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UK NEQAS for Molecular Genetics Scheme 2009 Report for CMGS

UK NEQAS for Molecular Genetics provided a large number of External Quality Assurance (EQA) schemes covering a wide range of disorders during 2009. Ten of these schemes were developed for core diseases and involved full genotyping/interpretation exercises. The following diseases were assessed:

- Cystic fibrosis (CF)
- Familial breast and ovarian cancer (*BRCA1* and *BRCA2*)
- Fragile X syndrome (FRAX)
- Friedreich Ataxia (FRDA)
- Hereditary Motor and Sensory Neuropathy and Hereditary Neuropathy with Liability to Pressure Palsies (HSMN/HNPP)
- Huntington disease (HD)
- Medium chain acyl coA dehydrogenase (MCADD)
- Mitochondrial diseases (mtDNA)
- Myotonic dystrophy type 1 (DM)
- Spinal muscular atrophy (SMA)

There were two UK laboratory poor performers for genotyping (one in the CF scheme and one in the SMA scheme). There was also a poor performing UK laboratory in the CF for non-submission of results. All other schemes were completed to a high standard.

Following the success of the Maternal Cell Contamination pilot EQA during 2008, this scheme was expanded to include sexing of the samples. The EQA was also upgraded to a full scheme and performance was assessed for genotyping and interpretation. This EQA was performed to a high standard by the UK participants.

To reflect the nature of the gene screening work performed by diagnostic laboratories, a full *BRCA1* gene screening pilot EQA scheme was offered during 2009. This scheme involved the testing of one sample within a 12 week testing period. This was a genotyping only exercise and fully interpretative reports were not required. This pilot scheme was completed to a high standard and proved useful to participants. This format will be repeated in 2010 for the *BRCA2* gene.

As a diagnostic service is provided by regional laboratories for rarer diseases then sample swap genotyping only EQAs were offered for the following diseases:

- Achondroplasia
- Familial Hypercholesterolaemia
- Multiple Endocrine Neoplasia type 2
- *MUTYH*-associated polyposis
- Rett syndrome
- Spinal Bulbar Muscular Atrophy
- Von Hippel-Lindau disease

Again the participants completed these schemes well. There was one poor performing laboratory in the Familial Hypercholesterolaemia scheme.

The collaboration with UK NEQAS for Clinical Cytogenetics continued during 2009 and the Molecular Rapid Aneuploidy EQA provided to 50 laboratories (UK and non-UK). The assessment was completed well and no UK based laboratories performed poorly.

The Cystic fibrosis molecular testing on blood spots EQA was provided in collaboration with NGRL (Manchester). Two sample distributions were dispatched to 19 participants. This EQA will be upgraded to meet the UK Newborn Screening Laboratory Quality Assurance Development Group EQA Specifications during 2010 and will be transferred to UK NEQAS for Molecular Genetics. We would like to thank NGRL (Manchester) for all their help and support over the years with this EQA.

The gastro-intestinal stromal tumour (GIST) molecular testing pilot EQA scheme was successfully launched during 2008 in collaboration with NGRL (Manchester) and continued as a pilot EQA for 2009. There was an increase in the number of participants in this second year of the pilot; however there continued to be a variation in the reporting of mutations in these samples. The scheme will continue as a pilot during 2010 and interpretation will be assessed.

In response to demand from participants, a new pilot EQA was launched in 2009 for the molecular genetic analysis of *KRAS* in metastatic colorectal cancer. Samples were provided either as paraffin embedded tumour tissue or slide mounted paraffin tumour tissue (or both). The standard of results submitted varied between laboratories and indicated the need for future molecular based EQA in this field. A second year of this pilot scheme will be provided during 2010.

A successful pilot scheme was completed for the molecular testing for Cystic fibrosis in Preimplantation Genetic Diagnosis (PGD). This pilot scheme included two stages; Stage 1 required participants to perform a DNA feasibility study to determine if PGD could be provided by their laboratory for the "EQA couple", and Stage 2 required the testing of single lymphocyte cells to mimic the testing of embryo cells. The standard of performance by the PGD laboratories was very high and this new type of EQA was received well. This pilot EQA will be offered during 2010 and will be opened to all interested parties.

Full details of 2010 EQA schemes can be found on the UK NEQAS for Molecular Genetics website (www.ukneqas-molgen.org.uk).

Dr Sandi Deans
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