2020 EQA

GenQA GENOMICS QUALITY ASSESSMENT

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

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**	**✓**

Please see overleaf for more EQA information

Email: info@genqa.org

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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)**

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

The EQA Cycle