

ERNDIM and the Eurogentest Project

"Genetic Testing in Europe

– Network for test development, harmonization, validation and standardization of services"

- "Quality is the overall denominator of the EuroGentest NoE"

Aims:

- To **structure**, **harmonize** and **improve** the overall quality of medical genetic testing services
 - while paying attention to issues resulting from testing:
 - legal, health policies and health economic impact
 - IPR (Intellectual Property Rights)
 - confidentiality, informed consent, employment, insurance...



Network of Excellence in Genetic Testing

- Supported by the European Union 2005-2009
 - contract FP6-512148
- Thirty-two participating centres from 17 countries
- "Network of networks"
 - Almost 200 additional expert networks, centres and participants

Leader of the project

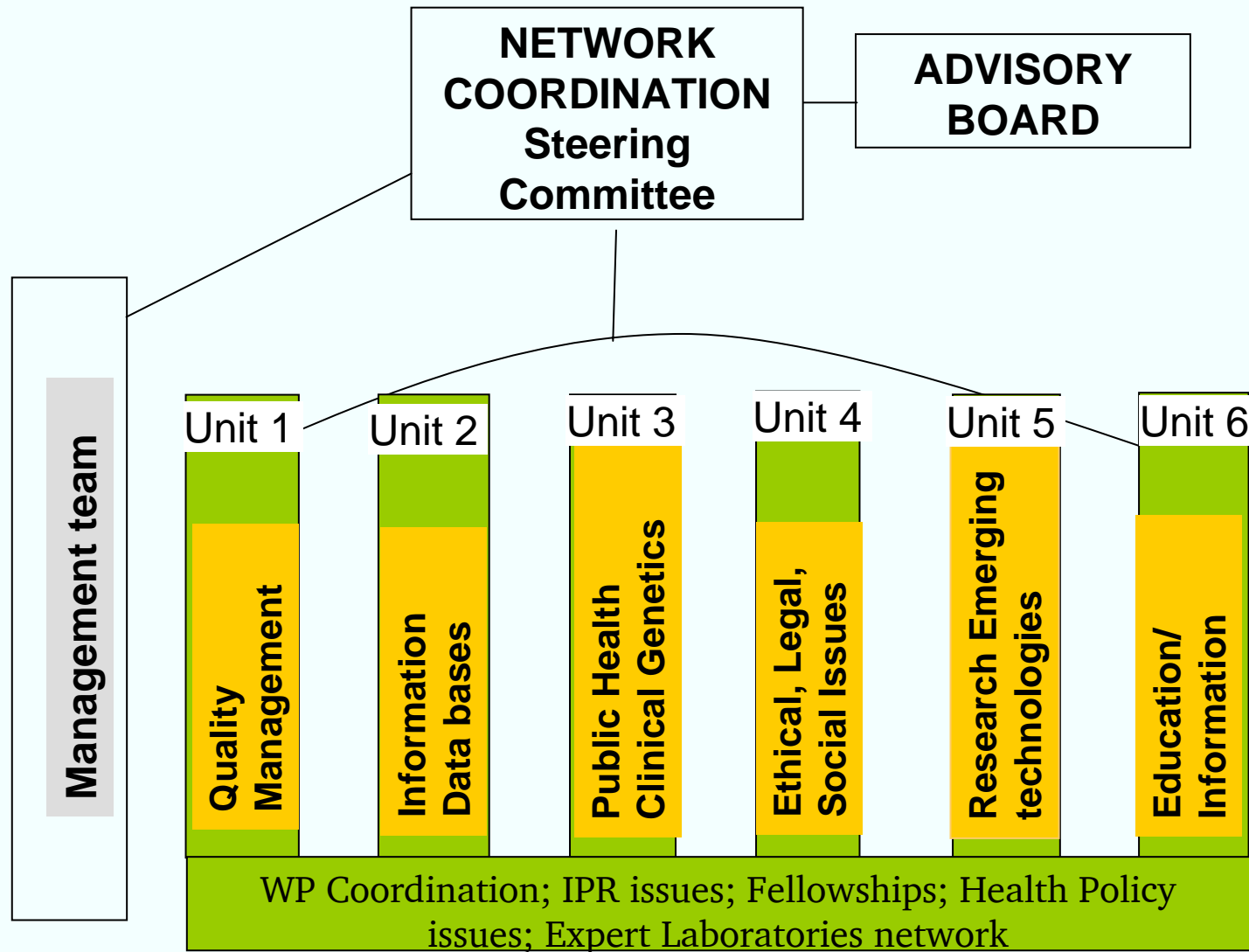
Jean-Jacques Cassiman, Leuven

Management under pressure



From David Barton

Network of Excellence: "EUROGENTEST"



The Eurogentest website

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EuroGentest HARMONIZING GENETIC TESTING ACROSS EUROPE

Home Medical Professionals Laboratories Students Patients & Family Industry About Us Documents Events & News

What is EuroGentest?

EuroGentest is an EU-funded Network of Excellence (NoE) with 5 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 harmonization of standards and practice in all these areas throughout the EU and beyond. [More...](#)

How to register?

Find out more about the world of Genetic Testing by registering! You will get access to additional information and receive a two monthly newsletter if you [register here](#).

News [More news...](#) [Newsletter](#)

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"Gene Patents and Collaborative Licensing Models"-book now on the market

16 Jul 2009

A book on patent pools, clearinghouses, open source models and liability regimes [More...](#)

New on EuroGentest.org


Read the latest articles, webpages and documents that have been added to "eurogentest.org". [More...](#)

Contact us

If you have any questions, suggestions or want to be involved in EuroGentest, please feel free to [contact us](#).


Medical Professionals

- Genetic Counselling
- Documents & guidelines
- Patient Leaflets
- Find a genetic lab [□] / [□] test [□]
- Education
- Ethical & Legal aspects
- Quality Management (QM)
- Databases and Resources
- Professional organizations




Laboratories

- Quality Management (QM)
- Accreditation
- External Quality Assessment (EQA)
- Reference Materials
- Diagnostic Validation
- Research & emerging Technologies
- Documents & Guidelines
- Training/Workshops on quality
- Bio Informatics
- Databases & Resources
- Find a genetic lab [□] / [□] test [□]




Students

- Fellowships
- Education & Training
- Educational Courses
- Educational Software
- Documents & Guidelines
- Publications
- Definitions & Terminology
- Professional Organizations



Patients & Family

- Patient Information
- Genetic Glossary
- Patient Rights
- Genetic Counselling
- Find a support group [□]
- Find a genetic disease [□]
- Find a genetic test [□]
- Professional Organizations



The Eurogentest website



Laboratories

- [Quality Management \(QM\)](#)
- [Accreditation](#)
- [External Quality Assessment \(EQA\)](#)
- [Reference Materials](#)
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Unit 1: Quality Management and Accreditation of Genetic Testing

- Unit leaders: Els Dequeker, Ros Hastings

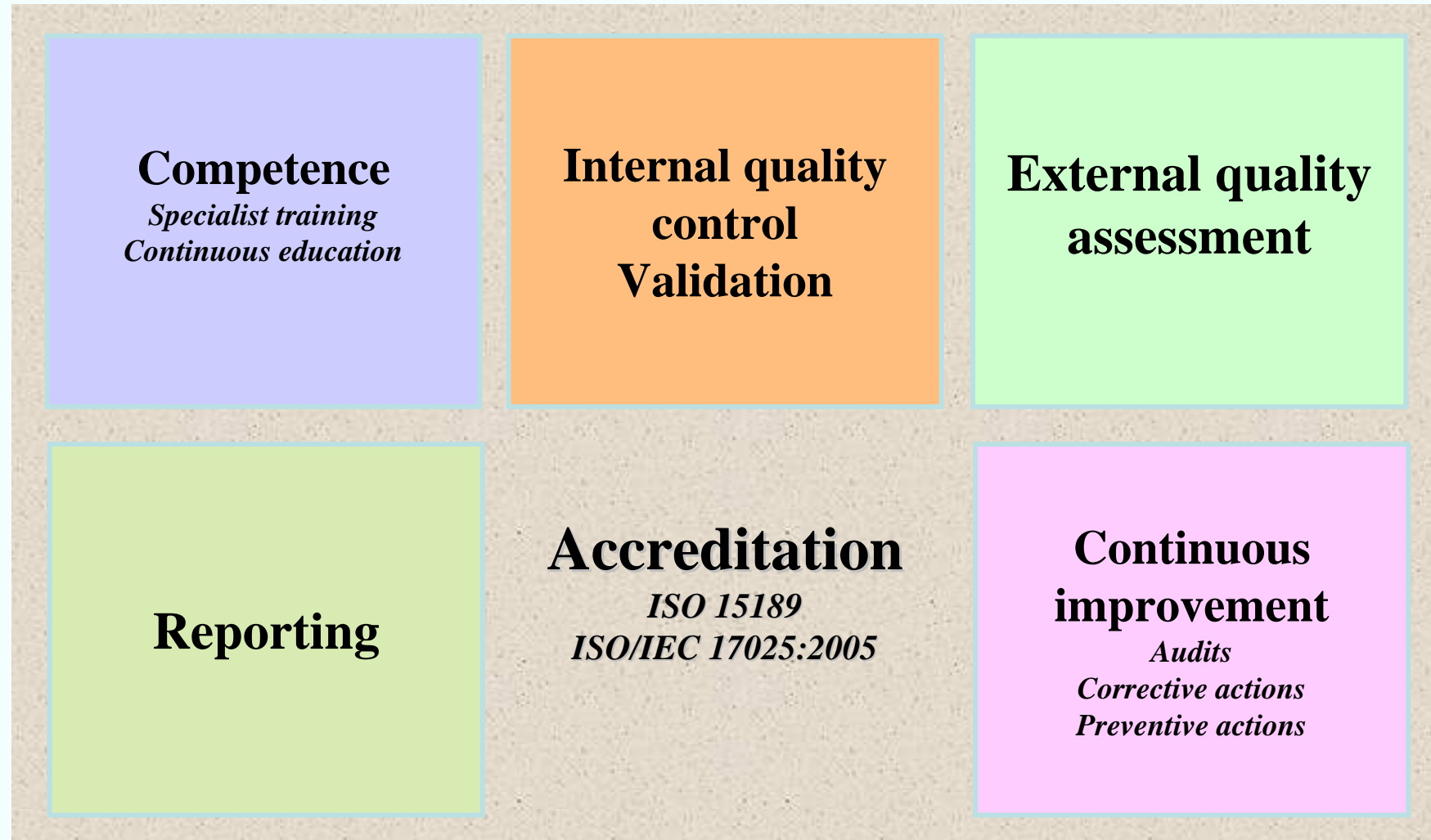
Aims:

- To **measurably improve the quality of management and provision** of genetic laboratory services, for the benefit of patients;
- **Laboratory accreditation** to be considered as the norm.

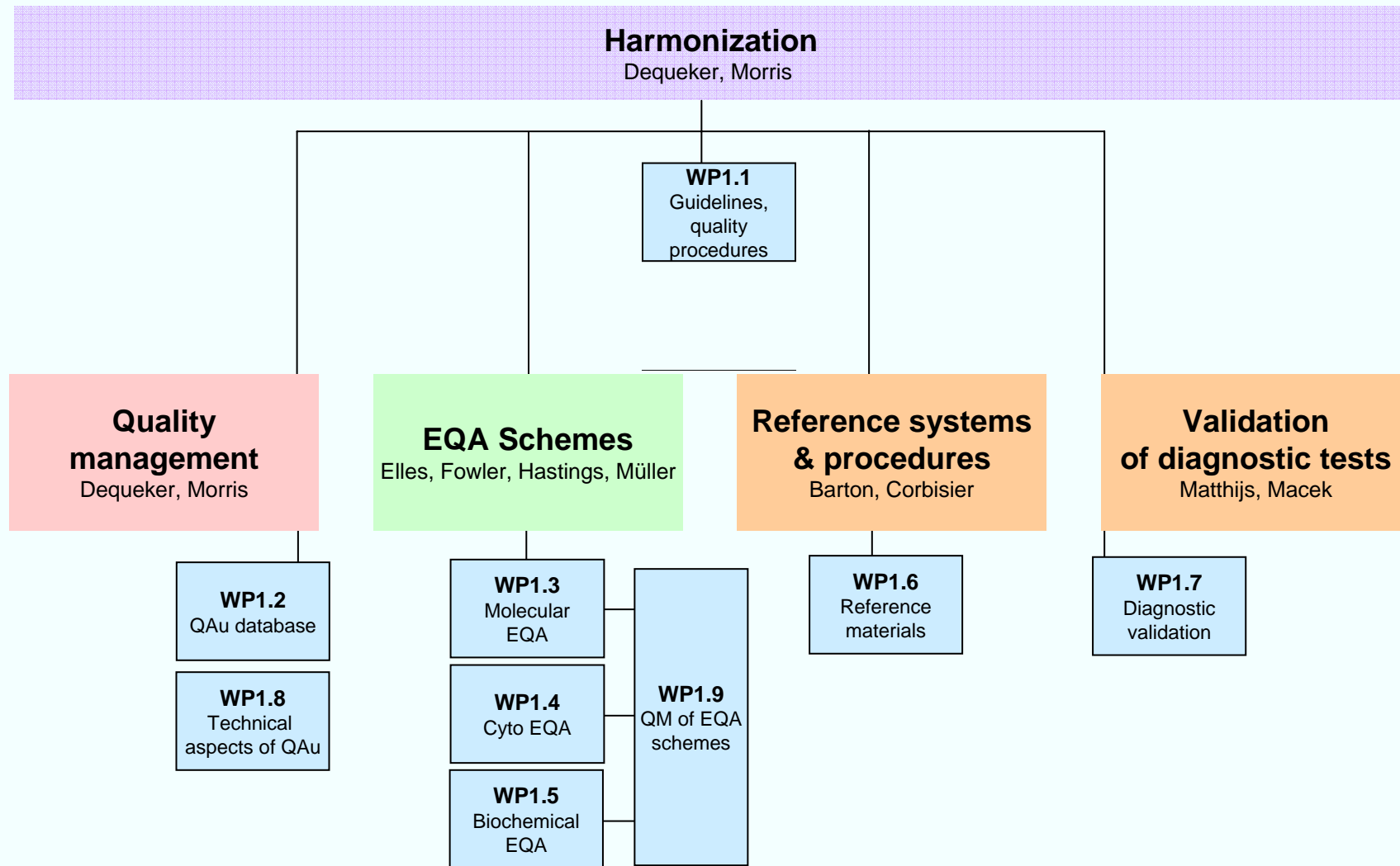
Definitions:

- *Accreditation*: formal recognition of a laboratory's competence to perform a test.
- *Certification*: attestation that a laboratory complies with the requirements of a quality management system.
- *Licensing*: official or legal permission to perform testing.

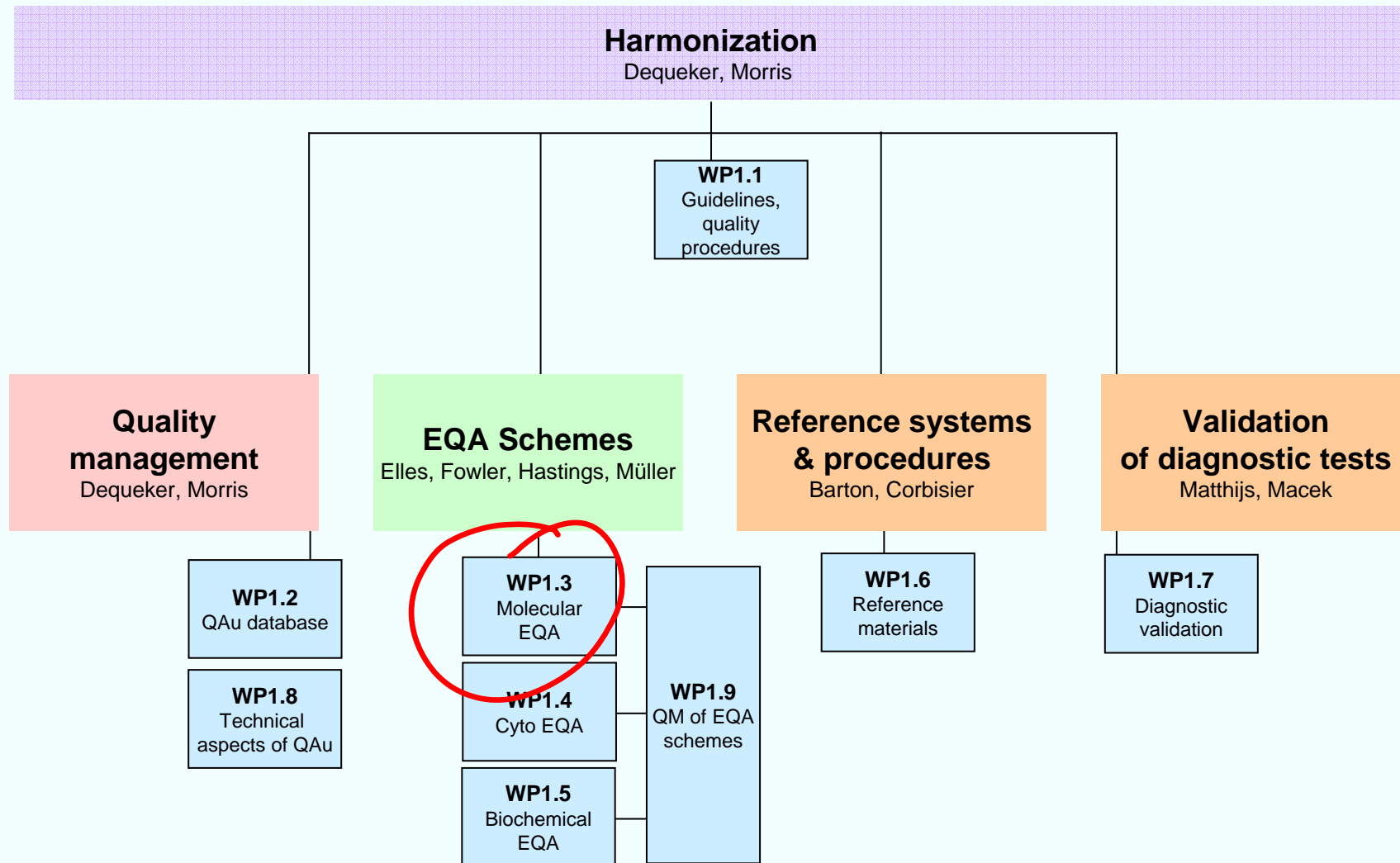
Key elements of quality management and assurance



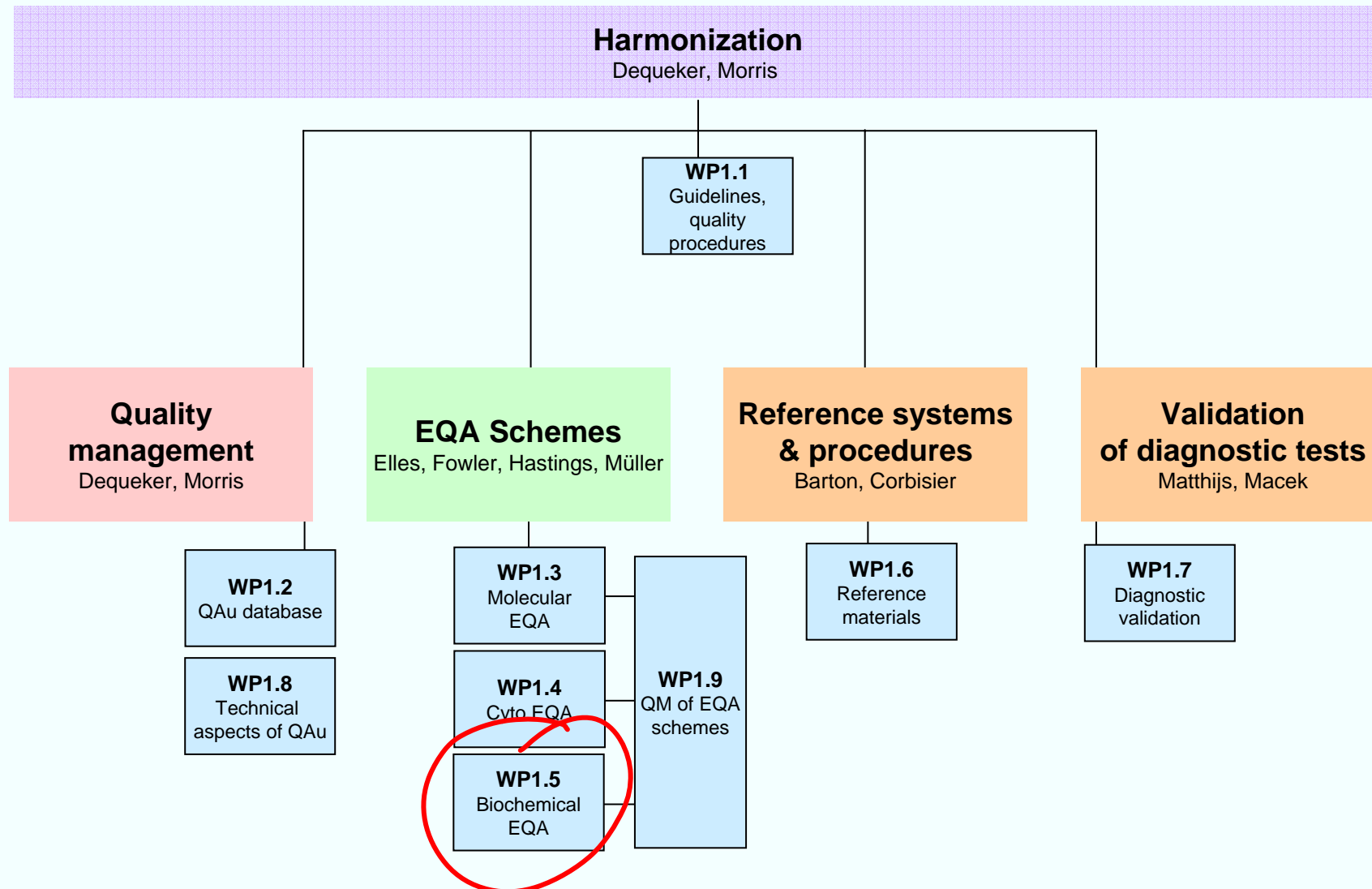
Unit 1: Quality management and accreditation



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Unit 1: Quality management and accreditation



Eurogentest: cooking up quality in 2007

- Clinics
 - Recommendations for genetic counselling
- Labs
 - OECD Guidelines
 - Quality workshops
 - EQA and Best Practice
 - QAu database



Eurogentest: work package for Biochemical Genetic Testing

ERNDIM responsible for Work package

"Organize and harmonize EQA for biochemical Testing"

Objectives

- 1) To expand opportunities for Biochemical Genetic testing laboratories in EU25 to participate in EQA.
- 2) To link Internal Quality Control and EQA through consensus discussions on best practice.

Biochemical Genetics EQA WP1.5

Katharina Honegger, Brian Fowler (ERNDIM)

Aims :

- To expand opportunities for Biochemical Genetic Testing laboratories in EU25 to participate in EQA.
- To link Internal Quality Control and EQA through consensus discussion on best practice.
- Inclusion of complete data on biochemical genetics labs in the ERNDIM web site directory.
- Best practice meetings and development of poor performance criteria.
- Scoping and expansion of paediatric biochemical genetic EQA in EU25.
- To explore standard methodologies and standard reporting procedures.
- Coordination of IntQA, EQA with accreditation
- Accreditation of Biochemical EQA schemes in collaboration with Cytogenetic and Molecular Genetic EQA

Biochemical Genetics EQA WP1.5

Expansion of Biochemical Genetic Testing and EQA in EU27

- Consolidation of 9 existing ERNDIM schemes
- Pilots for Lysosomal enzymes / glycosylation → established schemes
Pilot for Mucopolysaccharides
- Diagnostic Proficiency Testing (DPT) centre Basel:
Now 5 centres
Website submission/ evaluation
- 3rd Best practice meeting with National Representatives (from 27 countries) in Basel, May 9, 2008: Provisional guidelines have been established together with National Representatives for
Recommendations to overcome deficits in BGT services and Provision of BGT services

Biochemical Genetics EQA WP1.5

Expansion of Biochemical Genetic Testing and EQA in EU27

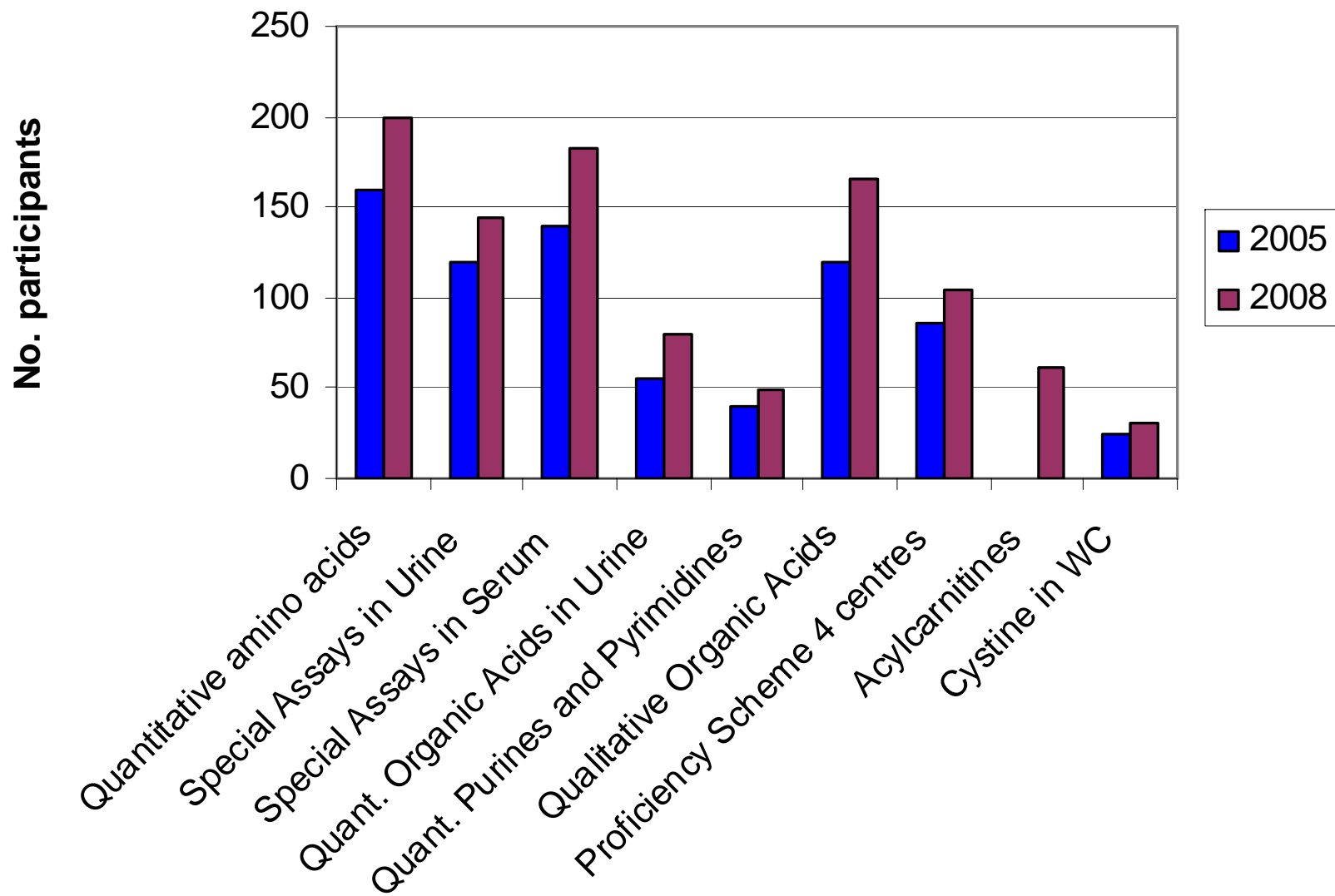
- SSIEM academy training workshops
 - amino acid disorders at SSIEM 2008 (36 participants from 18 countries)
 - organic acids Basel 2009 (42 participants, 60 applicants)
- ERNDIM Workshops on best practice held at SSIEM
 - 2005: Purines/Pyrimidines
 - 2006: Various at Prague
 - 2007: Peroxisomal disorders
 - 2008: Creatine synthesis disorders (web-site)
 - 2009: Neurotransmitters
- National Representatives for all EU countries + Switzerland (see website Erndim.org)

Total Participants

1994: 123

2008: 265

Participation 2005 / 2008



Biochemical Genetics EQA WP1.5

Harmonization of EQA performance assessment

- Best practice on EQA meetings of the ERNDIM Scientific Advisory Board and Executive Committee
Twice yearly
- New participation certificate with performance: effective 2008 for 2007

Biochemical Genetics EQA WP1.5

Standard methodologies and standard reporting procedures

Work continuing with ERNDIM board and Scheme providers

- Guidelines for scoring of DPT schemes completed.
- Draft documents for acyl carnitines, purine and pyrimidines discussed and evaluated need to be finalised
- Methodologies for Lysosomal enzymes and mucopolysaccharides planned

- Publications: Journal of Inherited Metabolic Disease, 2008
 - "Qualitative urinary organic analysis: methodological approaches and performance"
 - "The role of ERNDIM in improving the Quality of Biochemical Genetic Testing"
- Questionnaire for best practice criteria for amino acid analysis received from 63% of participants (125 labs)

Biochemical Genetics EQA WP1.5

Consolidation and expansion of BGT laboratories directory with links to Unit 2 (Orphanet) and National directories

- Inclusion of complete data on biochemical genetics labs in ERNDIM website directory
- ERNDIM laboratory directory: modified information page with accreditation status to harmonize with QAu database
- New and existing labs have been contacted. Existing labs have updated their information, 12 new labs have joined in 2008-9
- Intensive chasing up of all BGT labs for inclusion of complete data in ERNDIM laboratory directory.
- Transfer of data to QAu database
- Linking of analytes in ERNDIM lab directory to diseases in Orphanet database
- New ERNDIM domain: www.erndim.org

Biochemical Genetics EQA WP1.5

Accreditation of biochemical genetic EQA schemes

- Roadmap for accreditation of ERNDIM agreed at Best practice meeting in Lyon, April 3/4, 2008
- Search for physical location/central office for ERNDIM and consolidation of ERNDIM administration in central office.
- Establishment of post for ERNDIM activities /Training (SSIEM Academy) at St. Mary's Hosp. Manchester (Dept. Molecular Genetics, Rob Elles
- Aim is / **was** accreditation end 2010

Biochemical Genetics EQA WP1.5

Training workshops and meetings

- Meeting of the Baltic States on inherited metabolic disorders was held in Tallinn on March 10-11, 2009.
- The first South African meeting on Inherited Metabolic disorders was sponsored in part by ERNDIM. This took place in Cape Town, March 27-29, 2009
- Training grants
 - Vilnius, Lithuania
 - Cape Town, South Africa

Quality Management of EQA schemes WP1.9

Els Dequeker, Clemens Müller, Rob Elles, Ros Hastings, Brian Fowler

Aims :

- Improve the awareness of the importance of organizing EQA schemes related to the international requirements for EQA scheme providers (ISO, ILAC, CPA, IFCC).
- Accreditation of individual Genetic EQA schemes
- Accreditation of EQA umbrella organisations

QM of EQA schemes WP1.9

Best Practice meetings of WP1.9 Partners

- Minutes available
- Planning of 1st meeting of the quality committee of ESHG
- Guide to certify/accredit EQA schemes relating to existing standards
-
- Generic Quality Manual for EQA providers (thanks to Ros Hastings)

QM of EQA schemes WP1.9

Formation and Consolidation of sub-committee of ESHG for
Quality issues

European Society for Human Genetics Sub-Committee for Quality Issues

Aims

- Identify gaps in quality issues within diagnostic genetic testing services
- Identify where there can be harmonisation between the biochemical genetic, cytogenetic and molecular genetic disciplines
- To commission and approve new documents relating to quality in genetic testing
- Give recommendations for those countries where no guidance is currently available.

It was agreed that the 4 Schemes would submit their annual Management Reviews for the QC meeting each May after their EQA year had completed.

European Society for Human Genetics Sub-Committee for Quality Issues

Members

Ros Hastings

Brian Fowler

Viktor Kozich

Els Dequeker

Rob Elles

David Barton

Chair & CEQA

ERNDIM

SSIEM and ERNDIM Board

CF Network

EMQN

Reference Materials expert

Jacques Beckmann

Claude Giroud

Mireille Claustres

ESHG Board

EDMA (industry)

Diagnostic Laboratory

Lidia Larizza / Konstantin Miller

Peter Farndon

Orsetta Zuffardi

Cor Oosterwijk

ECA Board representative replacing

Clinical Geneticist & UKGTN

Genetic Research Community

EGAN (patient group)

QM of EQA schemes WP1.9

Formation and Consolidation of sub-committee of ESHG for Quality issues

- 1st meeting of the committee at the ESHG meeting in Barcelona, June 1
- 2nd meeting December 16th. 2008
- 3rd meeting of the committee at the ESHG, Vienna May 2009 (esp. presentation of annual reports of EQA organisers)

The Future

Eurogentest follow up project

much smaller than previous

Strong emphasis on EQA

Build on the contacts we have built up during Eurogentest

(Main benefit of the project)

Consolidate the strong position of Biochemical Genetic Testing (=ERNDIM) within the Genetics field

ERNDIM /EUGT meeting October 5 and 6, 2006, Prague

The Role of QA in the diagnosis and dietary management of IEM The European perspective

Programme

Thursday October 5

9:00 – 12:30 Session 1

- 9:00 Opening
Brian Fowler (Basel)
Prof. Tomas Zima (Dean, First Faculty of Med.-Charles Univ.)
- 9:10 Special Guest Lecture: Diagnosis of Lysosomal storage diseases
Milan Elleder (Prague)
- 9:55 Results obtained with samples from Lysosomal storage diseases in Diagnostic Proficiency Testing
DPT scheme organisers
- 10:10 Lysosomal enzymes pilot EQA scheme
Otto van Diggelen (Rotterdam)

10:30 Coffee

- 11:00 Eurogentest: activities: Overview
Brian Fowler (Basel)
- 11:15 Perspective of Cytogenetics
Rod Howell (Oxford)
- 11:35 Eurogentest NoE: towards analytical and diagnostic validation
Milan Macek (Prague)



ERNDIM = uality

We provide 9 external quality control schemes for:

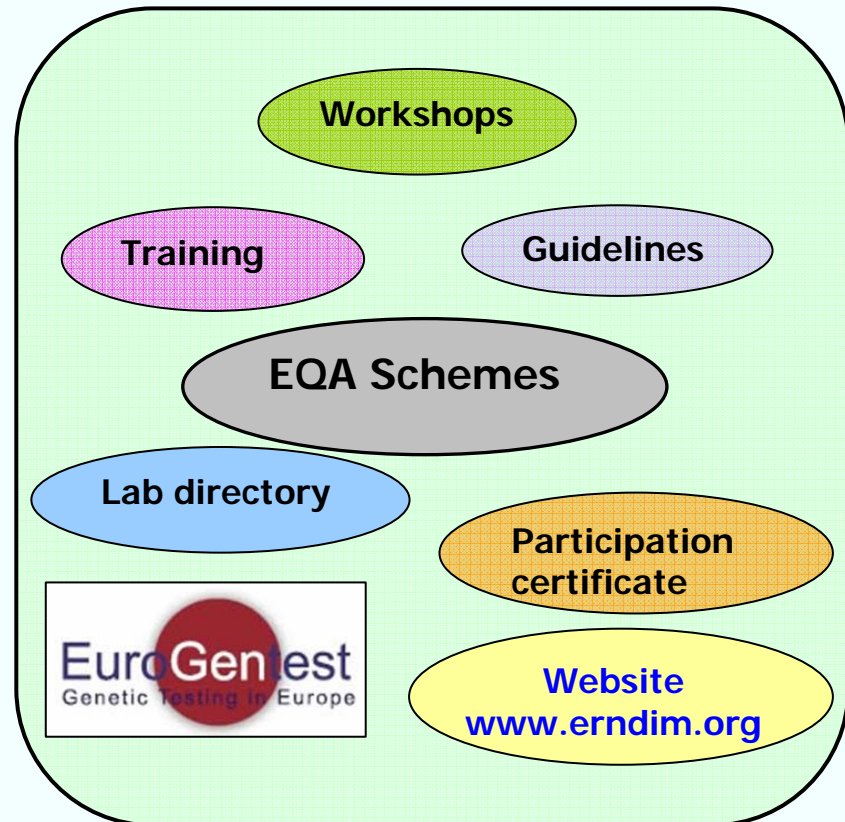
- Quantitative Amino Acids
- Special Assays in Urine
- Special Assays in Serum
- Quantitative Organic Acids in Urine
- Qualitative Organic Acids
- Quantitative Purines / Pyrimidines in Urine
- Acyl Carnitines
- Cystine in White Cells
- Diagnostic Proficiency Schemes

Number of participants range from 28 to 200 per scheme.

Schemes are organized according to the latest scientific and regulatory standards.

We have 1050 scheme subscriptions with 270 participants from mainly European countries but also worldwide.

The ERNDIM board, scientific advisor board and executive committee work together to strive for improvement of laboratory services to the benefit of our patients.



Partners



SSIEM

Eurogentest: work package for Biochemical Genetic Testing

Description of work

- (1) To research the current situation in EU25 including needs and deficits.
- (2) A program of meetings to discuss best practice and agree consensus guidelines in Biochemical Genetic practice.
- (3) To instigate the links between internal quality control, EQA and accreditation.
- (4) To explore standard methodologies and standard reporting procedures.
- (5) Co-ordination with other work packages through correspondence and meetings.