2024 EQA Price List

(includes EQAs from 1st April 2024 - 31st March 2025)

For 2024, GenQA are delighted to offer 120 genomics external quality assessments (EQA) covering thirteen different disciplines which encompass the sample and patient journey. In addition we are offering a number of assessments for individual competency.

Free GenQA membership is required to purchase EQA. If you have any questions regarding membership or enrolment, please email info@genqa.org.

Please see our live calendar at www.genqa.org/calendar for EQA distribution/assessment dates.

For full details of all EQA offered go to www.genqa.org/eqa

Please browse this directory and contact us at info@genqa.org if you require any further information.

Clinical Genetics & Genetic Counselling

These multi-stage case scenarios follow the patient pathway from first consultation and request of appropriate genetic test(s) based on family history and clinical examination, through interpretation of test results, and finally to diagnosis and counselling of the patient.

- Cases designed to assess appropriate genomic testing requested based on clinical presentation and family history, interpretation of test results and relevant genetic counselling.
- 6 weeks to submit results
- Submitted results are assessed by an expert panel against recommended guidelines
- Each participant receives an Individual Centre Report (ICR) and EQA summary report

External Quality Assessment (EQA)	EQA Code	Sample	Results submission/assessment	Distribution month	ISO 17043 Status	Fee (£)
Clinical Genetics - Cardiovascular disorders	2024 CGC	Case scenario	Online interpretation of cardiovascular disorders with a genetic aetiology.	November	Not accredited	155
Clinical Genetics - Dysmorphology	2024 CGD	Case scenario	Online interpretation of syndromes involving dysmorphism with a genetic aetiology.	May	Not accredited	155
Clinical Genetics - Monogenic disorders	2024 CGM	Case scenario	Online interpretation of syndromes involving single gene(s) disorders.	November	Not accredited	155
Clinical Genetics - Oncogenetic disorders	2024 CGO	Case scenario	Online interpretation of cancers with a genetic predisposition or inherited.	November	Not accredited	155
Clinical Genetics – Inherited Metabolic disorders POSTPONED	2024 CGU	Case scenario	Online interpretation of metabolic disorders with a genetic aetiology.	November	Pilot	155
Genetic counselling	2024 CGG	Case scenario	Online interpretation of pre and post- test counselling for Cancer, Prenatal Diagnosis, and Genomic disorders/ diseases.	May	Not accredited	155
Genomic multi-disciplinary team (MDT) working^	2024 CGMDT	Case scenario	Online assessment of the full MDT process including pre-meeting, post-meeting and reporting.	November	Pilot	155

^This EQA is now listed under the Multidisciplinary team (MDT) working specialty.

For full details of all EQA offered go to www.genqa.org/eqa

Please browse this directory and contact us at info@genqa.org if you require any further information.

Genomic and Inherited disorders

Assess testing and reporting of different genomic and inherited disorders, using various sample types and/or online case scenarios e.g. diagnostic and carrier testing. Cases based on real clinical scenarios, using validated samples.

- Suitable for molecular and/or cytogenomic techniques
- Each EQA may contain some or all the disorders/genes indicated.
- 6-16 weeks to submit results
- Submitted laboratory reports are assessed, by an expert panel, using international guidelines/recommendations.
- Each participant receives an Individual Laboratory Report (ILR) and EQA summary report

Suitable for both cytogenomic and/or molecular testing (including Clinical Pathway EQAs):

External Quality Assessment (EQA)	EQA Code	Sample	Assessment[#]	Results submission	Distribution month	ISO 17043 Status	Fee (£)
Chromosome instability syndromes (previously Chromosome breakage syndromes) **UPDATED** Fanconi anaemia, Bloom syndrome, Ataxia telangiectasia and Nijmegen syndrome, Robert syndrome, ICR, Cornelia de Lange syndrome, mosaic variegated aneuploidy and Seckel syndrome	2024 GCI	Images and DNA	Full	Laboratory report	April	Accredited	390
Developmental delay (previously Severe Developmental Delay)	2024 GDD	Case scenario	Full	Laboratory report	April	Accredited	390
Differences in Sex Development (DSD) (previously Disorders of Sex Development) Androgen insensitivity syndrome, Congenital adrenal hyperplasia, cytogenomic abnormalities and other disorders associated with a DSD NGS panel	2024 GDSD	Case scenario / DNA	Full	Laboratory report	April	Accredited	390
Hypotonic Infant Spinal Muscular Atrophy type 1 (SMA), Prader Willi Syndrome (PWS) and Myotonic Dystrophy type 1 (DM1)	2024 GHI	DNA	Full	Laboratory report	April	Accredited	390
Imprinting disorders Angelman Syndrome (AS), Beckwith-Wiedemann Syndrome (BWS), Silver Russell Syndrome (SRS), Wilms tumour and Temple syndrome	2024 GIM	DNA	Full	Laboratory report	April	Accredited	390
Infertility Chromosomal mosaicism, *CFTR*, *FMR1*, Y-deletions	2024 GINF	Case scenario	Full	Laboratory report	April	Accredited	390
Microdeletion syndromes Prader-Willi syndrome, Angelman syndrome, Williams syndrome and Di-George syndrome	2024 GDEL	Fixed cells/DNA	Full	Laboratory report	May	Accredited	390
Postnatal constitutional copy number variants (CNV) detection[1]	2024 GCNV	DNA	Full	Laboratory report	May	Accredited	455

[#]Full assessment of analytical/genotyping processes and clinical interpretation of results

[1]Provided in collaboration with EMQN

For full details of all EQA offered go to www.genqa.org/eqa

Please browse this directory and contact us at info@genqa.org if you require any further information.

Suitable for cytogenomic testing only:

External Quality Assessment (EQA)	EQA Code	Sample	Assessment[#]	Results submission	Distribution month	ISO 17043 Status	Fee (£)
Postnatal karyotyping Structural chromosome abnormalities	2024 GPK	Images	Full	Laboratory report	May	Accredited	390

[#]Full assessment of analytical/genotyping processes and clinical interpretation of results

Suitable for molecular testing only:

External Quality Assessment (EQA)	EQA Code	Sample	Assessment[#]	Results submission	Distribution month	ISO 17043 Status	Fee (£)
Ataxia, including Hereditary Spastic Paraplegia (HSP) Friedreich ataxia, spinocerebellar ataxia and hereditary spastic paraplegia	2024 GATAX	DNA	Full	Laboratory report	April	Accredited	390
Calcium disorders Hypercalcaemia and hypocalcaemia including: Familial hypoparathyroidism, Albright hereditary osteodystrophy, Pseudohypoparathyroidism, Pseudopseudohypoparathyroidism, Isolated hyperparathyroidism, Hypocalciuric hypercalcaemia, Calcium sensing receptor phenotypes	2024 GCAL	DNA	Full	Laboratory report	June	Accredited	390
Cardiac disorders Cardiomyopathies, arrhythmia and aortic dissection: Brugada syndrome, Long QT syndrome, Catecholaminergic polymorphic ventricular tachycardia (CPVT), Marfan syndrome and Ehlers Danlos syndrome	2024 GCARD	DNA	Full	Laboratory report	April	Accredited	390
Charcot Marie Tooth disease and related sensory and motor neuropathies *PMP22*, *GJB1*, *MPZ*, *MFN2* and other associated genes	2024 GCMT	Case scenario	Full	Laboratory report	April	Accredited	210
Cystic fibrosis and *CFTR*-related disorders	2024 GCF	DNA	Full	Laboratory report	June	Accredited	390
Epilepsy disorders Tuberous sclerosis, Rett syndrome, Dravet syndrome and focal seizures	2024 GEP	Case scenario	Full	Laboratory report	April	Accredited	210
Eye disorders Retinopathies, structural eye disorders, cataracts and albinism	2024 GEYE	DNA	Full	Laboratory report	April	Accredited	390
Familial colorectal cancer and polyposis Lynch syndrome, Familial Adenomatous Polyposis and *MUTYH*-associated Polyposis (MAP)	2024 GCRC	DNA	Full	Laboratory report	April	Accredited	390
Familial endocrine tumour predisposition disorders Von Hippel-Lindau disease (VHL), Multiple Endocrine Neoplasia (MEN) and Familial medullary thyroid carcinoma (FMTC)	2024 GEND	DNA	Full	Laboratory report	June	Accredited	390
Familial Hypercholesterolaemia *LDLR*, *APOB* and *PCSK9*	2024 GFH	DNA	Full	Laboratory report	June	Accredited	390

For full details of all EQA offered go to www.genqa.org/eqa

Please browse this directory and contact us at info@genqa.org if you require any further information.

External Quality Assessment (EQA)	EQA Code	Sample	Assessment[#]	Results submission	Distribution month	ISO 17043 Status	Fee (£)
Fragile X syndrome and *FMR1*-related disorders	2024 GFRAX	DNA	Full	Laboratory report	June	Accredited	390
Gastroenterology and hepatology disorders Hereditary Pancreatitis, Gilbert, Cholestasis, Hirschsprung disease, Polycystic liver disease.	2024 GGH	Case scenario	Full	Laboratory report	April	Accredited	210
Hereditary breast and ovarian cancer disorders Familial Breast and Ovarian Cancer (*BRCA1* and *BRCA2*), Cowden Syndrome and Li-Fraumeni	2024 GHBOC	DNA	Full	Laboratory report	April	Accredited	390
Huntington disease and DRPLA	2024 GHD	DNA	Full	Laboratory report	June	Accredited	390
Inborn errors of metabolism MCADD, Fabry disease, galactosaemia and lysosomal storage disease	2024 GIEM	DNA	Full	Laboratory report	April	Accredited	390
Linkage analysis Autosomal recessive, Autosomal dominant and X-linked disorders	2024 GLINK	Case scenario	Full	Laboratory report	April	Accredited	210
Mitochondrial disease Mitochondrial and *POLG*-related disorders	2024 GMT	DNA	Full	Laboratory report	April	Accredited	390
Muscular dystrophies DMD-related and other muscular dystrophies	2024 GMD	DNA	Full	Laboratory report	April	Accredited	390
Neurodegenerative disorders Alzheimer disease, Frontotemporal dementia, Motor neurone disease/Amyotrophic lateral sclerosis (ALS), Parkinson disease and Spinal and bulbar muscular atrophy (SBMA)	2024 GND	DNA	Full	Laboratory report	April	Accredited	390
Neurofibromatosis and rasopathies Neurofibromatosis (types 1 and 2), Noonan syndrome and schwannomatosis	2024 GNF	DNA	Full	Laboratory report	April	Accredited	390
Primary Immunodeficiency disorders (PID) Severe Combined Immunodeficiency (SCID), Agammaglobulinaemia, Hereditary angioedema, Chronic granulomatous disease and Hyper IgE syndrome	2024 GPID	DNA	Full	Laboratory report	April	Accredited	390
Renal disorders Haematuria, tubulointerstitial kidney disease, cystic renal disease and Alport syndrome	2024 GREN	Case scenario	Full	Laboratory report	April	Accredited	210
Respiratory disorders Pneumothorax, respiratory insufficiency, bronchiectasis (ciliopathies/PCD and surfactants) and pulmonary arterial disease	2024 GRESP	Case scenario	Full	Laboratory report	April	Accredited	210
Skeletal Dysplasias *FGFR2/FGFR3* related disorders, OI and other skeletal dysplasias	2024 GSKEL	DNA	Full	Laboratory report	April	Accredited	390
X-inactivation Determination of X-inactivation ratios	2024 GXI	Case scenario	Full	Laboratory report	April	Accredited	210

[#]Full assessment of analytical/genotyping processes and clinical interpretation of results

For full details of all EQA offered go to www.genqa.org/eqa

Please browse this directory and contact us at info@genqa.org if you require any further information.

Haematological Neoplasms

Assess your testing and reporting of a broad range of haematological neoplasms, using various sample types and/or online case scenarios. Cases based on real clinical scenarios, using validated samples.

- Any suitable technique can be used
- 6 weeks to submit results
- Submitted laboratory reports are assessed, by an expert panel, using international guidance/recommendations
- Each participant receives an Individual Laboratory Report (ILR) and EQA summary report

External Quality Assessment (EQA)	EQA Code	Sample	Assessment[#]	Results submission	Distribution month	ISO 17043 Status	Fee (£)
Acute Lymphoblastic Leukaemia (ALL) Images for G-band chromosome and FISH analysis, DNA for whole genome copy number analysis	2024 HALL	DNA/ Images	Full	Laboratory report	May	Accredited	390
Chronic Lymphocytic Leukaemia (CLL) Images for G-band chromosome and FISH analysis. whole genome copy number analysis	2024 HCLLC	DNA/ Fixed cells/ Images	Full	Laboratory report	September	Accredited	390
Chronic Lymphocytic Leukaemia (CLL) IGHV mutation status[2]	2024 HCLLI	DNA/ cDNA[3]/ lyophilised cells	Full	Laboratory report	September	Accredited	390
Chronic Lymphocytic Leukaemia (CLL) *TP53* analysis[2]	2024 HCLLT	DNA	Full	Laboratory report	September	Accredited	390
Haematological Technical FISH AML, CML, ALL, MDS, MPN	2024 HTF	Fixed cells	Summary of test results	Proforma	May	Accredited	390
Lymphoma Images for G-band chromosome and supplementary FISH analysis. Fixed cells and FFPE for FISH analysis	2024 HLYM	Fixed cells/ FFPE/ Images	Full	Laboratory report	September	Accredited	390
Lymphoma Technical NGS SNVs/CNVs/fusions associated with lymphoma	2024 HLYT	FFPE	Summary of test results	Proforma	September	Pilot	210
Myeloid disorders Images G-band chromosome and FISH analysis. whole genome copy number analysis	2024 HMD	DNA/ Images	Full	Laboratory report	May	Accredited	390
Myeloma Images and fixed cells for FISH analysis and DNA for whole genome copy number analysis	2024 HMM	DNA/Fixed cells/ Images	Full	Laboratory report	May	Accredited	390

[#]Full assessment of analytical/genotyping processes and clinical interpretation of results
[2]Provided in collaboration with UKNEQAS LI. [3]Please contact GenQA for this sample type

For full details of all EQA offered go to www.genqa.org/eqa
Please browse this directory and contact us at info@genqa.org if you require any further information.

Molecular Newborn Screening

Assess the quality of your molecular testing using blood spotted onto neonatal screening cards. Multiple rounds of EQA per annum included. Cases based on real clinical scenarios, using validated samples.

- Any suitable technique can be used
- 4 distributions per annum
- 4 weeks to submit results
- Submitted results are assessed, by an expert panel.
- Each participant receives an Individual Laboratory Report (ILR) and EQA summary report

External Quality Assessment (EQA)	EQA Code	Sample	Assessment[#]	Results submission	Distribution month	ISO 17043 Status	Fee (£)
Molecular testing for cystic fibrosis (CF) *CFTR* variants	2024 NBSC	Neonatal screening cards	Genotyping	Proforma or laboratory report	April, June, October, January 2025	Accredited	870
Molecular testing for Medium-chain acyl-CoA dehydrogenase deficiency (MCADD) c.985A>G p.(Lys329Glu) variant only	2024 NBSM	Neonatal screening cards	Genotyping	Proforma or laboratory report	April, June, October, January 2025	Accredited	870
Molecular testing for Severe Combined Immunodeficiency (SCID) T-cell Receptor Excision Circles (TRECs)	2024 NBST	Neonatal screening cards	Full	Proforma	April, June, October, January 2025	Not accredited	870
Molecular testing for Spinal Muscular Atrophy (SMA) *SMN1* deletion	2024 NBSS	Neonatal screening cards	Genotyping	Proforma	April, June, October, January 2025	Not accredited	210

Molecular Pathology

Assess your molecular testing and reporting of a variety of genes in different tumour types. Cases based on real clinical scenarios, using validated samples.

- Any suitable molecular technique can be used
- 6 weeks to submit results
- Submitted laboratory reports are assessed, by an expert panel, using international recommendations
- Each participant receives an Individual Laboratory Report (ILR) and EQA summary report

External Quality Assessment (EQA)	EQA Code	Sample	Assessment[#]	Results submission	Distribution month	ISO 17043 Status	Fee (£)
BRCA testing for ovarian and prostate cancer - somatic[1] UPDATED	2024 TBS	FFPE	Full	Laboratory report	September	Accredited	0[4]
BRCA testing for ovarian, breast, prostate and pancreatic cancer - germline[1]	2024 TBG	Germline DNA	Full	Laboratory report	September	Accredited	200[4]
BRCA testing in prostate cancer – cfDNA[1]	2024 TBP	Artificial plasma	Full	Laboratory report	September	Pilot	0[4]
Breast cancer (*PIK3CA* testing)	2024 TBCP	FFPE	Full	Laboratory report	October	Accredited	275
Breast Cancer - Tumour expression profiling	2024 TEP	FFPE	Full	Laboratory report	January 2025	Pilot	210
Central Nervous System (CNS) tumours	2024 TCNS	FFPE	Genotyping	Laboratory report	January 2025	Accredited	390
Cholangiocarcinoma *FGFR2* fusions and/or *IDH1* hotspots	2024 TCC	FFPE	Full	Laboratory report	January 2025	Not accredited	390
Colorectal cancer – core *KRAS*, *NRAS* and *BRAF*	2024 TCRC	FFPE	Full	Laboratory report	April	Accredited	695
Colorectal cancer - MMR *KRAS*, *NRAS* and *BRAF*, MSI and *MLH1* promoter methylation	2024 TCMMR	FFPE	Full	Laboratory report	April	Accredited	830
Endometrial tumours MSI, *MLH1* promoter methylation, *POLE* and *P53*	2024 TET	FFPE	Full	Laboratory report	January 2025	Not accredited	390
Gastrointestinal Stromal Tumours (GIST) *KIT* and *PDGFRA*	2024 TGT	FFPE	Genotyping	Laboratory report	October	Accredited	275
Genomic molecular tumour board (MTB) for lung cancer^ NEW	2024 TMTB	Case scenario	Online assessment of the full MDT process including pre-meeting, post-meeting and reporting.		November	Pilot	100
Lung cancer - *EGFR*	2024 TLE	FFPE	Full	Laboratory report	April	Accredited	695
Lung cancer – comprehensive molecular *EGFR*, *KRAS*, *BRAF*, *PIK3CA*, *ERBB2*, MET exon 14 skipping (DNA only), *TP53*, *STK11 and KEAP1*	2024 TLCM	FFPE	Full	Laboratory report	April	Accredited	830

^This EQA is now listed under the Multidisciplinary team (MDT) working specialty.

Molecular Pathology (cont.)

External Quality Assessment (EQA)	EQA Code	Sample	Assessment[#]	Results submission	Distribution month	ISO 17043 Status	Fee (£)
Lung cancer – fusions molecular *ALK, ROS1, RET, MET* (amplification and exon 14 skipping)	2024 TLFM	FFPE	Full	Laboratory report	October	Accredited	695
Lung cancer – fusions FISH/IHC *ALK, ROS1, RET, MET* (amplification)	2024 TLFF	FFPE	Full	Laboratory report	October	Accredited	275
Lung cancer - Circulating free (cf) DNA in lung cancer[1]	2024 TCFD	Artificial plasma	Full	Laboratory report	September	Accredited	275
Melanoma *BRAF, KIT +/- NRAS*	2024 TM	FFPE	Full	Laboratory report	April	Accredited	695
Microsatellite Instability (MSI) MSI, *BRAF, MLH1* promoter methylation	2024 TMSI	FFPE	Full	Laboratory report	April	Accredited	275
Molecular Tissue identification	2024 TMT	FFPE	Full	Laboratory report	January 2025	Accredited	275
NTRK fusions	2024 TNTRK	FFPE	Full	Laboratory report	January 2025	Accredited	275
Renal tumours *TFE3* rearrangements	2024 TRT	FFPE	Full	Laboratory report	January 2025	Accredited	275
Sarcoma	2024 TSA	FFPE	Full	Laboratory report	October	Accredited	275
Thyroid cancer *HRAS, NRAS, KRAS, BRAF, RET, TP53, TERT* promoter	2024 TT	FFPE	Genotyping	Laboratory report	October	Accredited	275

[##]Full assessment of analytical/genotyping processes and clinical interpretation of results. [1]Provided in collaboration with EMQN
[4]Sponsored by AstraZeneca and MSD

Individual Competency Assessment:

Tumour content estimation and annotation of tissue samples	Tissue-I Individual online competency assessment	Spring Autumn	Educational only	555 for 3 users

For full details of all EQA offered go to www.genqa.org/eqa
Please browse this directory and contact us at info@genqa.org if you require any further information.

Pharmacogenomics

Assess your testing and reporting of clinically actionable pharmacogenomic variants/alleles. Cases based on real clinical scenarios, using validated samples.

- Any molecular technique can be used
- 6 weeks to submit results
- Submitted laboratory reports are assessed, by an expert panel
- Each participant receives an Individual Laboratory Report (ILR) and EQA summary report

External Quality Assessment (EQA)	EQA Code	Sample	Assessment[#]	Results submission	Distribution month	ISO 17043 Status	Fee (£)
Pharmacogenomics: Aminoglycoside ototoxicity Includes m.1555A>G, m.1095T>C and m.1494C>T variants	2024 PGXA	DNA	Full	Laboratory report	August	Accredited	390
Pharmacogenomics: *APOE* NEW	2024 PGXE	DNA	Genotyping	Laboratory report	TBC	Pilot	100
Pharmacogenomics: panel	2024 PGXP	DNA	Genotyping	Proforma	August	Pilot	210
Pharmacogenomics: *CYP2C19* NEW *CYP2C19* variants	2024 PGXC	DNA	Full	Laboratory report	August	Pilot	100
Pharmacogenomics: *DPYD* *DPYD* variants	2024 PGXD	DNA	Full	Laboratory report	August	Accredited	390
Pharmacogenomics: *TPMT and NUDT15* *TPMT* and *NUDT15* variants	2024 PGXT	DNA	Full	Laboratory report	August	Not accredited	390

[#]Full assessment of analytical/genotyping processes and clinical interpretation of results

Preimplantation Genetic Testing (PGT)

Assess a range of preimplantation genetic testing scenarios, using various sample types, including embryo testing.

- Any suitable technique can be used
- 3-10 weeks to submit results
- Submitted laboratory reports are assessed, by an expert panel, using international recommendations.
- Each participant receives an Individual Laboratory Report (ILR) and EQA summary report

External Quality Assessment (EQA)	EQA Code	Sample	Assessment[#]	Results submission	Distribution month	ISO 17043 Status	Fee (£)
PGT for aneuploidies (PGT-A)	2024 PGTA	DNA	Full	Laboratory report	September	Accredited	390
PGT for blastomere FISH (Stages 1 and 2) Structural rearrangements <10Mb	2024 PGTBF	Images	Full	Laboratory report	May: stage 1 June: stage 2	Accredited	390
PGT for monogenic disorders (Stages 1 and 2)[$] Beta thalassaemia	2024 PGTM	DNA/Cells	Full	Laboratory report	May	Accredited	390
PGT for structural rearrangements (PGT-SR) Structural rearrangements >10Mb	2024 PGTSR	DNA	Full	Laboratory report	September	Accredited	390

[#]Full assessment of analytical/genotyping processes and clinical interpretation of results

[$]If your laboratory is unable to perform Stage 2, then the cost of Stage 2 (£260) will be refunded.

Reproductive Genetics

Assess various clinical pathways associated with reproductive genetics, using different sample types. Cases based on real clinical scenarios, using validated samples.

- Any suitable technique can be used
- 4-12 weeks to submit results
- Submitted laboratory reports are assessed, by an expert panel, using international recommendations
- Each participant receives an Individual Laboratory Report (ILR) and EQA summary report

External Quality Assessment (EQA)	EQA Code	Sample	Assessment[#]	Results submission	Distribution month	ISO 17043 Status	Fee (£)
Abnormal ultrasound Range of molecular and cytogenomics tests involved in the prenatal pathway	2024 RABU	Case scenario	Full	Laboratory report	April	Accredited	390
Carrier screening	2024 RPCS	DNA	Full	Laboratory report	October	Pilot	210
Maternal cell contamination (MCC) and fetal sexing	2024 RMCC	DNA	Full	Laboratory report	April	Accredited	390
Non-invasive prenatal testing (NIPT) for common aneuploidies[1] Chromosomal aneuploidies for 13, 18, 21, X and Y and fetal sexing	2024 NIPTA	Plasma	Full	Laboratory report	October	Accredited	455
Non-invasive prenatal testing (NIPT) for common microdeletions	2024 NIPTM	Plasma	Full	Laboratory report	October	Accredited	390
Non-invasive prenatal testing (NIPT) for fetal *RhD* status[1] Rhesus D antigen status	2024 NIPTR	Plasma	Full	Laboratory report	October	Pilot	300
Non-invasive prenatal testing (NIPT) for fetal sexing[1]	2024 NIPTS	Plasma	Full	Laboratory report	October	Accredited	455
Pregnancy loss Images for G-band chromosome and FISH analysis, DNA for whole genome analysis and targeted aneuploidy	2024 RNPL	Images/ DNA	Full	Laboratory report	September	Accredited	390
Prenatal constitutional copy number variant (CNV) detection Whole genome analysis	2024 RCNV	DNA	Full	Laboratory report	September	Accredited	390
Prenatal karyotyping Images for G-band chromosome and FISH analysis,	2024 RK	Images	Full	Laboratory report	September	Accredited	390
Rapid prenatal testing for common aneuploidies Chromosomal aneuploidies for 13, 18, 21, X and Y and fetal sexing	2024 RRA	Fixed cell suspension / DNA	Full	Laboratory report	October	Accredited	390

[#]Full assessment of analytical/genotyping processes and clinical interpretation of results
[1]Provided in collaboration with EMQN

For full details of all EQA offered go to www.genqa.org/eqa
Please browse this directory and contact us at info@genqa.org if you require any further information.

Sample Handling

Assess the quantity and quality of your DNA extraction methods for different sample types and quantification processes.

- Any method can be used
- 3 weeks to submit results
- Each participating laboratory receives an Individual Laboratory Report (ILR) and EQA summary report

External Quality Assessment (EQA)	EQA Code	Sample	Assessment	Results submission	Distribution month	ISO 17043 Status	Fee (£)
cfDNA extraction from plasma[#] **NEW**	2024 DNAC	Artificial plasma	DNA quality and quantity	Proforma and DNA	TBC	Pilot	0
DNA extraction from venous blood	2024 DNAB	Blood	DNA quality and quantity	Proforma and DNA	June	Accredited	1135
DNA extraction from blood for long read sequencing **NEW**	2024 DNAL	Blood	DNA quality and quantity	Proforma and DNA	June	Pilot	0
DNA extraction from formalin-fixed paraffin embedded (FFPE) tissue	2024 DNAP	FFPE	DNA quality and quantity	Proforma and DNA	November	Accredited	610
DNA extraction from fresh tissue	2024 DNAF	Fresh tissue	DNA quality and quantity	Proforma and DNA	June	Not accredited	610
DNA extraction from saliva	2024 DNAS	Saliva	DNA quality and quantity	Proforma and DNA	November	Not accredited	610
DNA quantification Measurement of DNA concentration	2024 DNAQ	DNA	Accuracy of DNA quantification	Proforma	April	Not accredited	350

[#]Offered in collaboration with SeraCare

For full details of all EQA offered go to www.genqa.org/eqa

Please browse this directory and contact us at info@genqa.org if you require any further information.

Technical

Next Generation Sequencing: Assess the quality of your NGS sequencing using any platform or technology, in germline and somatic samples.

- Use any platform and technology
- Test single gene, gene panels, exome sequencing and/or whole genome sequencing
- Submit up to 3 different data files per EQA (excluding germline CNV EQA)
- 8 weeks to submit results
- Each participating laboratory receives a review of their submitted data and EQA summary report

Optical Genome Mapping: Assess the accuracy of your OGM bioinformatic algorithms and pipelines for structural and copy number aberrations in both haematological neoplasms and rare diseases.

- Data files provided
- 6 weeks to submit results
- Each participating laboratory receives an Individual Laboratory Report (ILR) and EQA summary report

External Quality Assessment (EQA)	EQA Code	Sample	Assessment	Results submission	Distribution month	ISO 17043 Status	Fee (£)
Next Generation Sequencing (NGS) for germline SNVs and indels[1,2] NGS of single genes, panels, exomes and whole genomes	2024 NGSGS	Germline DNA	NGS data quality for SNV/indels	Proforma and data files	September	Accredited	700
Next Generation Sequencing (NGS) for germline CNVs[1,3] **NEW**	2024 NGSGC	Germline DNA	NGS data quality for CNV	Proforma and data files	September	Pilot	500
Next Generation Sequencing (NGS) for somatic SNVs and indels – tumour testing only[1,2,4] NGS of hot spot panels	2024 NGST	Tumour DNA	NGS data quality (SNV/indels only)	Proforma and data files	September	Not accredited	700
Next Generation Sequencing (NGS) for somatic SNVs and indels – tumour with germline subtraction analysis[1,2,4] NGS and germline subtraction	2024 NGSM	Matched tumour and germline DNA	NGS data quality (SNV/indels only)	Proforma and data files	September	Not accredited	1,100
Optical Genome Mapping (haematological neoplasms and rare diseases)	2024 OGM	Data files	Analysis and summary of test results	Proforma	To be confirmed	Pilot	210

[1]Provided in collaboration with EMQN,
[2]Indels <50bp
[3]CNVs ≥50bp
[4]May include CNVs as a pilot

For full details of all EQA offered go to www.genqa.org/eqa
Please browse this directory and contact us at info@genqa.org if you require any further information.

Variant Classification & Interpretation

Assess the accuracy of your SNV and CNV classification and interpretation in germline and somatic scenarios. Cases created by State registered Clinical Scientists, based on real clinical scenarios, using validated samples.

- Use your standard procedures
- 6-16 weeks to submit results
- Submitted results are assessed by an expert panel against recommended guidelines (where applicable)
- Each participant receives an Individual Laboratory Report (ILR) and EQA summary report

External Quality Assessment (EQA)	EQA Code	Sample	Assessment[#]	Results submission	Distribution month	ISO 17043 Status	Fee (£)
Pathogenicity of somatic solid tumour SNVs, CNVs and SVs	2024 VSC	Case scenario	Full	Proforma	October	Pilot	250
Pathogenicity of haematological neoplasm variants SNVs, CNVs and SVs	2024 VHC	Case scenario	Full	Proforma	October	Pilot	250

Suitable for both molecular and cytogenomic interpretation

Exome trio sequencing – prenatal UPDATED	2024 VEXE	DNA	Full	Laboratory report	May	Not accredited	390
Exome trio sequencing - postnatal (including neonatal and paediatric) **UPDATED**	2024 VEXO	DNA	Full	Laboratory report	May	Not accredited	390
Pathogenicity of postnatal CNVs[3]	2024 VPCNV	Case scenario	Full	Proforma	October	Not Accredited	250
Pathogenicity of prenatal CNVs[3]	2024 VCNV	Case scenario	Full	Proforma	October	Not Accredited	250
Variant validation Validation of variants (SNV and CNV) detected on a research basis and associated family studies	2024 VVAL	DNA	Full	Laboratory report	May	Accredited	390

Suitable for molecular interpretation

Classification of germline SNVs and indels[2]	2024 VGC	Case scenario	Summary of test results	Proforma	October	Accredited	200
Classification and Interpretation of germline SNVs and indels[2]	2024 VGI	Case scenario	Full	Laboratory report	October	Accredited	250
Interpretation of RNA splicing variants Interpretation of RNA analysis results to investigate the pathogenicity of splicing variants	2024 VRNA	Case scenario	Full	Laboratory report	October	Not accredited	250

[#]Full assessment of analytical/genotyping processes and clinical interpretation of results
[2]indels <50bp, [3]CNVs ≥50bp

Nomenclature assessment

ISCN Determine the correct ISCN for the abnormalities described	2024 VISCN	Case scenario	Summary of test results	Proforma	May October	Pilot	250

Viral Sequencing

Assess the accuracy of SARS-CoV-2 sequencing using any platform or technology. Quality data will also be assessed where provided. A collaborative EQA run jointly by GenQA and UK NEQAS for Microbiology. Participants will be required to register with UK NEQAS for Microbiology for the 2024 EQA (a GenQA account will then be created). All invoicing and shipment costs will be via UK NEQAS for Microbiology and registration will open in early 2024.

External Quality Assessment (EQA)	EQA Code	Sample	Assessment[#]	Results submission	Distribution month	ISO 17043 Status
SARS-CoV-2 sequencing **UPDATED**	2024 SCOV	Freeze-dried inactivated SARS-CoV-2	Sequencing accuracy	Survey and sequencing data files	June, August, November, March 2025	Pilot

[1]Provided in collaboration with UKNEQAS for Microbiology

Individual Competency Assessment

BRCA and HRR gene variant classification	GENie GENOMIC ONLINE INDIVIDUAL EDUCATION See www.genqa.org/genie	April October	Educational	0[4]
Germline SNV classification		Continuous	Educational Variant assessment module	£300 for 10 users[$]
Germline CNV classification				
HGVS nomenclature		Trial version available	Educational nomenclature assessment module	0[5]
ISCN nomenclature				

[4]Sponsored by AstraZeneca

[$]£300 for up to 10 user licences per year, £500 for 11-20 licences and £230 per additional 10 licences per year. Individuals can also purchase for £35 per year.

[5] When the continuous modules become available in early 2024, the cost will be £300 for up to 10 user licences per year, £500 for 11-20 licences and £230 per additional 10 licences per year. Individuals can also purchase for £35 per year.

Published 19th September 2023

Updated 3rd September 2024 to align with 2025 EQA directory