



European Society of Human Genetics

Synopsis of Minutes Quality Committee

Tuesday, 16th December 2008

10:30 – 15:30

Skyport Centre Brussels

Attendees:

Ros Hastings (RH) – Chair & CEQA

Brian Fowler (BF) – ERDIM

Jacques Beckmann – ESHG Board

Claude Giroud – EDMA (industry)

Mireille Claustres – Diagnostic Laboratory

Viktor Kozich – SSIEM and ERNDIM Board

Els Dequeker (ED) – CF Network

Konstantin Miller – ECA Board representative replacing Lidia Larizza

Bert Bakker - EMQN replacing Rob Elles

Apologies

Rob Elles (RE) – EMQN

Peter Farndon – Clinical Geneticist & UKGTN

Orsetta Zuffardi – Genetic Research Community - flight cancelled the day of the meeting

Lidia Larizza – ECA Board representative

Cor Oosterwijk – EGAN (patient group)

1. Minutes of previous meeting to be agreed

The minutes were approved as an accurate reflection of the meeting in June 2008 pending the correction of some spelling mistakes.

2. Matters arising

Question was raised if high throughput screening should be an issue of the QC (JB).

3. Adoption of reference materials by WHO

RH

WHO has adopted the Fragile X Reference Materials (RMs) following statements from the Q.C. and other professional bodies.

The PWS/AS RMs have not been adopted to date.

Action: The QC will send a letter to the WHO so that a decision can be made on the PWS/AS RMs.

4. Governance of EQA and review of annual Management Review by different European EQA Schemes.

It was agreed that the 4 Schemes would submit their annual Management Reviews for the QC meeting in May after their EQA year had completed.

Because all the Management Review submitted by the different Schemes varied in format and content, a discussion ensued on what the Q.C required to be in the report. The following items were agreed.

- Introduction and basic structure of the EQA Scheme.
- Plans for the future, staff changes.
- Marking criteria (basic)
- Minimum satisfactory performance criteria.
- Total number of participants and number of participants per EQA.
- EQAs offered that year.
- Who is responsible for each Scheme/ EQA.
- Assessors involved for each EQA
- Number labs with a satisfactory performance per EQA.
- Number of non-submissions.
- How many labs have poor performance
- What were the errors in poor performing labs
- How many labs had a poor performance in more than one EQA or over several years
- Problems with a particular method/technique
- Appeals process.
- Improvements in the EQA scheme.

Concern was raised about exchanging information about poor performing laboratories with other professional bodies such as the Q.C committee. Anonymity would be kept. The QC should be informed if the same laboratories made mistakes/poor performance year on year.

All 4 schemes produce participation certificates.

Customers need to know which lab is working well and the performance in any EQA scheme. As only participation can be public knowledge, one way forward is to encourage accreditation as assessors look at the EQA performance data.

The need for 2-3 assessors per EQA was discussed. **Action:** ERNDIM to look into changing the number of assessors per EQA from one to two in the first instance.

Governments do not know enough about the role of quality management and the different initiatives such as EMQN, CEQA, Eurogentest etc. The need to publicize the role of quality management through the ESHG was recognized by the Q.C committee. Suggested Q.C. mentions that the OECD guidelines and ISO 15159 should be adhered to by all genetic labs and labs should be accredited.

JB – Other EQA schemes offer EQA for genetics in addition to CEQA, ERNDIM, EMQN and CF Network. The need to interface with these EQA schemes was identified. Possible interfaces were discussed.

There is a need for a harmonized consent form. Patient consent is needed prior to any publication e.g. J Med Genetics.

5. ISO standards

ED

ISO 15189 will feature no separate annexe for genetics. The guidelines for the professions are under review and genetic laboratories are invited to comment. ISO 17043 (Conformity assessment – General requirements for proficiency testing) is now ready for voting and then will be released for consultation.

6. Identification of gaps in quality that need addressing – identify a way forward

Informed Consent (MC)

Clinical utility of genetic tests (JB)

1. Audit satisfactory performance across the three disciplines
 - 1.1 *Number of critical errors.*
 - 1.2 *Set standards for satisfactory performance.*
 - 1.3 *Risk calculations for genes vary within EQAs*
 - 1.4 *Alert all countries to EQA scheme provision.*
2. Genetic testing not done by genetic labs.
 - 2.1 *Need to make these labs aware EQA and Q.C. exists.*
 - 2.2 *Pharmacogenetics and haemochromatosis – contacts needed.*
3. Publicize information from the QC report.
 - 3.1 *To labs, ESHG, non genetic labs.*
 - 3.2 *European networks.*
4. Emerging EU countries.
 - 4.1 *Identify their specific needs.*
5. Best Practice Guidelines.
 - 5.1 *Best practice guidelines – identify which need updating. Review/ratify new versions*
 - 5.2 *Need for minimum organization provision for genetic labs in BP guidelines.*
 - 5.3 *Minimum population covered by a genetic service.*
 - 5.4 *Need for guidance on interpretation and risk assessment for CNVs, missense mutations.*
6. Changing landscape of genetic testing.
 - 6.1 *Whole genome analysis - Future planning, changes to genetic services.*
 - 6.2 *Suggest to the ESHG a symposium to discuss way forward.*
7. Rare variants.
 - 7.1 *Pathological missense mutations, CNVs etc.*
8. Newborn screening.
 - 8.1 *Gap in genetic testing –more attention required for pre-genetic counseling and testing prior to screening.*
 - 8.2 *Quality issues related to neonatal screening.*
9. Pre-implantation diagnosis – issues consist of:-
 - 9.1 *PGS screening- clinical utility.*
 - 9.2 *Single cell mutation and haplotype analysis.*
 - 9.3 *Whole genome analysis.*

7. Definition of poor performance in genetic laboratories.

- (i) Setting satisfactory/poor performance standards at an EQA level.

8. Feedback on the EUGT- Orphanet Quality Assurance (QAu) database ED on the status of accreditation and EQA participation in Europe.

Database publicly available since 1/2/08 via Orphanet.

Laboratories need to submit data on QA assurance. It was suggested that the EQA schemes could urge laboratories to register with the QA database. To date there are about 300 validated laboratories.

9. Any other business.

- (i) Need for Reference Materials expert on Committee – **it was agreed that** the QC will invite David Barton to join the committee.
Action: RJH to invite DB onto the Committee.
- (ii) It was suggested that the Q.C. make a synopsis of information relevant to the diagnostic labs e.g. Aims; QA database; EQA schemes available; reference materials.
- (iii) Publicity of Q.C outcomes/ information through the ESHG newsletter; EQA providers; ESHG journal; ESHG board and national societies.
- (iv) Marking criteria for EQA schemes. There was some discussion of the various marking categories used and total points allocated per category. **Action:** The 4 EQA schemes to document their scoring criteria (allocation of points) for the next Q.C. meeting.
- (v) BB mentioned the EMQN feedback questionnaire. **Action:** EMQN to feedback responses for the next Q.C. meeting.

10. Date of next meeting.

ESHG Conference May 25th 2009 in Vienna.