## Molecular Testing of Blood Spot Cards for Newborn Screening EQA

- 12 bloodspots in total
- 4 distributions
- Varying genotypes included
- 4 weeks to submit results

All EQA samples validated
- Detailed instructions provided
- Results assessed by expert panel
- Marking criteria based on professional guidelines

### EQA Details

<table>
<thead>
<tr>
<th>EQA</th>
<th>Type of EQA</th>
<th>Sample Type</th>
<th>ISO17043 Accreditation</th>
<th>Testing</th>
<th>Techniques</th>
</tr>
</thead>
<tbody>
<tr>
<td>Molecular testing for cystic fibrosis (CF)</td>
<td>Genotyping</td>
<td>Blood spot cards</td>
<td>✓</td>
<td>CFTR gene</td>
<td>Any molecular technique</td>
</tr>
<tr>
<td>Molecular testing for MCADD</td>
<td>Genotyping</td>
<td>Blood spot cards</td>
<td>✓</td>
<td>ACADM c.985A&gt;G p.(Lys329Glu) pathogenic variant</td>
<td>Any molecular technique</td>
</tr>
</tbody>
</table>

### Other Molecular Genetics EQAs for 2019

- Arrhythmia & Cardiomyopathy
- Ataxia and Hereditary Spastic Paraplegia
- Breast and Ovarian Cancer
- Dementia
- Cystic Fibrosis
- Charcot Marie Tooth Disease
- Duchenne & Becker Muscular Dystrophies
- Fabry Disease
- Fragile X Syndrome
- Huntington Disease
- Hypotonic Infant (PWS, SMA and DM1)
- Imprinting & UPD

- Lynch Syndrome
- MCADD
- Maternal cell contamination (MCC) & sexing
- Mitochondrial Disorders
- Neurofibromatosis & schwannomatosis
- Pathogenicity of Sequence Variants
- Phaeochromocytoma
- Polyposis
- Retinal Disorders
- Skeletal Dysplasia
- Variant Validation
- X-inactivation

Genotyping and interpretation EQAs with a single distribution per year.
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**

<table>
<thead>
<tr>
<th>Molecular Genetic Disorders</th>
<th>Molecular Rapid Aneuploidy (MRA)</th>
<th>Molecular Pathology</th>
<th>Sample Handling: DNA extraction &amp; quantification</th>
</tr>
</thead>
<tbody>
<tr>
<td>Newborn Screening</td>
<td>Non-Invasive Prenatal Testing (NIPT)</td>
<td>Haematological Neoplasms</td>
<td>Technical: Next Generation Sequencing</td>
</tr>
<tr>
<td>Constitutional Postnatal Testing</td>
<td>Constitutional Prenatal Testing</td>
<td>Clinical Genetics</td>
<td>Individual Competency Assessment (G-TACT / Tissue-i)</td>
</tr>
</tbody>
</table>

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

[Diagram showing the EQA Cycle]

For more information, visit [www.genqa.org](http://www.genqa.org) or contact [info@genqa.org](mailto:info@genqa.org)