## 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

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

Email: info@genqa.org
<table>
<thead>
<tr>
<th>EQA</th>
<th>Disorders/Genes included</th>
</tr>
</thead>
<tbody>
<tr>
<td>Ataxia, including Hereditary Spastic Paraplegia (HSP)</td>
<td>Friedreich ataxia, spinocerebellar ataxia and hereditary spastic paraplegia.</td>
</tr>
<tr>
<td>Cardiac disorders</td>
<td>Cardiomyopathies, arrhythmia and aortic dissection: Brugada syndrome, Long QT syndrome, Catecholaminergic polymorphic ventricular tachycardia (CPVT) and Marfan syndrome</td>
</tr>
<tr>
<td>Charcot Marie Tooth disease and related sensory and motor neuropathies</td>
<td>PMP22, GJB1, MPZ and other associated genes.</td>
</tr>
<tr>
<td>Cystic Fibrosis</td>
<td>CFTR-related disorders.</td>
</tr>
<tr>
<td>Disorders of Sexual Development (DSD)</td>
<td>Androgen Insensitivity syndrome and Congenital Adrenal Hyperplasia</td>
</tr>
<tr>
<td>Epilepsy disorders</td>
<td>Tuberous sclerosis, Rett syndrome and Dravet syndrome.</td>
</tr>
<tr>
<td>Eye disorders</td>
<td>Retinopathies, structural eye disorders and albinism.</td>
</tr>
<tr>
<td>Familial Colorectal Cancer and Polyposis</td>
<td>Lynch syndrome, Familial Adenomatous Polyposis and MUTYH-associated Polyposis (MAP)</td>
</tr>
<tr>
<td>Familial Endocrine tumour predisposition disorders</td>
<td>Von Hippel-Lindau disease (VHL), Multiple Endocrine Neoplasia (MEN), Familial medullary carcinoma (FMTC)</td>
</tr>
<tr>
<td>Familial Hypercholesterolaemia</td>
<td>LDLR, APOB and PCSK9.</td>
</tr>
<tr>
<td>Fragile X syndrome</td>
<td>FMR1-related disorders.</td>
</tr>
<tr>
<td>Hereditary breast and ovarian cancer disorders</td>
<td>Familial Breast and Ovarian Cancer (BRCA1 &amp; BRCA2), Cowden Syndrome, Li-Fraumeni, Peutz Jeguer syndrome</td>
</tr>
<tr>
<td>Huntington Disease</td>
<td>Huntington Disease (HTT).</td>
</tr>
<tr>
<td>Hypotonic Infant</td>
<td>Spinal Muscular Atrophy type 1 (SMA), Prader Willi Syndrome (PWS) and Myotonic Dystrophy type 1 (DM1)</td>
</tr>
<tr>
<td>Imprinting disorders</td>
<td>Angelman Syndrome (AS), Beckwith Wiedemann Syndrome (BWS) and Silver Russell Syndrome (SRS)</td>
</tr>
<tr>
<td>Inborn Errors of Metabolism</td>
<td>Fabry syndrome, Tay Sachs and Gaucher disease.</td>
</tr>
<tr>
<td>Infertility (pilot) – online only</td>
<td>CFTR, FMR1, Y-deletions and karyotyping.</td>
</tr>
<tr>
<td>Linkage analysis (pilot) – online only</td>
<td>Autosomal recessive, Autosomal dominant and X-linked disorders.</td>
</tr>
<tr>
<td>Maternal Cell contamination and sexing</td>
<td>DNA sexing and determination of level of maternal cell contamination</td>
</tr>
<tr>
<td>Mitochondrial and POLG-related disorders</td>
<td>Mitochondrial disorders and POLG</td>
</tr>
<tr>
<td>Muscular Dystrophies</td>
<td>DMD-related and other muscular dystrophies.</td>
</tr>
<tr>
<td>Neurodegenerative disorders</td>
<td>Alzheimer Disease, Frontotemporal Dementia, Motor Neurone Disease, ALS and Parkinson disease</td>
</tr>
<tr>
<td>Neurofibromatosis and Rasopathies</td>
<td>Neurofibromatosis (types 1 and 2) and Noonan syndrome.</td>
</tr>
<tr>
<td>Osteogenesis Imperfecta</td>
<td>COL1A1 and COL1A2 analysis.</td>
</tr>
<tr>
<td>Renal disorders</td>
<td>Alport syndrome and polycystic kidney disease.</td>
</tr>
<tr>
<td>Respiratory disorders</td>
<td>FLNC-related disorders and Pulmonary Arterial Hypertension</td>
</tr>
<tr>
<td>Skeletal dysplasias, including FGFR2/FGFR3-related disorders</td>
<td>FGFR2/FGFR3 related disorders and other skeletal dysplasias.</td>
</tr>
<tr>
<td>X-inactivation</td>
<td>Determination of X-inactivation ratios.</td>
</tr>
</tbody>
</table>

Please see overleaf for more EQA information

Email: info@genqa.org
**2020 EQA**

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

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**Other Molecular Genetic Disorder EQAs for 2020**

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

1. Receive validated EQA material plus clinical questions
2. Analyse material using routine procedures
3. Upload genotyping results
4. Expert evaluation of submissions
5. Compare laboratory performance (benchmark)
6. Education/Internal review/Quality improvement

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