The Royal College of Pathologists
Pathology: the science behind the cure

National Quality Assurance Advisory Panel - Genetics

Annual Report 2015-16

Meetings

The Panel met twice during this period (30th March 2015 and 5th October 2015) with this frequency of meetings set to continue for 2016-17. In addition panel business is conducted by email between meetings.

Panel Membership

At the beginning of this period, Dr Jonathan Waters took over from Prof Mike Griffiths as Chair. Prof Griffiths kindly agreed to remain as Deputy Chair until the end of 2016. Two professional bodies with representation on the committee, AC C and CMGS have now merged to form the ACGS (Association of Clinical Genetic Science) and are identified accordingly (three nominees).

Current membership of the panel at the end of this period is as follows:

Dr Jonathan Waters (Chair)	RCPath Representative
Prof Mike Griffiths (Deputy Chair)	ACGS Representative
Paul Roberts	ACGS Representative
Dr Kathy Mann	ACGS Representative
4th ACC/CMGS representative	*post rescinded in agreement with the relevant professional bodies*
Dr Shirley Henderson	BSH Representative
Dr Gill Rumsby	ACB Representative
Ms Fiona Coyne (secretary)	Genetic Technologists Rep
Dr Jane Moorhead	IBMS Representative

In addition specialist advice continues to be provided by the scheme organisers or their delegated deputies for schemes which report to NQAAP for Genetics:

Dr Ros Hastings	NEQAS in Clinical Cytogenetics Scheme Organiser
Dr Sandi Deans	NEQAS in Clinical Molecular Genetics Scheme Organiser
Dr Simon Patton	EMQN Scheme Organiser
Prof David Barnett	NEQAS for LI (Molecular Haematology)
Jerry Hancock	UKMRD Network and EuroMRD Network

The following EQA or similar schemes currently report to or are monitored by NQAAP for Genetics:

- UK NEQAS for Clinical Cytogenetics
- UK NEQAS for Molecular Genetics
- UK NEQAS for Molecular Pathology
- UK NEQAS for Leucocyte Immunophenotyping (Molecular Haemato-oncology).
- Cytogenetics European Quality Assessment Schemes (CEQAS)
- European Molecular Genetics Quality Network (EMQN)
- UKMRD Network and EuroMRD Network

Scope of Report

This NQAAP for Genetics Annual Report is for 2015-16, covering NQAAP meetings and activity within this period. The scheme reports to NQAAP generally cover the 2015 calendar period but where this is not the case, the time period covered is clearly stated.

Overview of Schemes

Significant growth in cross disciplinary EQA has taken place during this period, particularly in Molecular Pathology. In addition Scheme Organisers are developing EQAs across traditional Organiser domains.

CEQAS (UK NEQAS for Clinical Cytogenetics)

No performance criteria were submitted for approval or review in 2015.

UK NEQAS for Clinical Cytogenetics offered 14 full EQAs and nine pilot EQAs, run over 2015.

Poor performance in 2015 is summarised below:

Poor Performance 2015

EQA	No. of participants	Non-submission	No. of countries	Total amber analysis	Total amber interpretation	Total red		UK participants	Total amber analysis	Total amber Interpretation	Total Red
ALL	73	1/73	24	6	1	3		26	1	0	0
Amniotic Fluid	118	2/118	28	3	0	0		24	0	0	0
AMN (1p/19q)	24	0	6	2	0	0		19	1	0	0
Blood	175	1/175	39	23	3	3		25	1[1]	0	0
CVS	110	1/110	23	4	2	2		24		0	0
Constitutional array (postnatal) *	107	0	25	2	0	0		22	1	0	0
FISH RA	41	2 (plus 1 not marked)	20	2	0	0		6	0	0	0
Mature B&T neoplasms (G-banding)	45	0	15	11	0	3[2]		15	3	0	0
Mature B&T neoplasms (FISH only)	66	1/66	19	7	2	3[2]		26	2	0	0
MRA (QF-PCR/MLPA) ***	85	0	20	1***	0***	0		14***	1[1]***	0	0
Myeloid (AML/MDS/CML)	117	1	30	32	11	5[2]		26	1	0	0
PGD blastomere (FISH)	18	1	12	0	0	0		3	0	0	0
Products of conception	21	0	8	0	0	0		12	0	0	0
Tumour	27	1	9	2	4	1		20	1	2	0

*In collaboration with EMQN (European Molecular Genetics Quality Network), only CEQAS data given here
**In collaboration with UK NEQAS for Molecular Genetics, 56 labs from CEQAS and 29 labs from UK NEQAS for Molecular Genetics
***Only CEQAS data
[1] Same lab
[2] One lab received poor performance for Mature B&T neoplasms (online), Mature B&T neoplasms (FISH) and Myeloid (AML/MDS/CML) in 2015 so has been designated as persistent poor performance for acquired in 2015

There were 12 instances of Poor Performance (amber) in UK Laboratories in 2015.

A number of outstanding poor performance issues that had been previously brought to the NQAAP's attention were closed off during this time period. At the time of this report (June 2016) there were no outstanding persistent poor performance issues pertaining to UK laboratories.

UK NEQAS for Molecular Genetics

No performance criteria were submitted for approval or review.

There were 16 instances of Poor Performance (amber) in UK Laboratories in 2015 (data compiled from two reports: April-September 2015 and October 2015-2016.

EQA run	UK Participants	UK amber (genotype)	UK amber (interpretation)	UK Red (genotype and interpretation)
PGD for Myotonic Dystrophy – single cell testing	6	0	0	0
Lung Cancer – run 1	28	5	0	1
Colorectal cancer – run 1	34	5	0	0
Melanoma cancer – run 1	32	3	0	0
Huntington disease	17	1	0	0
Myotonic Dystrophy	16	1	0	0
GIST molecular testing	12	1	0	0

Performance issues were over-represented in the 'Molecular Pathology' schemes – partly because these EQAs are relative new, the inevitable tissue heterogeneity of the samples and the quality of DNA obtained from archive tissue samples. These issues are gradually been worked through as part of the educational aspects of the EQA schemes.

There were three laboratory referrals for Persistent Poor Performance to the NQAAP. At the time of this report (June 2016) there were no outstanding persistent poor performance issues pertaining to UK laboratories.

European Molecular Genetics Quality Network

Only performance in the genotyping aspects of the EQA schemes by UK labs is currently reported to the NQAAP panel.

The scheme ran 37 full EQAs and five pilot EQAs during this time period. For UK laboratories there was a single amber poor performance marker assigned. In particular the EQMN has been working to harmonise its Performance/Scoring Criteria in line with comparable NEQAS schemes.

EMQN reported no UK cases of Poor Performance, or Persistent Poor Performance in 2015.

UK NEQAS for Leucocyte Immunophenotyping (Molecular Haemato-oncology)

There were 12 EQA programmes together with a further seven identified pilot programmes.

There were six amber performance incidents in UK laboratories in 2015 out of a total of 306 scheme participations (2.0%) – see table below).

Four persistent poor performers came to the NQAAP's attention and these have now been resolved.

The following changes in performance monitoring were considered by the NQAAP this year.

- A New z-score based ISO 17043/13528 compliant performance monitoring systems have now been implemented for all quantitative UKNEQAS LI programmes.
- Amendment to current scoring system submitted for the IGH/TCR clonality programme to take into account BIOMED guidelines (Langerak 2013) that advocated a more descriptive molecular conclusion was also being implemented The NQAAP was receiving regular reports on their implementation.
- New performance monitoring system submitted for UKNEQAS LI's KIT D816V and *BRAF* V600E EQA programmes.

There were four laboratory referrals for Persistent Poor Performance to the NQAAP. At the time of this report (June 2016) there were no outstanding persistent poor performance issues pertaining to UK laboratories.

UK NEQAS-Li Schemes summary

8. Accredited EQA Programmes issued 30th March 2015 – 5 Oct 2015

Programme	Trial ID	No. of Participants	No. of UK Participants	Total Non Returns	UK Non returns	No. Amber performance	No. UK Amber performance
BCR-ABL1 + AML Translocation Identification	141503	121	24	4 (3.3%)	0 (0%)	13 (10.7%)	2 (8.3%)
BCR-ABL1 + AML Translocation Identification	151601	127	30	4 (3.1%)	0 (0%)	2 (1.6%)	0 (0%)
Major *BCR-ABL1* Quantification	141502	181	28	12 (6.7%)	1(3.6%)*	21 (11.7%)	2 (7.1%)
Major *BCR-ABL1* Quantification	141503	181	28	11 (6.1%)	2 (1.1%)*	12 (6.6%)	0 (0%)
Post-SCT Chimerism Monitoring	141503	107	25	4 (3.7%)	0 (0%)	11 (10.2%)	1 (4.0%)
IgH/TCR Clonality Status	141503	104	29	1 (1.0%)	0 (0%)	3 (2.9%)	0 (0%)
IgH/TCR Clonality Status	151601	103	29	1 (1.0%)	0 (0%)	6 (5.9%)	0 (0%)
JAK2 p.Val617Phe (V617F) Mutation Status	141503	202	39	12 (5.9%)	3* (7.7%)	5 (2.5%)	0 (0%)
NPM1 Mutation Status	141502	97	25	3 (3.1%)	1 (4.0%)*	0 (0%)	0 (0%)
NPM1 Mutation Status	141503	99	24	3 (3.03%)	0 (0%)	1 (1.01%)	0 (0%)
FLT3 Mutation Status	141503	98	25	2 (2.0%)	1 (1.0%)*	7 (6.9%)	1 (1.0%)
Totals	**11 TRIALS**	**1420**	**306 (21.5%)**	**57 (4.0%)**	**8 (2.6%)**	**81 (5.7%)**	**6 (2.0%)**

* participant(s) not currently offering a clinical service

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UKMRD Network and the EuroMRD Network QC scheme

EuroMRD scheme is developing performance scoring and poor performance criteria.

Complaints

No complaints were received by the panel in 2015-16.

The Chair of the Panel is grateful for all the hard work of all the EQA Organisers, their support staff, Panels and SACs together with members of the Genetics NQAAP itself in this year.

Dr Jonathan Waters,
Chair, NQAAP for Genetics,
June 2016