

2019 EQA Scheme for Molecular testing of BRCA genes in Ovarian Cancer

1. Who are EMQN and GenQA?

The European Molecular Genetics Quality Network (EMQN) is the largest external quality assessment network for molecular genetic testing in the world. Membership is open to public and private testing laboratories, commercial manufacturers of relevant instruments and reagents, and to pharmaceutical and other laboratories. The EMQN is based in the Department of Genomic Medicine in St Mary's Hospital, Manchester, UK.

Genomic Quality Assessment (GenQA) is a collaboration between CEQAS and UK NEQAS Molecular Genetics, both of whom are members of the UK NEQAS consortium. The organisation provides a comprehensive world-wide EQA service for genomics laboratories. GenQA is based in the Department of Laboratory Medicine in The Royal Infirmary of Edinburgh, Edinburgh, UK.

External Quality Assessment (EQA) schemes are designed to test the whole analytical process of molecular testing procedures including the ability to interpret data in the light of clinical information, and to produce a clear and accurate report. By participating in these schemes' laboratories gain recognition for their activities, may help drive quality improvements and innovation, and gain access to help and advice from a network of international experts.

Both EQA providers are ISO 17043 accredited and this EQA scheme is covered within each organisation's accreditation scope.

EMQN and GenQA are collaborating to deliver two different BRCA Ovarian Cancer schemes; one for Germline mutation testing and another for Somatic mutation testing.

2. Description of the EQA schemes

Participant laboratories register for one or both schemes and receive 3 samples (per scheme) with corresponding mock clinical cases.

The laboratories have 8-10 weeks to perform mutation analysis of the *BRCA1* and *BRCA2* genes using their routine procedures and report their findings. Assessment includes genotyping, and biological and clinical interpretation within the context of the clinical case scenarios provided relating to targeted PARP inhibitor treatment.

All reports are assessed anonymously and independently by at least 2 expert assessors against peer ratified criteria. Participants receive their marks and a final report showing any general take home messages and improvement needs. All laboratories will receive a certificate of participation on completion of the assessment

2.1. Target

- Mutations in the *BRCA1* and *2* genes.

2.2. Sample Material

- **Germline scheme:** g.DNA in TE buffer
- **Somatic scheme:** Rolled sections of artificial reference materials. **NOTE** we cannot provide slide mounted materials.

2.3. Scheme Format

- Suitable for any molecular based assay e.g. PCR, MLPA and/or NGS based testing methodologies.
- 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.
- We only require clinically relevant pathogenic (disease-causing) mutations to be reported, and not common SNPs / variants.
- Maximum of 300 participant laboratories per scheme.
- Open to laboratories from ALL countries.
- 3 mock clinical cases with matching samples (per scheme)

2.4. Reporting Language

- Reports accepted in English ONLY

2.5. Performance criteria

- Performance criteria DO apply.

2.6. Accreditation

This scheme is covered by the scope of EMQN and GenQA's accreditation.

3. How to participate

The schemes are a collaboration between both EQA providers and therefore identical. There is NO difference in the cost, or assessment process.

Laboratories are advised to register for the germline and/or somatic schemes via **ONE of the EQA providers only** – please do not register twice via EMQN and GenQA

Laboratories **MUST be members of EMQN or GenQA to gain access to the scheme.** Non-members must join one of the providers first to gain access to the scheme.

3.1. Already a member of EMQN or GenQA?

- Login to your EQA providers account via www.emqn.org or www.genqa.org
- Goto the "Purchase" page and select / register for the "BRCA Ovarian (vGermline) EQA Scheme" and/or "BRCA Ovarian (vSomatic) EQA Scheme".
- Don't forget to "Complete" you purchase to ensure that we receive it.

3.2. Not a member of EMQN or GenQA?

- Join your preferred EQA provider by registering via their website (<https://www.emqn.org/joining-emqn/> or <https://www.genqa.org/d/AccountApplication.do>).
- Please allow 5 working days for approval of your new account.
- You will receive an email from the EQA provider once your account has been activated. If you have not had a confirmation email from us after 5 working days, then please contact the EQA providers office office@emqn.org or info@genqa.org.
- Once the EQA provider has approved your account, then please register for the scheme as per the procedure 3.1 above for existing members.

4. Cost of participation

- The costs of your participating in this EQA scheme will be covered by AstraZeneca.
- **Please note** that labs must cover their own costs for any other EQA schemes they may wish to participate from the EQA providers catalogues. AstraZeneca is only sponsoring participation in the two BRCA Ovarian Cancer EQA schemes.

5. Key Dates and Timelines

- | | |
|---|---|
| • Application period: | 1st September 2018 – 28th February 2019 |
| • Sample dispatch dates: | September 2019 |
| • EQA testing period: | September – November 2019 |
| • EQA results assessment: | November 2019 – January 2020 |
| • Results released to participant laboratories: | February 2020 |

6. Confidentiality

- The fact that your laboratory participates in the scheme is not confidential. However, the raw data and performance scores are.
- Further information on our terms and conditions and privacy / confidentiality policies can be found on the EQA providers website (www.emqn.org or www.genqa.org).

7. Contact information

- office@emqn.org or info@genqa.org for any queries regarding registration or participation in the scheme.



EMQN Office, Genetic Medicine, 6th Floor, St Mary's Hospital, Oxford Road, Manchester M13 9WL, United Kingdom. **Tel:** +44 161 276 6741 **Email:** office@emqn.org



GenQA [Edinburgh Office], Department of Laboratory Medicine, Royal Infirmary of Edinburgh, Edinburgh, EH16 4SA, United Kingdom. **Tel:** +44 131 242 6898 **Email:** info@genqa.org