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## European Workshop on Genetic Testing Offer in Europe

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# European Workshop on Genetic Testing Offer in Europe

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## Introduction

Genetic testing has been facing an exponential growth in the last years. Further recent advances in molecular medicine are associated with even larger prospects and expected impacts on care management. When integrating these technologies into the healthcare system, it is important that their application is implemented within a responsible framework of accompanying measures and activities. As such, genetic testing has become also the subject of policy debates at different institutional and international levels.

In order to help decision-makers at all levels to rapidly introduce the necessary requirements, the European Commission has organized a number of initiatives involving different services that follow the subject from different perspectives.

Firstly, genetic testing currently falls under the scope of the Directive 98/79/EC of the European Parliament and of the Council on in vitro diagnostic medical devices. In the current system, patients, healthcare professionals and other interested parties do not have sufficient access to essential information on how medical devices have been assessed, and what clinical evidence there is to show they are safe and effective. After several procedures of consultation, on 26 September 2012, the European Commission adopted a package on innovation in health consisting of the (a) Communication on safe, effective and innovative medical devices and in vitro diagnostic medical devices for the benefit of patients, consumers and healthcare professionals, (b) Proposal for a Regulation of the European Parliament and of the Council on medical devices, and amending Directive 2001/83/EC, Regulation (EC) No 178/2002 and Regulation (EC) N 1223/2009 and (c) Proposal for a Regulation of the European Parliament and of the Council on in vitro diagnostic medical devices.

This new text proposes to clarify and extent the scope of the IVD Directive on in vitro diagnostics, medical devices and clarifies the regulation of genetic tests. Target for its adoption is 2014 and the new rules would then gradually come into effect from 2015 to 2019.

Secondly, on 11 November 2008 the Commission Communication on Rare Diseases was adopted, that sets out an overall Community strategy to support Member States in diagnosing, treating and caring for the 36 million EU citizens with rare diseases (1). This Communication focuses on three main areas: a) improving recognition and visibility of rare diseases, b) supporting policies on rare diseases in Member States for a coherent overall strategy, and c) developing cooperation, coordination and regulation for rare diseases at

EU level. As of rare diseases, there were several real and perceived obstacles to the transition from disease gene discovery to accessible, high-quality clinical laboratory testing, the major one being the perceived lack of financial incentives for individual laboratories to invest in the development and validation of a test with very low volume. In addition, genetic testing in research laboratories has significant issues related to access and quality of testing for this important group of disorders. Here the European cooperation aims to bring together the scarce resources for rare diseases fragmented across EU Member States and the European Commission works with the aim of promoting joint actions which may help patients and professionals to share expertise and information across borders. Quality management of diagnostic laboratories is one of the aspects discussed in the Communication. It recognizes that given the large number of tests and the need to design and validate a specific set of diagnostic assays for each, no single country can be self-sufficient in the provision of testing and in an efficient external quality assessment of the provided tests. Therefore, there is a need to enable and facilitate the exchange of expertise through clearly stated, transparent, EU agreed standards and procedures of guaranteed quality and utility.

Thirdly, the Council Recommendation on an action in the field of rare diseases was adopted on 8 June 2009 (2). The Recommendation engages the responsibility of Member States and concentrates on supporting and strengthening the adoption before the end of 2013 of national plans and strategies for responding to rare diseases, on improving recognition and visibility of rare diseases, on encouraging more research into rare diseases and forging links between centers of expertise and professionals in different countries through the creation of European reference networks in order to share knowledge and expertise and, where necessary, to identify where patients should go when such expertise cannot be made available to them. The role of patients' organizations is also highlighted as particularly important. Delegated and implementing acts will define the criteria to establish the methodology, including the process of selection and designation of the healthcare providers to be considered members of the European Reference Networks and several categories of criteria for the adequate management, monitoring and evaluation of the networks. One of the recommended areas to foster gathering the expertise on rare diseases at European level is the development of European guidelines on diagnostic tests or population screening, while respecting national decisions and competences.

Fourthly, the Directive 2011/24/EU on patients' rights in cross-border healthcare, clarifies rules regarding the right of patients to be treated outside their home country and reimbursed under certain conditions. Documents have specific provisions in regards to rare

diseases patients. The Directive will have no impact on the rights of each Member State to determine which health services they will provide.

Fifthly, the European Commission Decision C (2009)9181 of 30 November 2009 formally established a European Union Committee of Experts on Rare Diseases (EUCERD) (3). This new structure, evoked in Point 7 of the Communication from the Commission to the European Parliament, the Council, the European Economic and Social Committee and the Committee of the Regions on Rare Diseases: Europe's Challenges, adopted on 11 November 2008, recommends that the European Commission be assisted by a European Union Advisory Committee on Rare Diseases:

*"The preparation and implementation of Community activities in the field of rare diseases require close cooperation with the specialised bodies in Member States and with the interested parties. Therefore, a framework is required for the purpose of regular consultations with those bodies, with the managers of projects supported by the European Commission in the fields of research and public health action and with other relevant stakeholders acting in the field."*

*Thus, "the Committee acting in the public interest shall assist the Commission in formulating and implementing the Community's activities in the field of rare diseases, and shall foster exchanges of relevant experience, policies and practices between the Member States and the various parties involved".*

A number of documents and publications have been produced by the EUCERD and by other EC funded projects such as EUROPLAN ([www.europlanproject.eu](http://www.europlanproject.eu)), which are of relevance for shaping collaboration on genetic testing between Member States and for defining points for action at EU level. Among those are: "Recommendations on Quality Criteria for Centers of Expertise for Rare Diseases in Member States" (4), "EUCERD Recommendations for European Reference Networks for Rare Diseases" (5), "State of the Art of Rare Disease Activities in Europe" (6), "Recommendations for the Development of National Plans for Rare Diseases" (7).

In the light of these initiatives and the apparent need of expert input for further policy development European Commission's Joint Research Centre in Ispra (Italy) in collaboration with EUCERD and the EC funded project Eurogentest ([www.eurogentest.org](http://www.eurogentest.org)) convened a workshop on 19-20 November 2012 in Ispra. The objective of the workshop was to provide expertise-based knowledge and views with regard to the quality and organization of genetic testing in Europe in order to support points of actions and decision-making initiatives at the research, clinical and policy levels. Furthermore, it aimed to explore future visions on the use, value and integration of genomic medicine into clinical practice.



The meeting received input from experts from European genetic testing laboratories; representatives of the Joint Research Centre, DG Research & Innovation, and DG Sanco; clinical and laboratory geneticists; experts on quality management and external quality assessment; representatives of rare disease patients' associations; representatives of national and/or international professional societies in genetics; experts in Health Technology Assessment; and experts in ethical, legal and social aspects of genetics and genomics.

Considering the broadness of the field, the organizers decided to frame the scope of the workshop and provided the participants with six topics. Groups were formed and were each assigned a given topic to be discussed during interactive and bottom-up sessions. The goal of this set up was to stimulate expert perspectives in an open way. This approach provided opportunities for information to come forward through a process of discussion and interaction. This approach highlights commonalities in collective understanding and also the points at which perspectives differ. The outcomes of parallel sessions were then discussed at plenary meetings in the presence of all participants.

The following six topics were discussed during the meeting:

**(1) Regional distribution of genetic services**

Nations - especially the smaller ones - cannot easily or reasonably offer all clinical and laboratory services for all rare diseases. Expertise is rare as well, especially when it is highly specialized. Hence, cross-border activities and international exchanges are a requisite, underlying the importance of national and international organization of rare disease networks, the organization of cross-border testing and the interoperable and federated knowledge-bases.

**(2) Suggestions for the rationalization of services**

Clinical geneticists and genetic services are primarily dealing with rare diseases. It has to be decided which genetic tests are useful and which are not. The plethora of offers needs to be assessed from a Health Technology and Health Care Provision viewpoint. This includes an analysis of the current genetic testing offer and its anticipated evolution, the analysis of clinical utility and the assessment of the needs for prioritization.

**(3) How to improve (national) accreditation of genetic laboratories?**

Accreditation is the best vehicle to ensure, assess and control quality in diagnostic testing. Accreditation is currently a matter that falls under the competence of national Member

States. In view of this, initiatives to support and promote quality assurance at the European level would be welcome. In particular, the exchange of knowledge and experience between Member States would be valuable, surely in relation to the fact that to obtain and maintain quality standards is a costly process.

**(4) (Long term) Organization of External Quality Assessment (EQA)**

Annual participation in EQA is an essential feature of the quality assurance in a laboratory and an obligation according to ISO norms for accreditation. EQA providers and scheme organizers must be equally subject to quality standards.

**(5) Validation of Next Generation Sequencing (NGS) platforms and applications**

The emerging 'massive parallel sequencing' technologies have revolutionized genetic research, and are increasingly applied in diagnostics. Initiatives are needed to validate those applications and to issue guidelines for the clinical application of exome sequencing and whole genome sequencing. Issues such as informed consent procedures, incidental findings, targeted versus exome versus total genome analysis, and interlaboratory/collaborative evaluation has to be addressed.

**(6) Perspectives on direct-to-consumer genetic testing**

In recent years various companies have started to advertise, sell or provide genetic tests directly to consumers. These companies cover a very broad spectrum of tests, including carrier tests for recessive genetic disorders, "life style"-related genetic traits, pharmacogenomics, genomic risk profiles for many conditions, paternity tests, and other relationship testing such as ancestry and invasive and non invasive prenatal paternity or gender tests. While the majority of the large companies are operating from the United States, the number of companies operating in Europe is increasing. This leads to the question which governance systems should be in place to deal with this emerging offer of genetic tests outside the healthcare system. Some Member States have considered these issues, for example in the UK, through the "Human Genetic Commission".

This final report which provides an overview of all inputs gathered during the meeting and feedback received after circulation of a draft report should be considered as the result of a cross-fertilization exercise in which experts from different backgrounds and different areas were able to share their knowledge, concerns and views on necessary measures to be taken. The final outcome of this meeting is an overview of points of action which we hope will be taken into account by all stakeholders involved in the development, implementation

and monitoring of future initiatives addressing the organization and quality of genetic testing in Europe.

## 1- Organization of genetic testing for rare diseases in Europe

The organization of genetic testing services in European countries follows different models due to the differences in the healthcare structure, due to the way clinical genetics developed historically as a specialty or not, and due to the level of development of basic research in human genetics.

According to data from Orphanet ([www.orphanet.com](http://www.orphanet.com)), the number of laboratories providing clinical tests is not proportional with the size of the population to serve and the scope of tests they offer varies widely between countries, somewhat in proportion with the size of the population but also according to a west-east geographical gradient. The exact number of diseases for which there is a test available at the country level can be found in the 2012 report on the state of art of rare disease activities in Europe of the EUCERD (6). These numbers do not reflect the number of tests which are accessible to the citizens of a given country. Some countries routinely purchase tests abroad, while for other countries, this may be impossible, for financial reasons. Some countries have bilateral agreements, which handle the purchase of tests from one country to another (Germany and Austria, for instance); in other countries, specific regulations for the reimbursement of genetic testing abroad are in place (Belgium and Portugal, for instance). The size of the cross-border testing activity is not exactly known. The last report on it can be found in the OECD Guidelines for Quality Assurance in Molecular Genetic Testing (8).

The balance between private and public laboratories differs widely between countries as well, with a majority of private (albeit social security contracted) laboratories in Germany for instance, and none in the UK where currently the activity is normally embedded in the National Health System. The commercial offer increases rapidly at world level. It becomes hard to know where the testing activity is really performed as many companies subcontract their tests without releasing this information. The number of laboratories which are already accredited and/or participating to EQA schemes is quite different from one country to another. The funding process is quite different as well, genetic tests being considered as other types of tests and listed on the purchase list (fee schedule) with a defined price in some countries, while other countries provide a global budget to their diagnostic centers to run the activity. Finally genetic testing is regulated by law in some countries like France or Portugal, by professional guidelines in others like UK, or by law and professional guidelines as in Germany.

→ **This disparity of situations is a contribution to the inequity of access to genetic testing across Europe which request corrective measures by policymakers.**

**Points for action:**

- Document the differences in the organization of genetic testing services to help identify where the specific bottlenecks are for each type of organization, which is preventing equitable access to tests and provide a basis for discussion between MS on ways to improve their own system;
- Ensure that MS include provision for trans-border testing in their national plan;
- Ensure that undiagnosed patients with RD have timely access to newly developed NGS diagnostic testing services as clinically appropriate and when available at national level. The access to laboratories providing NGS should be ensured within European Reference Networks for RD when appropriate.

Genetic tests are generally considered as costly investigations the utility of which is not always perceived favorably when it comes to test for rare diseases. Clinical utility refers to the ability of a genetic test to significantly affect the clinical setting and patient outcome. A major challenge is to balance clinical validity, clinical utility and cost-benefit issues. In some cases a test is performing superbly in the laboratory, but is not viable from the economical point of view. On the other hand, some tests are limited in their validity, but nevertheless have great impact on patient and family management. Therefore it is important that the requirements for a test are defined in the context of their impact on the clinical setting and that the laboratory genetic test is only one of the components of an overall evaluation.

Clinical Utility Gene Cards have been developed in the context of EuroGentest (9) to enable quick guidance to all stakeholders, including clinicians, geneticists, referrers, service providers and payers. Many care providers look only at the price of a test when deciding to reimburse or not, or when allowing for getting a test from another institution, when the price has to be appreciated in relation with the overall quality of the test and its suitability for the specific purpose, as often commercial companies offer look cheaper when compared to academic offer, but have a narrower scope (fewer mutations, exons and/or genes analysed, and no complementary techniques, such as MLPA or TP-PCR in appropriate cases), having therefore a lower relevance.

→ **Economic constraints impose the consideration of all possible scale economies and prioritization of services on the basis of patient needs, cost/efficiency and evidence of clinical utility**

### **Points for action:**

- Support the development of the evidence of the clinical utility of tests as part of the care pathway, in a collaborative manner, based on the experience of countries which have already developed initiatives in this area (such as Germany, France and the UK), and disseminate widely this information to health care providers and decision makers. Post this information in Orphanet, EuroGentest website, national institutional websites (e.g. MoH and their scientific bodies) and any other relevant information portal and disseminate it to appropriate third parties to ensure that reimbursement is linked to clinical utility;
- Support the establishment of an EU/international registry of genetic tests with evidence-based claims, including those to be offered by DTC genetic testing companies;.
- Promote the appropriate use of genetic testing and the allocation of resources to genetic tests with clinical utility only. Referrals for genetic testing should better comply with the recommendations produced when generating evidence for the assessment of the clinical utility. If so, the number of tests performed will decrease significantly as the inappropriate use of genetic test has been well documented and subrogated against. This measure is not to limit the autonomy of the physicians but to guide them;.
- Document in the Orphanet databases the price of the tests and the scope of the testing offered (techniques used and scope of investigated mutations). This will provide the ground for selecting the most cost/efficient laboratories when having to order a test abroad;
- Support an increase in the health economy evaluation of genetic tests.

The evolution of technology will very soon permit the incorporation of NGS into clinical laboratory activities. Among the many challenges generated by NGS, the interpretation of genomics data is the most burning one. Interpretation requires an expert knowledge both of the genomics field but also of the specialized medical field to which the disease of the investigated patient belongs. Networking between experts and between laboratories and clinical centres has emerged gradually these last years and proved to be very efficient in boosting research and improving clinical care and appropriate use of resources. In addition pooling of data generated by sequencing must be organized and encouraged in order to provide evidence allowing the interpretation of variants for the benefit of patients, as quickly as possible.

**→ The right level of organization to maximize resources and expertise is the European, not the national one. The organization of the collaboration between expert laboratories should be set within the context of the European Reference Networks (ERNs), as described in the cross border directive. These networks should be specialized in a subset of RD corresponding to a subspecialty field.**

**Points for action:**

- Recommend to ensure access to a laboratory performing NGS in any ERN if relevant for the disease group in question, and include all the laboratories offering test for this group of diseases;
- Recommend to provide these ERNs with a set of services, to be organized at European level, to support their activity, including support services and support tools for the governance, the legal and ethical requirements and for the informatics;
- Ease early translation of genomics discoveries into quality services. This requires agreement on when a technology is sufficiently developed and validated enough to be used in a clinical setting, an issue of particular importance with NGS. Professional guidelines should be developed in support of the use of new diagnostic testing methods;
- Ensure bridging between research data and clinical data by supporting the development of knowledge engineering. Make use of all open source tools already developed in Europe, like e.g. by Gen2Phen;
- Ensure that clinical data are available for research purposes; at the minimum, aggregated data should be released, including by private laboratories;
- Recommend principles under which to operate when establishing databases including phenotype and genotype data to ensure interoperability

## 2- Quality Assurance

### **2.1- Accreditation of laboratories**

Over the past decades, the international quality standards for demonstrating the technical competence of medical laboratories (ISO15189) have been issued and refined, that deal with the provision of laboratory services. Specific points of interest are e.g. that test and methods have to be analytically and clinically validated before they are offered for diagnostics, and that an interpretation of a technical result is an integral and necessary part of a genetic laboratory report. (8, 10, 11)

Laboratories thus put quality assurance systems in place to improve the quality of their services and to warrant patient safety. The ISO standards, used for accreditation, also focus on the technical competence of the laboratory and the professionals involved in diagnostics. It is accepted that good quality systems result in better services, even without external evaluation. However, the external evaluation (e.g. by an external quality assessment scheme, recognized national accreditation bodies) of a laboratory service is a requisite if one wants to ensure the quality and safety of diagnostic services for patients,

compare the quality among laboratories, and enable equity of access and standardization of services over an entire sector (12, 13)

Accreditation bodies are established in many countries with the primary purpose of ensuring that conformity assessment bodies are subject to oversight by an authoritative body.

Those accreditation bodies, that have been evaluated by peers as competent, sign arrangements that enhance the acceptance of products and services across national borders, thereby creating a framework to support international trade through the removal of technical barriers. These arrangements are managed by the International Laboratory Accreditation Cooperation (ILAC), in the field of laboratory and inspection accreditation, and the International Accreditation Forum (IAF), in the fields of management systems, products, services, personnel and other similar programs of conformity assessment. Both organizations, ILAC and IAF work together and coordinate their efforts to enhance the accreditation and the conformity assessment worldwide.

Accreditation is a national matter, and there is only one recognized national accreditation body (NAB) in each country that assesses laboratories against internationally agreed standards (Regulation (EC) No 765/2008). The European cooperation for Accreditation (EA, <http://www.european-accreditation.org>) a non-profit association, is the European network of the recognized NABs located in the European geographical area. One of its purposes is to develop and promote accreditation criteria and guidelines that will ensure harmonized performance of national accreditation bodies throughout the European economic area. ILAC is the international umbrella organization that covers all national and regional accreditation organizations. In principle, it is not the laboratory or an institute per se that is accredited, but the tests are included in the so-called 'scope for accreditation'. This scope is published for the respective laboratories by the national accreditation bodies.

A study conducted by EuroGentest in 2012 (13) in 31 European countries has revealed that 23 % of the surveyed genetic testing laboratories were inspected by official accreditation bodies and a further 26% were certified. Notably, the accredited laboratories were located in only 12 different countries. This survey was the first large comprehensive update of the quality status in European genetic testing laboratories since the European projects on quality assurance were initiated in the early 2000s and literature was published that aimed to improve the quality in genetic testing. The previous study had been conducted in 2003 and included 15 European countries and US (14). An exact comparison cannot be made, but a quick look at the Orphanet/EuroGentest database suggests that only 183 out of 1645 (11.1%) laboratories providing testing for rare diseases are effectively accredited and that 544 (33%) of them participated in at least one EQA in 2011. It is generally accepted that

EU-sponsored projects like e.g. CRMGEN, CF-Network and especially EuroGentest, have significantly promoted accreditation. Hence, the fact that only a small minority of the laboratories are effectively accredited reveals that significant hurdles exist on the road towards accreditation. These include the mere fact that the installation of a quality system and the subsequent maintenance of it, in combination with regular external control schemes and the audits by the accreditation bodies, cost time, effort and money. Moreover, laboratories may not see or be aware of the advantages of being accredited, as long as other laboratories can continue to offer similar services without taking that extra burden. In some Member States, accreditation is – or will soon become – a requisite for the provision and/or reimbursement of genetic diagnostic tests. This requirement definitely provides an impetus for laboratories and institutes to move forward and invest in quality, and should become the norm in all Member States.

→ **It is necessary to ensure the quality of the genetic diagnostic laboratories, for the sake of the patients and the benefit of the community. However, the wish to offer quality and peer pressure does not seem to drive the laboratories towards accreditation. The only way forward is to make accreditation the norm, i.e., that all diagnostic laboratories in Europe should be accredited. To further guarantee equity, regulation should include the requirement for all methods used in genetic testing to be within the scope of accreditation. Practical solutions do exist to deal with the case of extremely rare and thus low volume tests.**

However, several problems exist or become apparent:

- Preparing for accreditation costs money, and in general, the national health care systems do not 'reward' laboratories for the extra costs. On the contrary, in many countries, the reimbursement rates for genetic tests have been or may soon be reduced, under economic constraints.

- In general, bigger laboratories (higher number of personnel and higher number of samples received) were more likely to be accredited than smaller laboratories, even though, in principle, size per se should not be a hurdle or impediment (13). Some very small laboratories may not be able to redeem the effort or the costs to establish a quality system and go for accreditation. Such laboratories should perhaps close their doors, because if they are too small to cope with formal quality requirements, they are probably not fit for offering genetic testing at all. Nevertheless, for some very rare diseases, it is possible that only one or a few (often academic or research) laboratories in Europe (or even in the world) offer a specific test. Such laboratories would be identifiable through the ERNs for rare diseases.



- **Some laboratories may be eligible for help in bearing the costs and achieving the transition towards accreditation. A transition period could be proposed, with specific funding for that transition.**

Some of the national accreditation bodies have difficulties to gear up towards auditing the genetic laboratories. The lead assessors need to accustom themselves with this new field, but more critically, more specialized technical experts are needed to evaluate the services on behalf of the accreditation body. In all EU countries, National Accreditation Bodies (NAB) are non-for-profit organizations that work under direct supervision of National Authorities. Hence, the national governments have to warrant that sufficient funds and people are available to provide good technical assessment. In general, the burden of an accreditation is significant, but the costs for the audits, i.e. the formal visits by the auditors and experts, are only a fraction of the total costs that the establishment and the maintenance of a quality system entail.

- **Governments may have to put mechanisms in place to alleviate the costs of the audits, for the laboratories as well as for the NABs. Measures have to be taken and systems have to be put in place to warrant that the accreditation requirements are equivalent in all instances.**

An EA Working Group for laboratory medicine meets every 6 months. All the issues related to one specific field are discussed with the ad hoc stakeholders. EuroGentest is the representative for human genetics, and other stakeholders could be added, if necessary. Nevertheless, specific guidelines for the evaluation and accreditation of specialized methods and novel technologies are needed. They have to be generated with the expert input from laboratory specialists, manufacturers, quality managers and representatives of the accreditation bodies.

- **The ISO norm provides a basis for harmonization but interpretation by experts may still vary. It would be good to increasingly liaise with European Accreditation (EA) about this issue.**

All too often, research data are transferred directly to the medical file of a patient without passing through the 'quality filter' of a diagnostic service. This is a dangerous practice, that may put patients and families at risk, e.g. when carrier and prenatal testing is offered on the basis of results that have not been confirmed independently in a laboratorial diagnostic context.

Diagnostic laboratories, that use research results or involve research scientists to generate (part of) a report, are responsible for the quality of those data that are eventually included in the clinical report. In practice, this means that the diagnostic laboratory has to effectively check the data that are generated in a research laboratory, or preferably repeat the essential part of the analysis in the diagnostic viz. accredited context. Generic Standard Operation Procedures (SOPs) are essential, however it has to be clear that the context may vary, and SOPs shall thus not provide an invitation to install a superficial system.

### **Points for action**

- Clarify the ISO standard to laboratories, through training sessions, workshops and personal guidance
- Facilitate the transit towards accreditation by encouraging the laboratory geneticists to share their interpretation of the norm and existing guidelines into the clinical practice.
- Write guidelines to complement the general ISO standards.
- Establish a link with EA to ensure the uniformity of the implementation of the norm and the adoption of guidelines.
- Ensure that accreditation bodies are able to handle the requests of accreditation of genetics laboratories and propose to pool international experts to visit the laboratories together with the lead assessors.
- Verify at the national level the norm that it is necessary to validate the tests and the data before using them in a clinical context. Research data should not be returned to patients or their physicians, unless they have undergone the same scrutiny in terms of analytical and clinical validation as the tests, offered by diagnostic laboratories.

### **2.2- External Quality Assessment**

External quality assessment is a tool for monitoring the quality of the laboratories and an integral part of the Quality Assurance. Several organizations are offering these services to the laboratories, either at national or international level. The European Commission has fostered the development of international EQA schemes through different projects like, e.g., EuroGentest, of which EMQN was a partner.

Some current issues are:

- **Poor performers:** there are laboratories which are repeatedly failing to perform well in the analysis and interpretation of the samples that are circulated by the EQA providers. EQA

providers identify those laboratories, but they can only try and invite the collaborators from those laboratories to participate in training programs, to improve their service. However, it seems to be very difficult to motivate those laboratories to effectively seek support, for several reasons. It is not within the remit of the EQA providers, nor of EuroGentest, nor of the ESHG, to reprimand those laboratories. This can only be done at the national level, either by the competent authorities, by the department of health or by the healthcare payers. Only in a few countries, such a feedback mechanism to address poor performance is yet effectively in place. Especially the healthcare payers should be informed of the importance of accreditation in the provision of quality and safe services in genetic testing. They can make accreditation a requirement of the services which they will fund. This can be a powerful encouragement for compliance to EQA participation and improve performance.

- **Participation to EQA is voluntary**, except for an accredited laboratory that is strictly obliged to participate in inter-laboratory comparison such as EQA schemes. External evaluation could be organized in different ways, but, in practice, EQA schemes are often the most convenient way. This, in combination with the fact that only a minority of the laboratories are accredited, results in a variable uptake of EQA among regions and laboratories. For most EQA schemes, the participation has been growing over the past years, and the laboratories engaged in EQA seldom withdraw. But a significant percentage of the laboratories does not participate at all (13).

- **Participation to EQA schemes is expensive**. The ISO norm and the accreditation process insist on external assessment for as many different tests within the laboratory's scope as possible, and this for the entire examination process including pre and post examination. Not all EQA providers offer schemes which cover the whole process. The full costs of EQA are a combination of the price for the subscription to the scheme, as well as the processing and reporting time within the lab. It is part of the burden of quality assurance in general. Given that the laboratories are funded nationally, significant differences exist in the way that laboratories can cope with these costs and efforts. These costs associated with quality initiatives should be included in any laboratory's budget.

Suggestions have been made to train people by offering the possibility to host the collaborators from the poorly performing labs in better established laboratories. However, this has not been very successful, not in the least to due to the stigmatization which is associated with such activities.

Laboratories could share and exchange samples. However, this is a low scale initiative that only partly deals with the problem of quality in the laboratory, and that is hampered by regulations on cross-border transfer of samples.

Both national and international schemes exist. There are advantages and disadvantages associated with this situation:

- The national schemes allow laboratories to report in their own language. To some extent, this is also possible for the international schemes.
- For both the national and the international schemes, a certain critical mass is necessary to sustain the activity. Hence, scale is an issue that may affect the quality of the EQA provider per se.
- International schemes are important for rare diseases, i.e. most countries would not be able to support such schemes except for the more frequent genetic diseases, and probably no country would be able to offer the complete panel.
- Some governments have effectively approved specific schemes. This is only acceptable if the schemes have the same standards as the international, accredited scheme. It should not be a mere protective measure for a national activity. For instance, since 2001 the National Centre for Rare Diseases (Istituto Superiore di Sanità), upon commitment by the Italian MoH, coordinates the National EQA schemes for public and private genetic laboratories, according to the international standards (<http://www.iss.it/cnmr/tege/index.php?lang=1>) (15, 16).

At present, the international EQA providers seem to be financially break-even. This is partly due to the fact that they are embedded in existing (academic) structures and it is only possible because the organizers of the schemes as well as the assessors donate their time. As the schemes get bigger, this will become problematic. Also, hospitals and academic institutions start to ask about the 'value' of this voluntary work and may request the scheme organizers to reimburse the people for their work. The fear is that this will very soon undermine the quality and provision of EQA. One of the strengths of existing EQA schemes is that the assessors are working scientists who understand the technicalities of the tests and the requirements for reporting. This especially applies to the EQA providers that were fostered by EuroGentest.

**→ EQA providers have to be accredited, to confirm the quality of their services.**

Commercial EQA schemes do exist, and some EQA providers are capable of offering their services at a lower cost than that of the no-for-profit schemes. This is partly due to the fact that they run more basic schemes (e.g. genotyping only), and that interpretation of the lab reports is not commonly included in the evaluation. It is observed that the EQA schemes have an important educational role and help the laboratories not only to monitor their technical performance but also to improve the quality of their reports.

- **The evaluation of the interpretation of test results in laboratory reports by the scheme organizers and assessors should be mandatory. Efforts and support are needed to make the EQA schemes sustainable without increasing the financial burden on the laboratories.**

Currently, the fees include the costs for the preparation and distribution of the samples, the administration, the assessment by experts, and the organization of pilot schemes together with best practice meetings. Each of these activities could be sponsored and/or centralized to reduce the expenses on behalf of the EQA providers. This would be a better option than to subsidize individual laboratories to cope with the costs of EQA.

- **For many laboratories, the costs for EQA are already too high. Ways to subsidize the activity of the EQA process should be explored, as it would lead to the reduction of the costs of the EQA participation.**

The uptake of EQA has to be promoted for quality monitoring and assurance in general. To deal with poor performances in particular, specific educational and regulatory measures have to be taken.

- **Ideally, the EU could issue regulation on uptake of EQA.**

#### **Points for action**

- Establish a registry of EQA schemes where providers submit their data to monitor the use of the EQA schemes as well as the performance of the laboratories. Harmonizing and reporting should be made mandatory, to warrant the value of the registry.
- Develop an offer of educational meetings and courses to help the laboratories to improve their quality. EuroGentest has developed such courses, however, funding is limited and the

project ends in 2013. Issue a directive or regulation to make the accreditation of EQA providers mandatory.

- Make an analysis of the costs of the EQA schemes to identify those costs that could be borne by a central national or European agency or organization. The latter would alleviate the costs of participation in the schemes. Suggestions are the funding of pilot schemes and best practice meetings, logistic support to the generation, distribution and validation of samples, organization support for the assessment of the laboratory reports, etc.

### 3- Next Generation Sequencing

Targeted sequencing, exome sequencing and, to a lesser extent, whole genome sequencing, are frequently and increasingly used for research. Several laboratories are already offering targeted sequencing and exome sequencing (for targeted analysis) in a diagnostic context.

→ **It is generally believed that the use of panels of genes, that allows for comprehensive testing for the genetically heterogeneous diseases, will pass the threshold for clinical utility. Exome sequencing and total genome analysis for unknown diseases remains a research activity for the time being.**

Opening the "exome" or "genome" is currently outside the scope of what most of the diagnostic laboratories should offer, both in terms of quality standards as well as in operational terms. Also, in most countries, a specific regulation for the reimbursement of such comprehensive exploration of an individual's genome is not in place. A few laboratories in Europe have already been accredited for NGS applications, including exome sequencing (for targeted analysis), even if no clear guidelines or consensus criteria exist.

→ **It is important to specify the role of NGS for diagnostics.**

Several applications exist that effectively empower the specialized medical care, not in the least for the rapid identification of the genetic defect for rare disease patients. The fact that the new and especially the targeted approaches create a possibility to save resources should be exploited. On the one hand, the targeted analysis of genes with a known and proven role in a specific disease, should be supported. Exome or total genome analysis may be used as a tool to generate such panels, on the condition that the sensitivity and specificity of this approach are acceptable from a diagnostic standpoint.

On the other hand, exome sequencing for the identification of novel genes and defects is currently outside the scope of what can reasonably be offered in a diagnostic context.

The clinical utility of gene panels is a matter of discussion. It cannot be determined by the laboratories alone, it should be established by expert panels that include medical specialists as well as clinical and laboratory geneticists

Incidental findings are a side-product of genomic approaches to genetic testing. However, the mere fact that they exist and will be encountered should not overly impede the introduction of NGS into diagnostics. The following considerations should be made, and actions should be taken accordingly:

- The phenomenon of incidental findings is known in medicine, and in particular in radiology. Over the years, the radiologists have established procedure and guidelines for dealing with incidental findings, that are both accurate and flexible, and regularly updated as the technologies for imaging evolve. It is accepted though, that this issue of incidental findings is of a different order of magnitude in genetics, esp. if the entire exome or genome is scrutinized in a patient.
  - A 'red list' of genes and/or variants that disclose 'high risk' and potentially treatable genetic predisposition would be helpful to deal with incidental findings in practice. There is a consensus that, at this stage, there is no meaningful clinical use of low risk predictions and hence, these should in principle not be communicated to the patient. It is an aspect of consumer protection to warn the community against unnecessary medicalisation. The 'red list' would also be helpful to regulate a direct-to-consumer offer of total genome or exome sequencing, as the providers of such test could be requested to 'flag' abnormal results that belong to this list and be obliged to refer the consumer for medical follow-up.
  - An informed consent is necessary for a genomic diagnostic approach. It should allow the patient to decide beforehand whether or not to receive information other than that related to the disease under investigation.
  - Opinions and regulations vary as to whether a clinician can overrule the patient's or parents' opinion in case of severe risk alleles, e.g. highly penetrant cancer predisposition mutations.

NGS can be used to screen for fetal aneuploidies. In those applications, the technology is applied to quantify and determine the respective contribution of DNA fragments from e.g. chromosomes 18 and 21 to the free fetal DNA, circulating in the maternal circulation. This is a technical achievement, rather than a conceptual shift. However, total exome or genome

sequencing to determine an individual's complete genotype or predisposition to disease, is not a tool for neonatal, prenatal or population screening at this stage. Genetic counseling is a communication process, which deals with the occurrence, or risk of occurrence, of a genetic disorder in the family. The process involves an attempt by appropriately trained person(s) to help the individual or the family to understand the medical facts of the disorder and the options on how to deal with it.

→ **Total genome or exome testing should not be offered without such counseling.**

**Points for action**

- Produce guidelines for NGS sequencing in a diagnostic setting, including that the diagnostic specificity and sensitivity and the performance of these assays have to be determined, and minimal criteria have to be defined. This is important both from the standpoint of the laboratories, who have to ensure the quality of the service and the safety of the patient, and from the standpoint of the accreditation bodies, who need criteria according to which those NGS tests can be accredited in the laboratories. The latter is also important to guarantee quality and equity in cross border testing.
- It is important to create a knowledge basis for the interpretation of unclassified variants (UVs) i.e. variants for which the clinical significance is initially uncertain. An appropriately accessible international and comprehensive data basing environment is a necessary tool to allow the laboratories to reliably analyze the data.
- Gather data about the frequency of incidental findings, and about the attitude of patients and families towards such findings. Several project proposals have been filed, that deal with the aspects of data management and exchange, for research as well as for diagnostics.
- The national health care systems should put the systems in place for the reimbursement of 'gene panels' with a clinical utility.
- Aspects of and knowledge about the genetic constitution of an individual should be included in the proposal for an EU Regulation on consumer protection.
- Produce guidelines on the use of clinical information from NGS sequencing in the diagnostic setting. In particular, work should be supported on how to clinically manage incidental findings information from NGS. This is something that needs to be dealt with at the European or even global level



## 4- Direct-to-consumer genetic testing

In recent years various companies have started to advertise, sell or provide genetic tests directly to consumers.

In view of the quality standards advanced by the Council of Europe's Additional Protocol to the Convention on Human Rights and Biomedicine, concerning Genetic Testing for Health Purposes and the OECD Guidelines for Quality Assurance in Molecular Genetic Testing (17), the offer of genetic tests for health purposes directly-to-consumers has created various concerns. Firstly, there are significant concerns regarding the clinical validity and utility of various tests offered (18). Also in the context of carrier identification of autosomal recessive disorders, there are concerns with regard to clinical validity. Secondly, marketing strategies of such companies often overstate the predictive value of the tests advertised and overrate its potential health implications. - For most common complex disorders, genotype-phenotype associations are weak and selective genotypes bad or even misleading predictors with regard to the development of the phenotype.

Thirdly, the offer of genetic tests through the Internet by commercial companies runs the risk to disconnect these services completely from their usual embedding in a medically supervised context. The absence of medical supervision for most DTC tests may compromise or fail to foster patient health. However, medical supervision is not always a guarantee of quality of service provision. Some examples illustrate that some of these companies involve healthcare professionals. In this case impartial health advice might be compromised. Fourthly, testing of third parties, not having given or being incapable of giving informed consent becomes possible. Such testing will most often be at odds with best practice clinical guidelines. Other concerns with regard to the activities of DTC companies include the research activities of these companies performed on submitted samples and information without adequate informed consent or monitoring by a research ethics committee as well as the potential submission of samples without consent.

- ➔ **It would be a mistake, and ultimately an unsuccessful endeavour, to focus efforts on remedying the potential harms from DTC tests without considering the entire regulatory context.**

Without a system in which an upfront expert evaluation can be made with respect to the validity of genetic tests, it will be difficult if not impossible to make rational decisions about who can and should order the test and receive the results, and what claims are appropriate in advertising.

## **Points for action**

### **Information provision to health care professionals and the general public**

- Provide information to healthcare professionals and the general public that gives background on genetic testing and describes the provision of genetic testing services.
- Underline that clinically validated and medically appropriate genetic tests are offered in clinical services for those that need them and that these are reimbursed by the healthcare system.
- Make available the information about the limitations and concerns of the tests that are currently advertised, provided or sold through the internet.
- Take initiatives to stimulate public education (e.g. at school) and education of healthcare professionals.

### **Frameworks regarding the provision of genetic testing services**

- The embedding of genetic testing in a healthcare setting can ensure a context where due emphasis is being provided on the individualized medical supervision of patients, the presence of pre-test and post-test counseling, psychological evaluation and follow-up if appropriate and quality assurance of the tests performed.
- Analyze the legal models in place in some Member States in order to study the strengths and limitations of these approaches, and their potential for application in other Member States.

### **Legal frameworks regarding the use of predictive health information in other contexts**

- Concerns have been raised regarding the use of personal predictive (genomic) health information by third parties, such as insurers or employers. It is necessary to study various existing legislations on this issue in the various Member States, in order to generate best practices for harmonization across European Union.

### **Penalization of the submission of DNA samples of third parties without their consent**

- More efforts are necessary at a European level in order to sanction individuals that submit samples from a third person without his or her consent. Services through the internet are organized in such a way that there is no control over the origin of the samples being analyzed. Most companies send mouth swab kits as these are easier and more practical than having to send the client to a clinic where blood is drawn. Since the mouth swab is done in the privacy of the clients' home, there is no way of controlling for the identity of the sample provider.

*The report has been drafted by Ayme, Borry, Gribaldo and Matthijs before being circulated to the participants.*

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#### Abstract

The workshop was designed with the aim of bringing together experts and stakeholders in the field of genetic testing to discuss the (future) organization of genetic testing in Europe. Obviously since it is not be possible to adequately deal with all aspects of genetic testing within the framework of one workshop, a limited number of issues have been selected. The selection was based on the importance and urgency of the matter and the need and opportunity for action at the European level, and the likelihood for successful intervention. Primary deliverables of this workshop have been planned as to be able to define a vision on the use, value and integration of genomic medicine into clinical practice and to prepare a briefing note to highlight the specific points that deserve the Commission's interest. Quality of genetic testing and organization of genetic testing services were the two main themes of the scope of the workshop. To warrant the quality of the genetic diagnostic laboratories the way forward is to make accreditation the norm, i.e. the diagnostic laboratories in Europe should be accredited. To further guarantee equity, the regulation should include the requirement for all tests to be within the scope accreditation. The embedding of genetic testing in a healthcare setting can ensure a context where due emphasis is being provided on the individualized medical supervision of patients, the presence of pre-test and post-test counseling, psychological follow-up if appropriate and quality assurance of the tests performed. In light of growing number of companies selling and advertising genetic tests, it is crucial that information is available for healthcare professionals and the general public that gives background on genetic testing and describes the provision of genetic testing services.

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