

# Genomics Quality Assessment (GenQA)

## GenQA Latest

1922

Participating  
Laboratories

382

Expert  
Assessors

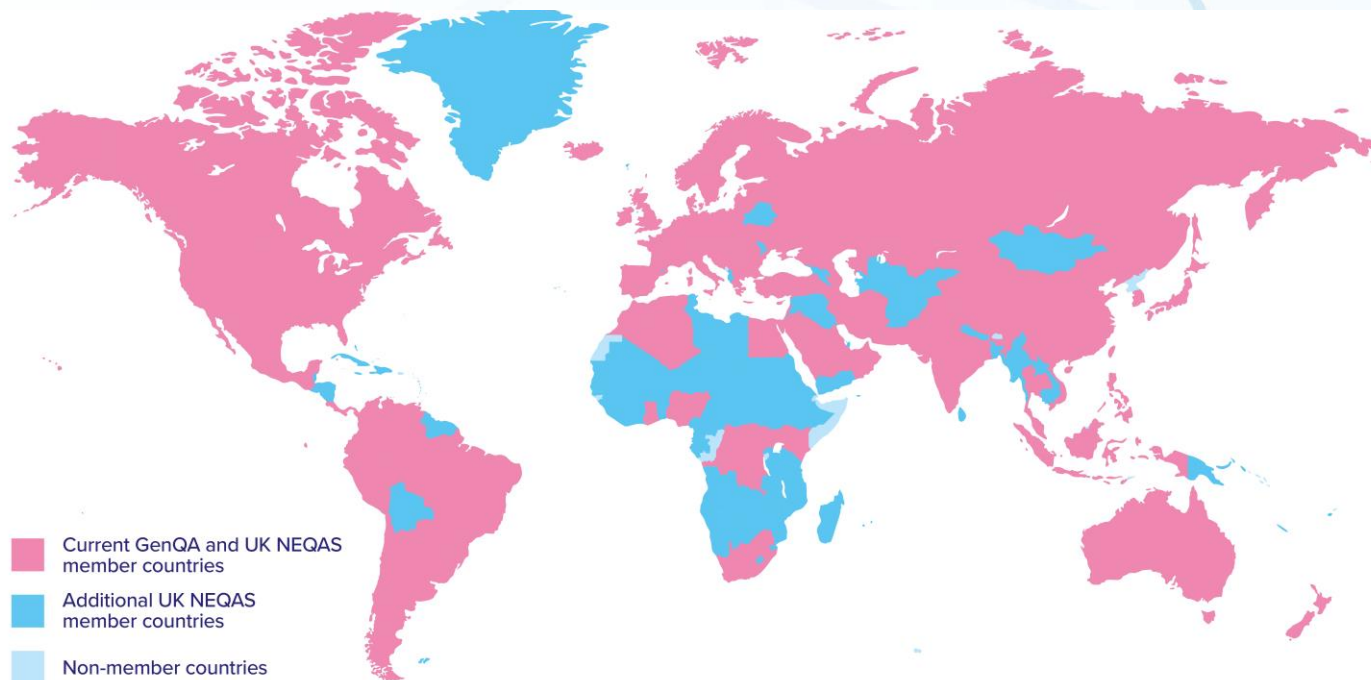
119

EQAs for 2023

89

Participating  
Countries

183  
Spanish  
Labs



GenQA is operated by Oxford University NHS Foundation Trust

## UK National External Quality Assessment Services

*“helping to ensure clinical laboratory test results are accurate, reliable and comparable wherever they are produced”*

- Charity organisation
- Self-funding, non-profit making schemes
- 25 specialist centres
- Over 50 years' experience providing EQA
- Participants worldwide
- ISO Accredited





Member of UK NEQAS consortium



World Health  
Organization

“EQA is defined as a system for objectively checking the laboratory’s performance using an external agency or facility.”



We’re driven to work in partnership with  
genomics centres  
- **both laboratories and clinicians**  
- **across the world**  
to provide high quality patient testing for  
genomic disorders and acquired diseases.



We know the **fast-evolving field of genomic medicine** will continue to advance, and our **scientific team** is focussed on delivering fit for purpose external quality assessments (proficiency testing) and individual competency assessments that continuously develop with the pace of change.

# EQAs cover the breadth of genomics services

**Reproductive  
Genomics**



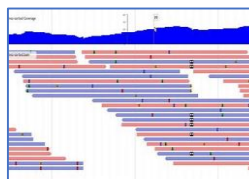
**Molecular Pathology**



**Genomic and  
Inherited Disorders**



**Technical:  
NGS and OGM**



**Haematological  
neoplasms**



**Clinical Genetics and  
Genetic Counselling**



**Sample Handling**



**Molecular Newborn  
Screening**



**Pharmacogenomics**

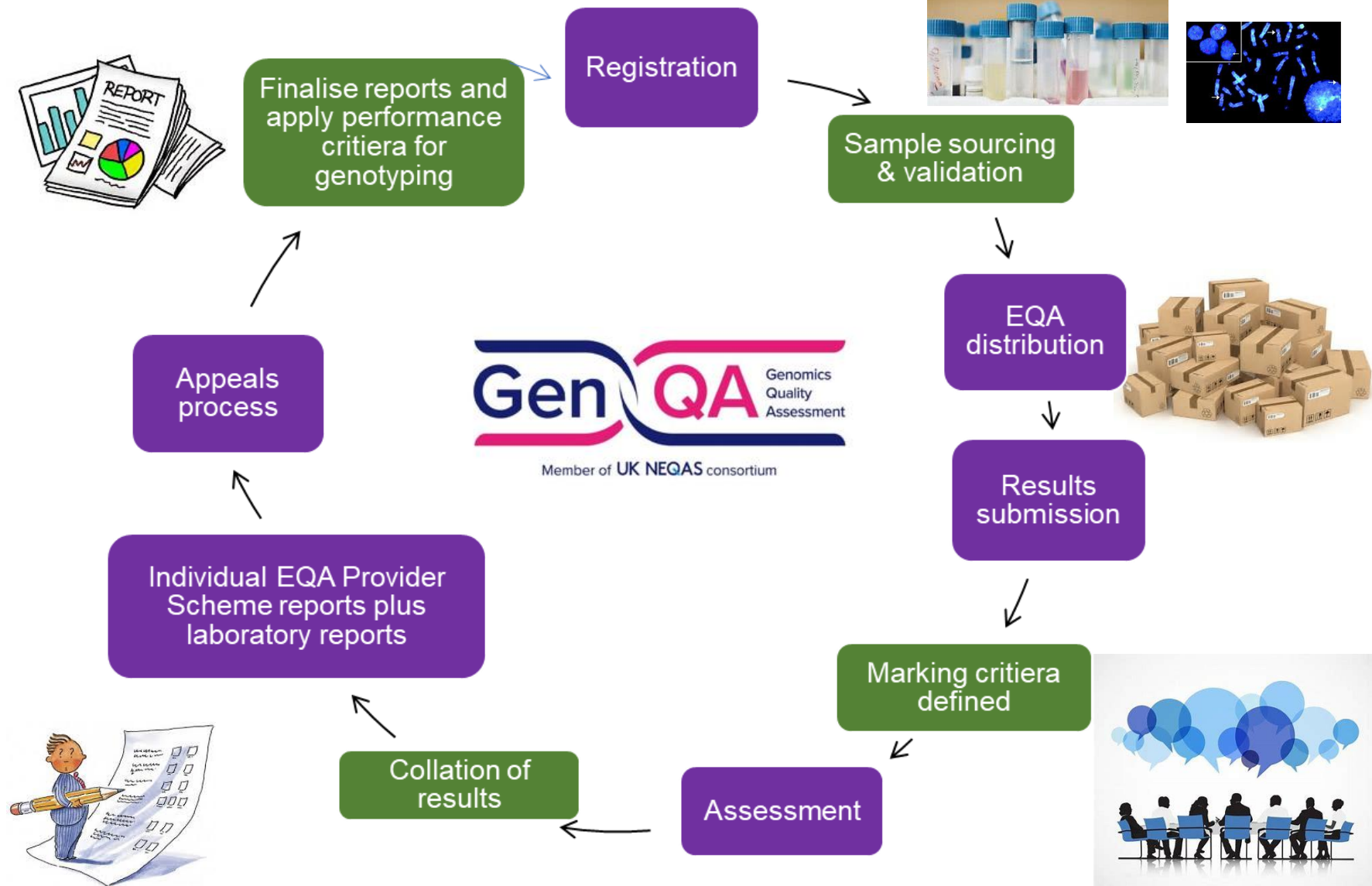


**Preimplantation  
Genetic Testing**





# Format of EQAs



# End to End testing

Pre-test  
consultation/  
Referral

Sample

Analysis

Interpretation

Reporting

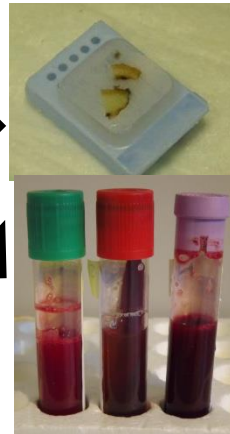
MDT

Consultation

## Pre-test referral



Consultation



Sample  
handling

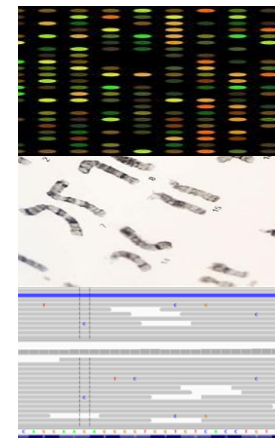


DNA extraction  
DNA quality

DNA quantity



Testing accuracy



Reporting



Interpretation



**Haematology SAG**

**Molecular Pathology SAG**



**Cancer Genomics  
Specialist Advisory Group**

**Co-chairs**  
**David Gonzalez de Castro – UK**  
**Blanca Espinet - Spain**

# Cancer Genomics EQAs

Acute Lymphoblastic Leukaemia
Chronic Lymphocytic Leukaemia
Haematological Technical FISH
Lymphoma
Lymphoma Technical NGS
Myeloid disorders (AML, MDS)
Multiple myeloma
CLL-IGHV mutation status
CLL- <i>TP53</i> analysis
Pathogenicity of haematological neoplasm variants
ISCN nomenclature
Optical Genome Mapping (pilot)

Breast Cancer - Tumour expression profiling
Central Nervous System tumours
Cholangiocarcinoma
Endometrial tumours
Gastrointestinal Stromal
Microsatellite Instability (MSI)
Molecular Tissue identification
<i>NTRK</i> fusions
Renal tumours
Sarcoma
Pathogenicity of somatic solid tumour SNVs,
BRCA testing for ovarian and prostate cancer - somatic
BRCA testing for ovarian, breast, prostate and pancreatic cancer - germline
BRCA testing in prostate cancer – cfDNA
Lung cancer - (cf) DNA in lung cancer
Lung cancer
Colorectal cancer
Melanoma



# Roles of an assessor

- To provide samples/images to aid in producing the EQAs (not mandatory)
- To aid in validating the EQA samples
- To assess EQAs using GenQA marking interface online – assessment meetings are held to discuss results following marking
- To provide advice for EQAs
- To potentially help with any participants that ask advice when they are having difficulties

# Advantages to being an assessor

- Chance to see ways different laboratories report results
- Can be used as evidence of CPD (GenQA will provide a certificate)
- Interaction with other experts in your field
- Potential to influence best practice in genomic medicine

## Scientific expert advisors

### Are you interested in:

- Finding out more about external quality assessment?
- Working with like-minded scientists worldwide?
- Supporting GenQA to shape the future of our EQAs?
- Reviewing your laboratory's report format against other anonymised laboratory reports?
- Gaining evidence for your continued professional development?

### To apply, you will require:

- Several years' experience of providing diagnostic testing and authorising clinical laboratory results/reports in the relevant EQA.

### The role may include:

- Sourcing/advising on suitable samples/images for EQA.
- Validating the test results for an EQA.
- Advise on content of clinical case scenarios.
- Advising on marking criteria.
- Assessing anonymised EQA reports/results submitted by participants.
- Advising on participant appeals.
- Contributing to EQA Summary reports.
- Attending the relevant Special Advisory Group (SAG) meetings.

Contact us: [info@genqa.org](mailto:info@genqa.org)  
providing a copy of your CV