EUROPEAN COMMISSION
DIRECTORATE-GENERAL
Joint Research Centre

Joint Research Centre

Availability of Control Materials for Genetic Testing in Europe

David Gancberg

Institute for Reference Materials and Measurements (IRMM)
Geel, Belgium

http://www.irmm.jrc.be
http://www.jrc.cec.eu.int

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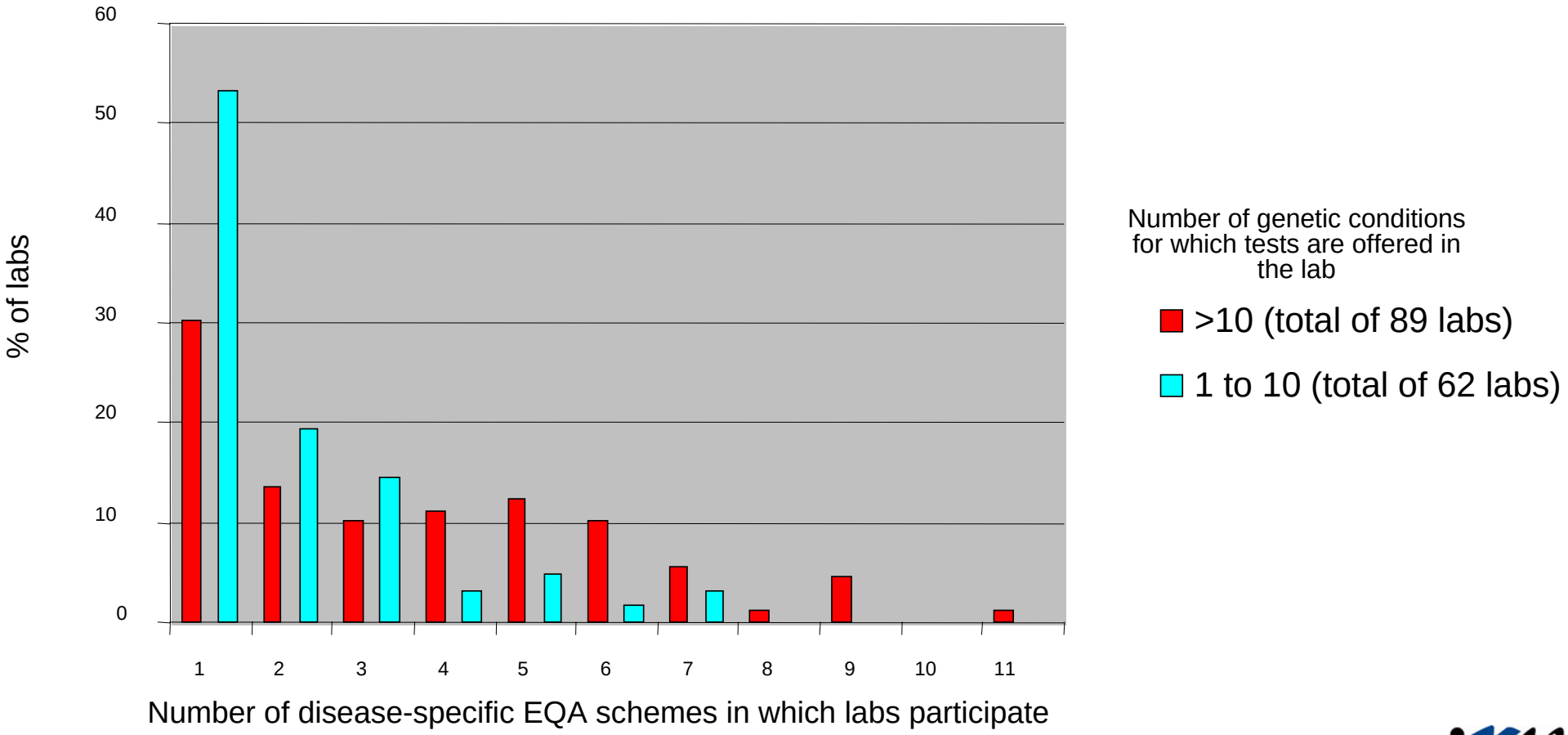
Current Weaknesses identified

1. Lack of quality assurance
2. Need for networking
3. Lack of certified reference materials
4. Need for normative and regulatory framework application
5. Impact of patents

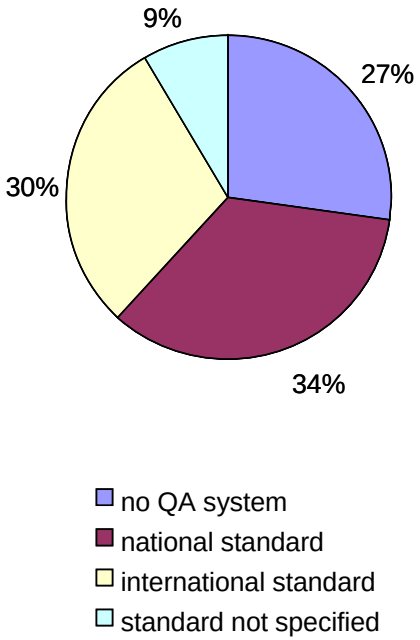
1. Lack of quality assurance

Participation in EQA schemes of different diseases during the years 1998-2003

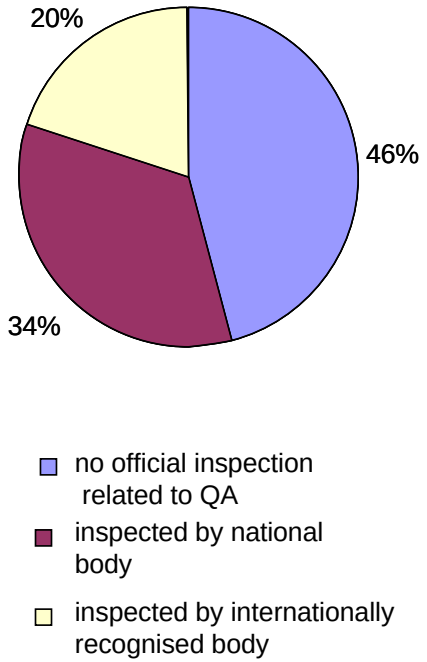


1. Lack of quality assurance

QA implementation



Lab inspection



*Need for **harmonisation of EQA schemes** bringing together existing schemes and for **reinforced accreditation (according to ISO 17025/ ISO15189)***

2. Networking/Collaboration

- **CRMGEN project**
- **EU Network of Excellence EUROGENTEST**

(Unit 1: Quality Management and Accreditation/certification of genetic testing)

- **IFCC, WHO**

3. Lack of Certified Reference Materials

NIST
SRM 2390 - DNA profiling
SRM 2391a - PCR-based DNA profiling
SRM 2392 - PCR and sequencing of mtDNA
SRM 2394 - Heteroplasmic mitochondrial DNA
SRM 2395 -Human Y DNA profiling
SRM 2399 - Fragile X triplet repeat

JRC-IRMM
IRMM/IFCC-490, IRMM/IFCC-491,IRMM/IFCC-492 for G20210A Factor II mutation under final characterisation

WHO
Factor V Leiden R506Q (Nov 2004)
Factor II G20210A mutation
Fragile X syndroma (CGG triplet expansion)

DNA-based candidate RMs evaluation (CRMGEN)

Disorder/gene	RMs to be developed
Cystic Fibrosis/CFTR	ΔF508, G542X, G551D, N1303K
Haemochromatosis/ HFE	C282Y, H63D
Fragile X syndrome/FMR1	Normal, premutation, expansion
Sickle cell anaemia, HBB	HbS
Beta thalassaemia, HBB	Codon 39 (C->T), IVSI-110 (G->A)
Factor V Defect/FV	R506Q, Factor V “Leiden"
Huntington Disease/HD	CAG Expansion
Hereditary non-polyposis colon cancer/ HMLH1-2	Representative nonsense mutations
Spinocerebellar ataxias/SCA1-7	CAG Expansions
Duchenne Musc.Dystr. /DMD	Deletions, duplications

Ethical issues in sourcing raw materials from human donors

Issue related to the choice and coverage of target populations

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4. Need for normative and regulatory framework application

- Lack of accreditation (ISO 17025/ ISO15189)
- Laboratory practice harmonisation (methods validation)
- Application/Interpretation of the IVD directive

The IVD Directive of the EC (98/79/EC) (12/2003), requires that " The traceability of values assigned to calibrators and/or control materials must be assured through available reference measurement procedures and/or available reference materials of a higher order" (Annex I A.3).

5. Impact of patents

- On development of control materials
- On development of home-brew methods
- Kit control materials