
**Summary
Document**

Clinical Molecular Genetics
External Quality Assessment

European National representatives meeting
Prague, Czech Republic
17th February 2006

June 2006
1st update Sept. 2006

EuroGentest



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The views expressed in this document are those of the meeting participants and do not necessarily reflect the policies of the institutions or companies they are affiliated to.

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Clinical Molecular Genetics External Quality Assessment
European National Representatives meeting
The situation of External Quality Assessment in Europe.

Summary

The EuroGentest project invited a group of experts in clinical molecular genetics from across Europe, to discuss how innovation and improvement of access to External Quality Assessment schemes (EQA) might most benefit molecular genetic testing laboratories. This report represents an overview of the discussion between representatives of European countries. EuroGentest recognises that because only a single representative from each country was able to attend, the views presented may not be wholly representative of an entire country. However, the intention was not to produce a detailed review of the situation in each country. If views from a larger number of representatives from each country were needed then it is hoped that this report may stimulate further meetings at a national or regional level.

The meeting highlighted variation between different countries in the legislation relevant to molecular genetic testing laboratories and in the regulations concerning EQA. Whilst EQA participation was not mandatory in any country there was agreement that disease specific EQA was essential for the most clinically relevant diseases. It was agreed that in disease specific EQA schemes the assessment of written interpretative reports is an essential element of quality. Disease specific EQA with written interpretation should best be organised at a national/regional level (according to language). It was also felt that technical EQA should be a complement to and not a substitute for disease specific EQA, and could better be organised internationally. At the present time, EQA is absolutely dependent on volunteer experts to act as scheme organisers, otherwise cost of participation would become a major barrier for laboratories. A lack of and need for up-to-date best practice guidelines was expressed, which should ideally be addressed through European cooperation.

Meeting programme

The meeting took place on Friday February 17th 2006 at the University Hospital Motol, Prague, Czech Republic.

Programme	
09:00 – 09:30	Clemens Müller-Reible (Würzburg, Germany), welcome, aims and format of workshop
	Milan Macek Jr (Prague, Czech Republic) Welcome by local host and technical announcements
	Tour de table
09:30 – 10:00	Els Dequeker (Leuven, Belgium) Presentation of the EuroGentest project
10:00 – 10:45	Round table discussion of the questionnaire items and other issues relevant to EQA Each item will be introduced by a brief summary of the questionnaire results (Clemens Müller-Reible). Participants are invited to comment on the situation in their countries and address other issues not covered by the questionnaire.
10:45 – 11:15	Coffee break
11:15 – 13:00	Round table continued
13:00 – 14:00	Lunch
14:00 – 14:30	Rob Elles (Manchester, United Kingdom) Presentation of EuroGentest work package 1.3 relating to the harmonisation of existing EQA schemes in Europe and the expansion of opportunities for molecular genetic testing laboratories in EU25 to participate in EQA
14:30 – 15:45	Round table discussion continued
15:45 – 16:15	Coffee break
16:15 – 17:00	Discussion (EuroGentest activities and beyond)

Meeting Participants

Participant	Institution	
Clemens Müller-Reible	EuroGentest WP1.3 co-leader Professional Association of German Human Geneticists (BVDH)	
Uta Malburg	EuroGentest project officer, Würzburg, Germany	
Rob Elles	EuroGentest WP1.3 co-leader, Coordinator European Molecular Genetics Quality Network, Manchester UK	
Kate Vickers	EuroGentest project officer, Manchester, UK	
Sandi Deans	United Kingdom National External Quality Assessment Service (UKNEQAS) for Molecular Genetics, Manchester, UK	
Els Dequeker	EuroGentest unit 1 leader Cystic Fibrosis Network, Leuven, Belgium,	
Paula Pacheco	Centre for Human Genetics, Instituto Nacional de Saude, Lisbon Portugal	
Elve Raukas	Tartu University Clinics United Laboratory, Tartu, Estonia	
Olaug Rodningen	Dept. Medical Genetics, Ullevaal University Hospital, Oslo, Norway	
Marianne Schwartz	Dept. of Clinical Genetics, Rigshospitalet, Copenhagen, Denmark	
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David Barton	Our Lady's Hospital for Sick Children, National Centre for Medical Genetics, Dublin, Ireland	
Radim Brdicka	Laboratory for Molecular Genetics, Institute of Haematology and Blood Transfusion, Prague, Czech Republic	
Maurizio Ferrari	Molecular Biology Laboratory, Ospedale S. Raffaele, Milan, Italy	
Michel Goossens	Hopital Henri Mondor, Creteil, France	
Franziska Joncourt	Childrens University Hospital, Bern, Switzerland	
Ludovit Kadasi	Genexpress Ltd. Bratislava, Slovak Republic	
Veronica Karcagi	National Centre for Public Health, Budapest Hungary	
Mauri Keinanen	LabQuality, Helsinki, Finland	
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Milan Macek Jr.	Dept. of Molecular Genetics, Institute of Biology and Medical Genetics, Prague, Czech Republic	

Introduction

Background

During the past 15 years, molecular genetic testing (MGT) has evolved into the fastest growing field in laboratory medicine, characterised by a rapid transition from research into diagnostic genetics services. Molecular genetic testing is being used in all areas of medicine. The number of genes that have been associated with monogenic Mendelian disorders is steadily increasing. Recently, genetic variation in individuals has come into focus as the basis for drug responses and predisposition to multi-factorial diseases. These new areas of pharmacogenetics and -genomics are likely to further expand the application of molecular genetic testing from rare, monogenic disorders to mainstream medicine and large at-risk populations.

Genetic testing stands out from other laboratory parameters as it covers – apart from cancer genetics – constitutional alterations. Therefore, a specific genetic test is usually carried out only once in a lifetime. Furthermore, a genetic variant or mutation not only concerns the patient, but as a heritable character may have important implications for their relatives. A third notable feature is the rarity and complexity of many genetic disorders, which therefore require a comprehensive written interpretation of the test result, in order to be useful to the patient and their doctor. This demands genetic tests to be of the highest possible accuracy and reliability and places an emphasis on the role of the genetics laboratory in adding value to the genotype analysis by the interpretation and communication of the result through the means of a clinical report.

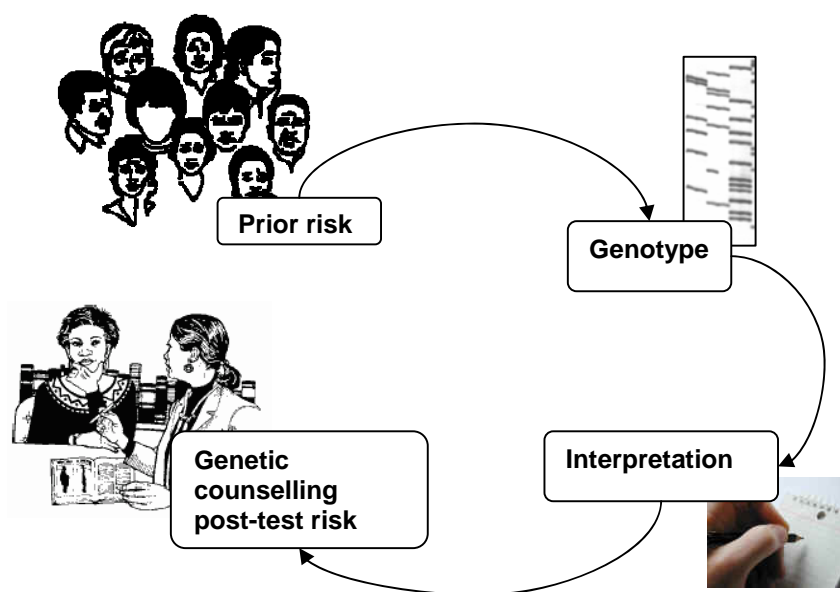


Figure 1. Diagram showing the cycle of clinical molecular genetic testing, a patient is referred to the laboratory with a prior genetic risk of an inherited disorder, after testing and interpretation of the results, the final output of the laboratory is a modified genetic risk to inform the counselling process.

There is a need to raise quality assurance in MGT to levels recognised in other areas of laboratory medicine. One of the key components of quality management and a cycle of quality improvement in the molecular genetic laboratory is External Quality Assessment (EQA). In this context, we define EQA, also called proficiency testing, as a procedure by which a well characterised sample (usually genomic DNA) is sent out to a group of laboratories in conjunction with a clinical request to analyse the sample for a specific genetic mutation/variation. In addition to the analytical performance

(genotyping), a written clinical genetic interpretation may be requested and form part of the assessment. Results from the laboratories are evaluated by a group of experts and a final report is returned to the laboratories with individual comments on their performance.

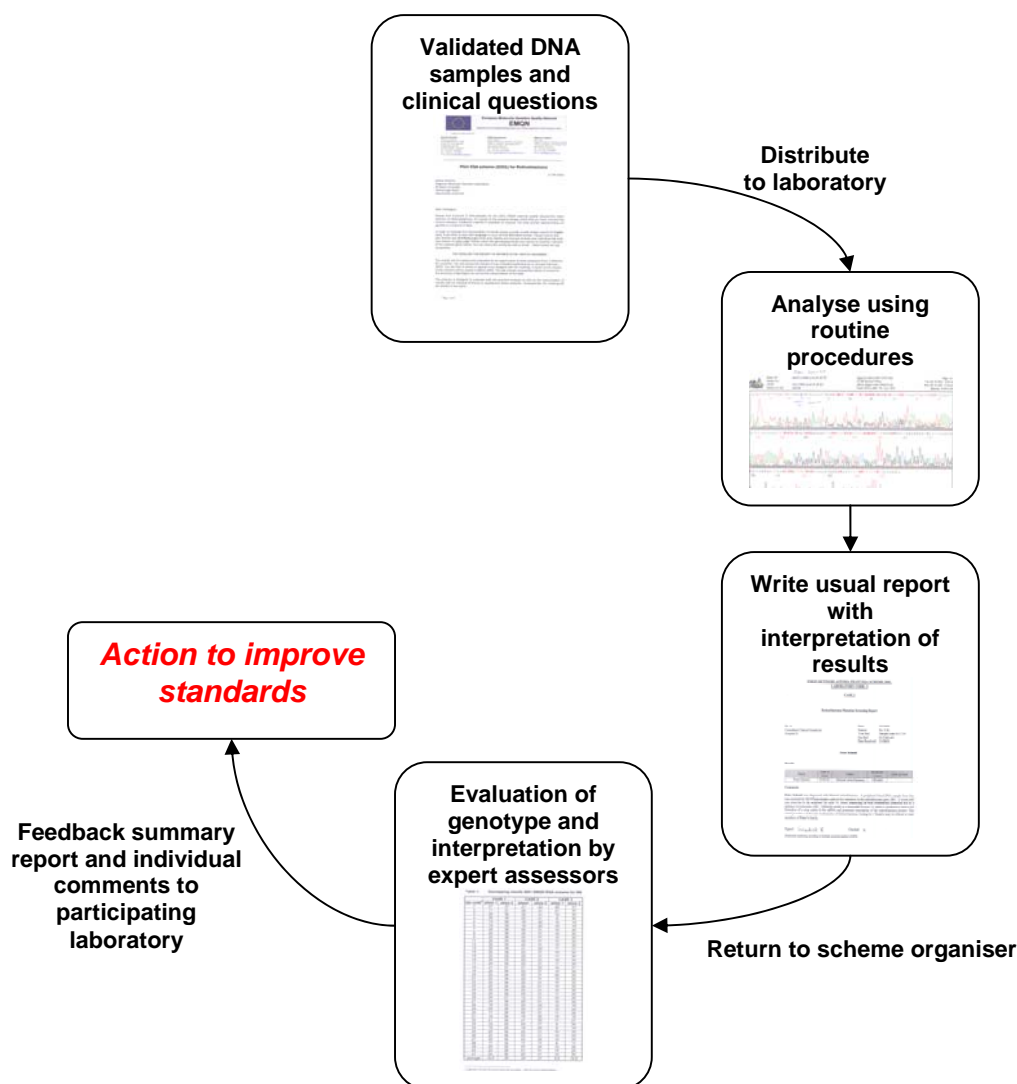


Figure 2. Diagram showing the external quality assessment scheme cycle.

Programmes for EQA using molecular genetic analyses have been implemented in a number of countries from the early 1990's onwards. Typically, these EQA schemes focus on a single disorder or gene and cover one of the major analytical techniques. Coded analytes (usually genomic DNA samples from patients' cell lines) are distributed and the questions include the technical analysis (the genotype) and a written interpretation.

Scope of this meeting

It was already clear that EQA schemes are not readily available in all EU countries for all services relevant to disorders, despite the need for external comparisons of laboratory performance.

Points of discussion at the meeting included:

- The current situation of EQA across Europe
- The barriers to EQA participation

- The demand for EQA schemes in different countries
- Ways to permit easier access to EQA
- The future of EQA in molecular genetic testing in view of the impact of new technologies

EuroGentest's aim is to review the present situation and explore the scope for future development of EQA for molecular genetic testing in Europe.

Representatives from 26 European countries were invited to attend this meeting. The invitee list was initially derived from EMQN National Partners and their collaborators. This group therefore had a prior interest in and knowledge of EQA and were able to act as representatives of their country on such issues.

Prior to this meeting a short questionnaire (Appendix I) was sent to each of the representatives, in order to assess the situation of EQA in each country and provide a framework to guide the discussions during the meeting. Twenty-four participants from 20 different European countries were present at the meeting. In addition, Turkey and The Netherlands, provided written feedback in the form of the questionnaire. The countries shown in green in figure 3 contributed to the discussion on the situation of EQA by returning a questionnaire and/or sending a representative to the meeting (six countries from the EU 25 were not represented these were: Latvia, Lithuania, Cyprus, Malta, Greece and Luxembourg).

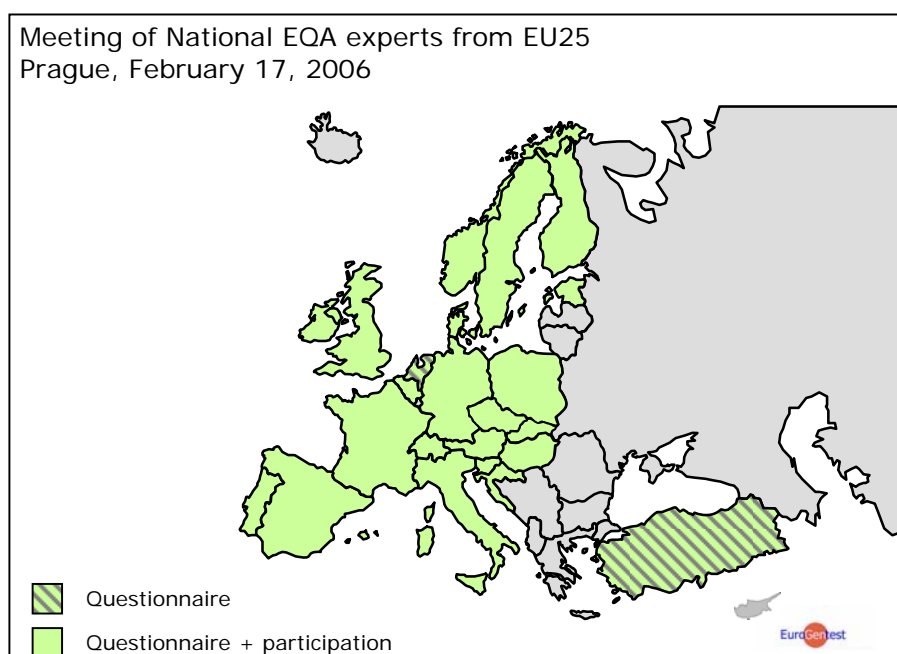


Figure 3. Map showing representation of countries at EQA meeting in Prague.

An extended discussion based on the responses to the questionnaire was held. This was found to be a successful way to allow everyone to contribute their own perspectives. This document summarises the situation of EQA in each of the individual countries that contributed, in the form of a structured summary of the main topics for each country. In addition this document provides a record of the main points of the discussion held in Prague.

The situation of Quality Assurance for Molecular Genetic Testing in Europe

Austria



Legal and regulatory background

In Austria molecular genetic testing is regulated by national legislation. The legislation is specific for medical genetics, including MGT. National guidelines and recommendations for MGT are issued from the government. Additionally the guidelines from EMQN and the Swiss Society of Medical Genetics are recommended.

Licensing, certification, accreditation

Licensing is mandatory for laboratories performing predictive/presymptomatic testing. The laboratory director has to make an application for a permit to the Federal Ministry of Health and Women. The laboratory, but not the laboratory director is licensed.

Certification and accreditation are not mandatory, but since December 2005 inspections are carried out by the government based on the checklists that are used by accreditation agencies.

Availability of EQA

Participation in EQA is required by national legislation/guidelines.

There is one national EQA provider for MGT in Austria. The Austrian government has compiled a list of relevant EQA providers. Laboratories also participate in EQA schemes provided from other countries.

ÖQUASTA

Österreichische Gesellschaft für Qualitätssicherung und Standardisierung medizinisch-diagnostischer Untersuchungen (ÖQUASTA)

Hörlgasse 18

A-1090 Wien

Austria

Tel: +43 (1) 319 8895

Fax: +43 (1) 319 8897

Web address: www.oequasta.at/

Belgium



Legal and regulatory background

In Belgium molecular genetic testing is regulated by national legislation. The legislation is specific for medical genetics, including MGT.

Guidelines and recommendations for MGT from EMQN, ACMG (American College of Medical Genetics) are used and the Swiss Society of Medical Genetics guidelines are recommended.

Licensing, certification, accreditation

Licensing is not mandatory, but is necessary for test reimbursement.

There is no specific accreditation requirement for molecular genetic diagnostic laboratories. But in order to obtain a government licence the laboratory must be accredited. Furthermore reference laboratories, which perform HIV testing and forensic DNA testing laboratories must be accredited by Beltest according to ISO 17025.

Availability of EQA

Participation in EQA is not required by any national legislation or guidelines, although it is a requirement of accreditation to ISO standards.

There is no national EQA provider in Belgium.

Laboratories participate in EQA schemes provided from other countries.

Croatia



Legal and regulatory background

In Croatia molecular genetic testing is regulated by national legislation. The legislation covers laboratory medicine in general, however there are no national guidelines or recommendations for MGT. Guidelines from EMQN are recommended.

Licensing, certification, accreditation

A licence is required to operate a MGT laboratory. Certification and accreditation are not mandatory.

Availability of EQA

Participation in EQA is not required by national legislation/guidelines. There is no national EQA provider for MGT in Croatia.

Czech Republic



Legal and regulatory background

In the Czech Republic molecular genetic testing is not regulated by national legislation. Guidelines and recommendations for MGT are issued from the Czech National Society of Medical Genetics. Additionally MGT guidelines from EMQN, UK CMGS and ACMG are recommended.

Licensing, certification, accreditation

There is no licence required to operate a MGT laboratory. Certification and accreditation are not mandatory for MGT laboratories.

Availability of EQA

Participation in EQA is not required by any national legislation or guidelines. There are plans to establish a national EQA system which will be organised by the National Society of Medical genetics. Laboratories participate in EQA schemes provided by national reference laboratories and/or by other countries (eg. CF-Network, EMQN).

Denmark



Legal and regulatory background

In Denmark molecular genetic testing is not regulated by national legislation. There are national guidelines and recommendations for MGT issued by the national society of human genetics. Additionally the guidelines from EMQN, UK CMGS and ACMG are recommended.

Licensing, certification, accreditation

A licence is not required to operate a MGT laboratory. Certification and accreditation are not mandatory.

Availability of EQA

Participation in EQA is not requested by national legislation/guidelines. There is one national EQA provider for MGT in Denmark. Laboratories also participate in EQA schemes provided from other countries.

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Denmark

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Fax: +45-4453 5369

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Estonia



Legal and regulatory background

In Estonia molecular genetic testing is not regulated by national legislation. There are no national guidelines or recommendations for MGT. Guidelines from EMQN are recommended.

Licensing, certification, accreditation

In Estonia a licence is required to operate a MGT laboratory. Certification and accreditation are not mandatory.

Availability of EQA

Participation in EQA is required by national legislation or guidelines. There is no national provider of EQA schemes for general laboratory medicine. Laboratories participate in EQA schemes provided from other countries.

Finland



Legal and regulatory background

In Finland molecular genetic testing is not regulated by national legislation. There are some informal national guidelines and recommendations for MGT issued from the national society of human genetics. Additionally the guidelines from EMQN, UK CMGS and ACMG are recommended in Finland.

Licensing, certification, accreditation

In Finland molecular genetic laboratories do not need a specific licence, but all private, clinical, diagnostic laboratories need a work permit, which is easily obtainable. Certification and accreditation are not mandatory.

Availability of EQA

Participation in EQA is not requested by any national legislation or guidelines. There is one national provider of EQA schemes for MGT in Finland. Laboratories also participate in EQA schemes provided from other countries.

LABQUALITY

Ratamestarinkatu 11

FIN-00520 Helsinki

Finland

Tel.: +358 9 2293320

Fax: +358 9 22933210

Web address: <http://www.labquality.fi>

France



Legal and regulatory background

In France molecular genetic testing is regulated by national legislation, which is specific for medical genetics, including MGT. There are no national guidelines for MGT, but it is recommended that laboratories follow the guidelines published by EMQN, UK CMGS, ACMG and the Swiss Society of Medical Genetics.

Licensing, certification, accreditation

In France two categories of tests are regulated by the Ministry of Health: prenatal tests and presymptomatic or predictive genetic tests. A licence is required to perform predictive tests. For prenatal tests, only authorised laboratories can deliver services (legislation 1994). The authorization to perform tests is restricted to a specific practitioner at a specified location. If an authorized practitioner moves to another institution, they have to re-apply. Conversely, if an authorized laboratory hires a new practitioner, it is also required to re-apply.

Certification and accreditation are not mandatory. However, all private laboratories performing clinical tests have to receive authorisation (legislation 1975) and are required to follow the procedures laid down in Guide de Bonne Execution des Analyses de Biologie Medicale. Public laboratories do not fall within this system, but must be located in a hospital. Public, university/research laboratories are not permitted to report to patients. However, if they are the only service provider for a genetic condition, they may have an unofficial agreement with an authorised laboratory to approve the results.

Availability of EQA

Participation in EQA is required by national legislation or guidelines.

There is currently one national provider of EQA schemes for MGT in France, there are plans from the government to establish another national EQA system. Laboratories also participate in EQA schemes provided from other countries.

French Health Products Safety Agency (afssaps)

143/147, bld Anatole France

93285 Saint-Denis Cedex

France

Web address: <http://afssaps.sante.fr>

Germany**Legal and regulatory background**

In Germany molecular genetic testing is not regulated by national legislation to date, but a gene diagnostic law is currently under discussion.

National guidelines and recommendations for MGT are issued from the German society of human genetics (GfH) and the Federal chamber of physicians (BÄK), who are authorised to define quality assurance measures in healthcare according to the Code of Social Law. Additionally the guidelines from EMQN are recommended.

Licensing, certification, accreditation

A general licence is required to operate a laboratory providing medical tests, but there is no specific licence for genetic testing. Certification and accreditation are not mandatory, but according to the national recommendations laboratories should have a quality system in place.

Availability of EQA

National guidelines from the Federal chamber of physicians (BÄK) require participation, once a quarter, in EQA for quantitative tests, but there are no adequate regulations for qualitative tests. If a laboratory is accredited, the German accreditation bodies demand regular participation in EQA. If no EQA scheme is available, the laboratory is asked to implement other external quality assessment measures, for example inter-laboratory comparison.

In Germany there are three national providers of EQA schemes for genetic testing. Currently, there are no plans to establish another EQA system in Germany.

Berufsverband Deutscher Humangenetiker e.V. (BVDH)

Inselkammerstr. 4

82008 München-Unterhaching, Germany.

Tel.: +49- 89 /55 02 78-55

Fax: +49- 89 /55 02 78-56

Web address: <http://www.bvdh.de/> or <http://www.hgqn.org/> (EQA scheme database)

INSTAND (Institute for Standardization and Documentation in the Medical Laboratory)

Ubier Str. 20

40223 Düsseldorf, Germany

Tel: +49 211-33 82 621 / -251

Fax: +49 221-33 82 603

Web address: <http://www.instand-ev.de/>

DGKL (German Society for Clinical Chemistry and Laboratory Medicine), RfB (Reference Institute for Bioanalytics)

Im Mühlenbach 52a

53127 Bonn, Germany

Tel: +49-228-21 50 25

Fax: +49-228-21 15 29

Web address: <http://www.dgkl-rfb.de/>

Hungary



Legal and regulatory background

In Hungary molecular genetic testing is regulated by national legislation, which covers laboratory medicine in general. There are national guidelines or recommendations for MGT issued by the government and the National Board of Clinical Genetics. External guidelines from EMQN are also recommended.

Licensing, certification, accreditation

In Hungary no licence is required to operate a MGT laboratory. Certification and accreditation are not mandatory.

Availability of EQA

Participation in EQA is requested by national legislation or guidelines.

There is one national provider of EQA schemes for general laboratory medicine and there are plans to establish a national EQA system through the National Board of Clinical Genetics. Laboratories also participate in EQA schemes provided from other countries.

QualiCont Ltd.

6720 Szeged, Somogyi
Hungary

Web address: www.tiszanet.hu/qualicont/

Ireland



Legal and regulatory background

In Ireland molecular genetic testing is regulated by national legislation (January 2006). There are no national guidelines or recommendations for MGT, but it is recommended that laboratories follow guidelines from EMQN, UK CMGS, ACMG and the Swiss Society of Medical Genetics.

Licensing, certification, accreditation

No specific licence is required for MGT laboratories. Certification and accreditation are not mandatory.

Availability of EQA

Participation in EQA is not requested by national legislation or guidelines.

There is no national provider of EQA schemes for MGT. Laboratories participate in EQA schemes provided from other countries. Ireland is formally represented on the Steering group of the UKNEQAS for molecular genetics scheme.

Italy



Legal and regulatory background

In Italy molecular genetic testing is regulated by national legislation, which covers laboratory medicine in general. National guidelines and recommendations for MGT are issued from the government. Additionally the guidelines from EMQN and UK CMGS are recommended.

Licensing, certification, accreditation

All clinical laboratories are required to have general approval from a regional authority. However, there is no specific requirement for genetic laboratories. Certification and accreditation are not mandatory for MGT laboratories.

Availability of EQA

Participation in EQA is requested by the national legislation/guidelines.

In Italy the Istituto Superiore di Sanità (ISS) is responsible for providing EQA for MGT. There are plans to expand the schemes organised by the ISS. Laboratories also participate in EQA schemes provided from other countries.

Istituto Superiore di Sanità (ISS)

Viale Regina Elena 299
00161 Roma
Italy

Tel. : +39 06 49901

Fax: +39 06 49387118

Web address: <http://www.iss.it>

The Netherlands



Legal and regulatory background

In the Netherlands molecular genetic testing is regulated by national legislation. There are no national guidelines or recommendations for MGT. However, guidelines from EMQN and UK CMGS are recommended.

Licensing, certification, accreditation

Only licensed laboratories are allowed to perform MGT. Licenses are only given to laboratories affiliated to Universities with a medical faculty. Other laboratories are allowed to perform MGT under the license of such a University laboratory (network structure). There is a registration for Clinical Molecular Geneticists.

Certification and accreditation are not mandatory yet, but the Dutch Society for Human Genetics and the professional organisation for clinical genetics laboratories are working on this.

Availability of EQA

Participation in EQA is not requested by national legislation/guidelines.

There is no national EQA provider for MGT in the Netherlands. Laboratories participate in EQA schemes provided from other countries. The Netherlands is formally represented on the Steering group of the UKNEQAS for molecular genetics scheme.

Norway



Legal and regulatory background

In Norway molecular genetic testing is regulated by national legislation. The legislation is specific for medical genetics, including MGT.

There are no national guidelines and recommendations for MGT, but the guidelines from EMQN are recommended.

Licensing, certification, accreditation

A licence is needed to become director of a genetic laboratory.

Certification and accreditation are not mandatory for MGT laboratories. However, all clinical laboratories have to receive formal approval by the competent authorities.

Availability of EQA

Participation in EQA is not required by national legislation/guidelines.

There is no national provider of EQA schemes in Norway.

Poland



Legal and regulatory background

In Poland molecular genetic testing is not regulated by national legislation. There are national guidelines/recommendations for MGT issued by the national society of human genetics. External guidelines from EMQN are recommended.

Licensing, certification, accreditation

In Poland no licence is required to operate a MGT laboratory.

Certification and accreditation are not mandatory.

Availability of EQA

Participation in EQA is not requested by national legislation or guidelines.

There is no national provider of EQA schemes for MGT in Poland. Laboratories participate in EQA schemes provided from other countries.

Portugal



Legal and regulatory background

In Portugal molecular genetic testing is regulated by national legislation. The legislation is specific for medical genetics, including MGT. National guidelines and recommendations for MGT are issued from the government. Additionally the guidelines from EMQN and UK CMGS are recommended in Portugal.

Licensing, certification, accreditation

In Portugal only private, molecular genetic laboratories need a specific licence to operate. Certification and accreditation are not mandatory.

Availability of EQA

Participation in EQA, when a suitable scheme is available, is requested by national legislation or guidelines. There is no national provider of EQA schemes for MGT in Portugal. Laboratories participate in EQA schemes provided from other countries.

Slovak Republic



Legal and regulatory background

In the Slovak Republic molecular genetic testing is not regulated by national legislation. There are no national guidelines or recommendations for MGT.

Licensing, certification, accreditation

In the Slovak Republic a licence is required to operate a MGT laboratory. Certification and accreditation are not mandatory.

Availability of EQA

Participation in EQA is not requested by national legislation or guideline. There is one national provider of EQA schemes for MGT in the Slovak Republic.

Genexpress – Institute of Molecular Physiology and Genetics

Ludovít Kadasi
Mlynsarovicova 22
85103 Bratislava
Slovak Republic
Tel: +421-2 602 96 444
Fax: +421-2 623 14 083

Slovenia



Legal and regulatory background

In Slovenia molecular genetic testing is regulated by national legislation. The legislation covers laboratory medicine in general. There are no national guidelines or recommendations for MGT.

Licensing, certification, accreditation

In Slovenia a licence is required to operate a MGT laboratory. Certification and accreditation are not mandatory.

Availability of EQA

Participation in EQA is not requested by national legislation or guidelines. There is no national provider of EQA schemes for general laboratory medicine in Slovenia, but the society of human genetics plans to establish a national EQA system.

Spain



Legal and regulatory background

In Spain molecular genetic testing is not regulated by national legislation. There are no national guidelines and recommendations for MGT, but the guidelines from EMQN are recommended.

Licensing, certification, accreditation

Clinical laboratories require a licence issued by the regional health authorities. However, there is no specific requirement for genetic laboratories. Certification and accreditation are not mandatory for MGT laboratories.

Availability of EQA

Participation in EQA is not requested by the national legislation/guidelines. There is no national EQA provider for MGT in Spain. Laboratories participate in EQA schemes provided from other countries.

Sweden



Legal and regulatory background

In Sweden molecular genetic testing is not regulated by national legislation. There are no national guidelines and recommendations for MGT.

Licensing, certification, accreditation

In Sweden molecular genetic laboratories do not need a specific licence. Certification and accreditation are not mandatory.

Availability of EQA

Participation in EQA is not requested by national legislation or guidelines. There is one national provider of EQA schemes for MGT in Sweden. Laboratories also participate in EQA schemes provided from other countries.

External quality assurance in laboratory medicine in Sweden (Equalis)

Box 977
751 09 Uppsala
Sweden
Tel.: +46-18 - 69 31 45
Fax: +46-18 - 69 31 46
Web address: <http://www.equalis.se/>

Switzerland



Legal and regulatory background

In Switzerland molecular genetic testing is regulated by national legislation. A new law specific for medical genetics, including MGT is expected to be in effect from January 2007.

There are no national guidelines and recommendations for MGT, but guidelines from the Swiss society of medical genetics (SGMG), EMQN and GfH are recommended.

Licensing, certification, accreditation

In Switzerland clinical laboratories require a licence issued by the competent federal authorities. For MGT Switzerland is in the process of converting to a new regulatory system. One of the conditions that must be met in order to receive a licence are not yet defined. Certification and accreditation are not mandatory, but in the near future laboratories may have to be accredited to ISO 15189:2003.

Availability of EQA

Participation in EQA is requested by national legislation or guidelines.

There are two national EQA providers with a limited number of schemes for MGT in Switzerland. Most laboratories performing MGT participate in EQA schemes provided from other countries.

MQ Verein für medizinische Qualitätskontrolle

Institut für Klinische Chemie

Universitätsspital

8091 Zürich

Switzerland

Tel.: +41-44 255 34 11

Fax: +41-44 261 12 83

Web address: <http://www.mqnet.ch>

CSCQ Schweizerisches Zentrum für Qualitätskontrolle

2, chemin du Petit-Bel-Air

1225 Chêne-Bourg

Switzerland

Tel: +41-22 305 52 36

Fax: +41-22 305 52 38

Web address: <http://www.cscq.ch>

Turkey



Legal and regulatory background

In Turkey molecular genetic testing is regulated by national legislation, which regulates medical practice. There are national guidelines and recommendations for MGT issued from the UNESCO National Committee for Bioethics. Additionally guidelines from EMQN and ACMG are recommended.

Licensing, certification, accreditation

In Turkey MGT laboratories require a licence issued by the Ministry of Health. Certification and accreditation are not mandatory.

Availability of EQA

Participation in EQA is not requested by national legislation or guidelines. The national quality assurance programme for MGT is organised jointly by the Department of Medical Biology of Hacettepe University and the Society for Medical Genetics.

Hacettepe University Faculty of Medicine

Dept. Medical Biology & TUBITAK DNA/Cell Bank

Professor Meral Ozguc

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TURKEY

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United Kingdom



Legal and regulatory background

In the UK molecular genetic testing is regulated by national legislation relating to general medical practice. An example is the Human Tissue Act (2004).

National guidelines are available through professional bodies for example from the Clinical Molecular Genetics Society (CMGS), the British Society for Human Genetics (BSHG) and others.

Licensing, certification, accreditation

There is no formal licensing system for molecular genetic testing laboratories in the UK. However, laboratories may register with the officially supported UK Genetic Testing Network (www.ukgtn.org) if they meet certain criteria, including accreditation. Accreditation in the UK is overseen by the UK Accreditation Service (UKAS) and Clinical Pathology UK Ltd. (CPA).

While accreditation is not mandatory it is very strongly encouraged. Through its policy document on Genetics in Health 'Our inheritance; Our future' (2003). the UK government set a date at the end of 2005 by which time it expected all

genetics laboratories to have obtained accreditation. In addition, wherever possible, laboratories in the UK must ensure that when they export samples the receiving laboratory is also accredited.

Availability of EQA

The UK accreditation bodies demand participation in EQA, where schemes are available. UKNEQAS for molecular genetics provides a rolling programme of 13 EQA schemes. Over 300 genetic tests are formally recognised by the UKGTN. Many UK laboratories also participate in EMQN schemes if the scheme for a certain disease or technique is not provided by UKNEQAS.

UKNEQAS for Molecular Genetics

Institute of Human Genetics

International Centre for Life

Newcastle upon Tyne

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Discussion

Legal and Regulatory background of molecular genetic testing.

Approximately half of the countries surveyed reported that national legislation affects molecular genetic testing in their countries, however, legislation is very variable and may be specific to medical genetics or more general to the practice of medicine. New legislation concerning molecular genetic testing was in preparation in several countries. For instance in Ireland there has been a recent change to the law on genetic testing in force from 1st January 2006 and in Switzerland there are proposals for changes to the law. A new law is also in preparation in Slovakia, it is thought that this will cover genetic testing and biobanking, whereas, in Estonia there are few regulations in the genetic testing field, except for where IVF or pre-natal diagnosis are concerned. Legislation is present in Portugal, but it is vague and relates more to private laboratories than state run laboratories, although state laboratories should also follow the same rules. A law governing genetic testing was prepared two years ago in Hungary, but it has not yet been accepted by parliament. In Italy there is an umbrella law for clinical chemistry, however, it is not specific to genetic testing. The overall conclusion was therefore that some countries have very specific regulations, while others have much broader regulations relevant to molecular genetic testing.

In about half of the countries no national guidelines for molecular genetic testing are in place. Of those countries where guidelines are in place, approximately half were professional guidelines from the national human genetics society, and half were regulations from the government. The Organisation for Economic Co-operation and Development (OECD) has produced framework guidelines for MGT from their 2003 survey, these guidelines are directed towards governments and professional bodies. Public consultation of these guidelines is expected in summer 2006, before they are finalised in autumn 2006.

In the majority of countries existing external guidelines were recommended. The guidelines most mentioned in the questionnaire responses were those issued by EMQN (European Molecular Genetics Network), CMGS (UK Clinical Molecular Genetics Society) and ACMG (American College of Medical Genetics) and the SSMG (Swiss Society for Medical Genetics). Although external guidelines are important it was felt that in some countries they are not likely to be adopted if not endorsed by national societies/authorities.

Licensing, certification, accreditation of molecular genetic testing laboratories.

The understanding of the terms licensing, certification and accreditation can vary. However, the formal definition of these terms specified by DIN EN 45020 (Standardisation and related activities – General vocabulary (ISO/IEC guide 2:1996)'

- *Licence*: **Permission**, permit from a governmental agency to operate a laboratory.
- *Certification*: Procedure by which a third party gives written assurance that a product, process or service **conforms** to specific requirements.
- *Accreditation*: A procedure by which an authoritative body gives formal recognition that a body or person is **competent** to carry out specific tasks.

Licensing is required in about half of the countries surveyed. In some countries licensing refers to a list of laboratories that is held by the government. In other countries, for example France and Austria, in order to obtain a licence to operate governments require laboratories to adhere to certification or accreditation procedures and therefore more stringent controls are maintained over licensing. In some countries, for example Estonia, Slovakia and Sweden, licensing is not specific to molecular genetic testing, but is a requirement of all medical laboratories. In Italy, a licence is required to open a molecular genetic testing lab, in 2004 the Italian Society of Human Genetics (ISHG) reported that 41% of laboratories were certified, however there is no formal accreditation for genetics laboratories in Italy. In Portugal, an operating licence

is only required by private laboratories. In Austria, a strict law was introduced in December 2005, although this law does not require laboratories to have formal assessment by an accreditation body, laboratories are subject to the same requirements as for accreditation, which will be checked by government inspectors. In Estonia, all laboratories in the medical field require a government licence, which is not merely a register of laboratories. In Slovakia, a licence is issued with requirements similar to certification, and is checked by a specialist, who is appointed by the regional government. A new law is expected to be passed in Switzerland in January 2007, which will specify that molecular genetic testing laboratories must be licensed, although conditions of licensing are not yet defined. While there is variation in the definitions, formal certification or accreditation is not mandatory in any EU country. However, governments may be applying significant pressure for medical and specifically genetics laboratories to achieve accreditation, for instance, in the UK and France. Specifically the UK government set a date at the end of 2005 when it expected all laboratories performing genetic testing to be accredited. In addition laboratories in the UK must ensure that wherever possible when they export samples to other UK laboratories, that the receiving lab is also accredited. In France a whole hospital is subject to accreditation, rather than individual laboratories.

Availability of EQA

EQA providers

In a minority of countries participation in EQA is specified in national legislation or guidelines. However, in most countries EQA participation was strongly encouraged, but not mandatory. In many countries there is pressure to participate in EQA, this may be as a result of general or specific pressure due to licensing or accreditation systems. ISO standards 17025 and 15189 state that laboratories must check the quality of their results via internal and external control-mechanisms. ISO 15189 specifies that the external control mechanisms can be the participation in EQA schemes or in inter-laboratory comparisons. In Germany there is strict pressure by the Federal Chamber of Physicians to participate in EQA for certain tests in the clinical chemistry field, but this does not as yet apply to molecular genetic testing.

More than half of the countries have national EQA schemes related to genetic testing. EuroGentest organised a meeting in Frankfurt in November 2005 that brought together many of these EQA scheme providers for the first time. A summary report arising from this meeting can be found on the EuroGentest unit 1 website (www.eurogentest.org).

In Switzerland there are two quality control centres, however, the Swiss Human Medical Society recommends participation in foreign EQA schemes that include assessment of reporting as well as genotyping. In Italy, the Istituto Superiore di Sanità (ISS) started a national EQA scheme, which is financially supported by the Italian Ministry of Health. The ISS scheme is open to private laboratories, but only 30-40 from more than two hundred laboratories known to offer genetic testing participate. It is estimated that approximately two thirds of Italian laboratories are not involved in IEQA. Some national/regional schemes limit participation, e.g. UKNEQAS for molecular genetics limits participation to laboratories from the UK, Ireland and the Netherlands, partly due to historical reasons. The Nordic countries collaborate with UKNEQAS for Cytogenetics and Factor V Leiden schemes. Of the three German EQA providers, only one (BVDH) deals exclusively with EQA for genetic tests (molecular and cytogenetics) and requests written interpretation of the genotype (in German). The German schemes are also used by laboratories from Austria and Switzerland and to a lesser extent other neighbouring countries. The issue of the language that the reports are written in is important if clinical interpretation forms part of the assessment, because this adds to the difficulty and work load for laboratories if reports produced specifically for EQA schemes need to be written in a language other than the native language.

Table 1. EQA providers in Europe and scope of participation.

EQA scheme	Country where scheme is based	Scope of participation
EMQN http://www.emqn.org/emqn/	UK	International
UKNEQAS for molecular genetics http://www.uknegas-molgen.org.uk	UK	UK, Ireland, The Netherlands
UKNEQAS for blood coagulation, thrombophilia http://www.uknegas.org.uk/	UK	International
UKNEQAS for blood coagulation, haemophilia genetics http://www.uknegas.org.uk/	UK	UK
European concerted action on thrombosis (ECAT) http://www.ecat.nl	The Netherlands	European
Dutch Foundation for Quality Assessment in Clinical Laboratories (SKML) http://www.skml.nl/	The Netherlands	European
Danish Institute for External Quality Assurance for Laboratories in Health Care (DEKS) http://www.deks.dk/	Denmark	Denmark
External Quality Assurance in Laboratory Medicine in Sweden (EQUALIS) http://www.equalis.se/	Sweden	European
French Health Products Safety Agency (AFSSAPS) http://afssaps.sante.fr	France	French language
INSTAND (Institute for Standardization and Documentation in the Medical Laboratory) http://www.instand-ev.de/	Germany	German language
DGKL (German Society for Clinical Chemistry and Laboratory Medicine) and RfB (Reference Institute for Bioanalytics) http://www.dgkl-rfb.de/	Germany	German language
BVDH (Professional Association of German Human Geneticists) http://www.hgqn.org/	Germany	German language
ÖQUASTA www.oequasta.at/	Austria	German language
CSCQ (Swiss Centre for Quality Control) http://www.cscq.ch	Switzerland	International
MQ Verein für medizinische Qualitätskontrolle Institut für Klinische Chemie http://www.mqnet.ch	Switzerland	German, French, Italian language
Cystic Fibrosis Network http://www.cfnetwork.be/	Belgium	European
The Italian External Quality Assessment in Molecular Genetics (IEQA) http://www.iss.it	Italy	Italy
EQUAL: Multi-National External Quality Assay (EQA) programmes http://www.ec-4.org/equal/	Italy	European
QualiCont Kht. http://www.tiszanet.hu/qualicont/	Hungary	Hungarian language
Genexpress Institute of Molecular Physiology and Genetics, Bratislava	Slovak Republic	
LabQuality http://www.labquality.fi	Finland	International
EAA (European Academy of Andrology) http://www.uni-leipzig.de/%7Eeaa/index.html	Germany	International

The survey carried out for the meeting showed that EMQN is the most commonly used international EQA provider. Others providers that are used in countries where the scheme does not originate from include UKNEQAS schemes, German schemes and US College of American Pathologists (CAP), which may be linked to their accreditation procedures.

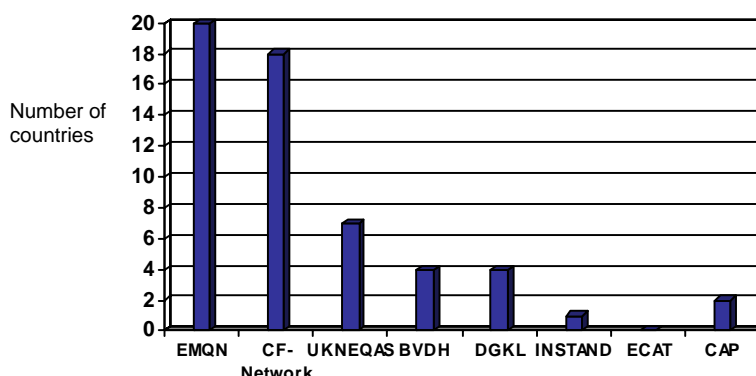


Figure 4. EQA providers that are used in countries other than the country where they are based.

There are plans to establish national EQA schemes in France, Italy, Czech Republic and others. In France there is central agency for all EQA for medical tests, but only four genetic schemes are offered. French geneticists have indicated that external EQA providers, such as EMQN could be recognised by the French Government. In Italy, the ISS scheme is likely to expand, encouraged by recent legal guidelines. In Denmark where five laboratories carry out molecular genetic testing, there are plans to establish an inter-laboratory comparison (ILC) involving mutual exchange of samples, rather than a national EQA scheme. In the Czech Republic there is only limited participation in EQA. They are working towards one centre acting as a reference centre, thus emulating the international system and acting as an organising centre to translate questions into the local language and distribute samples further.

Persistent poor performance in EQA

It was recognised that in the future there may be commercial or regulatory pressures to make performance in EQA public. It was difficult for EQA providers at the European level to address persistent poor performance except through offering advice and support as there is no European regulatory framework relevant to this area. However certificates issued by EQA agencies for satisfactory performance are a public way of distinguishing between laboratories that perform well or poorly. It was felt that performance was a national matter and should be discussed with professional bodies, regulators and governments. Accreditation is a system that can address poor performance in a non public way.

EQA can be used to monitor the performance of methods and commercial diagnostic products however it should be recognised that EQA materials are not certificated reference materials and there may be limits or challenges to the use of EQA data for product surveillance.

Cost of EQA participation

Representatives were surveyed about what they considered to be acceptable costs for EQA participation, the most frequent response was between 50 and 100 euros. This is well below the actual costs for administering and running an EQA scheme. EMQN is a non-profit making organisation, in 2005 the charge for participating in EMQN schemes was €200, per scheme, plus €50 yearly registration fee, in 2006 the charge for participating in EMQN schemes is €252, although the cost is reduced to €220 for laboratories registering early.

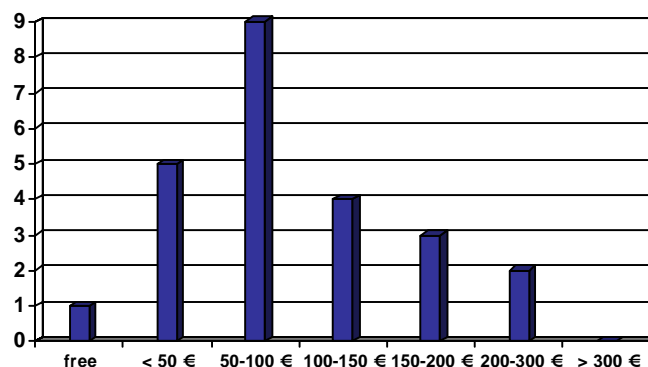


Figure 5. National representatives' opinion of the acceptable cost to participate in an EQA scheme.

As well as the costs of materials, shipping and administration, there are hidden costs of running EQA schemes: for example maintaining an internal quality management system, EQA volunteer scheme assessors and organisers provide their time free of charge, essentially their institution is supporting the scheme. If they were to charge for their time the actual cost of running a scheme would be much higher. Prof. E. Dequeker stated that she had estimated the true cost of a self-supporting EQA scheme with more than 100 participants to be ~€370 per participant. Charging participants actual costs may therefore be prohibitive for some developing countries. It was suggested that outsourcing production may help to reduce costs. Alternatively, larger schemes may subsidise smaller schemes, although it was pointed out that from a volunteer assessor's point of view significantly increasing participation may make the marking of reports a prohibitively time-consuming exercise.

Use of English language

The use of English as the reporting language can be a barrier to participation in EQA and may lead to laboratories participating in genotyping only schemes in preference. However, it was felt that loss of the reporting aspect of EQA would diminish the value of the schemes and de-value the role of the laboratory as an advisor to the referring doctor. If the most prevalent diseases were covered by EQA offered at a national level this would meet most of the need for an assessment of the quality of reporting. EQA reporting was felt to be the most important aspect of the EQA assessment process. If it is not possible to offer schemes for every language, the model suggested by Czech representatives of a national centre translating documentation and relaying materials might be a possibility. However, representatives from several countries expressed concerns that if a centre is dealing with a relatively small number of laboratories from their own country anonymity may be compromised, unless a national agency offering EQA on a large scale was involved, e.g. LabQuality, Finland.

Technique- versus disease specific EQA

It was suggested that technique specific schemes could be organised at the European level, and in English, because there is no need for interpretation of results into a clinical report. Technical EQA schemes may serve the needs of many laboratories without increasing the numbers or costs of EQA schemes excessively.

Technical EQA schemes could be supplemented by theoretical EQA to assess proficiency in reporting. It was thought that theoretical EQA would be acceptable to accreditation systems depending on how laboratories present EQA to the accreditation bodies, this is unlikely to be problem if laboratories participate in both disease specific and theoretical EQA. There was agreement that technique-specific schemes should complement rather than substitute for disease-specific EQA.

Recipients of the questionnaire prior to this meeting were asked to rank the following disorders/techniques in order of importance of the availability of EQA schemes, where one is the most important and eight the least. The overall ranking of the eight EQA topics is shown in table 2.

Table 2. Ranking of importance of EQA schemes: 1 most important, 8 least important.

Rank	Disorders/techniques
1	Cystic fibrosis
2	Fragile X Syndrome
3	Factor V Leiden
4	Haemochromatosis
5	Thalassaemias
6	DNA fragment length determination
7	DNA sequencing
8	Quantitative PCR

The national representatives were also asked to list two other disorders/techniques that were important for their country, those responses mentioned by more than one national representative were: Duchenne/Becker Muscular Dystrophy, Huntington's disease, Friedreich's Ataxia, Y-chromosome microdeletions, the MLPA technique, Spinocerebellar ataxias, Charcot-Marie-Tooth, Fascio Scapulo Humeral Dystrophy, and Familial breast cancer. When asked about how many EQA schemes per disease and per year the representatives regarded as necessary the overwhelming response was one scheme per disease per year. However, it was felt that the distribution of several schemes at the same time puts added pressure on laboratories, it was suggested that if schemes were distributed on the same date, that there could be different deadlines for each disease specific EQA return.

As a summary of the discussion, the following model of cooperation between national and international EQA providers was emerging:

1. EQA for common conditions (disease-specific, at national level and in national language wherever possible)
2. Technical EQA (covering the most common techniques, at European/ international level, in English)
3. Rare disease 'bundles' (covering rare diseases, at European/ international level, in English).

Best practice guidelines

At several points during the discussion, concern was raised about the availability, development and revision of best practice guidelines. Apart from the UK CMGS guidelines, there seems to be no mechanism in place to assure that

guidelines are regularly reviewed and updated. For some of the more frequently requested tests and clinically severe genetic disorders guidelines may not be available (an example is the Spinocerebellar Ataxias). Together with the availability of EQA, participants regarded competent guidelines as one of the most important factors for high quality laboratory output and as a pre-requisite for fair inter-laboratory comparisons through EQA.

Conclusions

This meeting was successful in bringing together a group of people from across Europe with an interest in External Quality Assessment (EQA), to discuss how advances in this field might most benefit molecular genetic testing laboratories. It is appreciated that only a single representative from each country was able to attend and that the views presented may not be wholly representative of the entire country. The intention was not to produce a detailed review of the situation in each country, rather to obtain an overview and encourage discussion between countries. If views from a larger number of representatives from each country were needed this would be best carried out at a national or regional level.

The meeting highlighted a great variation between different countries in the legislation pertaining to molecular genetic testing laboratories and in the regulations concerning EQA. While EQA participation was not mandatory in any country there was agreement that disease specific EQA was essential for the most clinically relevant diseases. It was agreed that in disease specific EQA schemes the assessment of written interpretative reports is an essential element of quality. Disease-specific EQA with written interpretation should best be organised at a national or language level. It was also felt that technical EQA should be a complement and not a substitute to disease specific EQA and could better be organised internationally. At the present time, EQA is absolutely dependent on volunteer experts to act as scheme organisers otherwise the cost of participation would become a major barrier for laboratories. A lack of and need for up-to-date best practice guidelines was expressed which should best be produced through European cooperation.

Glossary of terms

AFSSAPS	French Health Products Safety Agency
ACMG	American College of Medical Genetics
BÄK	Bundesärztekammer (Federal chamber of physicians in Germany)
BVDH	Berufsverband Deutscher Humangenetiker (Professional Association of German Human Geneticists)
CAP	College of American Pathologists
CF-Network	Cystic Fibrosis Network
CMGS	UK Clinical Molecular Genetics Society
CSCQ	Swiss Centre for Quality Control
CSLI	Clinical and Laboratory Standards Institute
DEKS	Danish Institute for External Quality Assurance for Laboratories in Health Care
DGKL	German Society for Clinical Chemistry and Laboratory Medicine
DIN	German Institute for standardisation
EAA	European Academy of Andrology
ECAT	European Concerted Action on Thrombosis
EMQN	European Molecular Genetics Quality Network
EQUAL	Multi-National External Quality Assay (EQA) Programmes in Clinical Molecular Diagnostics
EQAP	External Quality Assessment Programme
EQUALIS	External Quality Assurance in Laboratory Medicine in Sweden
EuroGentest	European Union Network of Excellence key words: genetic testing, quality, harmonisation.
External Quality Assessment (EQA)	Determination of laboratory testing performance by means of inter-laboratory comparisons.
GfH	German society of human genetics
IEQA	Italian External Quality Assessment in Molecular Genetics
ILC	Inter-laboratory comparison
INSTAND	Institute for Standardization and Documentation in the Medical Laboratory
ISHG	Italian Society of Human Genetics
ISO	International Organisation for Standardisation
ISO 15189	Standard relating to the particular requirements for quality and competence in medical laboratories
ISO 17025	General Requirements for the Competence of Testing and Calibration Laboratories
ISS	Istituto Superiore di sanità
MQ	Association for medical quality control (Verein für medizinische Qualitätskontrolle, Switzerland)
OECD	Organisation for Economic Co-operation and Development
ÖQUASTA	Austrian association for Quality Assurance and Standardization of medical-diagnostic tests
QM	Quality Management
QUALAB	Swiss Commission for Quality Control in the Medical Laboratory
SKML	Dutch Foundation for Quality Assessment in Clinical Laboratories
SSMG	Swiss Society for Medical Genetics
UKGTN	United Kingdom Genetic Testing Network
UK NEQAS	United Kingdom National External Quality Assessment Service

References

OECD (2005) Quality Assurance and Proficiency Testing for Molecular Genetic Testing: Summary results of a survey of 18 OECD member countries, Annex A

Appendix 1. Questionnaire distributed to country representatives prior to the meeting.

The situation of External Quality assessment schemes in Europe

With this questionnaire we want to learn about the quality procedures applied to molecular genetic testing (MGT) in the member states of the European Union. More specifically, we want to explore the availability of and the needs for 'external quality assessment' (EQA) in the EU countries.

Definition of EQA: In this context, we define EQA – also called 'proficiency testing' – as a procedure by which a well characterised sample (usually genomic DNA) is sent out to a group of laboratories with the question to analyse this sample for a specific genetic mutation/variation. In addition to the analytical performance (genotyping), a written clinical genetic interpretation may be requested. Results from the laboratories are evaluated by a group of experts and a final report is returned to the laboratories with individual comments on their performance.

How to fill in this questionnaire: You can move around within this Word document using the Tab key. Only the areas shaded in grey can be filled. Tick boxes (YES or NO) can be ticked by a left mouse click; free text may be added as required. Please save the document and return by e-mail to: uta.malburg@biozentrum.uni-wuerzburg.de.

Alternatively, you may print out the document and fill in by hand. Please return paper mail to:

Uta Malburg, Institut fuer Humangenetik, Biozentrum, Am Hubland, 97074 Wuerzburg, Germany.

Regulatory background	
Please indicate your country	
1. Is MGT* regulated by national legislation in your country?	YES <input type="checkbox"/> NO <input type="checkbox"/>
If Yes (tick the box):	
Is the legislation specific for medical genetics, including MGT?	<input type="checkbox"/>
Or does the legislation cover laboratory medicine in general?	<input type="checkbox"/>
Or does the legislation 'only' regulate medical practice?	<input type="checkbox"/>
2. Are there national guidelines/recommendations for MGT in your country?	YES <input type="checkbox"/> NO <input type="checkbox"/>
If Yes (tick the box):	
Who has issued the guidelines?	
The national society of human genetics.....	<input type="checkbox"/>
The government (e.g. department of health).....	<input type="checkbox"/>
Others (please specify).....	
3. Are guidelines for MGT from other countries ('external guidelines') recommended/followed in your country?	YES <input type="checkbox"/> NO <input type="checkbox"/>
If Yes (tick the box):	
Who has issued the guidelines?	
EMQN (The European Molecular Genetics Network).....	<input type="checkbox"/>
UK CMGS (UK Clinical Molecular Genetics Society).....	<input type="checkbox"/>
ACMG (American College of Medical Genetics).....	<input type="checkbox"/>
GfH (German Society of Human Genetics).....	<input type="checkbox"/>
Swiss Society of Medical Genetics.....	<input type="checkbox"/>
Italian guidelines.....	<input type="checkbox"/>
Others (please specify).....	

4. Does your national legislation/guideline request participation in EQA?	YES <input type="checkbox"/>	NO <input type="checkbox"/>
5. Is a licence (from the government or a governmental body) required to operate a MGT laboratory in your country?	YES <input type="checkbox"/>	NO <input type="checkbox"/>
6. Is certification (e.g. ISO 9000) for MGT labs mandatory in your country?	YES <input type="checkbox"/>	NO <input type="checkbox"/>
7. Is accreditation (e.g. ISO 17025/15189, CPA) for MGT labs mandatory?	YES <input type="checkbox"/>	NO <input type="checkbox"/>
Availability/access to EQA		
8. Is there a national provider of EQA schemes in your country?	YES <input type="checkbox"/>	NO <input type="checkbox"/>
If Yes: Please give the name(s) and address(es) of the provider(s).		
9. Do you know of laboratories in your country that participate in EQA of 'external/international' providers?	YES <input type="checkbox"/>	NO <input type="checkbox"/>
If Yes: Which EQA providers are being used?		
EMQN.....	<input type="checkbox"/>	
CF-Network.....	<input type="checkbox"/>	
UKNEQAS.....	<input type="checkbox"/>	
BVDH (Berufsverband Deutscher Humangenetiker).....	<input type="checkbox"/>	
DGKL (Deutsche Vereinte Gesellschaft für Klinische Chemie und Laboratoriumsmedizin)	<input type="checkbox"/>	
INSTAND.....	<input type="checkbox"/>	
ECAT.....	<input type="checkbox"/>	
CAP (College of American Pathologists).....	<input type="checkbox"/>	
Others (please specify).....		
10. Are there plans to establish a national EQA system in your country?	YES <input type="checkbox"/>	NO <input type="checkbox"/>
If Yes:		
Who is going to organise EQA in your country?	<input type="checkbox"/>	
Government or governmental body/agency?.....	<input type="checkbox"/>	
National society of human genetics?.....		
Independent body? (Please specify).....		

<p>11. If you had easy access to EQA, what would be an acceptable cost for participation?</p>	
<p>Less than 50 Euros.....</p>	<input type="checkbox"/>
<p>50 – 100 Euros.....</p>	<input type="checkbox"/>
<p>100 – 150 Euros.....</p>	<input type="checkbox"/>
<p>150 – 200 Euros.....</p>	<input type="checkbox"/>
<p>200 - 300 Euros.....</p>	<input type="checkbox"/>
<p>> 300 Euros.....</p>	<input type="checkbox"/>
<p>Can only participate if EQA is for free.....</p>	<input type="checkbox"/>
<p>12. Would the use of English language be a mayor hurdle for participation in international EQA schemes? YES <input type="checkbox"/> NO <input type="checkbox"/></p>	
<p>13. Please list and rank the 10 disorders/techniques for which you regard EQA schemes most desirable? (Please try to answer for the requirements of your country as a whole. Rank diseases by 1, 2, 3 etc.; with number 1 being the most desirable. You may delete given disorders and add on others.)</p> <p style="margin-left: 40px;"> Factor V Leiden..... Haemochromatosis..... Fragile X Syndrome..... Cystic fibrosis..... Thalassemias..... DNA sequencing..... DNA fragment length determination..... Quantitative PCR..... </p>	
<p>14. How many EQA schemes per disease and per year would you regard as necessary?</p> <p style="margin-left: 40px;"> Once every two years <input type="checkbox"/> Once a year <input type="checkbox"/> Twice a year <input type="checkbox"/> More frequently <input type="checkbox"/> </p>	
<p>15. Is availability and access to EQA in your country satisfactory? YES <input type="checkbox"/> NO <input type="checkbox"/></p> <p>If No: What is the single most important hurdle to EQA from your point-of-view? (Please specify)</p>	

Appendix 2. Results from the questionnaire presented by Clemens Mueller at the European External Quality Assessment National Representatives meeting.

