Prenatal Testing EQAs 2019

- Various sample types
- Technical, analytical 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>Amniotic Fluid</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>Chorionic Villus</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>Molecular Rapid Aneuploidy</td>
<td>Technical, analytical and interpretation</td>
<td>DNA</td>
<td>Targeted 13/18/21/X/Y</td>
<td>QF-PCR/MLPA/BoBs</td>
<td>✓</td>
</tr>
<tr>
<td>Maternal cell contamination and fetal sexing</td>
<td>Technical, genotyping &amp; interpretation</td>
<td>DNA</td>
<td>Fetal sexing and detection of maternal contamination</td>
<td>Any molecular techniques</td>
<td>✓</td>
</tr>
<tr>
<td>Prenatal constitutional CNV detection</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>Products of Conception (molecular methods)</td>
<td>Technical, analytical and interpretation</td>
<td>DNA</td>
<td>Targeted aneuploidy &amp;/or whole genome analysis CNV</td>
<td>QF-PCR/MLPA/Array/NGS</td>
<td>✓</td>
</tr>
<tr>
<td>Rapid Prenatal Aneuploidy FISH</td>
<td>Technical, analytical and interpretation</td>
<td>Fixed cell suspensions</td>
<td>Targeted 13/18/21/X/Y</td>
<td>FISH</td>
<td>x</td>
</tr>
<tr>
<td>Non-invasive prenatal testing (NIPT) for common aneuploidies</td>
<td>Technical, analytical and interpretation</td>
<td>Plasma</td>
<td>Chromosomal aneuploidies for 13, 18, 21 and fetal sexing</td>
<td>Any molecular techniques</td>
<td>x</td>
</tr>
<tr>
<td>NIPT for common microdeletions <em>PILOT</em></td>
<td>Technical, analytical and interpretation</td>
<td>Plasma/DNA</td>
<td>Microdeletions (defined by EQA at time of distribution)</td>
<td>Any molecular techniques</td>
<td>x</td>
</tr>
<tr>
<td>NIPT for fetal sex determination</td>
<td>Technical, analytical and interpretation</td>
<td>Plasma</td>
<td>Fetal sexing for early gestation</td>
<td>Any molecular techniques</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. 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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