Interpretation of Sequence Variants EQA 2019

➢ Test your variant interpretation
➢ Varying genotypes included
➢ 8-12 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>Type of EQA</th>
<th>Sample Type</th>
<th>Technique</th>
<th>Testing</th>
<th>ISO17043 Accredited</th>
</tr>
</thead>
<tbody>
<tr>
<td>Pathogenicity of Sequence Variants</td>
<td>Interpretation</td>
<td>N/A</td>
<td>N/A</td>
<td>Single clinical case with three variants supplied for interpretation and classification</td>
<td>✓</td>
</tr>
<tr>
<td>Variant Validation</td>
<td>Technical, Genotyping and Interpretation</td>
<td>DNA</td>
<td>Any molecular technique</td>
<td>Validation and interpretation of sequence variant(s) detected on a research basis and family follow-up studies</td>
<td>✓</td>
</tr>
</tbody>
</table>

Other Molecular Genetic EQAs for 2019

- Arrhythmia & Cardiomyopathy
- Charcot Marie Tooth Disease
- Dementia and ALS
- Fabry Disease
- Hereditary Breast and Ovarian Cancer
- Hypotonic Infant (PWS, SMA and DM1)
- Lynch Syndrome
- MCC & sexing
- Neurofibromatosis & schwannomatosis
- Polyposis
- Skeletal Dysplasia
- NGS (Germline testing)
- NGS (Somatic testing)

Ataxia and Hereditary Spastic Paraplegia
Cystic Fibrosis
Duchenne & Becker Muscular Dystrophies
Fragile X Syndrome
Huntington Disease
Imprinting & UPD
MCADD
Mitochondrial Disorders
Phaeochromocytoma
Retinal Disorders
X-inactivation

#Laboratories performing NGS gene panels, exome and whole genome sequencing are encouraged to participate.

For further information on these EQAs contact us at info@genqa.org

Please see overleaf for more EQA information

Email: info@genqa.org
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

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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