

Genetic testing in Europe -
A network for test development,
harmonization, validation and
standardization of services

FP6 2005-2009

www.EuroGentest.org

Ros Hastings, FRCPath
Oxford Radcliffe Hospitals NHS Trust



The most popular genetic tests

- Cytogenetics
- Cystic Fibrosis
- Hemochromatosis
- Fragile X mental retardation
- Factor V leiden
- BRCA 1 & 2



Potential impact of genetic tests on European health budget

30 million potential ? customers

- 500 – 4000 molecular tests/ million citizens/ year
- 4000 – 10.000 cytogenetic tests/ million citizens/ year

200 – 2000 € per test

900.000 – 28.000.000 €/ million citizens/ year



Survey Results Community Genet. 10(3):123-31, 2007

- **International exchange** is a widespread feature of Molecular Genetics service provision.
 - 64% of Laboratories received specimens from other countries
 - Over 18,000 specimens crossed borders in 2002
- Variables contributing to a high Quality Score ($p < 0.005$):
 - ✓ Accreditation of the laboratory
 - ✓ Participation in proficiency testing
 - ✓ Director having formal training in molecular genetics
 - ✓ Affiliation with a Genetics Unit

<http://www.oecd.org/dataoecd/25/12/34779945.pdf>



Issues to start from

- More than **1,000 laboratories/centers** in different settings
- More than **1,000 rare diseases** can be tested

- Lack of **centralized and uniform information** about services
- Limited networking

- Lack of harmonized and standardized PT/EQA
- Lack of reference materials
- Limited number of ISO 15189 **accredited labs**
- Limited number of **accredited EQA schemes**

- Limited analytical and clinical validation of tests
- Diverse regulatory environments

- Insufficient **counselling**/lack of harmonized approach

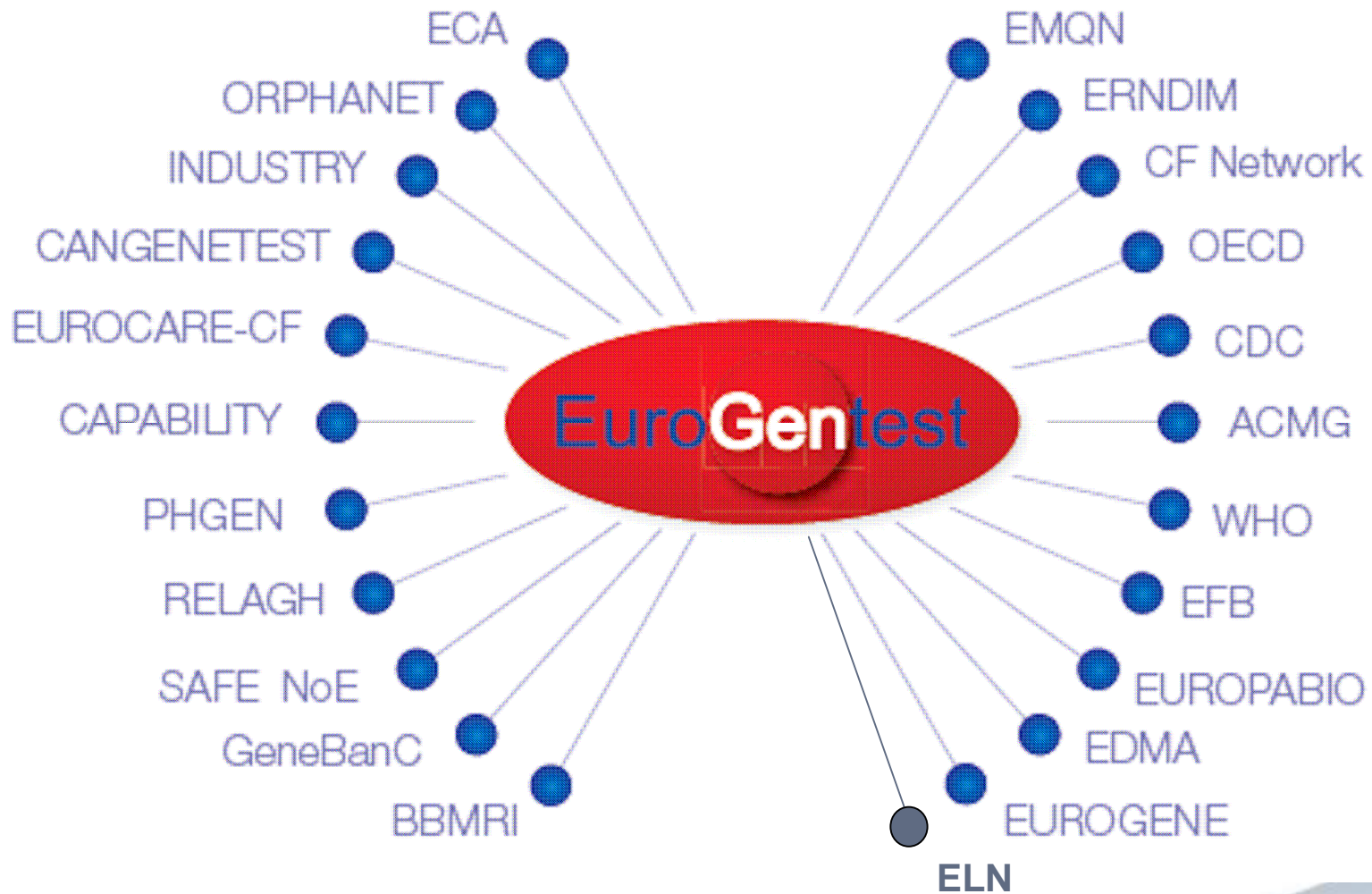
Unacceptable number of laboratory errors!!!!



IPTS report 2003

EuroGentest

Creating a network of networks



Major aims of the NoE

- Harmonization and quality improvement of genetic services
- Defining the European dimensions which are compatible with and complementary to National approaches

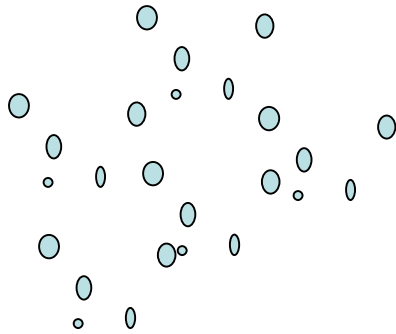
FOUR major domains of activities

- Quality of the genetic laboratories
- Quality of the Clinical aspects of the services
- Translation of Technologies into diagnostic practice
- Educational aspects



Present

individual facilities



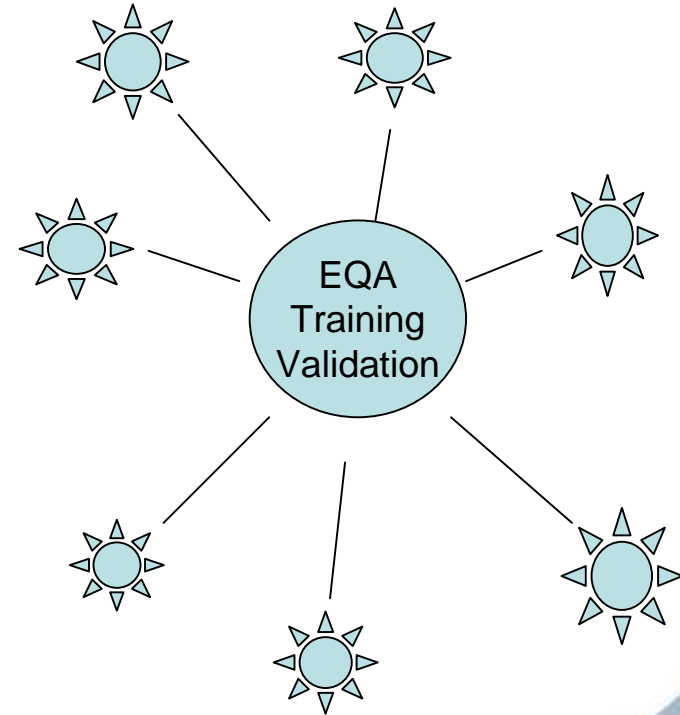
Highly variable quality
Individual rules and procedures



Future

Virtual Institute:

Structured Network of expert
and peripheral centers



Common goals for quality, criteria, training
Centralized information for all
EQA provision
Validation of new technologies



The ACCE model

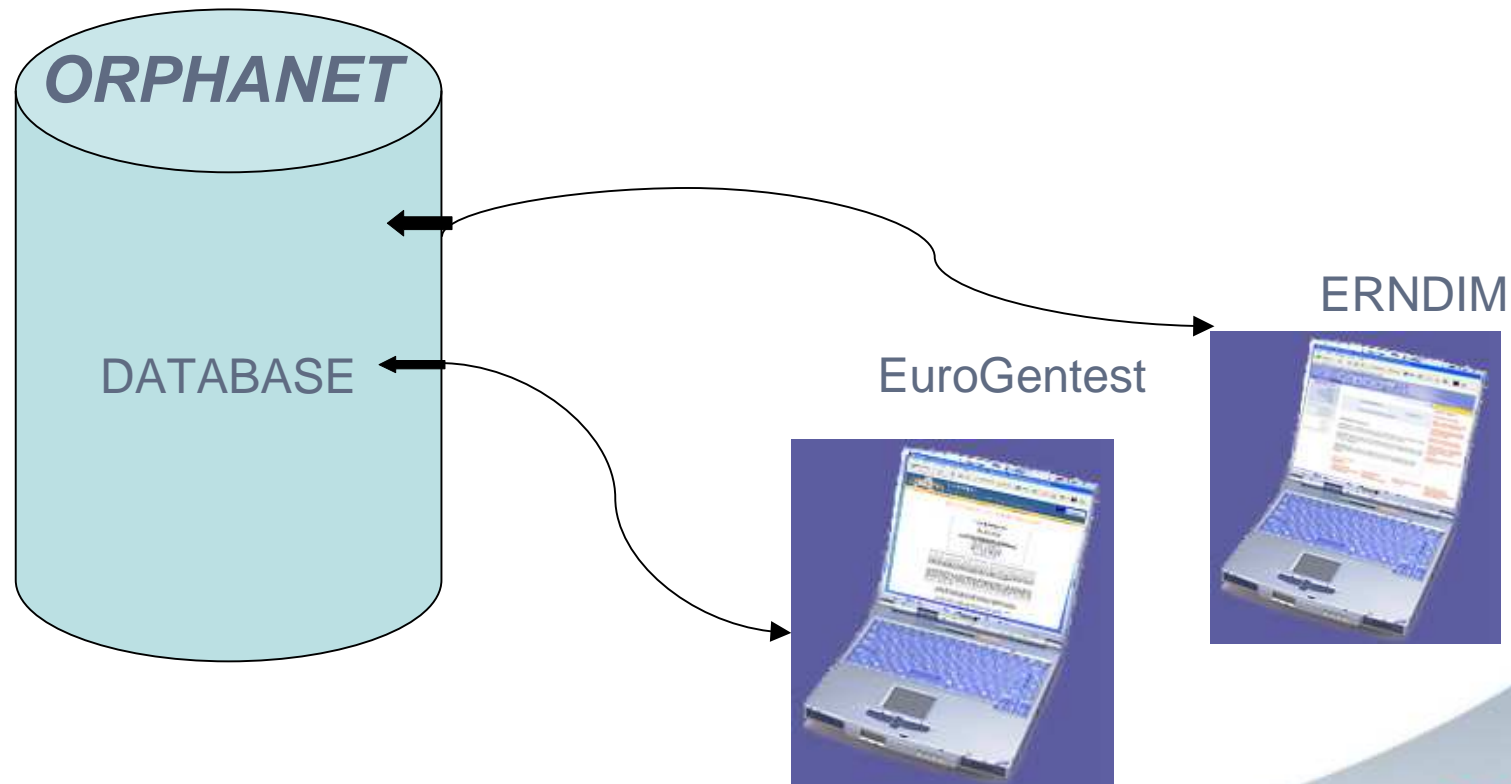
- **Analytical validity:** accuracy of test identifying biomarker
- **Clinical validity:** relationship between biomarker and clinical status
- **Clinical utility:** likelihood that test will lead to improved outcome
- **Ethical, legal and societal implications**



A comprehensive European database of labs and tests

Biochemistry
Cytogenetics
Molecular

- Operated by Orphanet in collaboration with EuroGentest
- Cross-referenced with SwissProt, OMIM and Medline
- Used daily by 25,000 users from 160 countries



Quality Assurance Database (since 2007)

- Access via www.orpha.net and www.eurogentest.org

UNIT 1 : QUALITY MANAGEMENT AND ACCREDITATION / CERTIFICATION OF GENETIC TESTING

SIMPLE SEARCH SEARCH

FILTERS

Filter

Country City

Laboratory/Institution Professional

EQA provider Accreditation body

Accredited Certified EQA participating

CLINICAL LABORATORIES **SEARCH**

Page: 1 Sort on: Accredited 0 - 14 of Labs - 20 labs/page

A	C	E	Country	City	Laboratory	Institution	Labdirector
■	●	▲	SWITZERLAND				
■	●	▲	SWITZERLAND				
■	●	▲	SWITZERLAND				
■		▲	SWITZERLAND				
■		▲	SWITZERLAND				
■		▲	SWITZERLAND				
■		▲	SWITZERLAND	Geneva	La		Morris, Michael

Certification ISO 2001

Accreditation ISO 15189

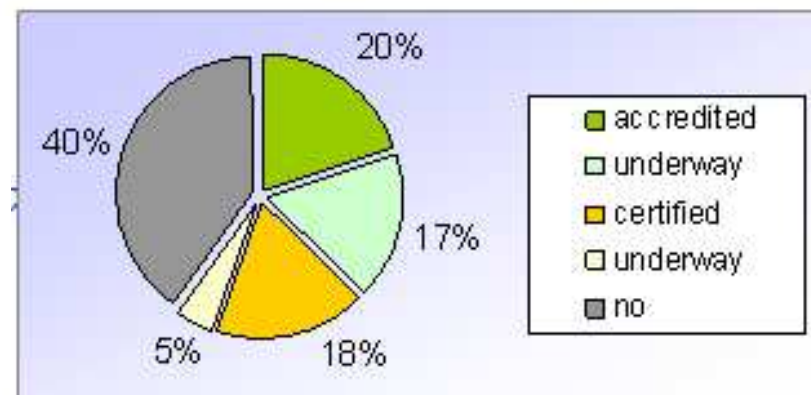
Forms under development



QA Survey EuroGentest (2006)

- Answers from ~350 testing labs from 32 countries:
 - Validation of information from questionnaire sent to 2,300 individuals
 - QAU data validated with EQA/accreditation providers
 - each lab receives unique, permanent EUGT number
 - data exchange with Orphanet
 - Orphanet completes test data

The screenshot shows a web-based form for quality management. It includes sections for EQA participation, accreditation, and certification, each with a 'Save' button and a 'No' radio button. The 'Accreditation' section features a bar chart with three bars of varying lengths, representing data for different categories.



Directory of EQA Schemes and Providers

UNIT 1 - QUALITY MANAGEMENT AND ACCREDITATION - CERTIFICATION OF GENETIC

Qm

- I. Constitutional mutations
- II. Methodological EQA
- Addresses of contacts
- Acknowledgements

EMQN and GfH EQA scheme merger



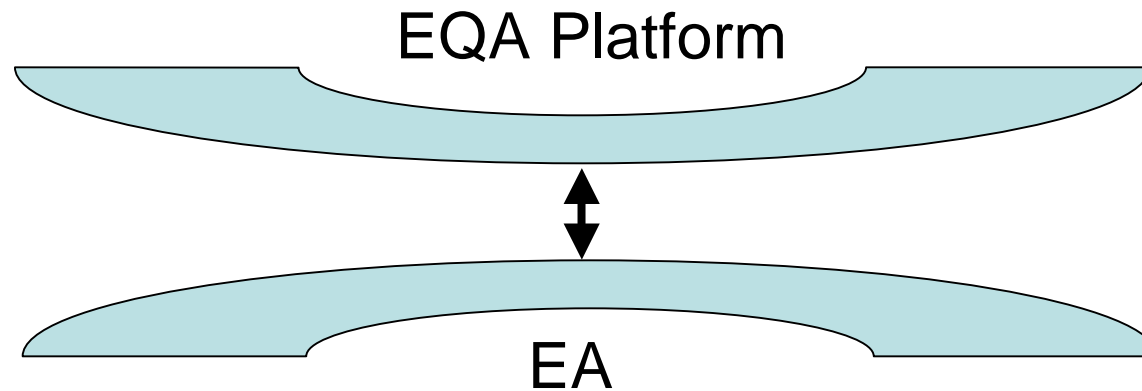
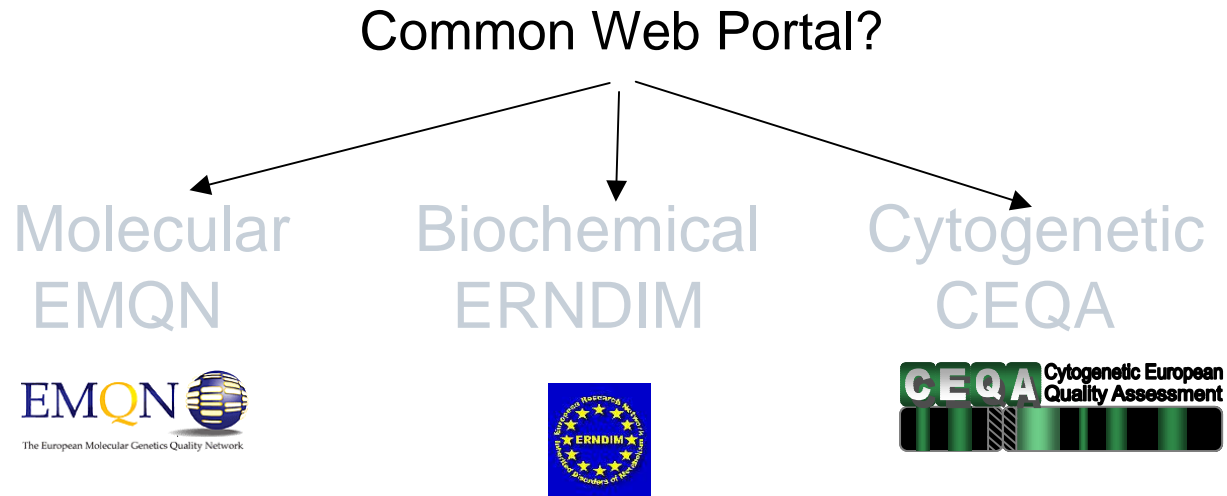
We are very pleased to announce that on December 20th 2007 the board of the German Society of Human Genetics (GfH) approved the merger of the EMQN and GfH External Quality Assessment schemes. (upd.18 Jan 2008) [More...](#)

MOLECULAR GENETIC TESTING - EXTERNAL QUALITY ASSESSMENT SCHEME PROVISION (2005)

I. Constitutional mutations

Disorder	Organisation	Contact Point
1. ACE (Angiotensin I Converting Enzyme)	DGKL ² (RIB) ¹²	M. Neumaier, Mannheim
	ÖQUASTA ⁴ (in collaboration with DGKL)	W.-M. Halbmayr, Ch. Mannhalter, Wien
	ECAT Fdn. ⁷ (in collaboration with DGKL)	P. Meijer, Leiden
	LABQUALITY ¹⁸	M. Keinänen, Helsinki
2. alpha-1-Antitrypsin (P1M, P1S, P1Z); (RV Nr.743)	INSTAND ¹¹	R.-R. Flörke, Düsseldorf
3. Alpha-1-Proteinase-Inhibitor (M5Z)	DGKL ² (RIB) ¹²	M. Neumaier, Mannheim
	ÖQUASTA ⁴ (in collaboration with DGKL)	W.-M. Halbmayr, Ch. Mannhalter, Wien
	ECAT Fdn. ⁷ (in collaboration with DGKL)	P. Meijer, Leiden
	LABQUALITY ¹⁸	M. Keinänen, Helsinki
4. Apolipoprotein B100 (ApoB100)	DGKL ² (RIB) ¹²	M. Neumaier, Mannheim
	ÖQUASTA ⁴ (in collaboration with DGKL)	W.-M. Halbmayr, Ch. Mannhalter, Wien
	ECAT Fdn. ⁷ (in collaboration with DGKL)	P. Meijer, Leiden
	LABQUALITY ¹⁸	M. Keinänen, Helsinki
5. Apolipoprotein E (ApoE): E2E3E4	DGKL ² (RIB) ¹²	M. Neumaier, Mannheim
	ÖQUASTA ⁴ (in collaboration with DGKL)	W.-M. Halbmayr, Ch. Mannhalter, Wien
	ECAT Fdn. ⁷ (in collaboration with DGKL)	P. Meijer, Leiden
	LABQUALITY ¹⁸	M. Keinänen, Helsinki
6. Apolipoprotein E (E2, E3, E4); (RV Nr.744)	INSTAND ¹¹	R.-R. Flörke, Düsseldorf
7. Azospermia (AZF/DAZ)	EAA ⁶ /EMQN ¹⁰	M. Simoni, Münster
8. Breast/ovarian cancer familial (BRCA1, BRCA2)	EMQN ¹⁰	S. Patton, Manchester

Common Platform for European EQAs



MS Accreditation agencies

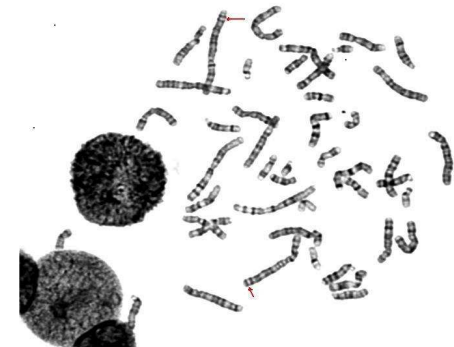


Cytogenetics EQA - CEQA



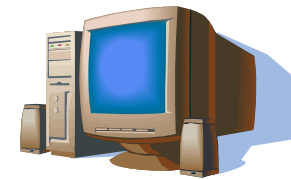
- **Online EQA – feasibility and viability demonstrated**

- Feasibility initially tested on UK NEQAS participants
- Two pilots and then extended to routine EQAs
- Registration, invoicing, submissions and results all online



- **European online EQA for Cytogenetics, CEQA**

- 80+ participants from 25 countries registered
- One postnatal and one prenatal EQA
- Leukaemia EQA and PGD/PGS pilots
- All European National Scheme Organisers are assessors
- Generic marking criteria
- European assessors/experts meeting
- Exemplary answers and annual summary report



Expert workshops and training:

9 workshops organized

Accreditation & Quality Systems

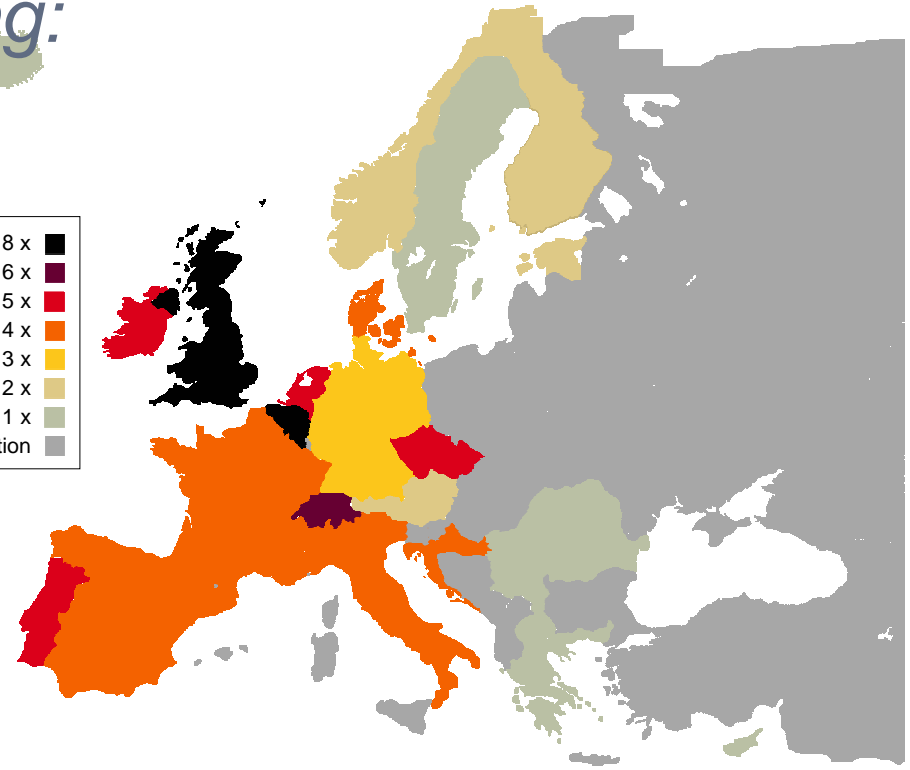
Internal Audit

IT support

Motivation & Change

Diagnostic Validation

Management review of IQC & EQC



- 27 different countries participated
(Europe: 24, North-America: 1, South-America: 1, Australia: 1)
- 86 different institutes participated
- 156 different participants, 29 of them participated more than once

<http://www.eurogentest.org/unit1/workshops.xhtml>



Types of workshops

Accreditation and quality management

General workshop for those who would like to start up or improve their quality system.

Internal audit

Specific workshop on the preparation, execution and evaluation of an internal audit.

Towards accreditation - Managing the human side of change

This workshop will give insight on the behavioral side of change and how to apply this in real life.

IT support

Learn what is on the market and which criteria are important when implementing electronic support for your QMS.

Fulfilling the requirements of ISO 15189

Workshop tackling specific topics of the ISO standard such as management review.

Diagnostic validation

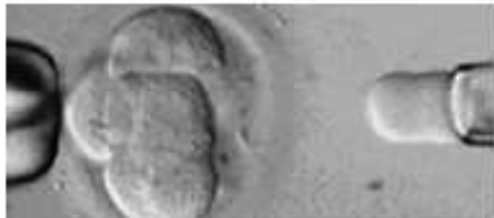
Workshop on the requirements for validation regarding ISO 15189 and related issues.

Round table sessions ESHG - Case studies on quality assurance and quality control issues in genetic testing laboratories

Short session to discuss on and share experiences with quality management and



Preimplantation Genetic Diagnosis in Europe: Current Practice and Future Needs



*A. Corveleyn, M. Morris, E. Dequeker, N. Nagels (EUGT);
EC Joint Research Centre, ESHRE, EPALAN*

Publications:

- *Survey results (submitted);*
- *Legal aspects (in preparation);*
- *IPTS Report (in press)*

- **First comprehensive, independent survey** of services, current practice and QAu status of European PGD providers
- Identification of providers:
 - Centres, clinics and labs in 17 countries
- Surveyed for
 - All PGD cycles performed in 2006
 - Including familial chromosomal anomalies, over 50 monogenic disorders (including adult-onset) and HLA typing.
- **Current practices** defined in :
 - Quality management
 - Qualifications
 - Internal quality control
 - External quality assessment
 - Traceability
 - Follow-up
 - International flow of patients and samples
- **A number of problems** of practice and quality assurance identified





Preimplantation Genetic Diagnosis in Europe

(Executive Summary)

Anniek Corveleyn, Eleni Zika, Michael Morris, Elisabeth Dequeker,
James Lawford Davies, Karen Sermon, Guillermo Antñolo,
Andreas Schmutzler, Jiri Vanecek, Fransesc Palau, Dolores Ibarreta



EUR 22764 EN - 2007



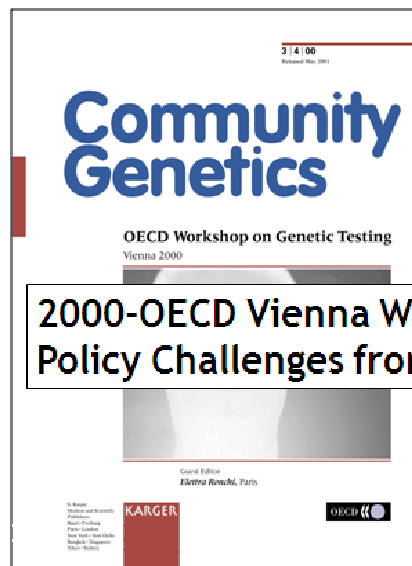
European
Science and
Technology
Observatory



www.eurogentest.org

EuroGentest

OECD Guidelines for Quality Assurance in Genetic Testing



2000-OECD Vienna Workshop:
Policy Challenges from the New Genetics



OECD Guidelines

- Guidelines are relevant to OECD and non-OECD countries
- Applies to ALL genetic disciplines
- Countries need to implement what they approved.
- Implement the Best Practice in your laboratory
- Spread the quality message!
- EuroGentest is available to help!
 - Workshops
 - Documentation
 - Validation
 - Advice
 - EQA

www.EuroGentest.org

www.OECD.org/biotechnology

OECD GUIDELINES FOR QUALITY ASSURANCE IN
MOLECULAR GENETIC TESTING



EuroGentest

What do the Guidelines do?

www.oecd.org/sti/biotechnology

- Set minimum standards
- Encourage best practice:
 - OECD countries are asked to implement the guidelines
 - Non-member countries invited to use them to set policies
- Progress report and review in 2011

Structure

- A. General principles of MGT
- B. Quality Assurance systems
- C. Proficiency Testing
- D. Result Reporting
- E. Education and training



Procedure for the development and follow-up of documents, guidelines etc

Survey – expert meeting
Patient organizations – Expert groups

Draft of leaflets, guidelines

Validation by the community

(Translation)

Distribution to Member States
Roadshows



Disease specific best practice guidelines

UNIT 1: QUALITY MANAGEMENT AND ACCREDITATION / CERTIFICATION OF GENETIC TESTING

[Disease specific guidelines](#)
[Non disease specific guidelines](#)
[List of institutions](#)

Qm

Short Glossary

[ACMG](#) : American College of Medical Genetics
[ASMG](#) : Association of the Scientific Medical Societies in Germany
[BvDG](#) : Professional Association of German Human Geneticists
[CF Network](#) : Cystic Fibrosis European Network
[CMGS](#) : Clinical Molecular Genetics Society
[EMQN](#) : The European Molecular Genetics Quality Network
[SfMG](#) : Swiss Society of Medical Genetics

BEST PRACTICE GUIDELINES FOR MOLECULAR GENETIC TESTING

Disease-specific guidelines

Disease	Date	Updated	Institution	Author	Comments	Language
Duchenne/Tourette disease	200		EMQN	Raatschenski E, Luzzi J, Tenenbaum Y		German
	1997	2004	ACMG	Raatschenski E, Nela E		German
Cystic Fibrosis	1991	200	EMQN	Boves M	1st edition archives *	English
	2002	2002	ACMG	Arns J, Fedner D, Wayne M		English
	1997	2002	EMQN	Stuhmann-Spannering H, Aulenta-Scholz C		German
	1997	2002	EMQN	Stuhmann-Spannering H, Aulenta-Scholz C		German
	2002		CF Network	Dequaker E, Coppens H, Dolsa J		English
Cystic Fibrosis Carrier Screening	2002		ACMG	Shay Wayne M, Cutting Gary R, Klinger K		English
Falco V Lysin	2004	2002	ACMG	Spicer E, Draly W, Watson C		English
Phenylketonuria Prevalence	1993	200	EMQN	McDonnell F	1st edition archives *	English
Fragile X Syndrome	2001	2002	EMQN	Barcelina Y, Bardach P, Barrowe D		English
	2002		EMQN	Wapleson J, Sawyer H		English
	1992	200	EMQN	Barton D	1st edition archives *	English

Reference materials for molecular genetic testing

Quality Control and Reference Material Producers websites

Name	Description
NIBSC	<p>NIBSC strategic aims:</p> <ul style="list-style-type: none"> • To respond to and advise on public health problems involving biologicals; • To provide a national scientific capacity in the field of biological medicine, and to maintain the flexibility, expertise and facilities needed to address new developments in science and medicine; • To operate, and be recognised, as a leading international authority on methods of assay such as those to quantify biological activity and to characterise and assess the safety of biologicals; • To maintain a central role in the development of the scientific basis for control and standardisation of biologicals within Europe; • To assist in the development of international consensus on scientific aspects of the regulation of biological medicines and, in this respect, work closely with the World Health Organisation; • To achieve and maintain Quality Accreditation in key areas of control and standardisation.
Coriell	<p>The Coriell Institute for Medical Research is an independent, non-profit research organization dedicated to understanding human genetic diseases and providing the highest quality genetic resources. In addition to conducting its own research in cancer (including breast cancer and leukaemia), human genetic variation, mechanisms of cellular differentiation, and other genetic disorders, the Coriell Institute for Medical Research also serves the entire scientific community by maintaining the world's largest collection of human cells for research. The identification of genes associated with Huntington Disease, cystic fibrosis, Alzheimer Disease, Down Syndrome and a severe form of manic depression, among many others, relied on cells from these collections.</p>
IRMM	<p>The Institute for Reference Materials and Measurements (IRMM) is one of the seven institutes of the Joint Research Centre (JRC), a Directorate-General of the European Commission (EC). The mission of the IRMM is to promote a common and reliable European measurement system in support of EU policies.</p> <p>The prime objective of the IRMM is to build confidence in the comparability of measurements by the production and dissemination of internationally accepted quality assurance tools, including validated methods, reference materials, reference measurements, interlaboratory comparisons and training.</p>
LGC	<p>LGC Promochem is Europe's most comprehensive source of reference materials. The UK's leading independent provider of analytical and diagnostic services, LGC offers chemical, biochemical and forensic analysis, DNA testing and genetic screening, research, method validation, consultancy and analytical outsourcing. LGC also distributes reference standards and ATCC biomaterials throughout Europe.</p>
	<p>BAM is the German Federal Institute for Materials Research and Testing. Its competences are to improve safety and reliability in chemical and materials technologies through research and</p>



Research and emerging technologies

Aim:

- To support and to guide the implementation of new technologies into diagnostic application.
- Set up a rigorous test evaluation program, with Beta testing in accredited laboratories, on selected clinical samples, to ease introduction of new technologies into the European diagnostic laboratories.

DNA extraction methods, MLPA, Conformation Sensitive Capillary Electrophoresis, High Resolution Melting Curve Analysis, Pyrophosphorolysis-Activated Polymerization

Long term goal:

Constitute a Network of Excellent laboratories (NoE) and SME's for Beta testing of new technologies following established procedures.



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- acting Networks

- VS & EVENTS
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- onal Details
- ut bakkee

UNIT 5 : RESEARCH AND EMERGING TECHNOLOGIES

Nt

SIMPLE SEARCH

SEARCH

(searches on all fields)

ADVANCED SEARCH

Filter

Abbreviation

Full name

Aliases

+ Legal Issues

PAGE

1

MAX RECORDS

25

RESULTS

19

NEW TECHNOLOGIES

Abbreviation	Full name	Aliases
ARMS	Amplification refractory mutation system	Allele-specific PCR (ASPCR) / PCR amplification of specific alleles (PASA)
ASO	Allele-Specific Oligonucleotide hybridization	Allele Specific Hybridization
CCM	Chemical Cleavage of Mismatch	Chemical Mismatch Cleavage, Amplification Mismatch Detection (AMD), HOT
DGGE	Denaturing Gradient Gel Electrophoresis	Constant denaturant gel-elpho (CDGE), perpendicular denaturing gradient gel-elpho
DHPLC	Denaturing High-Performance Liquid Chromatography	temperature-modulated heteroduplex analysis (TMHA)
FAMA	Fluorescence-assisted mismatch analysis	none
fiber-FISH	fiber-Fluorescence In Situ Hybridization	
FISH	Fluorescence In Situ Hybridisation	
HA	Heteroduplex analysis	Slab gel heteroduplex analysis (HDA)
Invader	Invader	Invasive cleavage
MAPH	Multiplex Amplifiable Probe Hybridisation	
MCA	Melting Curve Analysis	High-resolution melting curve analysis (HRMA), High-resolution DNA melting curve analysis
MLPA	Multiplex Ligation-dependent Probe Amplification	-
PAP	Pyrophosphorolysis-Activated Polymerization	PAP-A, BI-PAP (variant of PAP in which both primers are blocked at their 3' terminus with a ddNTP)
PCR	Polymerase Chain Reaction	Multiplex PCR (variant of PCR; using several primer pairs simultaneously amplify multiple sequences)
PTT	Protein Truncation Test	IVSP - In Vitro Synthesized Protein assay
RNAse Cl	RNAse cleavage	Non-isotopic RNAse cleavage assay (Nirca)

Mutalyzer - Sequence variant nomenclature check V1.0.1

Please insert the mutation specifications below:

Reference Insert a GenBank accession entry, or a GI number

Sequence Type Insert a gene symbol that is present in the Record AND has a CDS

Gene symbol Insert a variant number that is present in the Record

Variant (optional)

Start Position

End Position

Mutation Type

Old Sequence Insert a sequence. If you insert a sequence for a cDNA, which is coded by reverse complement, enter reverse complement code.

New Sequence

Comment

www.lovd.nl/mutalyzer

Mutalyzer - Sequence variant nomenclature check V1.0.1

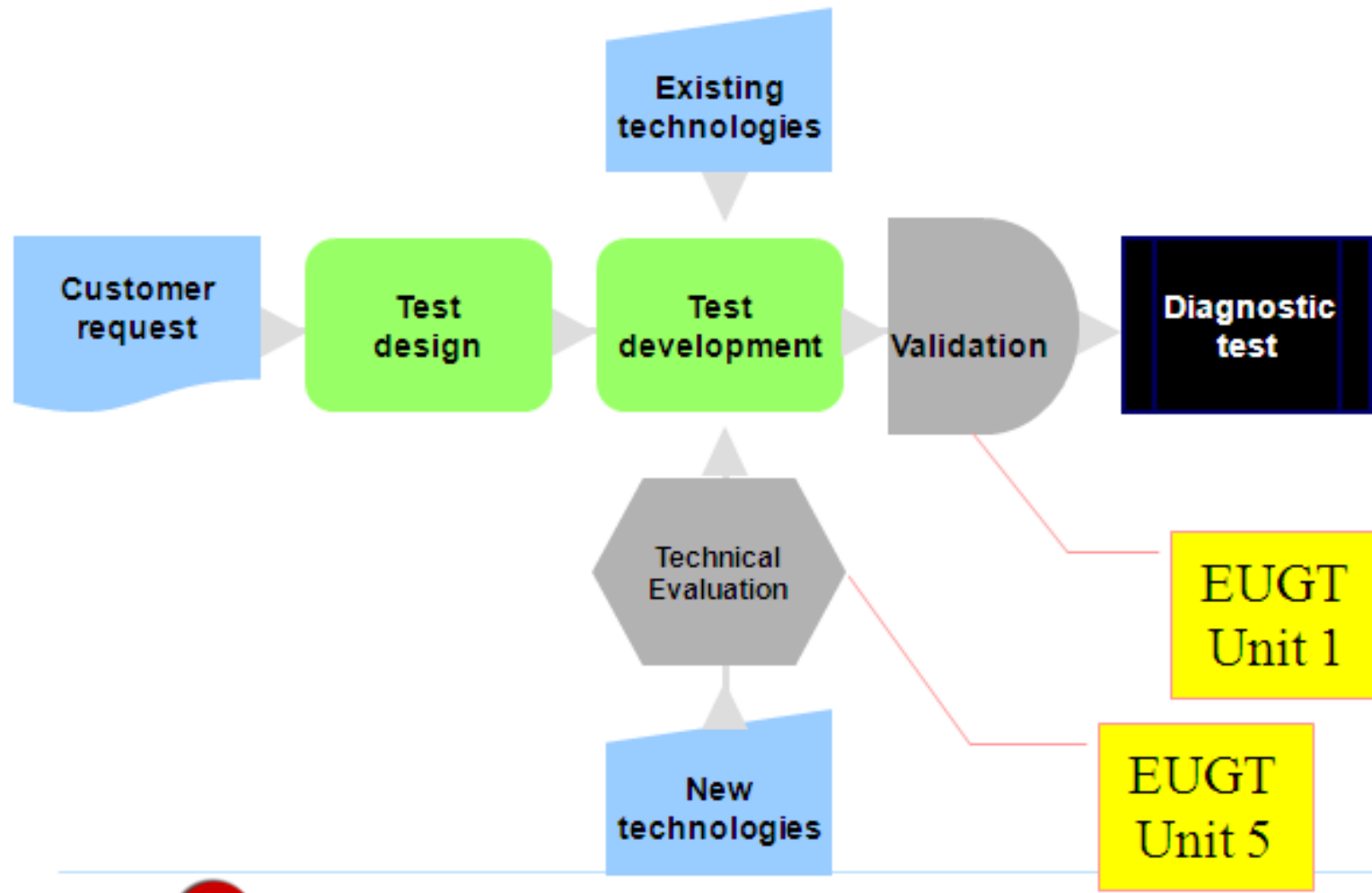
Please insert the mutation name using the format:
 <Accession Number>.<version number>(<Gene symbol>);<sequence type>.<mutation>

Example: AB026906.1:c.274G>T

Mutation Name

AL449423.14(CDKN2A_v002):c.5delA

What is validating a test ?



Definitions

- **"Demonstrate that the test is fit for the intended purpose"**
- ISO/CEI 17025 and 15189
- ""
 - "The confirmation by examination and provision of **objective evidence** that the particular requirements for a specific intended use are fulfilled"
 - "the validation shall be **as extensive as is necessary** to meet the needs of the given application"
 - "The methods and procedures selected for use shall be evaluated and **found to give satisfactory results before being used for medical examinations**".

What to validate?

- **Precision**
 - Repeatability
 - Ability to provide closely **similar results**, for repeated tests under the **same conditions**
 - Reproducibility
 - Ability to provide closely **similar results**, for repeated tests under **different conditions**
- **Ruggedness**
 - If relevant
 - Resistance to **changed conditions**
- **Uncertainty of measurement (mol. genetics becomes quantitative !)**
 - Required if the client requests it;
when it may be relevant to the validity or application of the results
when it may affect compliance to a specification limit
 - **'Estimation'** not **'Determination'** or **'Quantitation'**



Practically: “Validation”

- Repeatability
 - 10 different samples (normal + mutant)
 - 2 samples, tested 10 times
- Reproducibility
 - 2 samples, 10 times, on 3 different days
 - Same 2 samples, 10 times, by a second person
- Ruggedness
 - Adjust to the test
 - Different DNA concentrations?
 - Include “difficult” samples (uncultured amniocytes...)?
 - Different types of PCR machine and/or genescanner
 - Prealiquotted or diluted primers?
 - Frozen aliquots?

The devil is in the detail and... statistics



Clinical validity and utility of genetic testing

- Defining **clinical utility** criteria of genetic testing
- Investigating access to and utilisation of genetic services in EU
- Initiating an international collaborative network in order to promote internationally shared quality standards for clinical utility assessment
- Investigating criteria and mechanisms to maximize the output of genetic services, given limited resources
- Validate and translate the **utility guidelines** developed by the GSHG

Genetic Counselling

- Aim to improve the quality of genetic counselling

Specific Support Action to integrate partners from developing countries

“CAPABILITY Project” (2007-2009)



EuroGentest

Clinical genetics and community genetics

- Summary of Guidelines for Genetic Counselling
- What counselling in different testing categories?
- Who is qualified to give counselling?
- What aspects of the "ideal genetic counselling" should be included in genetic counselling in real life?



What is genetic testing?

	Context			Type of testing							Phenotypes			Mut	Object									
	Medical applications	Scientific research	Non-medical applications	Diagnostic testing	Presymptomatic testing	Disease prediction testing	Drug response	Carrier testing	Prenatal testing	PGD	Population screening	Paternity testing	Criminal identification	Mendelian trait	Gene prediction	Other polymorphic traits	Sequence variants (vs. germline only)	Chromosomes	Genes/DNA	Specific gene products	Routine blood test	Physiologom	Family history	
EC	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	○	○	
CoE	+	○	○	+	+	+	○	+	+	+	+	○	○	+	+	○	○	+	+	+	+	+	+	+
CDBI	+	+	○	+	+	+	+	+	+	+	+	○	○	+	+	○	○	+	+	+	○	+	+	
IPTS	+	○	○	+	+	+	+	○	+	+	+	○	○	+	+	○	○	+	+	+	+	+	+	○
ESHG	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	○	+	+	+	+	+	+	+
ASHG	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	○	+	+	+	○	○	○
OECD	+	○	○	+	+	+	+	○	+	+	+	○	○	+	+	○	○	+	+	+	+	+	+	○
WHO	+	○	○	+	+	+	+	+	+	+	○	○	○	+	+	○	○	○	+	○	○	○	○	○
UNESCO	+	+	○	+	+	+	+	+	+	+	+	○	○	+	+	○	○	+	+	+	○	○	○	○
CDC	+	○	○	+	+	+	+	○	+	+	○	○	○	+	+	○	+	+	+	○	○	○	○	○
NIH	+	○	○	+	+	+	+	+	+	+	+	○	○	+	+	○	+	+	+	+	○	○	○	○
HGP	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	○	+	+	+	○	○	○	○
Genetic Alliance	+	+	○	+	+	+	○	+	+	+	+	○	○	+	+	○	○	+	+	+	+	○	○	○
UK Cancer Incub. (UK)	+	○	○	+	+	+	○	○	+	+	○	○	○	+	+	○	+	○	+	○	○	○	○	○
NAHU	+	○	○	+	+	+	+	+	+	+	+	○	○	+	+	○	+	+	+	+	○	○	○	○
ACLU/ NTPOLW	+	○	+	+	+	+	+	+	+	+	+	+	+	+	+	+	○	+	+	+	+	+	+	+
CAP	+	○	○	+	+	+	○	+	+	+	+	○	○	+	+	○	○	+	+	○	○	○	○	○
ACGT	+	+	○	+	+	+	+	+	+	+	+	○	○	+	+	○	○	+	+	+	○	+	○	○
Nuffield	+	+	○	+	+	+	+	+	+	+	+	○	○	+	+	○	○	+	+	+	○	+	+	+



Ethical and Legal Issues

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**PATIENT RIGHTS
IN THE EU**



EUROPEAN ETHICAL - LEGAL PAPERS N° 1



**PATIENT RIGHTS
IN THE EU**



EUROPEAN ETHICAL - LEGAL PAPERS N° 2



Education and patient issues

- Updated list of National **Societies** of Genetics and others relevant ones
- List of European and National **genetic courses**
- List of other **educational material** and list of relevant institutions involved in genetics education
- Network of **national contact persons** in the European countries
- 1st Workshop on “Patient and Professional Perspective of Genetic Information/Education in Europe”
- **Core competences** for geneticists and allied health professionals



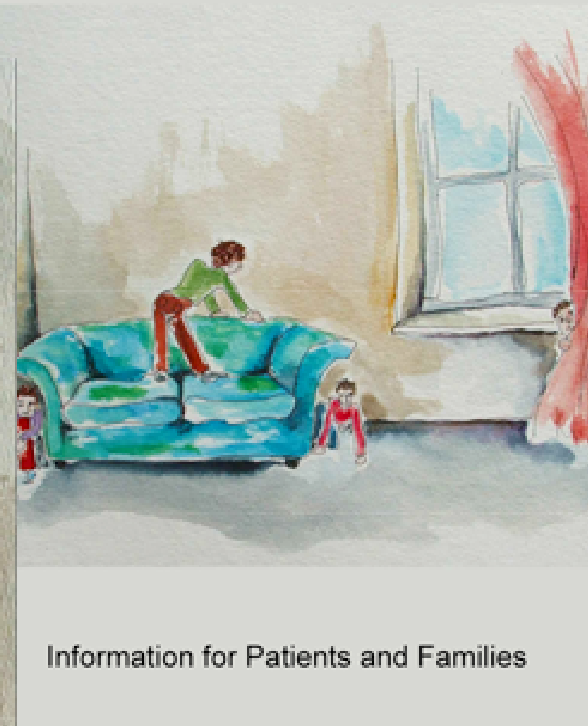
Developing informative guides

What is a Genetic Test?



Information for Patients and Families

Frequently Asked Questions



Information for Patients and Families

The Amniocentesis Test



Information for Patients and Families



Disseminating information: E-News

EuroGentest e-news

EuroGentest Harmonising genetic testing across Europe

Actions speak louder than words

Spreading the quality message
Our aim is to help all categories of genetic laboratories fully meet accreditation, certification and licencing criteria.

Already we have held several Expert Workshops on Laboratory Accreditation, which were well-subscribed. This work continues in 2007 with a further two workshops and Round-table sessions at the European Society of Human Genetics on specific, quality related, topics.

Inspiring professionals, reassuring patients and families

EuroGentest is a Network of Excellence funded by the EU to promote the harmonisation and improvement of genetic testing standards. After 4 years, the impact is more than meeting expectations. Our underlying actions range from coordinated and structured activities such as the production and evaluation of new testing methods to the organisation of training courses for genetic information professionals. Both activities will continue with ongoing projects and activities. Our first round-table sessions have enhanced the project in an even greater way. A European project can occur in many ways.

What can EuroGentest offer you?

Geneticists
The opportunity to join one of the most exciting groups

Patients
Reliability providing immediate evidence

Industry
A common evidence network for your new technologies

Legislators
Expert advice on ensuring the quality of your genetic actions

Patients and families
A comprehensive database of European genetic testing resources

Genetic testing in Europe

Facts and figures
No. of testing centres in Europe - 1000

Number of conditions tested - 1000

Annual growth in testing - 10-200%

Number of people in the EU suffering from genetic disease - 100 million

Over 100 health care professionals

Project coordinator:
Professor Jean-Jacques Cassiman

Holder of the Chair for Genetic Testing and the Director of the Center for Human Genetics in Leuven, Belgium.

For more information please visit www.eurogentest.org

EuroGentest e-news

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NOVEMBER 2008

19



Train The Trainer Workshop (WP1.8)

Belgium

October 2008

9 - 10



Workshop - Fulfilling the requirements of ISO 15189 for management review, internal quality control and external quality assessment (WP1.8)

Germany

Want to learn more on how to do a management review, on how to introduce internal quality controls in the lab or what participating in EQA means?

May 2008

31



Round table sessions - Case studies on quality assurance and quality control issues in genetic testing laboratories (WP1.8)

Spain

Discuss in small groups about concrete situations related to quality processes in genetic testing labs. Meet colleagues faced with similar challenges and problems.

30 - 31



Workshop - Towards accreditation: Managing the human side of change (WP1.8)

Spain

This workshop will tackle the human side of change. How do you motivate the lab to implement a new quality system? How do you manage the human side of this process of change?

30 - 31



Workshop - Accreditation in genetic testing laboratories (WP1.8)

Spain

Compare and share experiences of implementing and living with quality systems. Examine cases of concrete situations related to quality processes like non-conformities, reporting and training.

26



Training - Quality management in your laboratory: How you can get involved? (Dutch speaking) (WP1.8)

Belgium

Practical exercises, applied to the own works situation. Sharing experiences with colleagues and experts. Topics include the elements of a quality management system, aspects of ISO 15189 standard, non-conformities, ...

February 2008

7 - 8



Workshop - Towards accreditation: Managing the human side of change (WP1.8)

France

This new topic in a series of workshops related to quality management will tackle the human side of change. How do you motivate the laboratory to implement a new quality system? How do you manage the human side of this process of change?



Acknowledgements



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EuGT partners

