Genetic Services Quality Committee
GSQC

Ros Hastings
GSQC

2012/2013:

- Quality Assurance in Genetic Counselling survey and symposium
- Reporting Guidelines for genetic laboratories published (in press EJHG)
- Practical workshop on nomenclature and interpretation of laboratory results for genetics
- Meeting on European consensus on reporting unsolicited findings and unknown variation.
- Data on EQA poor performance
Participants

32 national societies were asked and reminded to complete the questionnaire.

22 questionnaires were returned (19 complete data)

Poster: P18:32
Workshop: WS07 15.15 today
GSQC

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GSQC

EQA unsatisfactory (poor) performance

- Genotyping critical errors
- Interpretation critical errors (CF network and CEQA)
- Some European labs do not follow OECD guidelines i.e. do not interpret the significance of the genetic results.
- Few countries have a surveillance body to which labs or EQA providers have to report unsatisfactory performance.

**Unsatisfactory performance** is different from **persistent unsatisfactory performance**.

- This data does not equate to the diagnostic error rate as there are 2-3 cases/EQA.
# GSQC

Mean critical error in genotyping and interpretation

<table>
<thead>
<tr>
<th>EQA</th>
<th>Year</th>
<th>Number EQA participations</th>
<th>EQA samples</th>
<th>Analytical critical error</th>
<th>Interpret. critical error</th>
<th>Total critical error</th>
<th>Unsatisfactory performance</th>
<th>Range</th>
</tr>
</thead>
<tbody>
<tr>
<td>CF network</td>
<td>2011</td>
<td>218</td>
<td>3</td>
<td>7</td>
<td>19</td>
<td>26</td>
<td>11.9%</td>
<td>n/a</td>
</tr>
<tr>
<td>EMQN</td>
<td>2011</td>
<td>2050</td>
<td>3</td>
<td>134</td>
<td>n/a*</td>
<td>134</td>
<td>6.5%</td>
<td>0-26%**</td>
</tr>
<tr>
<td>CEQA</td>
<td>2012</td>
<td>434</td>
<td>2</td>
<td>13</td>
<td>7</td>
<td>20</td>
<td>4.6%</td>
<td>0-11%***</td>
</tr>
</tbody>
</table>

This data does not include unsatisfactory performance due to non-submission.

*Critical interpretation errors not evaluated (except for UK labs and the joint CEQA/EMQN microarray EQA).

** 6/28 EQAs had >10% unsatisfactory analytical performance (11%, 15%, 18%, 18%, 19% & 26%)

*** 1 EQA had an 11% unsatisfactory performance (included analysis and interpretation critical errors)
A network for the harmonization, validation and standardization in genetic testing
- quality in the laboratory
- quality for genetic counselling
- policy on behalf of a genetic community

Some of the achievements
- More than 600 participants in Training courses on quality assurance
- Harmonization of External Quality Assessment schemes
- Guidelines on genetic counselling for prenatal diagnosis and PGD
- Annual conference on quality in genetic testing
  (Leuven, 2011; Nijmegen 2012; Prague 2013)
- Lobbying for IVD Directive/Regulation
- Representation at EUCERD, IRDiRC and other international forums
- Clinical Utility Gene Cards
- ......
Responsibilities at the national level...

- Accreditation of genetic laboratories is a national matter
  An obligation for accreditation should be written in national law (e.g. as a requisite for reimbursement).

- Workshops on quality assurance can be organized in conjunction with the national society meetings, please contact EuroGentest

- Information on genetic testing for professionals
  Leaflets for patients are available for free distribution via national society websites
  - guidelines on pre- and post-natal genetic counselling
  - Clinical Utility Gene Cards (CUGC, published in EJHG)
  - 15 leaflets in 30 languages
Accreditation status

Note 1: 65 accredited laboratories; 105 preparing for accreditation

Note 2: 58% (n=35) of laboratories achieved accreditation within 2 years; 93% (n=56) within 5 years

ARTICLE

Approaches to quality management and accreditation in a genetic testing laboratory

Sarah Berwouts¹,³, Michael A Morris²,³ and Elisabeth Dequeker⁺,¹,³

SPECIAL ARTICLE

Mutation Nomenclature in Practice: Findings and Recommendations from the Cystic Fibrosis External Quality Assessment Scheme

Sarah Berwouts,¹,⁶ Michael A. Morris,²,⁶ Emmanuelle Girodon,³,⁶ Martin Schwarz,⁴,⁸ Manfred Stuhrmann,³,⁶ and Elisabeth Dequeker¹,⁸⁺

REPRODUCTIVE GENETICS Review

Accreditation of the PGD laboratory

J.C. Harper¹,²,⁹ S. SenGupta¹, K. Vesela³, A. Thornhill⁴, E. Dequeker⁵,⁶, E. Coonen⁷, and M.A. Morris⁶,⁸
The future of EuroGentest

- Funding by the European Commission ends December 2013
- EuroGentest’s activities will be integrated with the ESHG

The European Society of Human Genetics and EuroGentest plan to create an association of genetic laboratories, health care and educational institutes in Europe.

= a new concept: institutional membership

Goals of the EuroGentest association:

- Provide a platform for collaboration between institutes and laboratories
- Be a voice and a lobbying group on behalf of genetic services and genetic laboratories,
- Continue to offer training and guidance in clinical and laboratory genetic testing (workshops, guidelines,...)