

Variant Classification and Interpretation

- **Test your variant interpretation**
- **Varying genotypes included**
- **8-12 weeks to submit results**
- All EQA samples validated
- Detailed instructions provided
- Results assessed by expert panel
- Marking criteria based on professional guidelines

EQA	Type of EQA	Sample Type	Technique	Testing	ISO17043 Accredited
Pathogenicity of germline Sequence Variants (Classification only)	Variant Classification	N/A	N/A	Classification of variants. Submit classification results on a <i>proforma</i> .	✓
Pathogenicity of germline Sequence Variants (Classification and Interpretation)	Variant Classification and Interpretation	N/A	N/A	Classification and clinical interpretation of variants. Submit a diagnostic report including clinical interpretation of the variants.	✓
Pathogenicity of somatic Sequence Variants (Classification only) (pilot) *NEW*	Variant Classification	N/A	N/A	Classification of variants. Submit classification results on a <i>proforma</i> .	X
Variant Validation	Technical, Genotyping and Interpretation	DNA	Any molecular technique	Validation and interpretation of sequence variant(s) detected on a research basis and family follow-up studies	✓

For further information on these EQAs contact us at info@genqa.org

Please see overleaf for more EQA information
Email: 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)

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

The EQA Cycle

