



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

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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

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- 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