EQA news for 2020

We are modifying our EQAs to adapt to the needs of our participants and the implementation of gene panel testing, so they are based on clinical indications rather than single gene test.

GenQA GENOMICS QUALITY ASSESSMENT
Member of UK NEQAS consortium

New for 2020

- **Disorders of Sexual Development (DSD)**
  (Congenital adrenal hypoplasia, Androgen insensitivity, *SHOX)*
- **Epilepsy Disorders**
  (includes tuberous sclerosis, Rett syndrome, Dravet syndrome)
- **Respiratory disorders**
  (*FLCN*-related disorders and Pulmonary Arterial Hypertension)
- **Renal Disorders**
  (Alport syndrome and Polycystic kidney disease)
- **Osteogenesis Imperfecta (OI)**
- **Infertility (CF, POF, Ydel)** (interpretation only)
- **Pathogenicity of Somatic Variants** (classification only)
- **NGS Copy Number Variant (CNV)**
- **SCID new-born screening** (TREC detection)
- **CLL *TP53* variant detection** (In collaboration with UK NEQAS LI)
- **CLL IGHV mutation status** (In collaboration with UK NEQAS LI)
- **Microdeletion syndromes**
- ***NTRK* fusions**
- **Endocrine tumours (somatic)**
- **Tumour mutation burden**
- **Genetic Counselling** (If interested please contact GenQA)
- **Linkage analysis for HD, CF and DMD** (interpretation only)

Updated EQA for 2020

- **Cardiac Disorders** (previously Arrhythmia/Cardiomyopathies)
  (also includes aortic dissection e.g. Marfan syndrome)
- **Neurodegenerative Disorders** (previously Dementia and ALS)
  (also includes Parkinson disease)
- **Muscular Dystrophies** (previously DMD/BMD)
  (also includes Limb-girdle, Emery Dreifuss and Congenital muscular dystrophies)
- **Inherited Colorectal Cancer and Polyposis syndromes**
  (combined Lynch syndrome and Polyposis syndromes)
- **Inborn Errors of Metabolism** (previously Fabry disease)
  (Fabry disease, Tay Sachs and Gaucher syndrome)
- **Neurofibromatosis type 1 and Rasopathies** (previously NF1/NF2)
  (Neurofibromatosis types 1/2 and Noonan syndrome)
- **Eye Disorders** (previously Retinopathies)
  (retinopathies, structural eye disorders and albinism)
- **Imprinting Disorders** (previously UPD/Imprinting)
  (Beckwith Wiedemann, Angelman and Silver Russell syndromes)
- **Lung Cancer/Additional Lung Cancer** choose one of:
  1) *EGFR* only, 2) core (*EGFR, ALK and ROS1,* 3) comprehensive (*EGFR, ALK, ROS1, KRAS, BRAF, PIK3CA, RET, MET*), 4) fusions (*ALK, ROS1, RET, MET)*
- **Colorectal Cancer** - choose one of:
  1) core (*KRAS, NRAS, BRAF,* 2) Mismatch repair (core + *MLH1* promoter methylation), 3) Extended mismatch repair (mismatch repair + MSI)
- **Pathogenicity of Sequence Variants** – choose either:
  Classification only: submit a proforma
  Classification and clinical interpretation: submit a clinical report

Further details on these EQAs, and the complete list of GenQA 2020 EQAs, is available at www.genqa.org/eqa