Postnatal Testing EQAs 2019

- Various sample types
- Technical, analysis and Interpretation required
- Various technique can be used
- 4-6 weeks to submit results
- All EQA samples validated
- Detailed instructions provided
- Results assessed by expert panel
- Marking criteria based on professional guidelines
- ISO17043 accredited

<table>
<thead>
<tr>
<th>EQA</th>
<th>Type of EQA</th>
<th>Sample Type</th>
<th>Testing</th>
<th>Techniques</th>
<th>ISO17043 Accredited</th>
</tr>
</thead>
<tbody>
<tr>
<td>Blood – Postnatal</td>
<td>Technical, analytical and interpretation</td>
<td>Online images</td>
<td>Whole genome analysis microscope</td>
<td>G-banded karyotype and FISH</td>
<td>✓</td>
</tr>
<tr>
<td>Chromosome Breakage syndromes</td>
<td>Technical, analytical and interpretation</td>
<td>Online images</td>
<td>Chromosome breakage and mutations for Fanconi anaemia, Blooms, Ataxia Telangiectasia, Nijmegen syndrome</td>
<td>Chromosomal and molecular techniques</td>
<td>X</td>
</tr>
<tr>
<td>Constitutional Copy Number Variation (CNV) - Postnatal</td>
<td>Technical, analytical and interpretation</td>
<td>DNA</td>
<td>Whole genome analysis (CNV)</td>
<td>Array/NGS</td>
<td>✓</td>
</tr>
<tr>
<td>IPS Stem cells <em>PILOT</em></td>
<td>Technical, analytical and interpretation</td>
<td>Online images</td>
<td>Whole genome analysis, microscope and CNV</td>
<td>G-banded and array</td>
<td>X</td>
</tr>
<tr>
<td>Severe developmental delay case scenario <em>PILOT</em></td>
<td>Interpretation only</td>
<td>Online images</td>
<td>Multiple tests/assays to detect genomic causes of severe developmental delay</td>
<td>Test results supplied – may include whole genome analysis, microscope and CNV interpretation</td>
<td>X</td>
</tr>
</tbody>
</table>

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. Education/Internal review/Quality improvement
3. Compare laboratory performance (benchmark)
4. Expert evaluation of submissions
5. Analyse material using routine procedures
6. Upload genotyping results

www.genqa.org
info@genqa.org