GenQA GENOMICS QUALITY ASSESSMENT

Collaboration between CEQAS and UK NEQAS Molecular Genetics
Members of UK NEQAS consortium

Molecular Genetic Disorders EQA 2019

- **Clinical case scenarios (e.g. diagnostic, carrier)**
- **Analysis, Genotyping and Interpretation required**
- **Any molecular technique can be used**
- **8-16 weeks to submit results**
- All EQA samples validated
- Detailed instructions provided
- Results assessed by expert panel
- Marking criteria based on professional guidelines
- ISO17043 accredited

EQA	ISO17043 Accredited
Arrhythmia & Cardiomyopathies	✓
Ataxia / Hereditary Spastic Paraplegia (HSP)	✓
Charcot Marie Tooth disease and Hereditary Liability to Pressure Palsies	✓
Cystic Fibrosis	✓
Dementia / Amylotrophic Lateral Sclerosis (ALS)	✓
Duchenne/Becker Muscular Dystrophies	✓
Fabry Disease	✓
Familial Hypercholesterolaemia *NEW*	✓
Fragile X Syndrome	✓
Hereditary breast and ovarian cancer (HBOC)	✓
Huntington Disease	✓
Hypotonic Infant	✓
Imprinting and uniparental disomy *NEW*	✓
Lynch syndrome	✓
Medium chain acyl-CoA dehydrogenase deficiency	✓
Mitochondrial Disease and POLG	✓
Neurofibromatosis and schwannomatosis *NEW*	✓
Phaeochromocytoma and Paraganglioma disorders *NEW*	✓
Polyposis syndromes	✓
Retinal disorders *NEW*	✓
Skeletal dysplasias	✓
Variant validation *NEW*	✓
X-inactivation *NEW*	✓

For disorders/genes included in these EQAs please see overleaf.

Please see overleaf for more EQA information

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EQA	Disorders/Genes included (each EQA may contain some or all of the disorders/genes indicated below)
Arrhythmia and Cardiomyopathies	Brugada syndrome, Long QT syndrome, Catecholaminergic polymorphic ventricular tachycardia (CPVT), general arrhythmia and cardiomyopathies.
Ataxia and HSP	Friedreich ataxia, spinocerebellar ataxia and hereditary spastic paraplegia
Cystic Fibrosis	*CFTR*-related disorders
Charcot Marie Tooth disease and Hereditary Liability to Pressure Palsies	*PMP22, GJB1, MPZ* and other associated genes
Dementia and ALS	Alzheimer Disease, Frontotemporal Dementia, Motor Neurone Disease and ALS
Duchenne and Becker Muscular Dystrophies	Dystrophin (DMD)-related disorders
Fabry Disease	GLA
Familial Hypercholesterolaemia	*LDLR, APOB* and *PCSK9*
Fragile X Syndrome	FMR1-related disorders
Hereditary Breast and Ovarian Cancer disorders	Familial Breast and Ovarian Cancer (*BRCA1* & *BRCA2*), Cowden Syndrome, Li-Fraumeni, Peutz Jeuger syndrome
Huntington Disease	Huntington Disease (HTT)
Hypotonic Infant	Spinal Muscular Atrophy type 1 (SMA), Prader Willi Syndrome (PWS) and Myotonic Dystrophy type 1 (DM1)
Imprinting and UPD	Angelman Syndrome and Beckwith Wiedemann Syndrome
Lynch Syndrome	*MLH1, MSH2, MSH6* and *PMS2*
MCADD	*ACADM*
MCC & sexing	DNA sexing and determination of level of maternal cell contamination
Mitochondrial Disease and POLG	Mitochondrial disorders and *POLG*
Neurofibromatosis and Schwannomatosis	Neurofibromatosis (types 1 and 2) and Schwannomatosis
Phaeochromocytoma and Paraganglioma	Von Hippel Lindau disease and other disorders associated with endocrine tumour predisposition
Polyposis syndromes	Familial adenomatous polyposis (FAP) and *MUTYH*-associated polyposis (MAP)
Retinal Disorders	Retinitis pigmentosa, Cone/Cone-rod dystrophies, Leber congenital amaurosis and macular degeneration
Skeletal Dysplasias	*FGFR2/FGFR3* related disorders and other skeletal dysplasias
X-inactivation	Determination of X-inactivation ratios

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Other Molecular Genetic Disorders EQAs for 2019

EQA	Type of EQA	Sample Type	Testing	Techniques	ISO1743 Accredited
Variant Validation	Technical, Genotyping and Interpretation	DNA	Validation of sequence variant(s) detected on a research basis and follow-up studies	Any molecular technique	✓
Pathogenicity of Sequence Variants	Interpretation only	N/A	One clinical case with three variants for interpretation and classification. Results to be submitted via a proforma.	N/A	✓
Severe Intellectual Disability	Interpretation only	N/A	One sequential online clinical case.	N/A	✗
Molecular testing for MCADD*	Genotyping	Blood spot cards	ACADM c.985A>G p.(Lys329Glu) pathogenic variant	Any molecular technique	✓
Molecular testing for cystic fibrosis (CF)*	Genotyping	Blood spot cards	CFTR gene	Any molecular technique	✓
Next Generation Sequencing (NGS): Germline	Technical	Germline DNA sample	One germline DNA sample supplied for NGS analysis. Up to three sets of data can be submitted.	NGS analysis	✗
Next Generation Sequencing (NGS): Somatic	Technical	DNA sample extracted from formalin-fixed paraffin embedded tumour tissue and matched germline DNA sample	NGS analysis of tumour tissue DNA sample and matched germline DNA sample (if appropriate). Up to three sets of data can be submitted.	NGS analysis	✗

*Four distributions of three bloodspot cards per year

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GenQA EQA Specialties

Molecular Genetic Disorders	**Molecular Rapid Aneuploidy (MRA)**	**Molecular Pathology**	**Sample Handling: DNA extraction & quantification**
Newborn Screening	**Non-Invasive Prenatal Testing (NIPT)**	**Haematological Neoplasms**	**Technical: Next Generation Sequencing**
Constitutional Postnatal Testing	**Constitutional Prenatal Testing**	**Clinical Genetics**	**Individual Competency Assessment (G-TACT / Tissue-i)**

For further information, please contact us at info@genqa.org

Registration for all GenQA EQAs for 2020 will open in September 2019

The EQA Cycle