

Elisabeth Dequeker, Sarah Berwouts, David Barton, Christine Brady, Jana Camajova, Philippe Corbisier, Anniek Corveleyn, Lieve Desmet, Rob Elles, Brian Fowler, David Gancberg, Tom Janssens, Outi Kämäräinen, Milan Macek Jr., Gert Matthijs, Michael Morris, Clemens Müller, Nick Nagels, Bettina Quellhorst-Pawley, Alexandra Stambergova, Elfriede Swinnen and Ros Hastings

EuroGentest Network of Excellence, Sixth Framework Programme Priority FP6-512148

UNIT AIMS

- provision of sustainable and harmonized EQA for all genetic labs through National and European EQA
- to help and encourage labs to implement a quality system
- to facilitate access to and production of (certified) reference materials
- to produce validated SOPs for diagnostic procedures

ACHIEVEMENTS & FUTURE PLANS

Harmonization

Aims:

- Up-to-date and informative website
- Develop, review and disseminate quality procedures and guidelines
- Strengthen internal networking
- Collaborate with other organizations:
 - OECD
 - EA and ISO
 - EQA providers
 - ESHG
 - Orphanet
 - ...

Focus:

- Bring all disciplines of quality management in medical laboratories together
- Create opportunities for close collaboration
- Start discussions



Future:

- On-line educational tool
- Document repository
- Experts' meeting for content of website

QAu Database

Aim: Make validated information on quality assurance in medical genetic testing laboratories easily available

EUROGENTEST QUALITY ASSURANCE DATABASE

- Including: Laboratory details
Full sharing and integration of data with Orphanet
Quality assurance data (quality manager, EQA participation and accreditation data)
All laboratories providing medical genetic testing: → molecular genetics
→ cytogenetics
→ biochemical genetics

Use: Find a lab performing test not available in your area
Identify labs investing in QAu
Watch the uptake of QAu in Europe

Reason to join the database: Valorize efforts and investment in quality assurance
Notification of the tests you perform.
It is for free

How: Update your data online via your lab page.
Add your data online: <http://www.eurogentest.org/QAuDatabase>
Or: <http://www.orpha.net>

More information: Contact gausurvey@eurogentest.org
See poster **P09.15**

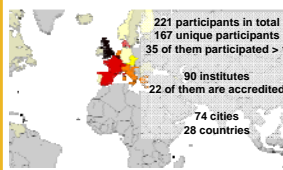


Workshops

Aim:

- To encourage accreditation
- To improve and harmonize quality systems

Participants of past 9 workshops (P09.71):



Share

Genetic testing labs all face the same problems
They have all found solutions in the past

Learn

What do the accreditation standards require for genetic testing?
How have accredited, expert and smaller labs dealt with similar situations?
How can we use other labs' experiences (and mistakes)?

Apply

Case studies and small-group discussion of common laboratory situations
Role-playing and videos of internal audits
Hands-on trials of quality management software

Future:

- Quality assurance and quality control**, 2 hrs during the ESHG, 31 May '08.
If there are places left, registration possible at EUGT booth #220
- Management review, internal quality control and EQA**, Berlin, 9-10 October '08.
Registration: www.eurogentest.org/unit1/workshops.xhtml
- Diagnostic validation** in genetic testing laboratories, Prague, 8-9 January '09.
Registration: www.eurogentest.org/unit1/workshops.xhtml

Reference materials

General Objective:

Promote the development and use of Reference Materials for Genetic Testing

Aims:

- Promote networking of developers and users of Reference Materials (RMs) to identify needs
- Develop a priority ranking of RMs required
- Support the development of new RMs for Genetic Testing
- Inform stakeholders of the implications of the IVD Directive for Genetic Tests

Ongoing Work:

- Two International Symposia on RMs in Genetic Testing organised (Geel 2005 and Dublin 2007)
- Prioritization score developed and publicized
- Financial and logistical support to NIBSC UK for the development of a panel of RMs for Prader-Willi and Angelman syndromes
- Participation in the development and characterization of WHO panels for Fragile X syndrome and hereditary haemochromatosis
- Field trials organised (in collaboration with EU CF Network) for 2 different commercial multiplex RMs for cystic fibrosis testing (published online *Human Mutation*, May 9, 2008)
- Release of a guidance document for the Use of RMs in Genetic Testing
- Proficiency testing study for Factor II mutations organised and evaluated
- Three IRMM/IFCC Certified Reference Materials for prothrombin G20210A mutation produced (*Clin Chem Lab Med* 2005, 43: 862-868 and 2008, 46:463-469 and see **Poster P09.61**)
- Workshop on the implications of the IVD Directive organised (Leuven, April 2007)
- Workshop on RMs needs for new genetic testing technologies e.g. microarrays (Geel, April 2008)



EQA schemes

Many laboratories do not have access to EQA because there is no National scheme or no EQA available for a specific test. EuroGentest has brought together all sectors of genetic testing and helped new initiatives:

Cytogenetics: after two years of pilot EQAs, CEQA now offers a full constitutional postnatal and prenatal EQA as well as two pilot EQAs, haematology and Preimplantation Diagnosis via the internet. Last year's pilot had participants from 23 countries. CEQA registration is open to all countries. The Forum of European EQA providers continues to meet. <http://www.ceqa-cyto.eu>



Molecular Genetics: EuroGentest brought together European molecular genetic EQA providers to discuss harmonization and following consultations with national representatives EUGT has helped the European Molecular Genetics Quality Network pilot new disease service specific and technical EQA schemes, promote a new series of meetings of scientists and pilot an electronic tool to draft best practice guidelines. <http://www.emgn.org>



Biochemical Genetics: The umbrella organisation ERNDIM networks with national and international societies and provides web administered EQA schemes with certified performance. ERNDIM offers a laboratory directory, methodologies, educational documents, best practice and training workshops and coordination of IQC, and accreditation. EUGT is allowing all these activities to be expanded. <http://www.erndim.unibas.ch>



Validation

General objective:

Establish procedures and guidelines for the analytical validation of methods and technologies

Aims:

- Issue guidelines for the analytical validation of molecular genetic tests
- Organize expert meetings on validation and train people during workshops
- Perform interlaboratory validation trials for MLPA, DNA extraction, ...

Ongoing work:

- Guidelines (more info see **P09.73**):
 - First draft 'Minimum acceptable standards for analytical validation of molecular genetic tests
 - Annexes providing detailed instructions for sequencing, Abbott Fragile X kit, ...
- MLPA:
 - Differences in MLPA protocol between laboratories and IT tools for the analysis of the MLPA results were evaluated: technical report in review
 - Interlaboratory validation of the MLPA-kit for BRCA1 : technical report in review
- DNA extraction:
 - Validation of DNA extraction (large blood volumes) on the Chemagen is ongoing, evaluation of other platforms will follow (Autogen, Gentra)