2020 EQA

GenQA GENOMICS QUALITY ASSESSMENT

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

Molecular Genetic Disorders

- **Clinical case scenarios (e.g. diagnostic, carrier)**
- **Analysis, Genotyping and Interpretation required**
- **Any molecular technique can be used**
- **12-16 weeks to submit results**
- All EQA samples validated
- Detailed instructions provided
- Results assessed by expert panel
- Marking criteria based on professional guidelines

EQA	ISO17043 Accredited
Ataxia, including Hereditary Spastic Paraplegia (HSP)	✓
Cardiac disorders *UPDATED*	✓
Charcot Marie Tooth disease and related sensory and motor neuropathies	✓
Cystic Fibrosis and *CFTR*-related disorders	✓
Disorders of Sexual Development (DSD) *NEW*	✗
Epilepsy disorders *NEW*	✗
Eye disorders *UPDATED*	✗
Familial Colorectal Cancer and Polyposis *UPDATED*	✓
Familial Endocrine tumour predisposition disorders	✓
Familial Hypercholesterolaemia	✓
Fragile X syndrome and *FMR1*-related disorders	✓
Hereditary breast and ovarian cancer disorders	✓
Huntington Disease	✓
Hypotonic Infant	✓
Imprinting disorders *UPDATED*	✗
Inborn Errors of Metabolism *NEW*	✓
Infertility (pilot) – online only *NEW*	✗
Linkage analysis (pilot) – online only *NEW*	✗
Maternal Cell Contamination and sexing	✓
Mitochondrial and POLG-related disorders	✓
Muscular Dystrophies *UPDATED*	✓
Neurodegenerative disorders *UPDATED*	✓
Neurofibromatosis and Rasopathies *UPDATED*	✗
Osteogenesis Imperfecta *NEW*	✗
Renal disorders *NEW*	✗
Respiratory disorders *NEW*	✗
Skeletal dysplasias, including *FGFR2/FGFR3*-related disorders	✓
X-inactivation	✗

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EQA	Disorders/Genes included (each EQA may contain some or all of the disorders/genes indicated below)
Ataxia, including Hereditary Spastic Paraplegia (HSP)	Friedreich ataxia, spinocerebellar ataxia and hereditary spastic paraplegia.
Cardiac disorders	Cardiomyopathies, arrhythmia and aortic dissection: Brugada syndrome, Long QT syndrome, Catecholaminergic polymorphic ventricular tachycardia (CPVT) and Marfan syndrome
Charcot Marie Tooth disease and related sensory and motor neuropathies	*PMP22, GJB1, MPZ* and other associated genes
Cystic Fibrosis	*CFTR*-related disorders
Disorders of Sexual Development (DSD)	Androgen Insensitivity syndrome and Congenital Adrenal Hyperplasia
Epilepsy disorders	Tuberous sclerosis, Rett syndrome and Dravet syndrome
Eye disorders	Retinopathies, structural eye disorders and albinism
Familial Colorectal Cancer and Polyposis	Lynch syndrome, Familial Adenomatous Polyposis and *MUTYH*-associated Polyposis (MAP)
Familial Endocrine tumour predisposition disorders	Von Hippel-Lindau disease (VHL), Multiple Endocrine Neoplasia (MEN), Familial medullary carcinoma (FMTC)
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 disorders	Angelman Syndrome (AS), Beckwith Wiedemann Syndrome (BWS) and Silver Russell Syndrome (SRS)
Inborn Errors of Metabolism	Fabry syndrome, Tay Sachs and Gaucher disease
Infertility (pilot) – online only	*CFTR, FMR1,* Y-deletions and karyotyping
Linkage analysis (pilot) – online only	Autosomal recessive, Autosomal dominant and X-linked disorders
Maternal Cell contamination and sexing	DNA sexing and determination of level of maternal cell contamination
Mitochondrial and POLG-related disorders	Mitochondrial disorders and *POLG*
Muscular Dystrophies	*DMD*-related and other muscular dystrophies
Neurodegenerative disorders	Alzheimer Disease, Frontotemporal Dementia, Motor Neurone Disease, ALS and Parkinson disease
Neurofibromatosis and Rasopathies	Neurofibromatosis (types 1 and 2) and Noonan syndrome
Osteogenesis Imperfecta	*COL1A1* and *COL1A2* analysis
Renal disorders	Alport syndrome and polycystic kidney disease
Respiratory disorders	*FLCN*-related disorders and Pulmonary Arterial Hypertension
Skeletal dysplasias, including *FGFR2/FGFR3*-related disorders	*FGFR2/FGFR3* related disorders and other skeletal dysplasias
X-inactivation	Determination of X-inactivation ratios

Please see overleaf for more EQA information

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Other Molecular Genetic Disorder EQAs for 2020

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 (classification only)	Interpretation only	N/A	Classification of sequence variants only. Results to be submitted via a proforma.	N/A	✓
Pathogenicity of Sequence Variants (classification and clinical interpretation)	Interpretation only	N/A	Classification and clinical interpretation of sequence variants. Results to be submitted as a diagnostic report.	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* gene including 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 fresh frozen 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	✗

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GenQA EQA Specialties

Molecular Genetic Disorders	**Variant Classification and Interpretation**	**Molecular Pathology**	**Sample Handling: DNA extraction & quantification**
Newborn Screening	**Preimplantation Genetic Testing**	**Haematological Neoplasms**	**Technical: Next Generation Sequencing**
Constitutional Postnatal Testing	**Prenatal (including Non-Invasive) Testing**	**Clinical Genetics**	**Individual Competency Assessment (G-TACT / Tissue-i)**

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

The EQA Cycle