

EuroGentest

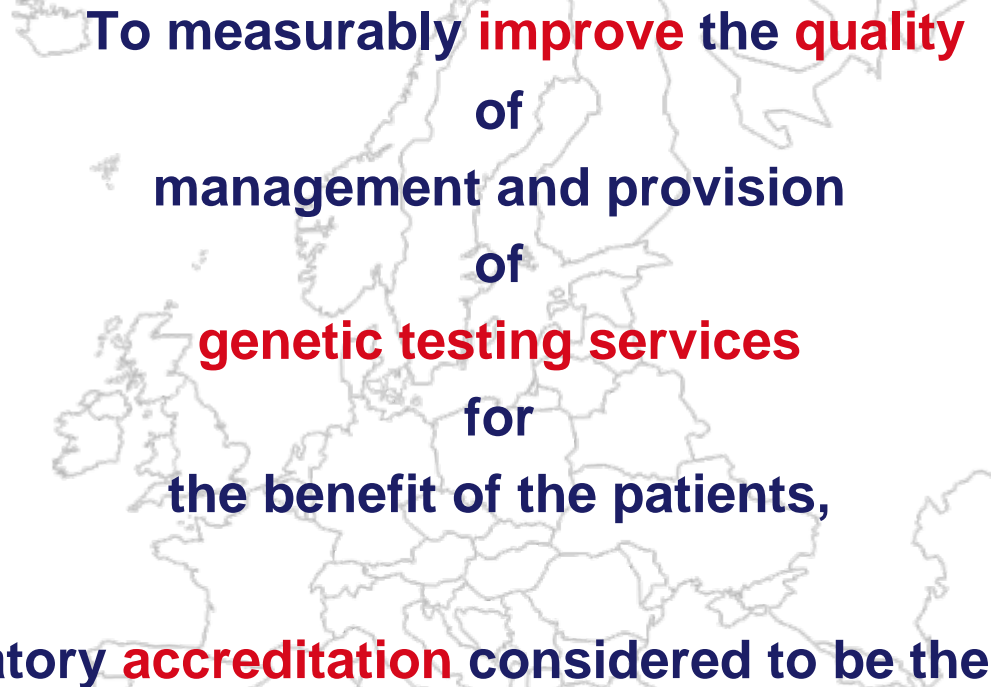
Unit 1: Quality Management and Accreditation of Genetic Testing

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www.eurogentest.org



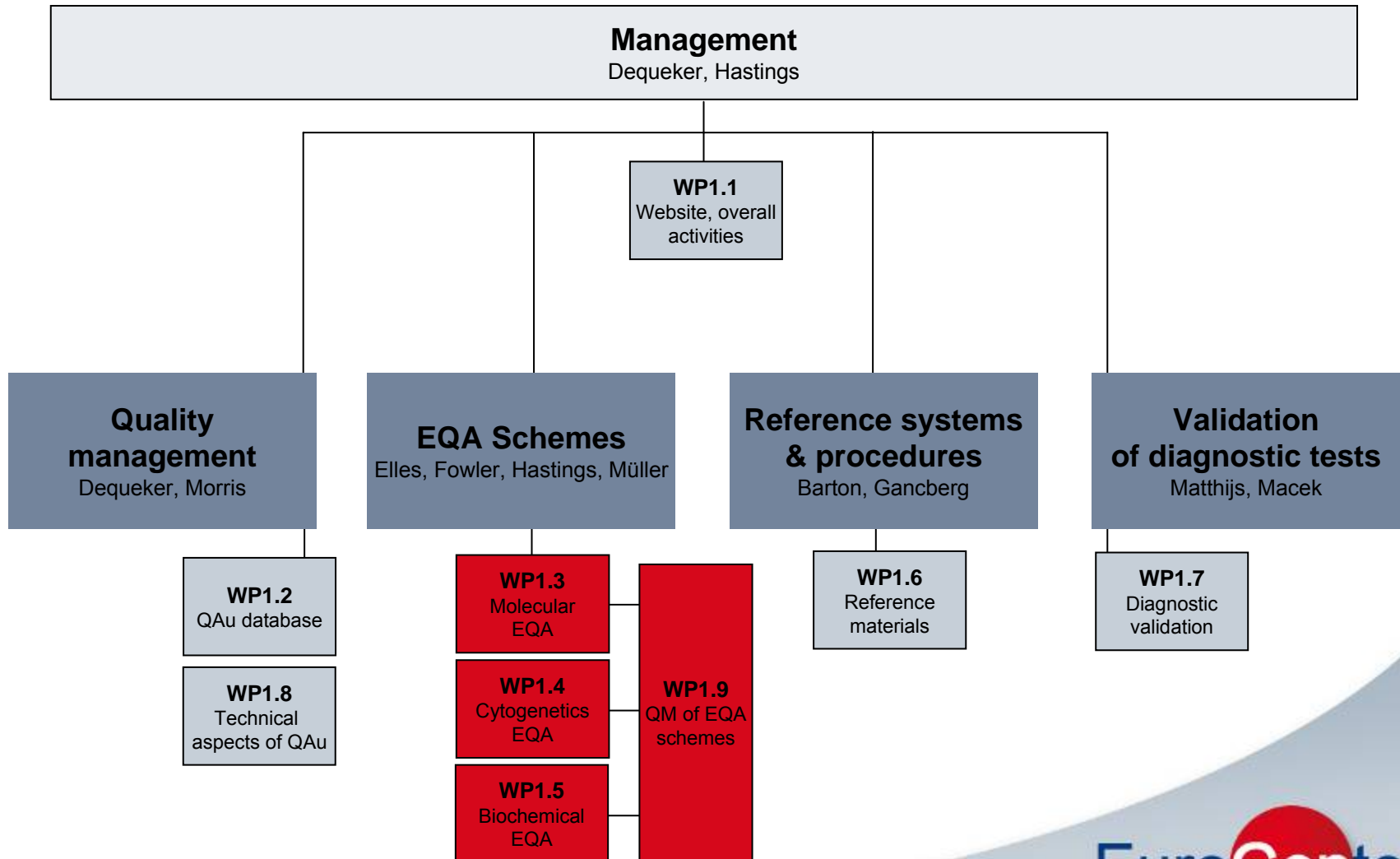
Overall Objective of Unit 1

A faint, light gray outline map of Europe is centered in the background of the slide. The text is overlaid on this map.

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








Unit 1: Quality Management and Accreditation



European and National EQA in Genetics

European Schemes:

ERNDIM (Biochem)		multiple EQAs
EMQN (Molecular)	 The European Molecular Genetics Quality Network	multiple EQAs
CF-Network (Molecular)	 Cystic Fibrosis European Network	single disease EQA
UK NEQAS (Cyto)		multiple EQAs
CEQA (Cyto)	 Cytogenetic European Quality Assessment	limited EQA at present

National/Multinational Schemes:

Germany, France, Italy, Finland, Spain, UK-
Holland-Ireland, Czech Republic

Aims for European EQA

Harmonizing existing EQA schemes for genetic testing in Europe:

- **Expand opportunities for genetic testing laboratories** in Europe to participate in EQA
- **Link Internal Quality Control and EQA** through consensus guidelines in genetic practice
- **Sustainability-** accreditation, financial stability and governance

How?

- **Forum** with all European EQA providers
- Expand number and scope of European EQA schemes
- Publish European **Guidelines**
- Collaborate and prepare for accreditation
- **Achieve self-financing** through subscriptions
- Form **joint** EQA governance body



EuroGentest – a forum for EQA discussions

Progress:

- ‘Forum’ Meetings for EQA providers

Biochemical Genetics	Bi-annual meeting
Cytogenetics	Annual meeting
Molecular Genetics	Multilateral and Bilateral discussions with other EQA schemes
Cross-discipline	Collaboration on governance and sustainability; Establishment of ESHG Genetic Services Quality Committee



EQA expansion of availability

Biochemical

7 scheme centres

9 EQAs

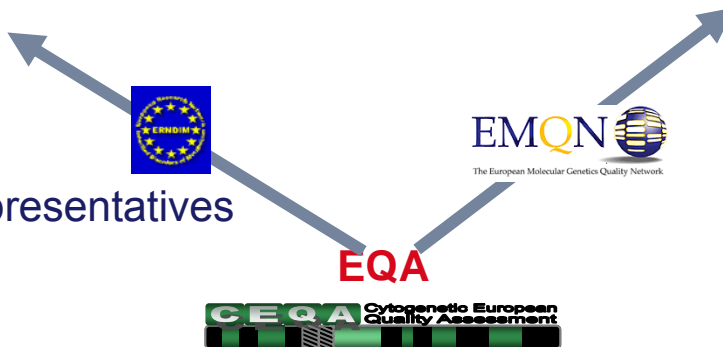
2 pilot EQAs

266 labs

18 countries

New participants

New national representatives



Molecular

New EQA: MODY

2 pilot EQAs: MD & mutation screening

464 labs

41 countries

Synthetic control material trials

Cytogenetics

Internet based EQA and registration

Pilots in blood and amniotic fluid → full EQAs

New EQAs for 2008:- Haematology/PGS

90+ labs

30 countries

National schemes expanding repertoire, New participants

Best Practice discussion

Biochemical

Best Practice meetings in 2007
New Guidelines
Workshops

Molecular

3 Best Practice meetings in 2007
MODY Guidelines published,
BRCA/SCA considered
for ratification



EQA



Cytogenetics

Summary of General B.P. Guidelines published
Best Practice Guidelines Microarrays
Exemplary reports given with CEQA



Improved availability of guidelines

Biochemical

ERNDIM Guidelines

- Guidelines for investigation of hyperammonaemia
- White cell preparation for cysteine
- 5 guidelines in preparation

Guidelines

- Published: ERNDIM & EUGT website



Molecular

Best Practice Guidelines

- 3 Best Practice meetings in 2007
- 2 Further BP meeting in 2008



EQA



Cytogenetics

European Cytogenetic Guidelines and Quality Assurance: A common European framework for quality assessment for constitutional and acquired cytogenetic investigations

- EJHG – a summary, March 2007

Microarray Best Practice

- EJHG – Summer 2007



Joint approaches - common web based platform for administration

- Cytogenetics: new, adaptation of EMQN registration website + integration of online EQA
- Molecular Genetics: updates
- Biochemical Genetics: link with Orphanet + QAU database for lab information



Joint approaches - accreditation and harmonisation

- Generic National Representatives job description
- Assisting National and European EQA Schemes to achieve accreditation
- Generic Quality Manual
- Meeting with EQA providers - report in preparation
- Standard EQA checklist translation approved
- Agreement on ESHG Genetic Services Quality Committee

- Merger between GfH (German Human Genetics Society Molecular Genetics EQA) and EMQN
- Discussion between ERNDIM and EMQN on sharing administrative platform



Future vision for EQA

The Future Vision:

CEQA, EMQN, ERNDIM, CF Network and National Schemes collaborate to give every laboratory in Europe access to EQA

Financial independence, Sustainability and Accreditation of all European EQA schemes



Plans for 2008

→ Establish a **governance structure** for CEQA, EMQN and ERNDIM through ESHG Quality Committee

→ Determine **poor performance** criteria

→ Organize **Best Practice Meeting** on Quality Management of EQA schemes

→ Prepare for **accreditation**: draft guide to accredit EQA schemes relating to existing standards



<p>New schemes for</p> <ul style="list-style-type: none"> - Acute leukaemia - PIGD <p>Pilot resulted in two full EQA schemes (prenatal and postnatal)</p>	<p>New schemes for</p> <ul style="list-style-type: none"> - Von Hippel Lindau - Congenital Adrenal Hyperplasia - Multiple Endocrine Neoplasia - Connexin 26 	<p>No new schemes for 2008</p>
<p>Establish a Steering Committee</p>	<p>Best Practice Guidelines</p> <ul style="list-style-type: none"> - Cardiac genetics - Myotonic Dystrophy - 2 others (open call) 	<p>Best Practice Guidelines</p> <p>Organisation of testing services at a national level</p>
<p>Open cytgenetic EQA to all participants</p>	<p>Consolidating merger with German EQA scheme</p>	<p>Development of standard methodologies and reporting procedures</p>
<p>Prepare for accreditation in 2009</p>	<p>Accreditation in 2008</p>	<p>Training workshops</p>
<p>Pilot a self-assessment system online (for CPD)</p>		<p>Link directory analytes to diseases in Orphanet database</p>

*Els Dequeker, Ros Hastings, Clemens Muller
Brian Fowler
and the team*

