you can participate.

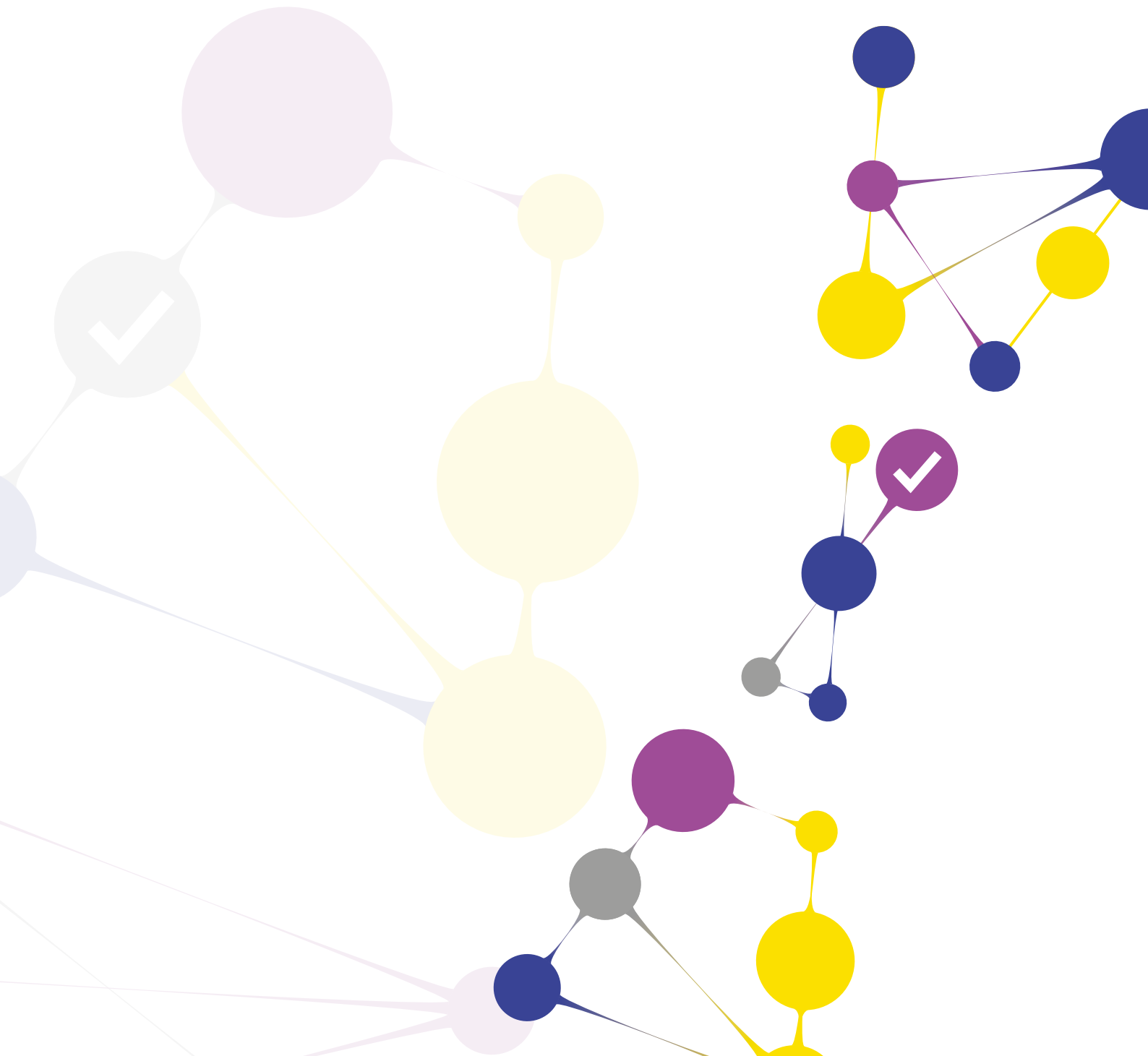




Germline mutation testing EQA schemes

Thirty two EQA schemes are being provided in 2018. These EQAs require 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.](#)





Arrhythmias



CARDIAC DISEASE

INHERITED

GERMLINE

Scheme Code	CARDIO(ARR)-18
Target	As testing approaches are still not standardized and vary between laboratories, the exact list of genes to be tested is not specified. This pilot EQA will enable the collection of information regarding the different panels in current use.
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. Aimed at laboratories using a panel based DNA sequencing strategy.
Reporting Language	Reports accepted in English ONLY.
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	<input checked="" type="checkbox"/> Performance criteria DO NOT apply.
Accreditation	<input checked="" type="checkbox"/> This scheme is NOT covered by the scope of EMQN's accreditation.
Collaborator	<input type="checkbox"/> None



Autosomal Dominant Polycystic Kidney disease

KIDNEY DISEASE

INHERITED

GERMLINE

Scheme code	ADPKD-18
Target	Mutations in the <i>PKD1</i> and <i>PKD2</i> genes.
Sample Material	DNA (in TE Buffer)
Scheme Format	<p>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.</p> <p>Full EQA scheme no restrictions on number of participants.</p> <p>Open to laboratories from ALL countries.</p> <p>Three mock clinical cases with matching samples. 3</p>
Reporting Language	Reports accepted in English, French and German ONLY.
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 <i>PKD1</i> and <i>PKD2</i> exons - 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



Beckwith-Wiedemann & Silver-Russell syndromes

IMPRINTING

DYSMORPHOLOGY

CANCER

INHERITED

GERMLINE

Scheme code	BWS/SRS -18
Target	BWS: Maternal hypomethylation at ICR2 (<i>KCNQ1OT1</i>), hypermethylation at maternal ICR1 (<i>H19</i>), copy number variants, segmental mosaic UPD11pat, and maternally-inherited mutations of <i>CDKN1C</i> . 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 and German ONLY.
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



Breast / Ovarian Cancer, familial (Full version)

CANCER

INHERITED

GERMLINE

Scheme Code	BRCA-18-Full
Target	Mutations in the <i>BRCA1</i> and <i>BRCA2</i> genes. New: other clinically relevant genes may also be included but performance criteria will only apply to the BRCA genes this year.
Sample Material	DNA (in TE Buffer)
Scheme Format	<p>Assessment of genotyping, and biological and clinical interpretation. For each case, participants are expected to return a clinical report which includes a complete (biological and clinical) interpretation of the results.</p> <p>No restrictions on number of participants.</p> <p>Open to laboratories from ALL countries.</p> <p>Three mock clinical cases with matching samples. 3</p>
Reporting Language	Reports accepted in English, French, German, Spanish or Italian ONLY.
Additional Information	<p>Suitable for sequence based analysis (e.g. NGS / Sanger Sequencing) and copy number analysis (e.g. MLPA, NGS based CNV analysis).</p> <p>Mutation screening in any BRCA exons required – scheme NOT suitable for labs testing for recurrent mutations only.</p>
Performance criteria	<input checked="" type="checkbox"/> Performance criteria DO apply.
Accreditation	<input checked="" type="checkbox"/> This scheme is covered by the scope of EMQN's accreditation.
Collaborator	<input type="checkbox"/> None



Breast / Ovarian Cancer, familial (Genotyping-only version)

CANCER

INHERITED

GERMLINE

Scheme code	BRCA-18-Geno
Target	Mutations in the <i>BRCA1</i> and <i>BRCA2</i> genes. New: other clinically relevant genes may also be included but performance criteria will only apply to the BRCA genes this year.
Sample Material	DNA (in TE Buffer)
Scheme Format	<p>Assessment of genotyping AND biological interpretation only. Participants are expected to return a report containing the genotyping results and the biological interpretation.</p> <p>Clinical interpretation of the results are not required for this scheme.</p> <p>No restrictions on number of participants.</p> <p>Open to laboratories from ALL countries.</p> <p>Three mock clinical cases with matching samples. 3</p>
Reporting Language	Reports accepted in English, French, German, Spanish or Italian ONLY.
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 – scheme 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



Charcot-Marie-Tooth disease

NEUROLOGICAL DISEASE

INHERITED

GERMLINE

Scheme code	CMT-18
Target	Mutation testing in the <i>PMP22</i> (deletion /duplication) and <i>GJB1</i> genes.
Sample Material	DNA (in TE Buffer)
Scheme Format	<p>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.</p> <p>No restrictions on number of participants.</p> <p>Open to laboratories from ALL countries.</p> <p>Three mock clinical cases with matching samples. 3</p>
Reporting Language	Reports accepted in English or German ONLY.
Additional Information	Suitable for copy number analysis of the <i>PMP22</i> gene (e.g. MLPA) and/or sequence analysis (Sanger/NGS) of <i>PMP22/MPZ/GJB1</i> genes.
Performance criteria	✔ Performance criteria DO apply.
Accreditation	★ This scheme is covered by the scope of EMQN's accreditation.
Collaborator	⊖ None



Congenital Adrenal Hyperplasia

HORMONE

INHERITED

GERMLINE

Scheme code	CAH-18
Target	Mutations in the <i>CYP21A2</i> gene
Sample Material	DNA (in TE Buffer)
Scheme Format	<p>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.</p> <p>No restrictions on number of participants.</p> <p>Open to laboratories from ALL countries.</p> <p>Three mock clinical cases with matching samples. 3</p>
Reporting Language	Reports accepted in English or German ONLY.
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	<input checked="" type="checkbox"/> Performance criteria DO apply.
Accreditation	<input checked="" type="checkbox"/> This scheme is covered by the scope of EMQN's accreditation.
Collaborator	<input type="checkbox"/> None



Duchenne / Becker Muscular Dystrophy

MUSCULAR DYSTROPHY

INHERITED

GERMLINE

Scheme Code	DMD-18
Target	Mutations in the Dystrophin gene
Sample Material	DNA (in TE Buffer)
Scheme Format	<p>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.</p> <p>No restrictions on number of participants.</p> <p>Open to laboratories from ALL countries.</p> <p>Three mock clinical cases with matching samples. 3</p>
Reporting Language	Reports accepted in English, Dutch or German ONLY.
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	<input checked="" type="checkbox"/> Performance criteria DO apply.
Accreditation	<input checked="" type="checkbox"/> This scheme is covered by the scope of EMQN's accreditation.
Collaborator	<input type="checkbox"/> None



Familial Adenomatous Polyposis Colon Cancer

CANCER **INHERITED** **GERMLINE**

Scheme code	FAP-18
Target	Mutations in the <i>APC</i> gene
Sample Material	DNA (in TE Buffer)
Scheme Format	<p>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.</p> <p>No restrictions on number of participants.</p> <p>Open to laboratories from ALL countries.</p> <p>Three mock clinical cases with matching samples. 3</p>
Reporting Language	Reports accepted in English and German 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	<input checked="" type="checkbox"/> Performance criteria DO apply.
Accreditation	<input checked="" type="checkbox"/> This scheme is covered by the scope of EMQN's accreditation.
Collaborator	<input type="checkbox"/> None



Familial autosomal dominant hypercholesterolemia

CARDIAC DISEASE

INHERITED

GERMLINE

Scheme code	FH-18
Target	Mutations in the <i>LDLR</i> , <i>APOB</i> and <i>PCSK9</i> genes
Sample Material	DNA (in TE Buffer)
Scheme Format	<p>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.</p> <p>Open to laboratories from ALL countries.</p> <p>Three mock clinical cases with matching samples. 3</p>
Reporting Language	Reports accepted in English, German, French and Spanish ONLY.
Additional Information	<p>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 <i>LDLR</i>, <i>APOB</i> and <i>PCSK9</i> exons required – scheme NOT suitable for labs testing for recurrent mutations only.</p>
Performance criteria	<input checked="" type="checkbox"/> Performance criteria DO apply.
Accreditation	<input checked="" type="checkbox"/> This scheme is covered by the scope of EMQN's accreditation.
Collaborator	<input type="checkbox"/> None



Familial SHOX-related disorders

INHERITED

GERMLINE

Scheme code	SHOX-18
Target	Mutations in the <i>SHOX</i> gene
Sample Material	DNA (in TE Buffer)
Scheme Format	<p>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.</p> <p>No restrictions on number of participants.</p> <p>Open to laboratories from ALL countries.</p> <p>Three mock clinical cases with matching samples. 3</p>
Reporting Language	Reports accepted in English and German 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	<input checked="" type="checkbox"/> Performance criteria DO apply.
Accreditation	<input checked="" type="checkbox"/> This scheme is covered by the scope of EMQN's accreditation.
Collaborator	<input type="checkbox"/> None



Fragile X Syndrome (Full version)

INTERLECTUAL DISABILITY

TRINUCELOTIDE REPEAT DISORDER

INHERITED

GERMLINE

Scheme code	FRAX-18-Full
Target	Triplet repeat expansions and methylation of Mutations the <i>FMR1</i> gene
Sample Material	DNA (in TE Buffer)
Scheme Format	<p>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.</p> <p>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.</p> <p>No restrictions on number of participants.</p> <p>Open to laboratories from ALL countries.</p> <p>Three mock clinical cases with matching samples. 3</p>
Reporting Language	Reports accepted in English, German or French ONLY.
Additional Information	Suitable for PCR-based and Southern blotting techniques. CCG repeat analysis ONLY.
Performance criteria	<input checked="" type="checkbox"/> Performance criteria DO apply.
Accreditation	<input checked="" type="checkbox"/> This scheme is covered by the scope of EMQN's accreditation.
Collaborator	<input type="checkbox"/> None



Fragile X Syndrome (Pre-screening only version)

INTERLECTUAL DISABILITY

TRINUCELOTIDE REPEAT DISORDER

INHERITED

GERMLINE

Scheme code	FRAX-18-Pre screen
Target	Triplet repeat expansions of Mutations in the <i>FMR1</i> gene.
Sample Material	DNA (in TE Buffer)
Scheme Format	<p>Applicable to labs which are NOT able to perform the full diagnosis in each case but perform a prescreen ONLY.</p> <p>Assessment of genotyping, and biological and clinical interpretation. No restrictions on number of participants.</p> <p>Open to laboratories from ALL countries.</p> <p>Three mock clinical cases with matching samples. 3</p>
Reporting Language	Reports accepted in English, German or French ONLY.
Additional Information	Suitable for PCR-based and Southern blotting techniques. CCG repeat analysis ONLY.
Performance criteria	<input checked="" type="checkbox"/> Performance criteria DO apply.
Accreditation	<input checked="" type="checkbox"/> This scheme is covered by the scope of EMQN's accreditation.
Collaborator	<input type="checkbox"/> None



Friedreich Ataxia

NEUROLOGICAL DISEASE

TRINUCELOTIDE REPEAT DISORDER

INHERITED

GERMLINE

Scheme code	FRDA-18
Target	Triplet repeat expansions of Mutations in the <i>FXN</i> gene
Sample Material	DNA (in TE Buffer)
Scheme Format	<p>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.</p> <p>No restrictions on number of participants.</p> <p>Open to laboratories from ALL countries.</p> <p>Three mock clinical cases with matching samples. 3</p>
Reporting Language	Reports accepted in English, French, German or Dutch ONLY.
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






Hereditary Deafness

DEAFNESS

INHERITED

GERMLINE

Scheme code	DFNB1-18
Target	Mutations in <i>GJB2</i> and <i>GJB6</i> genes (DFNB1)
Sample Material	DNA (in TE Buffer)
Scheme Format	<p>Assessment of genotyping, and biological and clinical interpretation.</p> <p>For each case, participants are expected to return a clinical report which includes a complete interpretation of the results.</p> <p>No restriction on the number of participants.</p> <p>Open to laboratories from ALL countries.</p> <p>Three mock clinical cases with matching samples will be supplied. 3</p>
Reporting Language	Reports accepted in English, Dutch, 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



Hereditary Haemochromatosis

METABOLIC

INHERITED

GERMLINE

Scheme code	HFE-18
Target	Mutations in the <i>HFE</i> gene
Sample Material	DNA (in TE Buffer)
Scheme Format	<p>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.</p> <p>No restrictions on number of participants.</p> <p>Open to laboratories from ALL countries.</p> <p>Three mock clinical cases with matching samples. 3</p>
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	<input checked="" type="checkbox"/> Performance criteria DO apply.
Accreditation	<input checked="" type="checkbox"/> This scheme is covered by the scope of EMQN's accreditation.
Collaborator	<input type="checkbox"/> None



Hereditary Recurrent Fevers

INFLAMMATORY DISEASE

INHERITED

GERMLINE

Scheme code	HRF-18
Target	Mutations in the <i>MEFV</i> , <i>MVK</i> , <i>TNFRSF1A</i> and <i>NLRP3</i> genes.
Sample Material	DNA (in TE Buffer)
Scheme Format	<p>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.</p> <p>No restrictions on number of participants.</p> <p>Open to laboratories from ALL countries.</p> <p>Four mock clinical cases with matching samples. ④</p>
Reporting Language	Reports accepted in English and German 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



Huntington Disease

NEUROLOGICAL DISEASE

TRIPLET REPEAT DISORDER

INHERITED

GERMLINE

Scheme code	HD-18
Target	Triplet repeat analysis of Mutations in the <i>HTT</i> gene
Sample Material	DNA (in TE Buffer)
Scheme Format	<p>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.</p> <p>No restrictions on number of participants.</p> <p>Open to laboratories from ALL countries.</p> <p>Three mock clinical cases with matching samples. 3</p>
Reporting Language	Reports accepted in English, Dutch or German ONLY.
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



Hypertrophic Cardiomyopathies



CARDIAC DISEASE

INHERITED

GERMLINE

Scheme code	CARDIO(HCM)18
Target	As testing approaches are still not standardized and vary between laboratories, the exact list of genes to be tested is not specified. This pilot EQA will enable the collection of information regarding the different panels in current use.
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)
Reporting Language	Reports accepted in English 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	(X) Performance criteria DO NOT apply.
Accreditation	(X) This scheme is NOT covered by the scope of EMQN's accreditation.
Collaborator	(-) None



Lynch Syndrome

CANCER

INHERITED

GERMLINE

Scheme code	HNPCC-18
Target	Mutations in the <i>MSH2</i> , <i>MLH1</i> , <i>MSH6</i> and <i>PMS2</i> genes.
Sample Material	DNA (in TE Buffer)
Scheme Format	<p>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.</p> <p>No restrictions on number of participants.</p> <p>Open to laboratories from ALL countries.</p> <p>Three mock clinical cases with matching samples. 3</p>
Reporting Language	Reports accepted in English, French or German ONLY.
Additional Information	Suitable for sequence based analysis (e.g. NGS / Sanger Sequencing) and copy number analysis (e.g. MLPA, NGS based CNV analysis). Scheme formerly known as the HNPCC scheme.
Performance criteria	<input checked="" type="checkbox"/> Performance criteria DO apply.
Accreditation	<input checked="" type="checkbox"/> This scheme is covered by the scope of EMQN's accreditation.
Collaborator	<input type="checkbox"/> None



Mitochondrial DNA (mtDNA) Metabolic Disorders

METABOLIC

INHERITED

GERMLINE

Scheme code	mtDNA-18
Target	Mutations in mtDNA (mitochondrial genome). 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.
Sample Material	DNA (in TE Buffer)
Scheme Format	<p>Assessment of genotyping, and biological and clinical interpretation.</p> <p>For each case, participants are expected to return a clinical report which includes a complete interpretation of the results.</p> <p>No restriction on the number of participants.</p> <p>Open to laboratories from ALL countries.</p> <p>Three mock clinical cases with matching samples will be supplied. 3</p>
Reporting Language	Reports accepted in English ONLY.
Additional Information	Suitable for sequence based analysis (e.g. NGS / Sanger Sequencing) and copy number analysis (e.g. MLPA, NGS based CNV analysis). Mutation variant screening for any mtDNA gene.
Performance criteria	X Performance criteria DO NOT apply.
Accreditation	X This scheme is NOT covered by the scope of EMQN's accreditation.
Collaborator	Royal College of Pathologists of Australasia Quality Assurance Programs – www.rcpaqap.com.au/.



Monogenic Diabetes

METABOLIC

INHERITED

GERMLINE

Scheme code	MONODIAB-18
Target	Mutations in the <i>GCK</i> , <i>HNF1A</i> , <i>HNF1B</i> and <i>HNF4A</i> genes
Sample Material	DNA (in TE Buffer)
Scheme Format	<p>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.</p> <p>No restrictions on number of participants.</p> <p>Open to laboratories from ALL countries.</p> <p>Three mock clinical cases with matching samples. 3</p>
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	<input checked="" type="checkbox"/> Performance criteria DO apply.
Accreditation	<input checked="" type="checkbox"/> This scheme is covered by the scope of EMQN's accreditation.
Collaborator	<input type="checkbox"/> None



Multiple Endocrine Neoplasia Type 2

CANCER

INHERITED

GERMLINE

Scheme code	MEN2-18
Target	Mutations in the <i>RET</i> proto-oncogene
Sample Material	DNA (in TE Buffer)
Scheme Format	<p>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.</p> <p>No restrictions on number of participants.</p> <p>Open to laboratories from ALL countries.</p> <p>Three mock clinical cases with matching samples. 3</p>
Reporting Language	Reports accepted in English and German 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	<input checked="" type="checkbox"/> Performance criteria DO apply.
Accreditation	<input checked="" type="checkbox"/> This scheme is covered by the scope of EMQN's accreditation.
Collaborator	<input type="checkbox"/> None



Myotonic Dystrophy

MUSCULAR DYSTROPHY

TRINUCELOTIDE REPEAT DISORDER

INHERITED

GERMLINE

Scheme code	DM-18
Target	Triplet repeat analysis of Mutations in the <i>DMPK</i> gene
Sample Material	DNA (in TE Buffer)
Scheme Format	<p>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.</p> <p>No restrictions on number of participants.</p> <p>Open to laboratories from ALL countries.</p> <p>Three mock clinical cases with matching samples. 3</p>
Reporting Language	Reports accepted in English, Danish or German ONLY.
Additional Information	Suitable for PCR-based and Southern blotting techniques. CTG repeat analysis ONLY.
Performance criteria	✔ Performance criteria DO apply.
Accreditation	★ This scheme is covered by the scope of EMQN's accreditation.
Collaborator	⊖ None



Osteogenesis imperfecta

CONNECTIVE TISSUE DISORDER

INHERITED

GERMLINE

Scheme code	OI-18
Target	Mutations in <i>COL1A1</i> and <i>COL1A2</i> 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 and Dutch ONLY.
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 <i>COL1A1</i> and <i>COL1A2</i> exons required – scheme 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



Phenylketonuria

METABOLIC

INHERITED

GERMLINE

Scheme code	PKU-18
Target	Mutations in the <i>PAH</i> gene
Sample Material	DNA (in TE Buffer)
Scheme Format	<p>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.</p> <p>No restrictions on number of participants.</p> <p>Open to laboratories from ALL countries.</p> <p>Three mock clinical cases with matching samples. 3</p>
Reporting Language	Reports accepted in English and German 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	<input checked="" type="checkbox"/> Performance criteria DO apply.
Accreditation	<input checked="" type="checkbox"/> This scheme is covered by the scope of EMQN's accreditation.
Collaborator	<input type="checkbox"/> None



Porphyrias

METABOLIC

INHERITED

GERMLINE

Scheme code	POR-18
Target	Mutations in the most frequently analysed porphyria genes (for example <i>PPOX</i> , <i>HMBS</i>).
Sample Material	DNA (in TE Buffer)
Scheme Format	<p>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.</p> <p>No restrictions on number of participants.</p> <p>Open to laboratories from ALL countries.</p> <p>Three mock clinical cases with matching samples. 3</p>
Reporting Language	Reports accepted in English and German 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	<input checked="" type="checkbox"/> Performance criteria DO apply.
Accreditation	<input checked="" type="checkbox"/> This scheme is covered by the scope of EMQN's accreditation.
Collaborator	<input type="checkbox"/> None



Prader-Willi and Angelman Syndromes

IMPRINTING

INHERITED

GERMLINE

Scheme code	PWAS-18
Target	15q11-q13 studies in order to diagnose Prader-willi or Angelman syndromes
Sample Material	DNA (in TE Buffer)
Scheme Format	<p>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.</p> <p>No restrictions on number of participants.</p> <p>Open to laboratories from ALL countries.</p> <p>Three mock clinical cases with matching samples. 3</p>
Reporting Language	Reports accepted in English and German ONLY.
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



Retinoblastoma

CANCER

INHERITED

GERMLINE

Scheme code	RB-18
Target	Mutations in the <i>RB1</i> gene
Sample Material	DNA (in TE Buffer)
Scheme Format	<p>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.</p> <p>No restrictions on number of participants.</p> <p>Open to laboratories from ALL countries.</p> <p>Three mock clinical cases with matching samples. 3</p>
Reporting Language	Reports accepted in English and German 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



Spinal Muscular Atrophy

NEUROLOGICAL DISEASE

INHERITED

GERMLINE

Scheme code	SMA-18
Target	Mutations in the <i>SMN1</i> gene
Sample Material	DNA (in TE Buffer)
Scheme Format	<p>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.</p> <p>No restrictions on number of participants.</p> <p>Open to laboratories from ALL countries.</p> <p>Three mock clinical cases with matching samples. 3</p>
Reporting Language	Reports accepted in English, Dutch and German ONLY.
Additional Information	Suitable for copy number analysis of the <i>SMN1</i> / <i>(SMN2)</i> 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



Spinocerebellar Ataxia's

NEUROLOGICAL DISEASE

TRINUCELOTIDE REPEAT DISORDER

INHERITED

GERMLINE

Scheme code	SCA-18
Target	Triplet repeat analysis of Mutations in the <i>ATXN 1-7</i> genes
Sample Material	DNA (in TE Buffer)
Scheme Format	<p>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.</p> <p>No restrictions on number of participants.</p> <p>Open to laboratories from ALL countries.</p> <p>Three mock clinical cases with matching samples. 3</p>
Reporting Language	Reports accepted in English, French, German, Dutch, Italian, Spanish or Portuguese ONLY.
Additional Information	Suitable for PCR-based techniques ONLY. CAG repeat analysis ONLY.
Performance criteria	<input checked="" type="checkbox"/> Performance criteria DO apply.
Accreditation	<input checked="" type="checkbox"/> This scheme is covered by the scope of EMQN's accreditation.
Collaborator	<input type="checkbox"/> None



Von Hippel Lindau Syndrome

CANCER

INHERITED

GERMLINE

Scheme code	VHL-18
Target	Mutations in the <i>VHL</i> gene
Sample Material	DNA (in TE Buffer)
Scheme Format	<p>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.</p> <p>No restrictions on number of participants.</p> <p>Open to laboratories from ALL countries.</p> <p>Three mock clinical cases with matching samples. 3</p>
Reporting Language	Reports accepted in English or German 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



Wilson Disease

METABOLIC

INHERITED

GERMLINE

Scheme code	WIL-18
Target	Mutations in the <i>ATP7B</i> gene
Sample Material	DNA (in TE Buffer)
Scheme Format	<p>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.</p> <p>No restrictions on number of participants.</p> <p>Open to laboratories from ALL countries.</p> <p>Three mock clinical cases with matching samples. 3</p>
Reporting Language	Reports accepted in English and German 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



Y-Chromosome Microdeletions

TBC

INHERITED

GERMLINE

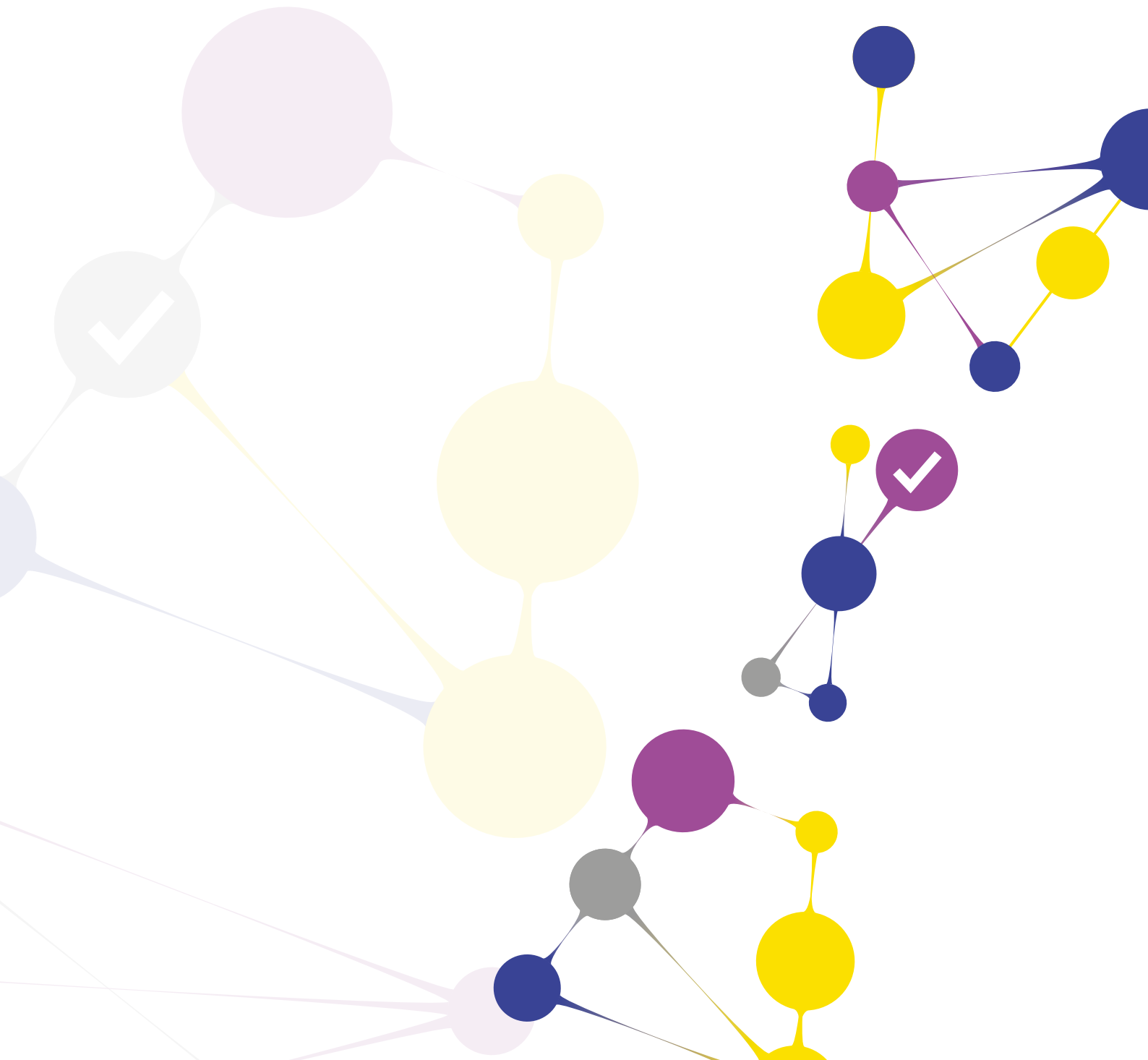
Scheme code	AZF-18
Target	Microdeletions of the Y-Chromosome
Sample Material	DNA (in TE Buffer)
Scheme Format	<p>Assessment of genotyping, and biological and clinical interpretation. For each case, participants are expected to return a clinical report which includes a complete interpretation of the results.</p> <p>No restrictions on number of participants.</p> <p>Open to laboratories from ALL countries.</p> <p>Three mock clinical cases with matching samples. 3</p>
Reporting Language	Reports accepted in English, French, German or Italian ONLY.
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



Somatic mutation testing EQA schemes

Eight EQA schemes are being provided in 2018. 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.](#)





Molecular testing for EGFR gene mutations in ctDNA (pilot)

CANCER

ACQUIRED

SOMATIC

PLASMA

Scheme code	LIQUIDBIOPSY(EGFR)-18
Target	Mutations in the EGFR gene (see information below on scheme format).
Sample Material	Plasma containing ctDNA
Scheme Format	<p>Assessment of genotyping, and biological and clinical interpretation.</p> <p>For each case, participants are expected to return a clinical report which includes a complete interpretation of the results.</p> <p>No restrictions on number of participants.</p> <p>Open to laboratories from ALL countries.</p> <p>5 mock clinical cases with matching samples. ⑤</p>
Reporting Language	Reports accepted in English ONLY
Additional Information	None
Performance criteria	⊗ Performance criteria DO NOT apply.
Accreditation	⊗ This scheme is NOT covered by the scope of EMQN's accreditation.
Collaborator	IQNPath (www.iqnpath.org) Liquid Biopsy working group



Molecular testing for RAS gene mutations in ctDNA (pilot)

CANCER

ACQUIRED

SOMATIC

PLASMA

Scheme code	LIQUIDBIOPSY (RAS)-18
Target	Mutations in the KRAS, NRAS, HRAS genes (see information below on scheme format).
Sample Material	Plasma containing ctDNA
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. 5 mock clinical cases with matching samples. (5)
Reporting Language	Reports accepted in English ONLY
Additional Information	None
Performance criteria	(X) Performance criteria DO NOT apply.
Accreditation	(X) This scheme is NOT covered by the scope of EMQN's accreditation.
Collaborator	IQNPath (www.iqnpath.org) Liquid Biopsy working group







Molecular testing in Lung Cancer

CANCER

ACQUIRED

SOMATIC

Scheme code	NSCLC-18
Target	Mutations in the <i>EGFR</i> , <i>PIK3CA</i> , <i>KRAS</i> and <i>BRAF</i> genes.
Sample Material	Mix of real tissue and artificial paraffin embedded (FFPE) materials designed to simulate a real patient sample. NOTE: Samples provided as provided as cut sections (rolled scrolls) ONLY we cannot provide slide mounted materials.
Scheme Format	<p>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.</p> <p>The minimum requirement is <i>EGFR</i> gene testing. If you provide a clinical service for <i>BRAF</i> and <i>KRAS</i> genes, these should be included in the interpretation of results. The testing of other genes is optional.</p> <p>We only require clinically relevant pathogenic (disease-causing) mutations to be reported, and not common SNPs / variants.</p> <p>No restrictions on number of participants.</p> <p>Open to laboratories from ALL countries.</p> <p>10 mock clinical cases with matching samples. </p>
Reporting Language	Reports accepted in English ONLY
Additional Information	Mutation allelic frequencies have been validated using ddPCR. We would like to thank AstraZeneca for the educational grant to deliver this EQA.
Performance criteria	 Performance criteria DO apply.
Accreditation	 This scheme is covered by the scope of EMQN's accreditation.
Collaborator	 None



Molecular testing in Melanoma

CANCER

ACQUIRED

SOMATIC

SKIN

Scheme code	MELANOMA-18
Target	Mutations in the <i>BRAF</i> , <i>NRAS</i> , <i>KIT</i> genes.
Sample Material	Mix of real tissue and artificial paraffin embedded (FFPE) materials designed to simulate a real patient sample. NOTE: Samples provided as provided as cut sections (rolled scrolls) ONLY we cannot provide slide mounted materials.
Scheme Format	<p>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.</p> <p>The minimum requirement is <i>BRAF</i> gene testing. If you provide a clinical service for the <i>NRAS</i> and <i>KIT</i> gene, these should be included in the interpretation of results. The testing of other genes is optional.</p> <p>We only require clinically relevant pathogenic (disease-causing) mutations to be reported, and not common SNPs / variants.</p> <p>No restrictions on number of participants.</p> <p>Open to laboratories from ALL countries.</p> <p>10 mock clinical cases with matching samples. ⑩</p>
Reporting Language	Reports accepted in English ONLY
Additional Information	Mutation allelic frequencies 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



Molecular testing in sporadic Colorectal Cancer

CANCER

ACQUIRED

SOMATIC

Scheme code	COLOREG-18
Target	Mutations in the <i>KRAS</i> , <i>BRAF</i> , <i>NRAS</i> and <i>PIK3CA</i> genes.
Sample Material	Mix of real tissue and artificial paraffin embedded (FFPE) materials designed to simulate a real patient sample. NOTE: Samples provided as provided as cut sections (rolled scrolls) ONLY we cannot provide slide mounted materials.
Scheme Format	<p>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.</p> <p>The minimum requirement is <i>KRAS</i>, <i>NRAS</i> and <i>BRAF</i> gene testing. If you provide a clinical service for the <i>PIK3CA</i> gene, this should be included. The testing of other genes is optional.</p> <p>We only require clinically relevant pathogenic (disease-causing) mutations to be reported, and not common SNPs / variants.</p> <p>No restrictions on number of participants.</p> <p>Open to laboratories from ALL countries.</p> <p>10 mock clinical cases with matching samples. (10)</p>
Reporting Language	Reports accepted in English ONLY
Additional Information	Mutation allelic frequencies 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



Molecular testing of BRCA genes in Ovarian Cancer (vGermline)

CANCER

INHERITED

GERMLINE

Scheme code	OVARIAN-18 (G)
Target	Mutations in the <i>BRCA1</i> and <i>BRCA2</i> genes
Sample Material	DNA (in TE buffer).
Scheme Format	<p>Assessment of genotyping, and biological and clinical interpretation (<i>BRCA1</i> and <i>BRCA2</i> 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.</p> <p>No restrictions on number of participants.</p> <p>Open to laboratories from ALL countries.</p> <p>3 mock clinical cases with matching samples. 3</p>
Reporting Language	Reports accepted in English ONLY
Additional Information	We would like to thank Astra Zeneca for the educational grant to deliver this pilot EQA.
Performance criteria	✔ Performance criteria DO apply.
Accreditation	☆ This scheme is covered by the scope of EMQN's accreditation.
Collaborator	UK NEQAS for Molecular Genetics.



Molecular testing of BRCA genes in Ovarian Cancer (vSomatic)

CANCER

ACQUIRED

SOMATIC

Scheme code	OVARIAN-18 (S)
Target	Mutations in the <i>BRCA1</i> and <i>BRCA2</i> genes (see information below on scheme format).
Sample Material	Artificial paraffin embedded (FFPE) materials designed to simulate a real patient sample.
Scheme Format	<p>Assessment of genotyping, and biological and clinical interpretation (<i>BRCA1</i> and <i>BRCA2</i> 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.</p> <p>No restrictions on number of participants.</p> <p>Open to laboratories from ALL countries.</p> <p>3 mock clinical cases with matching samples. 3</p>
Reporting Language	Reports accepted in English ONLY
Additional Information	We would like to thank Astra Zeneca for the educational grant to deliver this pilot EQA.
Performance criteria	✔ Performance criteria DO apply.
Accreditation	★ This scheme is covered by the scope of EMQN's accreditation.
Collaborator	UK NEQAS for Molecular Genetics.



Oncogene panel testing

CANCER

AQUIRED

SOMATIC

NGS

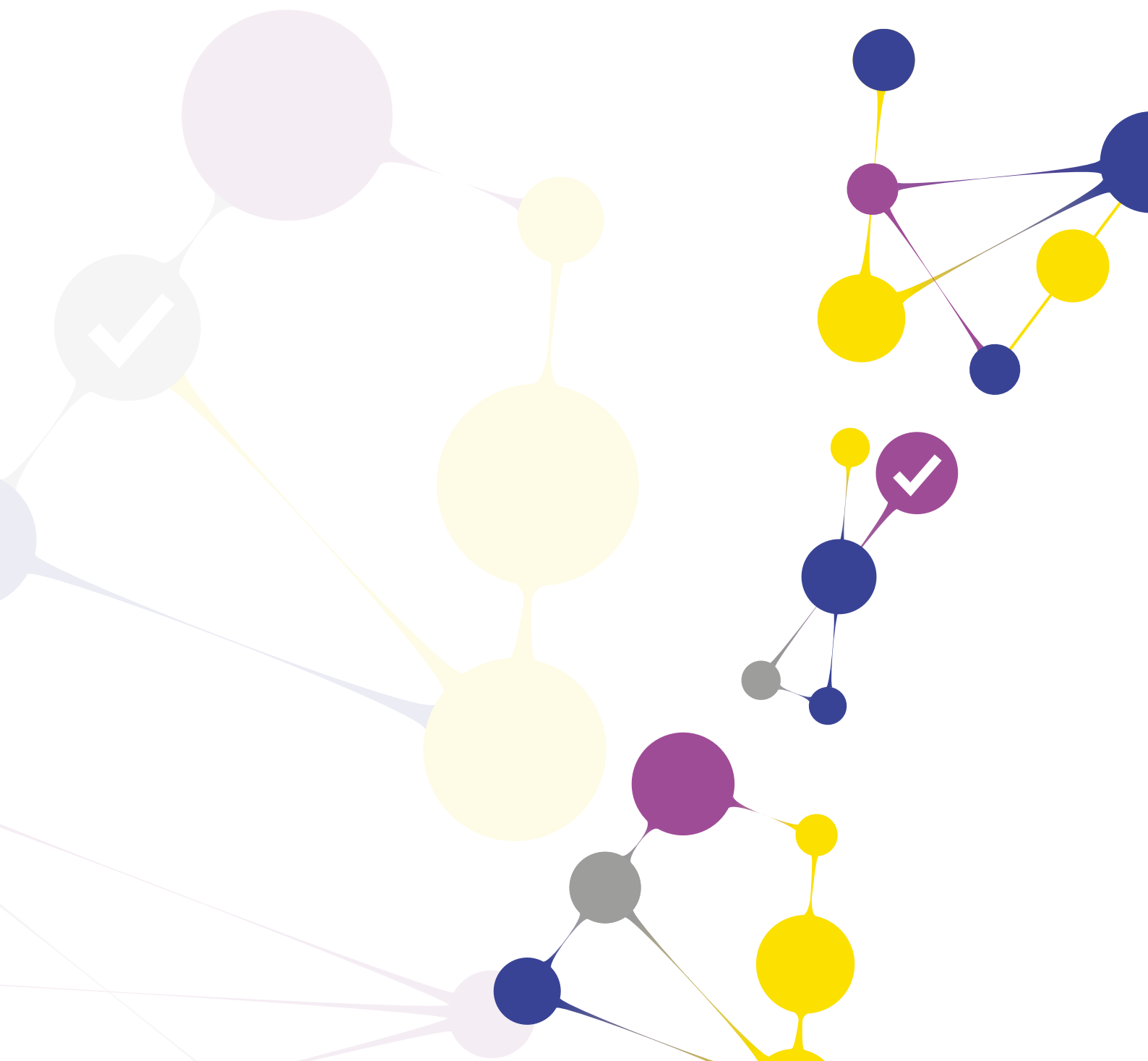
Scheme code	ONCOPANEL-18
Target	Mutations in the <i>EGFR</i> , <i>PIK3CA</i> , <i>KRAS</i> , <i>HRAS</i> , <i>NRAS</i> , <i>KIT</i> , <i>TP53</i> and <i>BRAF</i> genes.
Sample Material	Rolled sections of formalin fixed paraffin embedded (FFPE) materials designed to simulate a real patient sample. NOTE: Samples provided as provided as cut sections (rolled scrolls) ONLY we cannot provide slide mounted materials.
Scheme Format	<p>Assessment of genotyping ONLY.</p> <p>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.</p> <p>No restrictions on number of participants.</p> <p>Open to laboratories from ALL countries.</p> <p>3 mock clinical cases with matching samples 3</p>
Reporting Language	Reports accepted in English ONLY
Additional Information	This scheme is being offered to help labs using high thorough put technologies (e.g., NGS, MassArray etc) 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



Technique-specific EQA Schemes

Seven EQA schemes are being provided in 2018. 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.](#)





Constitutional Microarray analysis (Microarrays / arrayCGH)

INTERLECTUAL DISABILITY

TECHNICAL

INHERITED

GERMLINE

Scheme code	aCGH-18
Target	Genomic deletions and duplications
Sample Material	DNA samples (in TE Buffer)
Scheme Format	<p>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.</p> <p>-No restrictions on number of participants. Open to laboratories from ALL countries. 2 mock clinical cases with matching samples. ②</p>
Reporting Language	Reports accepted in English ONLY.
Additional Information	Platform independent - participants use their normal methodology.
Performance criteria	✔ Performance criteria DO apply.
Accreditation	★ This scheme is covered by the scope of EMQN's accreditation.
Collaborator	CEQAS



DNA Sequencing NGS (vGermline) (pilot)

SEQUENCING

TECHNICAL

INHERITED

GERMLINE

NGS

Scheme code	NEXTGEN (G) -18
Target	Not specified. The laboratory can choose.
Sample Material	g.DNA (in TE Buffer)
Scheme Format	<p>Assessment of genotyping and quality of raw data.</p> <p>Designed specifically for labs doing NGS based GERMLINE testing ONLY. Labs doing somatic mutation testing should register for the separate SOMATIC version of the scheme.</p> <p>Wet lab based exercise distributing g.DNA samples for testing.</p> <p>Any NGS strategy can be used (single gene, panel testing or exome sequencing).</p> <p>No restrictions on number of participants.</p> <p>Open to laboratories from ALL countries.</p> <p>1 mock clinical cases with matching samples. ①</p>
Reporting Language	English ONLY
Additional Information	None
Performance criteria	⊗ Performance criteria DO NOT apply.
Accreditation	⊗ This scheme is NOT covered by the scope of EMQN's accreditation.
Collaborator	UK NEQAS for Molecular Genetics (www.ukneqas-molgen.org.uk)



DNA Sequencing NGS (vSomatic) (pilot)

SEQUENCING

TECHNICAL

AQUIRED

SOMATIC

NGS

Scheme code	NEXTGEN (S) -18
Target	Not specified. The laboratory can choose
Sample Material	g.DNA sample derived from FFPE material. Matching control g.DNA sample also included.
Scheme Format	<p>Assessment of genotyping and quality of raw data.</p> <p>Designed specifically for labs doing NGS based SOMATIC testing ONLY. Labs doing germline mutation testing should register for the separate GERMLINE version of the scheme.</p> <p>Wet lab based exercise distributing DNA samples for testing Any NGS strategy can be used (single gene, panel testing, exome or genome sequencing).</p> <p>No restrictions on number of participants.</p> <p>Open to laboratories from ALL countries.</p> <p>1 mock clinical cases with matching samples. ①</p>
Reporting Language	English ONLY
Additional Information	None
Performance criteria	⊗ Performance criteria DO NOT apply.
Accreditation	⊗ This scheme is NOT covered by the scope of EMQN's accreditation.
Collaborator	UK NEQAS for Molecular Genetics (www.ukneqas-molgen.org.uk)



DNA Sequencing Sanger (Data quality assessment only version)

SEQUENCING

TECHNICAL

Scheme code	SEQ-18-QUAL
Target	Sanger DNA sequencing (gene independent)
Sample Material	DNA (in TE Buffer)
Scheme Format	<p>Assessment of raw data only.</p> <p>Laboratories which do genotype analysis should register for the full version of this scheme (see page 56).</p> <p>Wet lab based exercise distributing g.DNA samples for testing.</p> <p>No restrictions on number of participants.</p> <p>Open to laboratories from ALL countries.</p> <p>4 mock clinical cases with matching samples. 4</p>
Reporting Language	N/A
Additional Information	Suitable for Sanger sequencing technologies ONLY.
Performance criteria	<input checked="" type="checkbox"/> Performance criteria DO apply.
Accreditation	<input checked="" type="checkbox"/> This scheme is covered by the scope of EMQN's accreditation.
Collaborator	<input type="checkbox"/> None



DNA Sequencing Sanger (Full version)

SEQUENCING

TECHNICAL

Scheme code	SEQ-18-FULL
Target	Sanger DNA sequencing (gene independent)
Sample Material	DNA (in TE Buffer)
Scheme Format	<p>Assessment of genotyping, diagnostic interpretation, mutation nomenclature <u>and</u> quality of raw data.</p> <p>Wet lab based exercise distributing g.DNA samples for testing. No restrictions on number of participants. Open to laboratories from ALL countries. 4 mock clinical cases with matching samples. 4</p>
Reporting Language	N/A
Additional Information	Suitable for Sanger sequencing technologies ONLY..
Performance criteria	✔ Performance criteria DO apply.
Accreditation	★ This scheme is covered by the scope of EMQN's accreditation.
Collaborator	⊖ None



Non-invasive prenatal testing (NIPT) for common aneuploidies (pilot)

SCREENING

PLASMA

NGS

TECHNICAL

ACQUIRED

Scheme code	NIPT(ANEUPLOIDY)-18
Target	NIPT for the 3 most common aneuploidies (Chr 13, 18 and 21)
Sample Material	Plasma sample(s) containing cffDNA
Scheme Format	The final format of this pilot scheme is still to be confirmed.
Reporting Language	Reports accepted in English ONLY
Additional Information	Registration will be by invitation only please contact the EMQN office (office@emqn.org) for more information.
Performance criteria	⊗ Performance criteria DO NOT apply.
Accreditation	⊗ This scheme is NOT covered by the scope of EMQN's accreditation.
Collaborator	UKNEQAS for Molecular Genetics , and CEQAS



Non-invasive prenatal testing (NIPT) for fetal sexing (pilot)

INHERITED

NGS

PLASMA

SEQUENCING

TECHNICAL

Scheme code	NIPT(SEXING)-18
Target	NIPT for the fetal sexing
Sample Material	Plasma sample(s) containing cfDNA
Scheme Format	The final format of this pilot scheme is still to be confirmed.
Reporting Language	Reports accepted in English ONLY
Additional Information	<p>Registration will be by invitation only please contact the EMQN office (office@emqn.org) for more information.</p> <p>Please register an expression of interest by purchasing the scheme via our website</p> <p>Note this does not guarantee a place on the scheme. The final decision will be taken by the EMQN team based on data collected by us to ensure that the pilot scheme meets the needs of all labs and technical approaches</p>
Performance criteria	<input checked="" type="checkbox"/> Performance criteria DO NOT apply.
Accreditation	<input checked="" type="checkbox"/> This scheme is NOT covered by the scope of EMQN's accreditation.
Collaborator	UKNEQAS for Molecular Genetics



How to participate

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

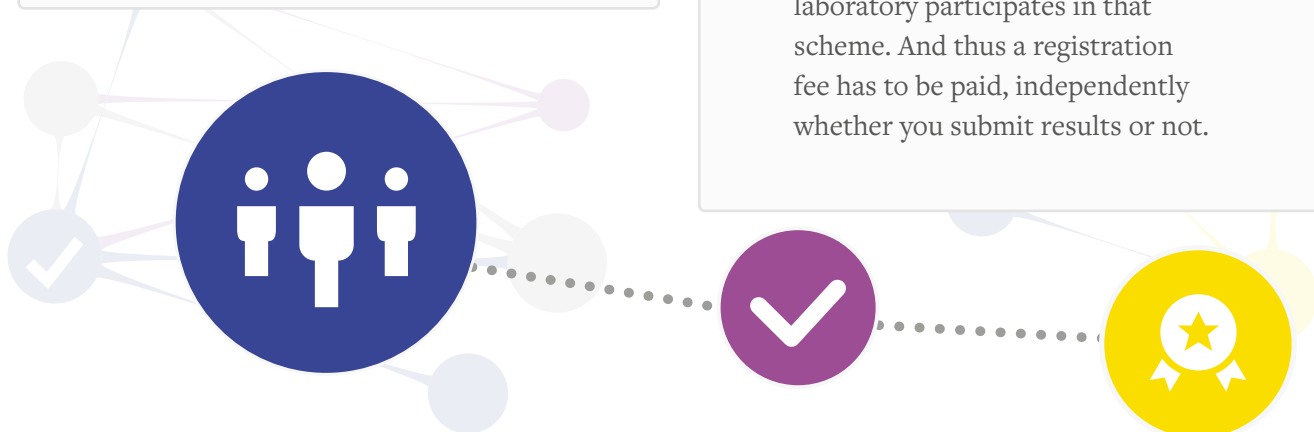
To participate in our EQA schemes you need to be a registered member of EMQN. There is a fee for this which is payable EVERY year.

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 on the EMQN website (www.emqn.org).
- Click the “Registration” button and select the scheme(s) in which you wish to participate.
- Please check that your laboratory details are up to date including the laboratory delivery address, primary contact email address and telephone number to ensure the smooth running of the scheme.
- Each laboratory will be charged an annual registration fee of £55 (regardless of the number of schemes applied for). This will be invoiced automatically.
- If you register to participate in a scheme, we assume that your laboratory participates in that scheme. And thus a registration fee has to be paid, independently whether you submit results or not.





Participation Fees

EMQN is a not-for-profit, self-funding organisation. We recognise the financial constraints being imposed upon many laboratories and therefore we keep our participation fees as low as possible.

Laboratories based within the European Union (EU) must supply their VAT/Tax number so the UK 20% charge is not applied to the invoice. All laboratories based out-with the EU will not be charged any VAT/Tax. If you require any further information then please contact the EMQN office (office@emqn.org).

Laboratories in developing / evolving economies are eligible to apply for our “Evolving Economies Discount “ that is offered to encourage participation. [Please see our website for more information](#)

MEMBERSHIP FEE	ANNUAL Fee (GBP, £)
All Laboratories	55
Additional staff users	50 per user
Germline mutation testing EQA schemes	ANNUAL Fee (GBP, £)
Arrhythmias	100
Autosomal Dominant Polycystic Kidney disease	285
Beckwith-Wiedemann and Silver-Russell syndromes	285
Breast / Ovarian Cancer, familial	285
Charcot-Marie-Tooth disease	285
Congenital Adrenal Hyperplasia	285
Duchenne / Becker Muscular Dystrophies	285
Familial Adenomatous Polyposis Colon cancer	285
Familial Autosomal Dominant Hypercholesterolemia	285
Fragile X Syndrome	285
Friedreich Ataxia	285
Hereditary Deafness	285
Hereditary Haemochromatosis	285
Hereditary Recurrent Fevers	285
Huntington Disease	285
Hypertrophic Cardiomyopathies	100
Lynch Syndrome	285
Mitochondrial DNA (mtDNA) Metabolic Disorders	285
Monogenic Diabetes	285
Multiple Endocrine Neoplasia (Type 2)	285
Myotonic Dystrophy	285
Osteogenesis Imperfecta	285



Germline mutation testing EQA schemes (cont.) **ANNUAL Fee (GBP, £)**

Phenylketonuria	285
Porphyrias	285
Prader-Willi and Angelman syndromes	285
Retinoblastoma	285
Short Stature Homeobox Gene Testing	285
Spinal Muscular Atrophy	285
Spinocerebellar Ataxias	285
Von Hippel Lindau Syndrome	285
Wilson Disease	285
Y-Chromosome Microdeletion testing	285

Somatic mutation testing EQA schemes **ANNUAL Fee (GBP, £)**

Molecular testing in Melanoma	432
Molecular testing in Lung cancer	432
Molecular testing in Colorectal cancer	432
Molecular testing for Oncogenes (panel testing).	432
Molecular testing (germline) of BRCA genes in Ovarian cancer	285
Molecular testing (somatic) of BRCA genes in Ovarian cancer	285
Molecular testing for EGFR gene mutations in ctDNA (pilot)	100
Molecular testing for RAS gene mutations in ctDNA (pilot)	100

Technique specific EQA schemes **ANNUAL Fee (GBP, £)**

Constitutional Microarray (arrayCGH)	360
DNA Sequencing (Sanger)	285
DNA Sequencing (NGS v Germline)	600
DNA Sequencing (NGS v Somatic)	600
Non-invasive prenatal testing (NIPT) for common aneuploidies (pilot)	360
Non-invasive prenatal testing (NIPT) for fetal sexing (pilot)	360

Invoices

EMQN is hosted within the Department of Genomic Medicine, St Mary's Hospital, Central Manchester University Hospitals NHS Foundation Trust (CMFT), Manchester, UK. Therefore all EMQN invoicing will be performed by CMFT. If you require a purchase order to register for the EQA scheme then please contact the EMQN Office (office@emqn.org) for account details. **Please ensure the annual registration fee is included in the total amount stated on the purchase order.**



EQA scheme timetable

R = registration period, S = survey period

Scheme	Page	September -1	October -1	November -1	December -1	January	February	March	April	May	June	July=y	August	September	October	November	December
GERMLINE MUTATION TESTING EQA'S																	
Arrhythmias	8	R	R	R		S	S	S									
Autosomal Dominant Polycystic Kidney disease	9	R	R	R		S	S	S									
Beckwith-Wiedemann and Silver-Russell syndromes	10	R	R	R		S	S	S									
Breast / Ovarian Cancer, familial	11	R	R	R		S	S	S									
Charcot-Marie-Tooth disease	13	R	R	R		S	S	S									
Congenital Adrenal Hyperplasia	14	R	R	R		S	S	S									
Duchenne / Becker Muscular Dystrophies	15	R	R	R		S	S	S									
Familial Adenomatous Polyposis Colon cancer	16	R	R	R		S	S	S									
Familial Autosomal Dominant Hypercholesterolemia	17	R	R	R		S	S	S									
Short Stature Homeobox Gene Testing	18	R	R	R		S	S	S									
Fragile X Syndrome	19	R	R	R		S	S	S									
Friedreich Ataxia	21	R	R	R		S	S	S									
Hereditary Deafness	22	R	R	R		S	S	S									
Hereditary Haemochromatosis	23	R	R	R		S	S	S									
Hereditary Recurrent Fevers	24	R	R	R		S	S	S									
Huntington Disease	25	R	R	R		S	S	S									
Hypertrophic Cardiomyopathies	26	R	R	R		S	S	S									
Lynch Syndrome	27	R	R	R		S	S	S									



R = registration period, S = survey period

Scheme	Page	September -1	October -1	November -1	December -1	January	February	March	April	May	June	July=y	August	September	October	November	December
Mitochondrial DNA (mtDNA) Metabolic Disorders	28	R	R	R		S	S	S									
Monogenic Diabetes	29	R	R	R		S	S	S									
Multiple Endocrine Neoplasia (Type 2)	30	R	R	R		S	S	S									
Myotonic Dystrophy	31	R	R	R		S	S	S									
Osteogenesis Imperfecta	32	R	R	R		S	S	S									
Phenylketonuria	33	R	R	R		S	S	S									
Porphyrias	34	R	R	R		S	S	S									
Prader-Willi and Angelman syndromes	35	R	R	R		S	S	S									
Retinoblastoma	36	R	R	R		S	S	S									
Spinal Muscular Atrophy	37	R	R	R		S	S	S									
Spinocerebellar Ataxias	38	R	R	R		S	S	S									
Von Hippel Lindau Syndrome	39	R	R	R		S	S	S									
Wilson Disease	40	R	R	R		S	S	S									
Y-Chromosome Microdeletion testing	41	R	R	R		S	S	S									
SOMATIC MUTATION TESTING EQA'S																	
Molecular testing for EGFR gene mutations in ctDNA (pilot)	43	R	R	R	R	R								S	S	S	
Molecular testing for RAS gene mutations in ctDNA (pilot)	44	R	R	R	R	R								S	S	S	



R = registration period, S = survey period

Scheme	Page	September -1	October -1	November -1	December -1	January	February	March	April	May	June	July=y	August	September	October	November	December
Molecular testing in Lung cancer	45	R	R	R	R	R					S	S	S				
Molecular testing in Melanoma	46	R	R	R	R	R					S	S	S				
Molecular testing in Colorectal cancer	47	R	R	R	R	R					S	S	S				
Molecular testing (germline) of BRCA genes in Ovarian cancer	48	R	R	R	R	R								S	S	S	
Molecular testing (somatic) of BRCA genes in Ovarian cancer	49	R	R	R	R	R								S	S	S	
Molecular testing for Oncogenes (panel testing).	50	R	R	R	R	R					S	S	S				
TECHNICAL EQA'S																	
Constitutional Microarray (arrayCGH)	52	R	R	R	R	R				S	S	S					
DNA Sequencing (NGS v Germline)	53	R	R	R	R	R								S	S	S	
DNA Sequencing (NGS v Somatic)	54	R	R	R	R	R								S	S	S	
DNA Sequencing (Sanger)	55	R	R	R	R	R					S	S	S				
Non-invasive prenatal testing (NIPT) for common aneuploidies (pilot)	57	R	R	R	R	R								S	S	S	
Non-invasive prenatal testing (NIPT) for fetal sexing (pilot)	58	R	R	R	R	R								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.ukas.com/wp-content/uploads/schedule_uploads/00013/4367Proficiency%20Testing%20Single.pdf. EQA schemes marked symbol are currently not included within the scope of accreditation.



4367





Terms and Conditions

The 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 as listed below.

Membership of EMQN (v7, reviewed January 2017)

An annual fee is charged to register as a member of the network. The membership period runs from 01 January to 31 December each year and the membership fee is payable every year. As a member of EMQN, you can participate in our EQA schemes and best practice meetings. Registered members of EMQN will receive a username and password which will allow them to access the member-only services (such as EQA, best practice etc.)

The registration fee to join the network is 55 GBP. There is a discount for any additional staff member registrations from the same laboratory 50 GBP.

Terms & conditions are:

1. There is an EMQN membership fee payable annually this must be paid every year to ensure lab membership remains valid.
2. To participate in our EQA schemes and best practice meetings, you must be a registered member of EMQN.
3. You must keep your password secure and confidential.
4. Your registration will only be processed when you have completed the registration and payment forms correctly.
5. Payments cannot be processed without correct billing information. Failure to provide this information accurately may result in delays to your participation. EMQN cannot accept responsibility for you not receiving our services if we do not get the correct information.
6. Payments should be made within 14 days of receipt of an invoice from EMQN (invoices will be sent by our host institution, Central Manchester and Manchester Children's NHS Trust). After this date we the host institution will send a reminder to you to pay but EMQN reserves the right to withdraw services after an appropriate time limit.
7. Payments by invoice or credit card are the only-options. Personal cheques are NOT accepted.
8. In order to use the EMQN services in the members only area you must have email which allows receipt of attachments and have Adobe Acrobat Reader installed on your PC (if you do not yet have it, go to <http://www.adobe.com/> to download a free copy).
9. Registered members of EMQN who have not paid their membership fees will have their accounts suspended until the outstanding fees have been paid. Account suspension will result in accessing being blocked to their EMQN website member's area.



The following additional terms and conditions apply to the evolving economies discount scheme available to labs who meet the criteria available from the EMQN office:

1. The laboratory wishes to apply for a discount in the cost of fees for EQA scheme participation.
2. The laboratory agrees to pay the minimum annual EMQN registration fee each year.
3. The laboratory agrees to participate for 5 consecutive years in each EQA scheme that they register for.
4. The laboratory agrees to pay the full EQA scheme fees for the fifth year of participation.
5. If the laboratory fails to participate in any of the five years without first notifying EMQN, then the full cost of participation for those years is payable.
6. The laboratory must notify EMQN immediately of any changes to their contact information.

EQA scheme participation (v7, reviewed January 2017)

Participation in the EMQN external quality assessment (EQA) schemes is on a voluntary, confidential basis. All laboratories are actively encouraged to participate in all relevant schemes.

Participation in our EQA schemes is open also to commercial manufacturers of relevant instruments and reagents, and to pharmaceutical, veterinary and other laboratories. Participation is anonymous laboratories are identified by a code number known only to the EMQN office staff.

Terms and conditions are:

1. The Head of the Laboratory will be responsible for registering the laboratory with the EMQN as a participant in the appropriate EQA Schemes. Any changes in the laboratory's requirements in this respect must be notified in writing to the EMQN Director.
2. Samples, reports and routine correspondence may be addressed to a named deputy, but correspondence from EMQN concerning poor performance or unsatisfactory return rates, will be sent directly to the Head of the Laboratory.
3. EQA samples must be treated in the same way as clinical samples.
4. EQA materials are supplied under the strict condition that they are used by the registered participant laboratory for external quality assessment tests only. The materials supplied must not be used as internal controls in molecular genetic tests or for any other purpose. Participant laboratories undertake to destroy EQA materials (please email office@emqn.org and request a DOC2918 Materials Certificate of Destruction form) or return them to EMQN within eight weeks of the published date of the closure of the scheme.



5. Participants must not collude with other laboratories on the results of their EQA scheme participation.
6. The EQA code number of the laboratory and the assessment of individual performance are confidential to the participant and will not be released by EMQN to any third party (except in circumstances described in conditions 6 and 7) without the written permission of the Head of the Laboratory.
7. The identity of participants (name of laboratory and Head of Laboratory) and the tests for which they are registered (but not details of performance, or the EQA code number of the lab) will be shared with the curators of the Orphanet Quality Assurance Database (<http://www.orpha.net/consor/cgi-bin/ClinicalLabs.php?lng=EN>). If a lab does not wish this information to be shared then they must notify the EMQN Director in writing.
8. Exceptions:
 - a. Individual participants' performance information may be shared with national/regional authorities with responsibility for laboratory standards and/or patient safety following approval by the EMQN board.
 - b. Aggregate performance information may be shared with a relevant national coordinators following approval by the EMQN board.
 - c. Performance information of laboratories registered with the HGSA/RCPA Quality Assurance Programs (QAP) is shared with HGSA/RCPA.
 - d. Performance information of Swiss laboratories is shared with CSCQ.
 - e. Performance information of United Kingdom laboratories is shared with NQAAP.
9. The EMQN sometimes collaborates with other EQA providers, for example, UK NEQAS Molecular Genetics, CEQAS etc. When this occurs, participant confidentiality is maintained.
10. When a laboratory shows unsatisfactory performance or fails to return results, the EMQN will make contact with the participant. Advice is then offered to the Head of the Laboratory by contact, usually in writing.
11. Problems relating to EQA schemes, including complaints from participating laboratories, should be referred directly to the EMQN office.
12. All documents issued by the EMQN, and the data they contain, are subject to copyright and may not be published in any form without the permission of the EMQN Board.
13. A special EMQN logo may be used on participants' publicity materials or web sites if permission has been granted by the EMQN. Please contact us on office@emqn.org for permission.



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 database by a code known only to the EMQN office staff.

Assessment

The genotypes of the scheme samples are published 7 days after the closing date for reporting.

Marks are allocated according to fixed criteria agreed by the scheme organiser 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 the scheme organiser and two expert assessors from different countries, at least one having experience of EQA schemes. 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 procedures. 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 and all marking is moderated and differences resolved at a meeting between the scheme organiser and 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. You must keep your results private and not collude with other laboratories. If you cannot complete your tests for any reason then contact EMQN. If the testing process fails you can request replacement samples.

Reporting

You are asked to prepare a report 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.

For general guidance on reporting, please see [European Journal of Human Genetics 2013;10.1038/ejhg.2013.125](https://doi.org/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 network 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 the 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.

EQA samples are validated by the scheme organiser and at least one other expert centre. Genotypes are generally checked by more than one analytical method.

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 be 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.

Sample receipt

Each sample dispatch will be accompanied with a dispatch note, a covering letter giving basic instructions, reporting dates and where to get more information.

We ship different sample materials depending on the EQA scheme. These can include DNA, plasma, and FFPE materials. The correct storage conditions for each material are clearly described in the instructions which accompany each scheme.

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.

When you receive the samples, please check the dispatch note carefully, and check the samples for damage. Notify the 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 and Management Board). Only your laboratory's allocated unique EMQN reference number will identify its scores within distributed summary reports. The fact that your laboratory participates in EMQN schemes is not confidential.



Please see the following documents for the relevant terms and conditions of membership and EQA scheme participation:

[Membership T&Cs](#)

[EQA scheme participation T&Cs](#)

Replacement Samples

Replacement samples cannot be guaranteed, but will be sent if available. EMQN will require a justification, and reserve the right to charge for replacement samples to cover reasonable costs. Testing of replacements must be within the same timeframe as the originals.

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, 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 re-occurring problems / mistakes is made available via the 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 results) to submit a written appeal to the EMQN Office. An appeal must be submitted online, using a form available from the website. The appeal will be considered by the scheme assessment team and a response communicated back to the the lab via the EMQN Office.

In cases where the scheme organiser and assessors cannot come to a conclusion, the appeal is forwarded to the EMQN board for a decision. **The EMQN cannot consider any appeal submitted after the set deadline**, so in this case the originally assigned marks will stand.





Feedback to EMQN

The EMQN office welcomes written comments about EMQN schemes at any time. Confidential communications about a scheme should be made to the EMQN Office. A customer satisfaction survey is sent out with each final scheme report.

Complaints procedure

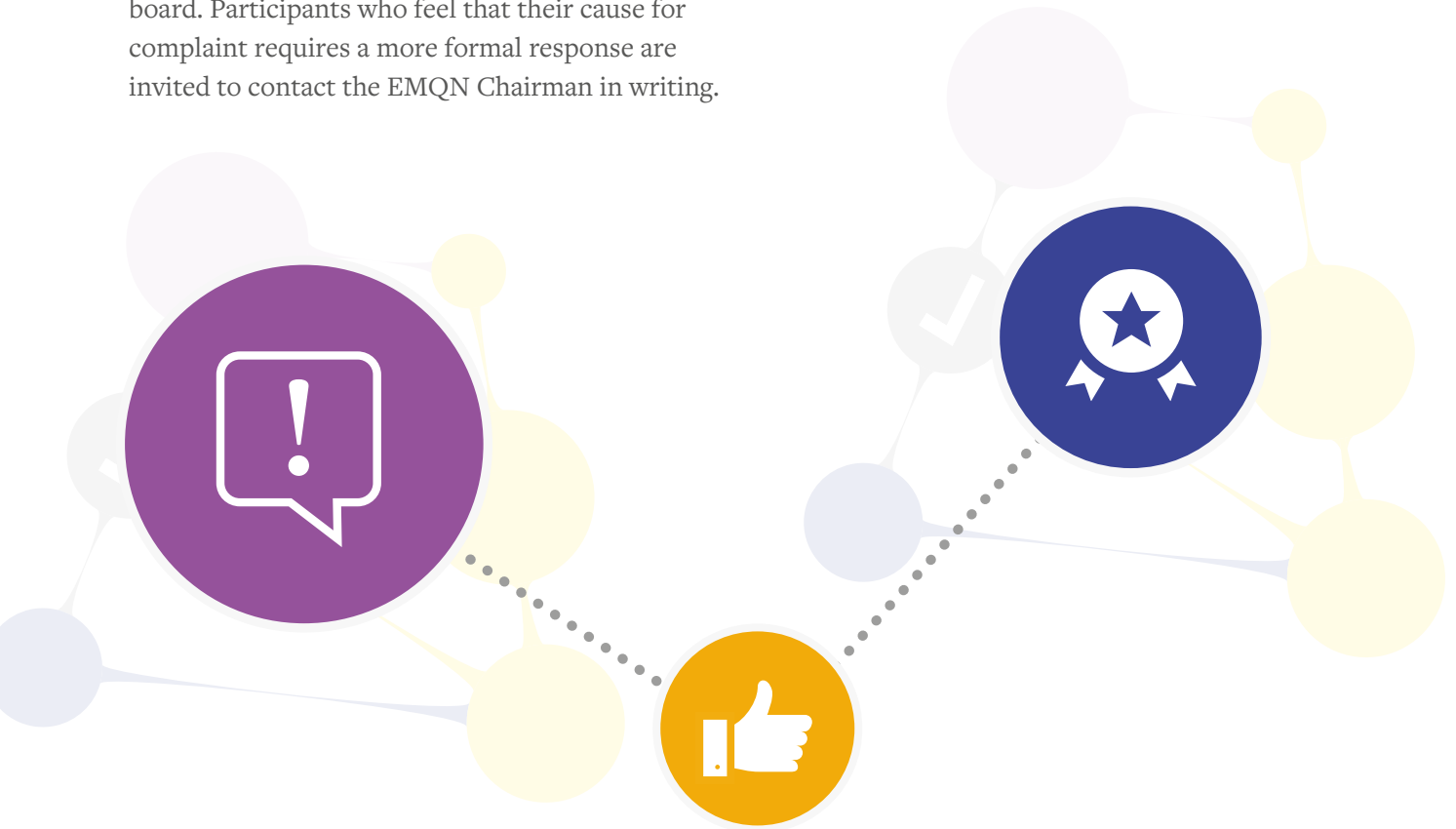
Most complaints received by the EMQN consist of minor misunderstandings or problems with specimens. These can usually be resolved over the telephone with the EMQN office staff. If a complaint is received it will be logged along with the action taken. The EMQN Office Staff will attempt to address the complaint as soon as possible by letter. If the participant is not satisfied with the response by the EMQN Office Staff then the matter will be brought to the EMQN management board at their next meeting. A response will be made in light of the advice given by the management board. Participants who feel that their cause for complaint requires a more formal response are invited to contact the EMQN Chairman in writing.

Collusion

Scheme participants are reminded that it is the duty of the scheme provider to prevent collusion between participating laboratories. Participants found colluding in their scheme returns may be excluded from participation in future scheme.

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 work by sub contracted activities.





The European Molecular Genetics Quality Network

European Molecular Genetics Quality Network

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