



# **Genetic Services Quality Committee GSQC**

Ros Hastings



# GSQC

## 2012/2013:

- Quality Assurance in Genetic Counselling survey and symposium
- Reporting Guidelines for genetic laboratories published (in press EJHG)
- Practical workshop on nomenclature and interpretation of laboratory results for genetics
- Meeting on European consensus on reporting unsolicited findings and unknown variation.
- Data on EQA poor performance



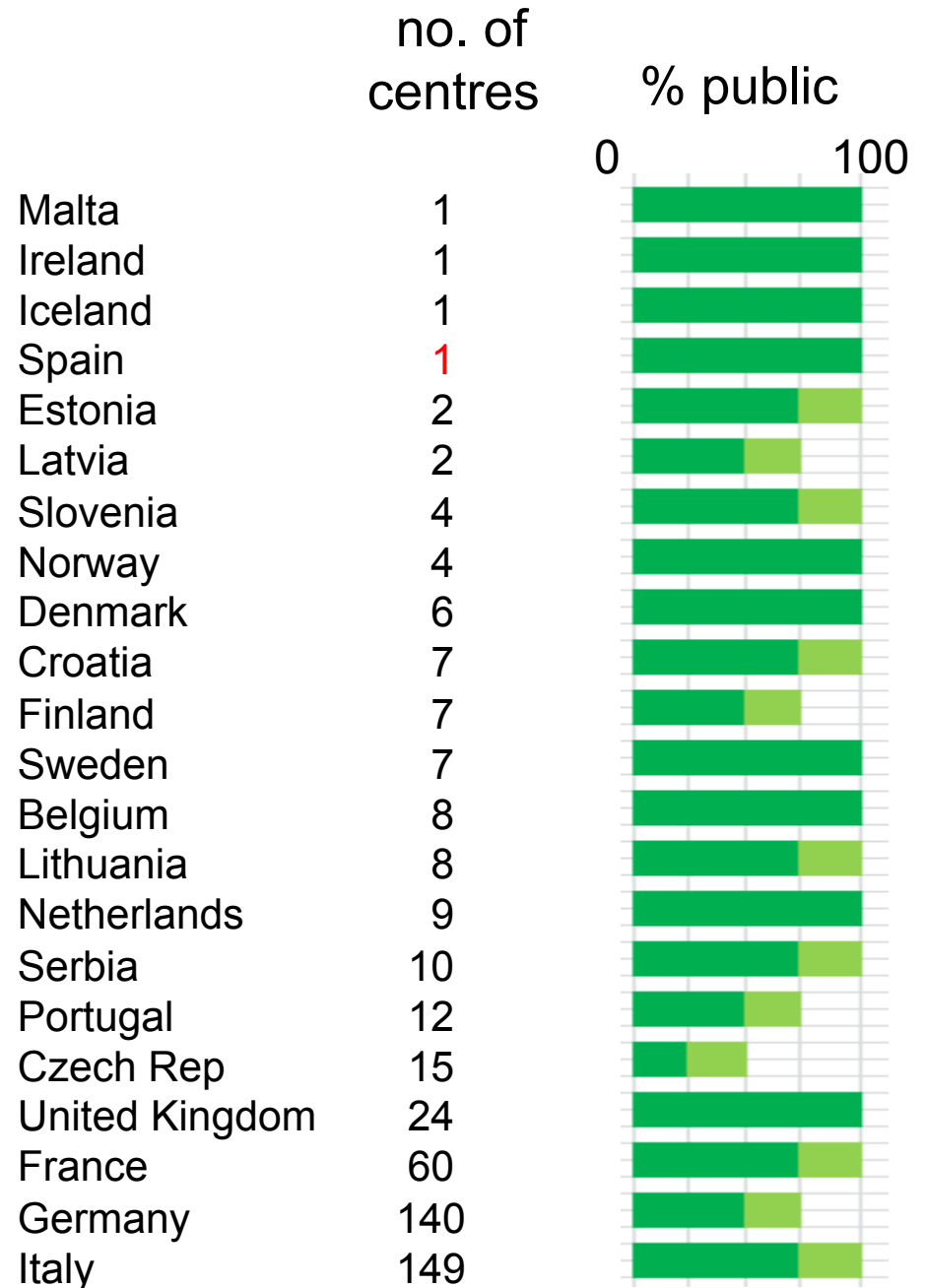
# Quality Assurance in Genetic Counselling survey

## Participants

32 national societies  
 were asked and  
 reminded to complete  
 the questionnaire.  
 22 questionnaires  
 were returned  
 (19 complete data)

Poster:P18:32

Workshop: WS07 15.15 today





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# GSQC

## EQA unsatisfactory (poor) performance

- Genotyping critical errors
- Interpretation critical errors (CF network and CEQA)
- Some European labs do not follow OECD guidelines i.e. do not interpret the significance of the genetic results.
- Few countries have a surveillance body to which labs or EQA providers have to report unsatisfactory performance.
- **Unsatisfactory performance** is different from **persistent unsatisfactory performance**.
- This data does not equate to the diagnostic error rate as there are 2-3 cases/EQA.



# GSQC

Mean critical error in genotyping and interpretation

EQA	Year	Number EQA participations	EQA samples	Analytical critical error	Interpret. critical error	Total critical error	Unsatisfactory performance	Range
CF network	2011	218	3	7	19	26	11.9%	n/a
EMQN	2011	2050	3	134	n/a*	134	6.5%	0-26%**
CEQA	2012	434	2	13	7	20	4.6%	0-11%***

This data does not include unsatisfactory performance due to non-submission.

\*Critical interpretation errors not evaluated ( except for UK labs and the joint CEQA/EMQN microarray EQA).

\*\* 6/28 EQAs had >10% unsatisfactory analytical performance (11%,15%, 18%, 18%, 19% & 26%)

\*\*\* 1 EQA had an 11% unsatisfactory performance (included analysis and interpretation critical errors)



# 2005 - 2013

A network for the harmonization, validation and standardization in genetic testing

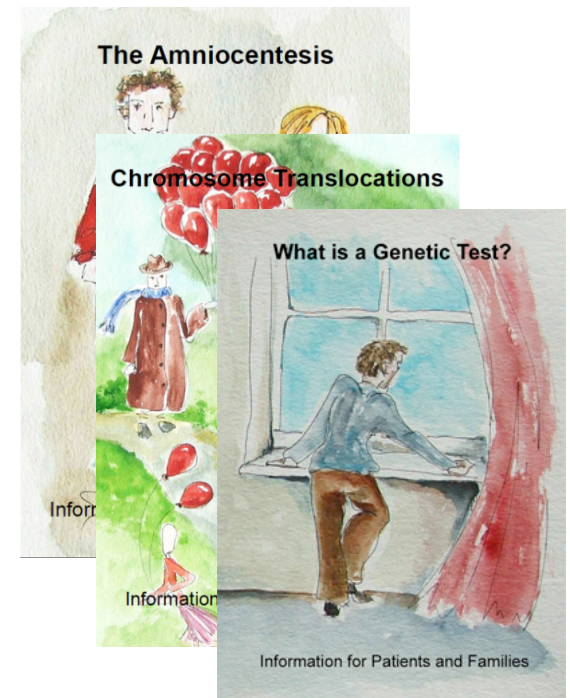
- quality in the laboratory
- quality for genetic counselling
- policy on behalf of a genetic community

## Some of the achievements

- More than 600 participants in **Training courses on quality assurance** **Poster 18.41**
- **Harmonization** of External Quality Assessment schemes
- **Guidelines on genetic counselling** for prenatal diagnosis and PGD
- **Annual conference** on quality in genetic testing (Leuven, 2011; Nijmegen 2012; Prague 2013)
- Lobbying for **IVD Directive/Regulation**
- Representation at **EUCERD, IRDiRC** and other international forums
- Clinical Utility Gene Cards
- .....

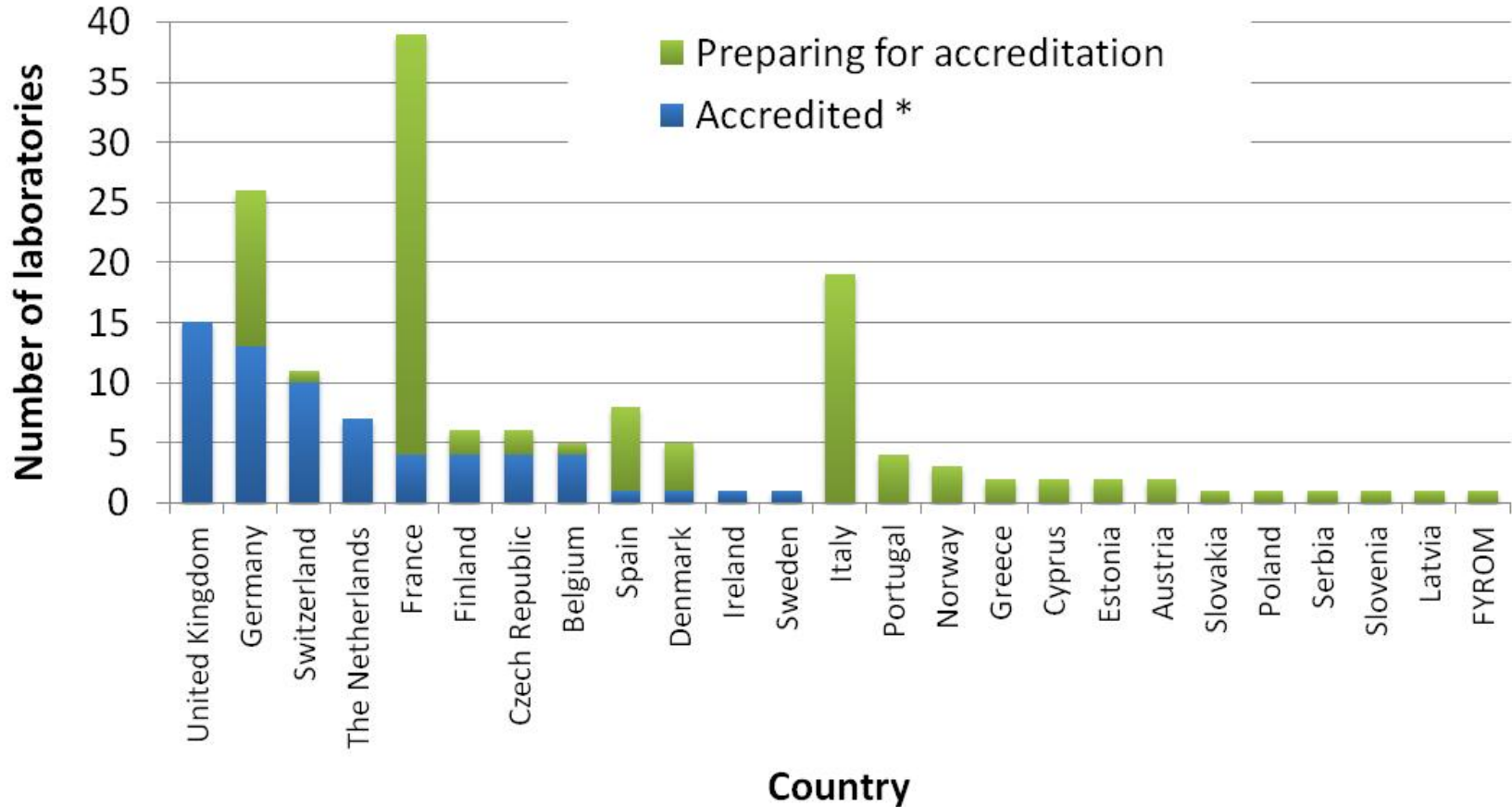
# Responsibilities at the national level... EuroGentest

- **Accreditation of genetic laboratories is a national matter**  
An obligation for accreditation should be written in national law (e.g. as a requisite for reimbursement).
- **Workshops on quality assurance** can be organized in conjunction with the national society meetings, please contact EuroGentest
- **Information on genetic testing for professionals**  
**Leaflets for patients** are available for free distribution via national society websites
  - guidelines on pre- and post-natal genetic counselling
  - Clinical Utility Gene Cards (CUGC, published in EJHG)
  - 15 leaflets in 30 languages





# Accreditation status



- › Note 1: 65 accredited laboratories; 105 preparing for accreditation
- › Note 2: 58% (n=35) of laboratories achieved accreditation within 2 years; 93% (n=56) within 5 years

Berwouts S, et al. Quality assurance practices in Europe: a survey of molecular genetic testing laboratories.

Eur J Hum Genet. 2012;20(11):1118-26.

## ARTICLE

# Approaches to quality management and accreditation in a genetic testing laboratory

Sarah Berwouts<sup>1,3</sup>, Michael A Morris<sup>2,3</sup> and Elisabeth Dequeker<sup>\*1,3</sup>



## SPECIAL ARTICLE

# Mutation Nomenclature in Practice: Findings and Recommendations from the Cystic Fibrosis External Quality Assessment Scheme

Sarah Berwouts,<sup>1,6</sup> Michael A. Morris,<sup>2,6</sup> Emmanuelle Girodon,<sup>3,6</sup> Martin Schwarz,<sup>4,6</sup> Manfred Stuhmann,<sup>5,6</sup> and Elisabeth Dequeker<sup>1,6\*</sup>

Human Mutation



human  
reproduction

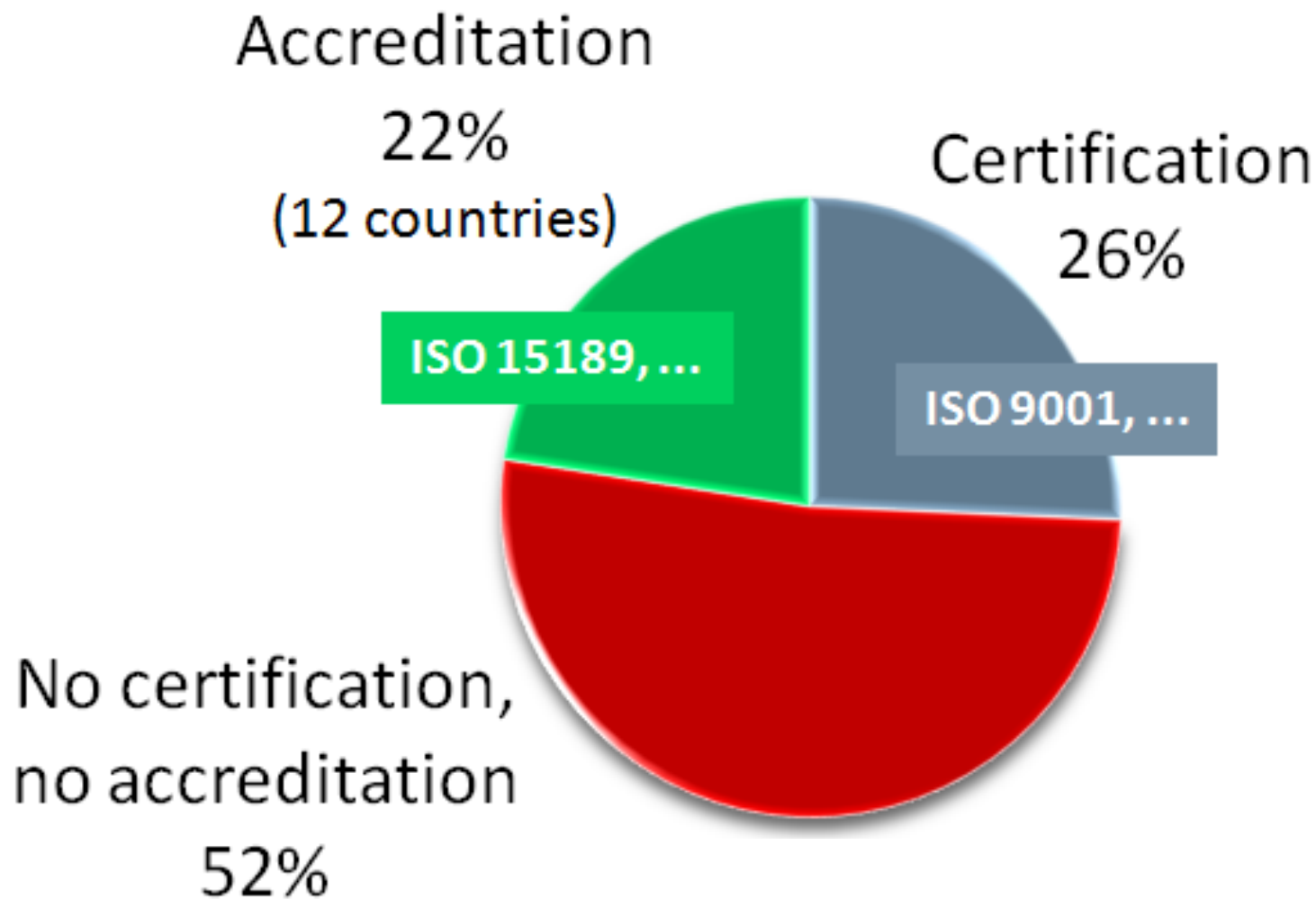
REPRODUCTIVE GENETICS *Review*



# Accreditation of the PGD laboratory

J.C. Harper<sup>1,2,9</sup>, S. SenGupta<sup>1</sup>, K. Vesela<sup>3</sup>, A. Thornhill<sup>4</sup>,  
E. Dequeker<sup>5,6</sup>, E. Coonen<sup>7</sup>, and M.A. Morris<sup>6,8</sup>





Berwouts S, et al. Quality assurance practices in Europe: a survey of molecular genetic testing laboratories. Eur J Hum Genet. 2012;20(11):1118-26.

# The future of EuroGentest



- Funding by the European Commission ends December 2013
- EuroGentest's activities will be **integrated with the ESHG**



The European Society of Human Genetics and EuroGentest plan to create an **association of genetic laboratories, health care and educational institutes in Europe.**

= a new concept: **institutional membership**

## Goals of the **EuroGentest** association:

- Provide a platform for collaboration between institutes and laboratories
- Be a voice and a lobbying group on behalf of genetic services and genetic laboratories,
- Continue to offer training and guidance in clinical and laboratory genetic testing (workshops, guidelines,...)



**The future of EuroGentest:  
towards an association of genetic  
laboratories, healthcare and educational institutes**



EuroGentest intends to create an association of genetic laboratories, health care and educational institutes in Europe, that will perpetuate the name and the key activities of EuroGentest. This will be a new concept in the field of genetics. It is meant to allow a better cooperation between genetic services. For this reason, laboratories, institutes and companies involved in genetic testing will be invited to become a member starting of the emerging association.

Negotiations are ongoing with the European Society of Human Genetics (ESHG) about a possible integration. This combination will provide the best platform for collaboration between all parties involved in genetic testing and to tackle the future challenges in genetics and genomic medicine.

We hope to welcome you to EuroGentest soon!  
We are convinced that the creation of the association will enable us to provide higher quality of care for patients with rare diseases and their families.

EuroGentest plays a leading role in assuring quality and harmonization of clinical and laboratory genetic services in Europe and has been funded as a European project since 2005.

This funding will end December 2013 but EuroGentest will not!

EuroGentest intends to be a voice and a lobbying group on behalf of genetic services and genetic laboratories, and will continue to offer training and guidance in clinical and laboratory genetic testing.

EuroGentest's website will remain available at

[www.eurogentest.org](http://www.eurogentest.org)