

## Interpretation of Sequence Variants EQA 2019

- **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
<b>Pathogenicity of Sequence Variants</b>	Interpretation	N/A	N/A	Single clinical case with three variants supplied for interpretation and classification	✓
<b>Variant Validation</b>	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	✓

## Other Molecular Genetic EQAs for 2019

Arrhythmia & Cardiomyopathy# Charcot Marie Tooth Disease Dementia and ALS# Fabry Disease Hereditary Breast and Ovarian Cancer# Hypotonic Infant (PWS, SMA and DM1) Lynch Syndrome MCC & sexing Neurofibromatosis & schwannomatosis Polyposis Skeletal Dysplasia  NGS (Germline testing)#	Ataxia and Hereditary Spastic Paraplegia Cystic Fibrosis Duchenne & Becker Muscular Dystrophies Fragile X Syndrome Huntington Disease Imprinting & UPD MCADD Mitochondrial Disorders Pheochromocytoma Retinal Disorders# X-inactivation  NGS (Somatic testing)#
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#Laboratories performing NGS gene panels, exome and whole genome sequencing are encouraged to participate.

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)



## 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](mailto:info@genqa.org)  
Registration for all GenQA EQAs for 2020 will open in September 2019

## The EQA Cycle

