



**Genetic testing in Europe:
a network for test development,
harmonization, validation and
standardization of services**

**Capability 1st Workshop, Trollenholm
Sweden, May 7-9, 2007**

**Milan Macek
Charles University, Prague**

Co-ordinator: Prof. Jean-Jacques Cassiman, KU, Leuven, Belgium



By Rebecca Kent



Quality issues in Europe

More than 1000 laboratories/centers in different settings

More than 1000 rare diseases can be tested

Lack of centralized and uniform information about services

Limited networking

Lack of harmonized and standardized EQA

Lack of reference materials

Limited number of accredited labs

Insufficient counselling (quantity and sometime even quality)

Source: IPTS/JRC-EC: Ibarreta et al. Towards quality assurance and harmonization of genetic testing services in the EU.

Report EUR20977, 2003

Major aims of the NoE “EUROGENTEST”

1. Harmonization and quality improvement of genetic services
2. Define the European dimensions which are compatible with and complementary to National approaches (reflecting historical and cultural differences)

EuroGentest - Project Activities - Microsoft Internet Explorer

Bestand Bewerken Beeld Favorieten Extra Help Koppelingen »

EuroGentest Genetic testing in Europe
Network for test development, harmonization, validation and standardization

Quality Management Information Databases Public Health Ethical & Legal New Technologies Education Search

MENU

- Homepage
- About EuroGentest +
- Project Management
- Fellowships/Jobs
- Genetic Support Groups
- Interacting Networks
- Calls

NEWS & EVENTS

- Current News +
- Previous News
- Newsletter Archive
- Meetings
- Press Releases

REGISTER

Find out more about the world of Genetic Testing!

LOGIN

Username

Password

Remember password

LOGIN [Forgotten password?](#)

Project Activities


Welcome to the EuroGentest website. We aim to provide you up to date general information on genetic testing in Europe, together with specific insight on how we are working to harmonise standards across the EU. Whatever your particular interest, click on the appropriate square to find out more.

Unit 1 Qm Quality Management	Unit 2 Id Information Databases	Unit 3 Ph Public Health	SSN-Capability
Unit 4 Et Ethics & Legal	Unit 5 Nt New Technologies	Unit 6 Ed Education	

Genetic testing touches us all

As science unravels the genetic basis of disease, the opportunities offered by increased genetic testing become more and more apparent. EuroGentest is an EU-funded project that aims to ensure these opportunities are secured for the benefit of all.

EuroGentest is organised as a Network of Excellence with 6 Units looking at all aspects of genetic testing – Quality Management, Information Databases, Public Health, New Technologies and Education. Through a series of initiatives EuroGentest encourages the harmonisation of standards and practice in



Testing for Genetic Disorders

Because of the rarity of many disorders and the complexity of tests, it is more and more necessary to **outsource samples** for testing:

But how can you find laboratories, and how can you know their quality?

- e.g. which are accredited?
- which participate regularly in external quality assessment (EQA) schemes?
- This information is important:
 - for **consumers**, to facilitate an informed choice of a quality partner;
 - for the **general public** and politicians, to educate them about the importance given to quality issues by our profession;
 - for the **laboratories and quality institutions**, to publicize and reward their efforts and investment in quality assurance.

Quality management and accreditation - Unit 1



Genetic testing in Europe
Network for test development, harmonization, validation and standardization



Quality Management
Information Databases
Public Health
Ethical & Legal
New Technologies
EducationSearch

MENU

- Homepage
- About EuroGentest
- Project Management
- Fellowships/Jobs
- Research Projects
- Genetic Support Groups
- Patient Information
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UNIT 1 : QUALITY MANAGEMENT AND ACCREDITATION / CERTIFICATION OF GENETIC TESTING



New in Quality Management (Unit 1)

- [Guidelines for Molecular Genetic Testing](#)
- [Current scope of Cytogenetics EQA schemes and key recommendations for harmonization of EQA in Europe](#)
- [National Representatives for Biochemical Genetic Testing](#)
- [First open genetic testing accreditation workshop proves a major success](#)
- [Cytogenetic EQA Report](#)

NEWS & EVENTS

Current News -

12-04-2007
EUGT Partner has article published in DS News

11-04-2007
Patient Information Booklets Now Available

05-04-2007
List of European National Contacts for Cytogenetics added to Unit 1 Website

16-03-2007

<p style="text-align: center;"><u>Quality Management</u></p> <ul style="list-style-type: none"> Quality Management System Accreditation Guidelines and standards related to QM Education Workshops Events 	<p style="text-align: center;"><u>EQA</u></p> <p><u>Molecular</u></p> <ul style="list-style-type: none"> Documents Events <p><u>Cytogenetics</u></p> <ul style="list-style-type: none"> Scope Events <p><u>Biochemical</u></p> <ul style="list-style-type: none"> Documents Events 	<p style="text-align: center;"><u>Quality Control Materials</u></p> <ul style="list-style-type: none"> Terminology Prioritization Events IVD Directive Resources
<p style="text-align: center;"><u>Validation</u></p> <ul style="list-style-type: none"> Diagnostic Technical 	<p style="text-align: center;"><u>QAu Databases</u></p> <ul style="list-style-type: none"> QAu Database European QAu Survey International laboratory databases 	<p style="text-align: center;"><u>Documents</u></p> <ul style="list-style-type: none"> Surveys Posters Publications

Best practice guidelines

National Genetics Reference Laboratories
 SUPPORTING THE GENETIC SERVICE NETWORK

Search: 

WELCOME TO THE JOINT WEB SITE OF THE NATIONAL GENETICS REFERENCE LABORATORIES

BASED IN MANCHESTER AND WESSEX (SALISBURY & SOUTHAMPTON)




EuroGentest Genetic testing in Europe
 Network for test development, harmonization, validation and standardization

Quality Management | Information Databases | Public Health | Ethical & Legal | New Technologies

EMQN 

The European Molecular Genetics Quality Network

- MENU**
- Homepage
 - About EuroGentest
 - Project Management
 - Fellowships/Jobs
 - Research Projects
 - Genetic Support Groups
 - Patient Information
 - Interacting Networks
 - Calls

UNIT 1 : QUALITY MANAGEMENT AND ACCREDITATION / CERTIFICATION OF GENETIC

Qm

- Disease-specific guidelines
- Non disease-specific guidelines
- List of institutions

Short Glossary

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

- NEWS & EVENTS**
- Current News
- 12-04-2007
EUGT Partner has article published in DS News
 - 11-04-2007
Patient Information Booklets Now Available
 - 05-04-2007
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 - 16-03-2007

BEST PRACTICE GUIDELINES FOR MOLECULAR GENETIC TESTING

Disease-specific guidelines

Disease	Date	Updated	Institution	Author	Comments	Language
Charcot-Marie-Tooth disease	2000		BVDH	Rautenstraß B., Lupski J., Timmerman V.		German
	1997	2004	AWMF	Rautenstraß B., Nelis E.		German
Cystic Fibrosis	1991	1996	CMGS	Schwarz M.	1st edition archives.*	English
	2002	2006	ACMG	Amos J., Feldman G., Wayne W.		English
	1997	2006	AWMF	Stuhrmann-Spangenberg M., Aulehla-		German



The European Molecular Genetics Quality Network

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Draft Best Practice Guidelines for Molecular Analysis in Fragile X Syndrome

Draft guidelines prepared by Valérie Biancalana, Peter Steinbach and Su Stenhouse following discussions at the EMQN workshop, 23-24 November 2001, Strasbourg, France. **These guidelines have been fully updated and were published in January 2006**

Address for correspondance :Valérie Biancalana, Laboratoire de Diagnostic Génétique, Faculté de Médecine CHRU, 11 rue Humann, 67085 Strasbourg Cedex, France. Email : valerie.biancalana@chru-strasbourg.fr

Unit 2



An encyclopedia

- ◆ About 1800 diseases
- ◆ 662 review articles English and French
- ◆ 1800 summaries in 6 languages
- ◆ Expert- authored
- ◆ Peer-reviewed:
International board
- ◆ **Free access, online**
publication: OJRD
- ◆ Partnership with EJHG


A directory of services

- ◆ 2,227 expert clinics
- ◆ 1,094 clinical labs
- ◆ 1,751 research labs
- ◆ 529 clinical trials
- ◆ 3,696 research projects
- ◆ 220 registries
- ◆ 367 networks
- ◆ 1,381 advocacy groups
- ◆ 7,920 professionals

Quality Assurance Database (from VI/2007)

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

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

SIMPLE SEARCH  

FILTERS

Filter

Country City

Laboratory/Institution Professional

EQA provider Accreditation body

Accredited Certified EQA participating

SEARCH

CLINICAL LABORATORIES

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

A	C	E	Country	City	Laboratory	Institution	Labdirector
<input checked="" type="checkbox"/>	<input checked="" type="checkbox"/>	<input checked="" type="checkbox"/>	SWITZERLAND				
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<input checked="" type="checkbox"/>	<input checked="" type="checkbox"/>	<input checked="" type="checkbox"/>	SWITZERLAND				
<input checked="" type="checkbox"/>	<input checked="" type="checkbox"/>	<input checked="" type="checkbox"/>	SWITZERLAND	Geneva	Laboratoire de Diagnostic Moléculaire	Hôpitaux Universitaires de Genève	Morris, Michael

Certification ISO 2001

Accreditation ISO 15189

Forms under development

QAu Survey 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

To assist us in processing your data, please note that the yellow fields are required.

Quality Management screen 2 of 4

EQA (External Quality Assessment) participation

Do you participate in EQA schemes? Yes No

If yes, please specify (for the last two years?)
(Please list all schemes you perform for each provider; only participation and not success or failure is requested).

Year	Activity	Provider	Schemes
<input type="text"/>	<input type="text"/>	<input type="text"/>	<input type="text"/>

Accreditation / Certification / Agrément

Is your laboratory accredited / certified? Yes Underway No

Accreditation

Is your laboratory accredited? Yes Underway No

If yes or underway, by which accreditation body?

Since when?

Under which norm?

What is your accreditation number?

For what kind of activity Molecular Genetics Cytogenetics Biochemical Genetics

Certification / Agrément

Is your laboratory certified? Yes Underway No

If yes or underway, by which certification body?

Since when?

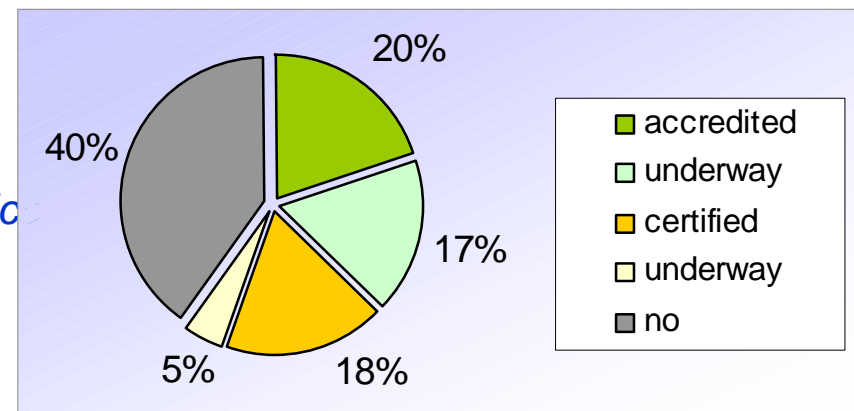
Under which norm?

What is your certification number?

For what kind of activity Molecular Genetics Cytogenetics Biochemical Genetics

- **Key presentations:**

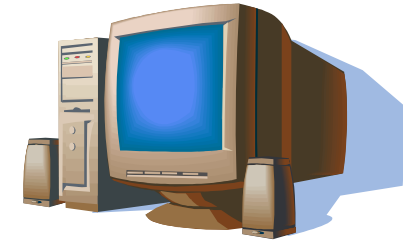
- *European Society of Human Genetics, Amsterdam*
- *International Congress of Human Geneticists, Brisbane*



Cytogenetics EQA - CEQA



- **On line EQA – feasibility and viability demonstrated**
 - Feasibility initially tested on UK NEQAS participants
 - Two pilots and then extended to routine EQAs
 - Subsequent improvements to the website
 - Administrative interface improved
 - Library of FISH images/EQA cases being created
- **First pilot European on line EQA for Cytogenetics, CEQA**
 - 25 countries registered, one postnatal and prenatal EQA case
 - European assessors/experts meeting
 - Summary report prepared - now with assessors for validation
 - EQA templates/registration documents revised for incorporation onto the line management system



Directory of EQA Schemes and Providers

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

Qm

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

Authors

Prof. Clemens Müller
Dr. Rob Elles

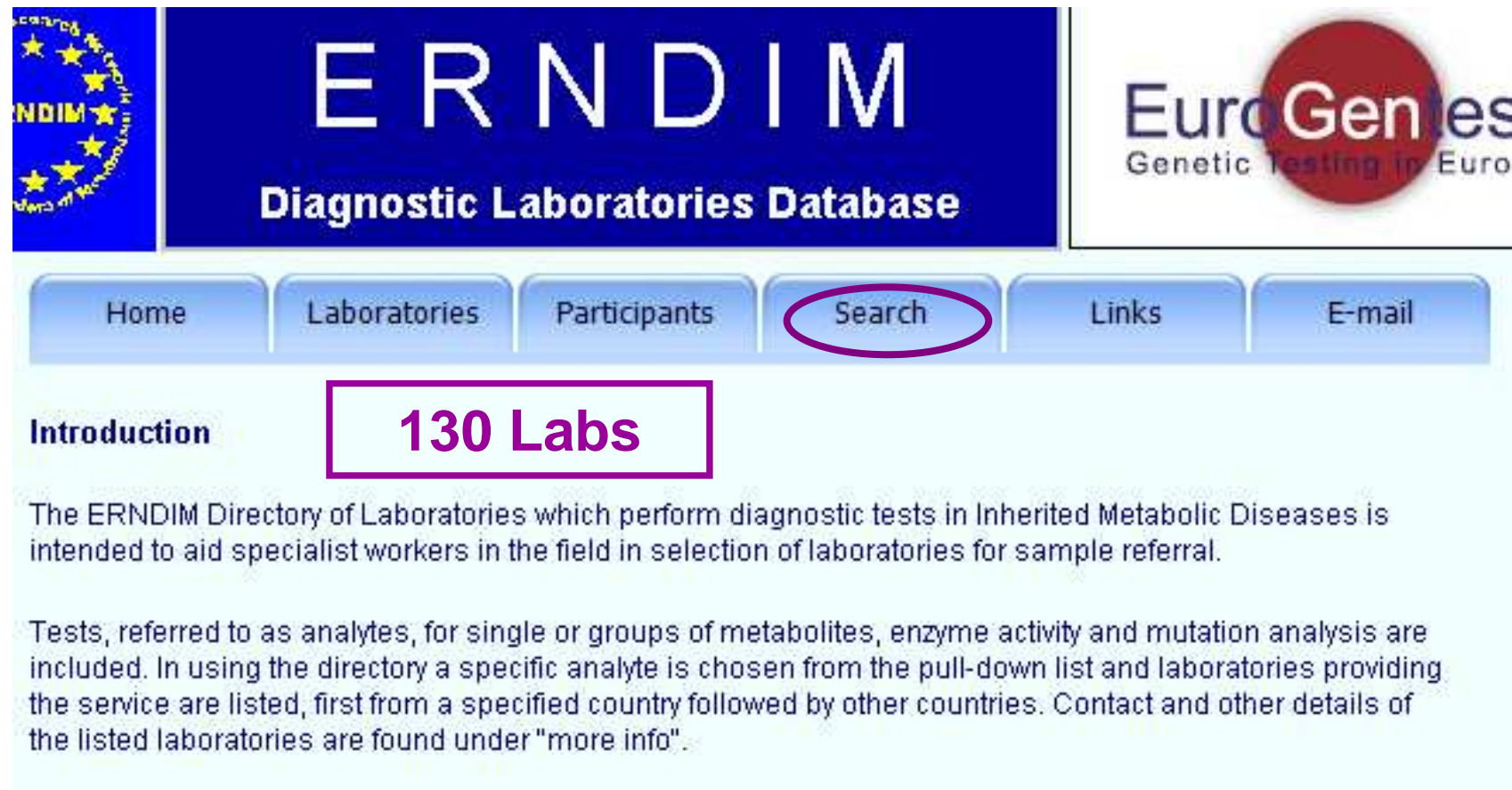
MOLECULAR GENETIC TESTING - EXTERNAL QUALITY ASSESSMENT SCHEME PROVISION (2005)

I. Constitutional mutations

Disorder	Organisation	Contact Point
1. ACE (Angiotensin I Converting Enzyme)	DGKL ² (RfB) ¹²	M. Neumaier, Mannheim
	ÖQUASTA ⁴ (in collaboration with DGKL)	W.-M. Halbmayer, Ch. Mannhalter, Wien
	ECAT Fdn. ⁷ (in collaboration with DGKL)	P. Meijer, Leiden
	LABQUALITY ¹⁶	M. Keinanen, Helsinki
2. alpha-1-Antitrypsin (PiM, PiS, PiZ); (RV Nr.743)	INSTAND ¹¹	R.-R. Flörke, Düsseldorf
3. alpha-1-Proteinase-Inhibitor (M/S/Z)	DGKL ² (RfB) ¹²	M. Neumaier, Mannheim
	ÖQUASTA ⁴ (in collaboration with DGKL)	W.-M. Halbmayer, Ch. Mannhalter, Wien
	ECAT Fdn. ⁷ (in collaboration with DGKL)	P. Meijer, Leiden
4. Apolipoprotein B100 (ApoB100)	DGKL ² (RfB) ¹²	M. Neumaier, Mannheim
	ÖQUASTA ⁴ (in collaboration with DGKL)	W.-M. Halbmayer, Ch. Mannhalter, Wien
	ECAT Fdn. ⁷ (in collaboration with DGKL)	P. Meijer, Leiden
5. Apolipoprotein E (ApoE): E2/E3/E4	DGKL ² (RfB) ¹²	M. Neumaier, Mannheim
	ÖQUASTA ⁴ (in collaboration with DGKL)	W.-M. Halbmayer, Ch. Mannhalter, Wien
	ECAT Fdn. ⁷ (in collaboration with DGKL)	P. Meijer, Leiden
	LABQUALITY ¹⁶	M. Keinanen, Helsinki
6. Apolipoprotein E (E2, E3, E4); (RV Nr.744)	INSTAND ¹¹	R.-R. Flörke, Düsseldorf
7. Azoospermia (AZF/DAZ)	EAA ⁶ /EMQN ¹⁰	M. Simoni, Münster
8. Breast/ovarian cancer familial (BRCA1, BRCA2)	EMQN ¹⁰	S. Patton, Manchester

Directory of Biochemical Genetics Laboratories

<http://www.erndim.unibas.ch>



ERNDIM
Diagnostic Laboratories Database

EuroGenes
Genetic Testing in Europe

Home Laboratories Participants **Search** Links E-mail

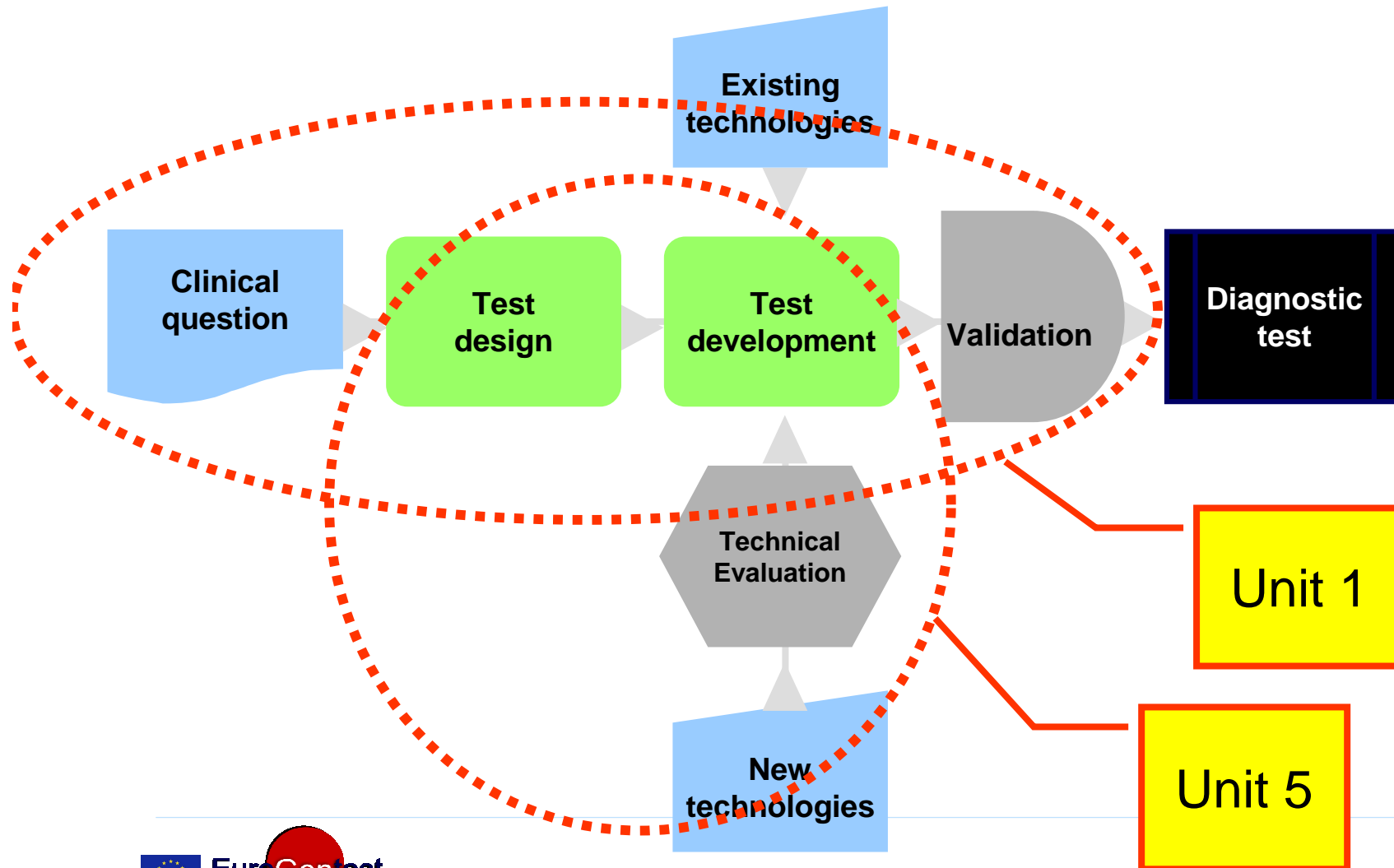
Introduction **130 Labs**

The ERNDIM Directory of Laboratories which perform diagnostic tests in Inherited Metabolic Diseases is intended to aid specialist workers in the field in selection of laboratories for sample referral.

Tests, referred to as analytes, for single or groups of metabolites, enzyme activity and mutation analysis are included. In using the directory a specific analyte is chosen from the pull-down list and laboratories providing the service are listed, first from a specified country followed by other countries. Contact and other details of the listed laboratories are found under "more info".

In collaboration Orphanet

Validation aspects of existing and new technologies – an interplay of Units 1 and Unit 5



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UNIT 5 : RESEARCH AND EMERGING TECHNOLOGIES

Nt

SIMPLE SEARCH

SEARCH (searches on all fields)

Filter
+ Legal Issues

Abbreviation

Full name

Aliases

PAGE
1
MAX RECORDS

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)

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ADVANCED SEARCH

Filter

Abbreviation

Full name

Aliases

NEW TECHNOLOGY: MCA

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[<< Back](#) | [\[Edit record\]](#) | [\[Create record\]](#) | [\[Drop record\]](#)

ID	3
Abbreviation	MCA
Full name	Melting Curve Analysis
Aliases	High-resolution melting curve analysis (HRMA), High-resolution DNA melting curve analysis
Description	A technique to scan genes or sequences of interest for mutations, e.g. single nucleotide polymorphisms or indels using differences in melting curves. Method: 1. Amplification of the target gene area is achieved by rapid cycle PCR using specific forward and reverse primers surrounding the area of interest and LC green saturating dye to generate a PCR product around 200-300 bp, 2. After PCR, the amplified fragments are melted at 0.1-0.3 °C/s in high-resolution melting instrument, 3. The fluorescence melting curves are recorded and analysed with HR-1 analysis software.
Scheme	http://www.idahotech.com/LifeScience/HR-App-Data.htm
Sample type	DNA RNA
Other sample type	
Sample amount	20-50ng/ul
Sample purity	requires accurate PCR amplification
Sample contaminants	dsDNA (primer dimers, non-specific products)
Hazardous constituents	No
Strong points	Rapidly establishment of identity of highly polymorphic loci; kind of mutation can be deduced from derivative plot; potential to detect most homozygous variants; no temperature optimization required; fast, inexpensive and easy-to-handle technique; close-tube system; High Throughput Screening (HTS); PCR fragments after melting can directly be used in sequencing reactions.
Weak points	Does not sequence or determine specific alleles
Part of certified protocol	No
Sensitivity	95% - 100%
Analysis equipment	melting analysis instruments (HR-1 or Lightcycler (HR 96/384)

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)*

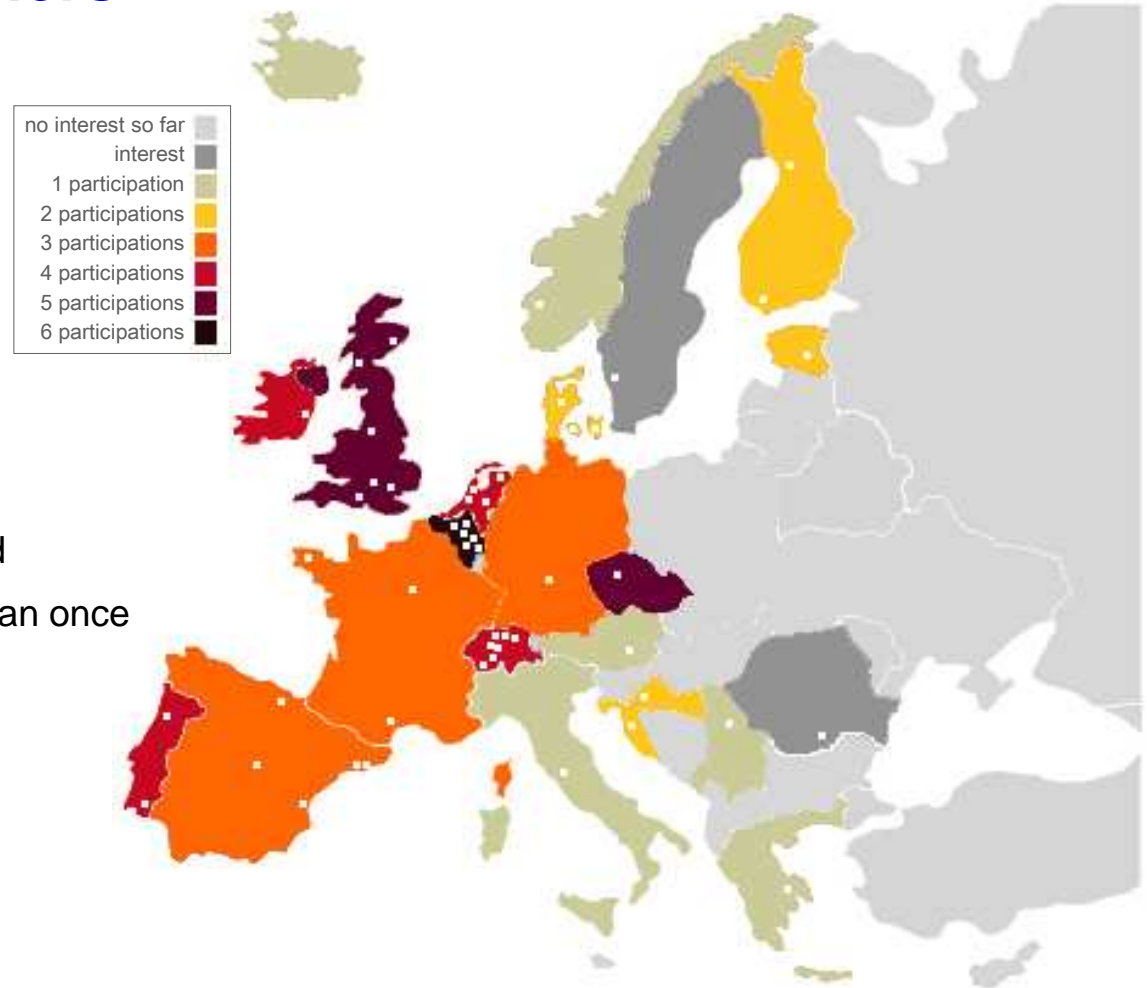
- **First comprehensive, independent survey** of services, current practice and QAu status of European PGD providers
- Identification of providers:
 - Centres, clinics and labs in 17 countries
- Surveyed for
 - All PGD cycles performed in 2006
 - Including familial chromosomal anomalies, over 50 monogenic 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

Expert workshops and training: a lot of interest, also from non-EuroGentest partners

6 workshops organized

- 20 different countries participated
- 56 different laboratories participated
- 16 laboratories participated more than once

www.eurogentest.org



Unit 3

Clinical validity and utility of genetic testing

Defining **clinical utility** criteria of genetic testing

- (1) Investigating **access to and utilisation** of genetic services in EU,
- (2) Initiating an **international collaborative network** in order to promote internationally shared quality standards for clinical utility assessment
- (3) Investigating criteria and mechanisms to maximize the output of genetic services, **given limited resources**.

Genetic Counselling-minimal criteria for genetic counselling!

Aim to improve the quality of genetic counselling

Specific Support Action to integrate partners from developing countries **“CAPABILITY Project”** (2007-2009)

Unit 4 – Ethical and Legal Issues

- P. Borry, JP. Fryns, P. Schotsmans & K. Dierickx, Attitudes towards [carrier testing in minors](#): a systematic [review](#), in Genetic Counseling 16 (2005) 4, p. 341-352.
- P. Borry, JP. Fryns, P. Schotsmans & K. Dierickx, [Carrier testing in minors](#): a systematic review of guidelines and position papers, in European Journal of Human Genetics 14 (2006) 2, 133-138.
- P. Borry, L. Stultiens, H. Nys, JJ. Cassiman, K. Dierickx, [Presymptomatic and predictive genetic testing in minors](#): a systematic review of guidelines and position papers, Clinical Genetics 70 (2006) 5, 374-81.
- P. Borry, J.P. Fryns, K. Dierickx, Predictive Genetic Testing in Children. A clinical-ethical analysis, in A. de Bouvet, P. Boitte, G. Aiguier, Questions éthiques en médecine prédictive, Montrouge, John Libbey and Company, 2006.

Unit 6 - Education

- 1) Updated list of National **Societies** of Genetics and others relevant ones (collaboration with **ESHG** – www.eshg.org)
- 2) List of European and National **genetic courses**
- 3) List of other **educational material** and list of relevant institutions involved in genetics education
- 4) Network of **national contact persons** in the European countries
- 5) Assessment of **written patient information** in initial 5 countries to identify the key issues patients are being informed
- 6) 1st Workshop on “Patient and Professional Perspective of Genetic Information/Education in Europe”

- Gentest
- Management
- Services/Jobs
- Projects
- Support Groups
- Information
- Networks

EVENTS

- News

Partner has article in DS News

Information Now Available

European contacts for genetics added to website

"Fully Yours"

Announces 'Call techniques'



INFORMATION FOR PATIENTS AND FAMILIES

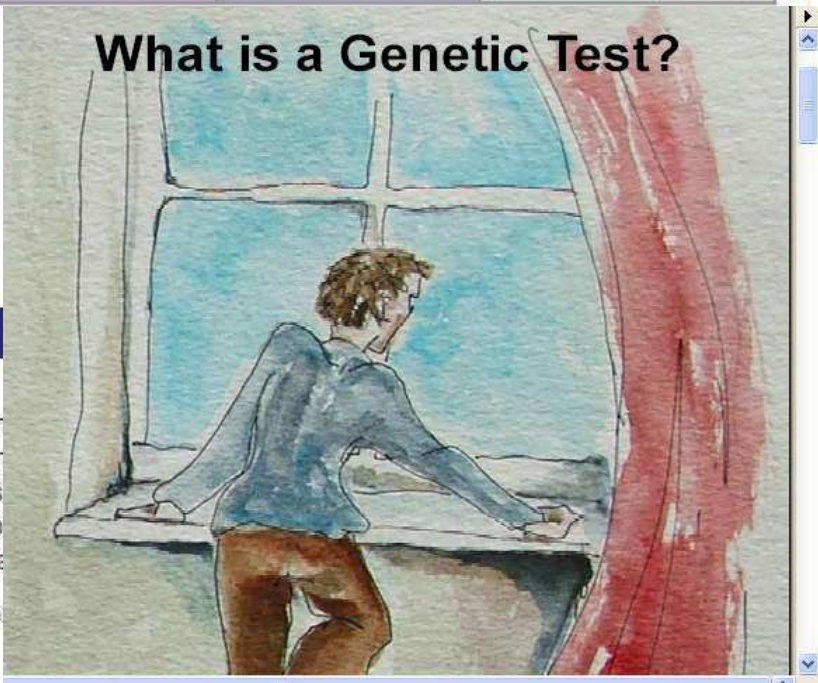
Below you will find information about Inheritance Patterns, Chromosome Appointment. There is also a Frequently Asked Questions leaflet which covers genetic testing, and a detailed Genetic Glossary. This information is designed for the specialist. It has been produced with the help of patients and families from across Europe and genetic testing. We hope therefore to have covered all the issues that you may have.

The information comes in 2 formats. If you want to look at the information online, click the 'view' format. If you want to print the information out as a booklet by clicking the 'booklet' format.

We would like to thank all the patients, families and support organisations from across Europe who have helped to create these leaflets. We are immensely grateful for their contribution to this work.

Please note that some of the files below are large (1MB) and may take a moment or two to download.

Print Format (Booklets)	View Format (A4)
Amniocentesis Test	Amniocentesis Test (A4)
Chromosome Translocations	Chromosome Translocations (A4)
Chromosome Changes	Chromosome Changes (A4)
Chronic Villus Sampling (CVS) Test	Chronic Villus Sampling (CVS) test (A4)
Dominant Inheritance	Dominant Inheritance (A4)
Frequently Asked Questions	Frequently Asked Questions (A4)



**Thank you very much for
your attention (and invitation) !**

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