### Molecular Testing for Newborn Screening

- 8 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

<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>Neonatal screening 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>Neonatal screening cards</td>
<td>✓</td>
<td>ACADM gene including c.985A&gt;G p.(Lys329Glu) pathogenic variant</td>
<td>Any molecular technique</td>
</tr>
</tbody>
</table>

### New for 2020

<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 Severe Combined Immunodeficiency (SCID) PILOT</td>
<td>Analytical</td>
<td>Neonatal screening cards</td>
<td>✓</td>
<td>T-cell Receptor Excision Circles (TRECs)</td>
<td>Any molecular technique</td>
</tr>
</tbody>
</table>

SCID Pilot EQA – single distribution of 3 bloodspot cards
No performance criteria will be applied

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>Variant Classification and Interpretation</th>
<th>Molecular Pathology</th>
<th>Sample Handling: DNA extraction &amp; quantification</th>
</tr>
</thead>
<tbody>
<tr>
<td>Newborn Screening</td>
<td>Preimplantation Genetic Testing</td>
<td>Haematological Neoplasms</td>
<td>Technical: Next Generation Sequencing</td>
</tr>
<tr>
<td>Constitutional Postnatal Testing</td>
<td>Prenatal (including Non-Invasive) 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](mailto:info@genqa.org)

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## The EQA Cycle

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

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