GenQA GENOMICS QUALITY ASSESSMENT

Collaboration between UK NEQAS Molecular Genetics and CEQAS

Members of UK NEQAS consortium

**www.genqa.org**

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# MOLECULAR GENETICS

DNA samples are provided for testing and full interpretative reports submitted for assessment of genotyping accuracy, interpretation of the result and clerical accuracy of the report.

The 28 EQAs offered for 2019 are:

- **Arrhythmia and Cardiomyopathies**[1]
- **Ataxia and Hereditary Spastic Paraplegia**[3]
- **Breast and Ovarian Cancer**[1,3]
- **Cystic Fibrosis**
- **Charcot Marie Tooth disease/ Hereditary Liability to Pressure Palsies**
- **Dementia**
- **Duchenne/Becker muscular dystrophy**
- **Fabry Disease**
- **Familial Hypercholesterolaemia**
- **Fragile X Syndrome**
- **Huntington disease**
- **Hypotonic Infant (SMA, HD and DM1)**
- **Imprinting and Uniparental disomy**[3]
- **Lynch syndrome**
- **Medium chain acyl-CoA dehydrogenase**
- **Maternal cell contamination & sexing**
- **Mitochondrial Disease**
- **Neurofibromatosis (types 1 & 2)**[3]
- **Osteogenesis Imperfecta**[3]
- **Pathogenicity of Sequence Variants**[2]
- **Phaeochromocytoma & Paraganglioma**[3]
- **Polyposis**
- **Retinal Disorders**[1,3]
- **Rett syndrome**
- **Skeletal dysplasias**
- **Variant Validation**[3]
- **X-inactivation**[3]

[1]**Gene panel testing** is included in the following EQAs: arrhythmia and cardiomyopathies, hereditary breast and ovarian cancer disorders and retinal disorders. Laboratories performing NGS gene panels, exome and whole genome sequencing are encouraged to participate.

[2]**Pathogenicity of sequence variants interpretation** will provide participants with a single clinical case scenario. Three corresponding genotyping results will be provided for interpretation and classification. Submission will be via a proforma.

[3]**NEW EQAs for 2019:**

- Ataxia and Hereditary Spastic Paraplegia (Friedreich ataxia, spinocerebellar ataxias and HSP)
- Breast & Ovarian Cancer disorders (includes Cowden, Li-Fraumeni and Peutz Jeuger Syndromes)
- Imprinting and Uniparental disomy (Angelman and Beckwith Wiedemann syndromes)
- Neurofibromatosis (types 1 and 2) and Schwannomatosis
- Osteogenesis Imperfecta
- Phaeochromocytoma and Paraganglioma (endocrine tumour predisposition and Von Hippel Lindau disease)
- Retinal Disorders
- Variant Validation (validation of a sequence variant detected on a research sample)
- X-inactivation