

Next Generation Sequencing (NGS) for SNV detection

- Assess data accuracy
- Use any platform/technology
- Test single gene(s), gene panel or whole genome
- Submit up to 3 different sets of data per EQA
- 8 weeks to submit results
- All EQA samples validated
- Detailed instructions provided
- Results assessed by expert panel
- Marking criteria based on professional guidelines
- Offered in collaboration with EMQN

EQA	Type of EQA	Sample Type	Testing	Files accepted	ISO17043 Accredited
NGS germline (inherited disorders)	Technical	Genomic DNA	Perform sequencing for the genes usually tested during your routine diagnostic NGS procedures	BAM BED FASTQ VCF	-
NGS somatic	Technical	DNA extracted from fresh frozen tumour tissue and matched germline DNA	Perform sequencing for the genes usually tested during your routine diagnostic NGS procedures	BAM BED FASTQ VCF	-

Next Generation Sequencing (NGS) for Copy Number Variant (CNV) detection

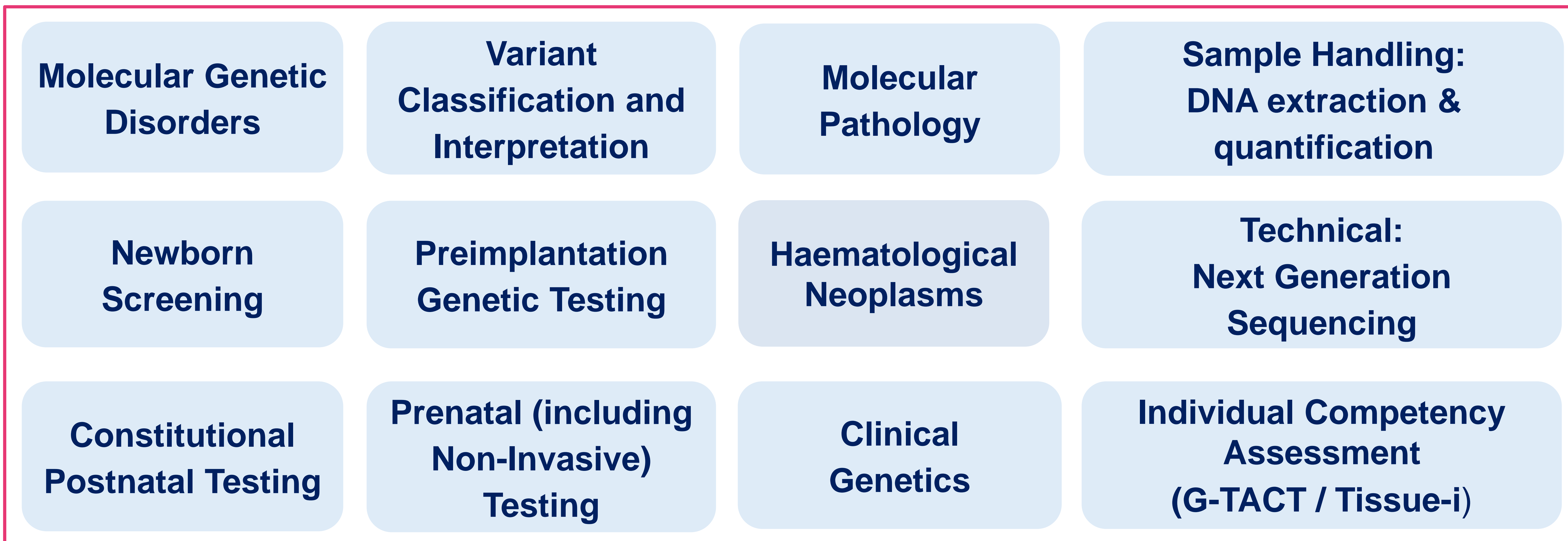
EQA	Type of EQA	Sample Type	Testing	Techniques	ISO17043 Accredited
Constitutional Copy Number Variation (CNV) - Postnatal	Technical, analytical and interpretation	DNA	Whole genome analysis (CNV)	Array/NGS	✓
Prenatal constitutional CNV detection	Technical, analytical and interpretation	DNA	Whole genome analysis CNV	Array/NGS	✓

2020 EQA



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

GenQA EQA Specialties



For further information, please contact us at info@genqa.org

The EQA Cycle

