Genetic Services Quality Committee
GSQC

Ros Hastings
GSQC

2013/2014:

• Reporting Guidelines for genetic laboratories published in EJHG (open access)
• Data on EQA poor performance
• Meeting on European consensus on reporting unsolicited findings and unknown variation – publication planned
• Quality Assurance in Genetic Counselling planning an exploratory EQA later this year
• Meeting on informed patient consent – pre-test counselling
• QC representative on the HGVS Sequence variant description committee
EQA unsatisfactory (poor) performance

- Genotyping critical errors that affect patient management
- Interpretation critical errors
- In some EQAs the poor performance >10%
- Some European labs do not follow OECD guidelines i.e. do not interpret the significance of the genetic results

Persistent performance – more than one poor performance in three years

- Swiss and UK have a surveillance body to which labs or EQA providers have to report unsatisfactory performance; not present in other countries

Still room for improvement
GSQC

- Quality Assurance in Genetic Counselling
- 4 Pilot cases
  - Monogenic
  - Cardiovascular genetics
  - Oncogenetics
  - Dysmorphology
- Checked by expert peers and patient representatives
- 11 genetic counselling depts (7 countries) registered so far
- Depts select two or more of the pilot cases
- If interested in participating please email c.m.a.van.ravenswaaij@umcg.nl
- EQA will be online