

## Prenatal (including Non-Invasive) Testing

- 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

EQA	Type of EQA	Sample Type	Testing	Techniques	ISO17043 Accredited
Amniotic Fluid	Technical, analytical and interpretation	Online images	Whole genome analysis microscope	G-banded Karyotype and FISH	✓
Chorionic Villus	Technical, analytical and interpretation	Online images	Whole genome analysis microscope	G-banded Karyotype and FISH	✓
Molecular Rapid Aneuploidy	Technical, analytical and interpretation	DNA	Targeted 13/18/21/X/Y	QF-PCR	✓
Maternal cell contamination and fetal sexing	Technical, genotyping & interpretation	DNA	Fetal sexing and detection of maternal contamination)	Any molecular techniques	✓
Prenatal constitutional CNV detection	Technical, analytical and interpretation	DNA	Whole genome analysis CNV	Array/NGS	✓
Pregnancy Loss (molecular methods)	Technical, analytical and interpretation	DNA	Targeted aneuploidy &/or whole genome analysis CNV	QF-PCR/MLPA/Array/NGS	✓
Pregnancy Loss (G-banding)	Technical, analytical and interpretation	Online images	Whole genome analysis microscope	G-banded Karyotype and FISH	✓
Rapid Prenatal Aneuploidy FISH	Technical, analytical and interpretation	Fixed cell suspensions	Targeted 13/18/21/X/Y	FISH	✓
Non-invasive prenatal testing (NIPT) for common aneuploidies (pilot)	Technical, analytical and interpretation	Plasma	Chromosomal aneuploidies for 13, 18, 21 and fetal sexing	Any molecular techniques	X
NIPT for common microdeletions (pilot)	Technical, analytical and interpretation	Plasma/DNA	Microdeletions (defined by EQA at time of distribution)	Any molecular techniques	X
NIPT for fetal sex determination	Technical, analytical and interpretation	Plasma	Fetal sexing for early gestation	Any molecular techniques	X

For further information on these EQAs contact us at [info@genqa.org](mailto:info@genqa.org)

Please see overleaf for more EQA information

Email: [info@genqa.org](mailto:info@genqa.org)

# 2020 EQA



Collaboration between CEQAS and UK NEQAS Molecular Genetics  
Members of UK NEQAS consortium

## GenQA EQA Specialties

Molecular Genetic Disorders

Variant Classification and Interpretation

Molecular Pathology

Sample Handling: DNA extraction & quantification

Newborn Screening

Preimplantation Genetic Testing

Haematological Neoplasms

Technical: Next Generation Sequencing

Constitutional Postnatal Testing

Prenatal (including Non-Invasive) Testing

Clinical Genetics

Individual Competency Assessment (G-TACT / Tissue-i)

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## The EQA Cycle

