

Guidelines for Quality Assurance in Molecular Genetic Testing

The Organization for Economic Cooperation and Development

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Agenda

1. US and International Laboratories / Testing
2. Journey to Guideline Development
3. Salient features of the Guideline
4. Potential implications for genetic testing
in the US

Test Availability: GeneTests

<http://www.genetests.org> as of 7/15/2007 - voluntary submission

- **Number of Diseases for which testing available: 1446**

Clinical Tests	1155	Research only Tests:	291
United States	934 (205 laboratories)		
Non US Only	221 (236 laboratories)		

- **Number of Clinical Tests listed: 1155**

- Testing for disease available from only 1 lab 329
- Testing for disease available from 2 – 5 labs 470
- Testing for disease available from > 5 labs 356

www.genetests.org, as of 7-15-2007

What is the Organization for Economic Cooperation and Development? (OECD)

- Governmental Organization - established in 1961
- Origin traced to post-WWII Marshal Plan
- 30 member countries + cooperative arrangements with 70+ others
- Focus is to promote economic growth and financial stability
- Biotechnology and health care viewed as key sectors
- Molecular genetic testing viewed as important and evolving
- Lack of an international framework



OECD Guidelines for Quality Assurance in Molecular Genetic Testing

- Promote minimum standards
- Facilitate mutual recognition of quality assurance frameworks
- Strengthen international co-operation to facilitate cross-border flow of patient specimens for medically necessary testing
- Increase public confidence in the governance of molecular genetic testing

The Journey to Guideline Development

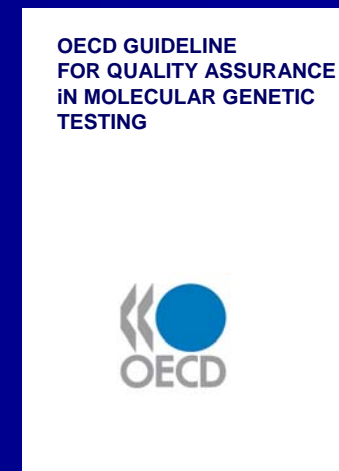
Focus: Molecular genetic testing for medical (disease-related) conditions



2000



2005



2007

Quality Assurance and Proficiency Testing for Molecular Genetic Testing *(Evidence for Policy Development)*



Total: 827 Responses

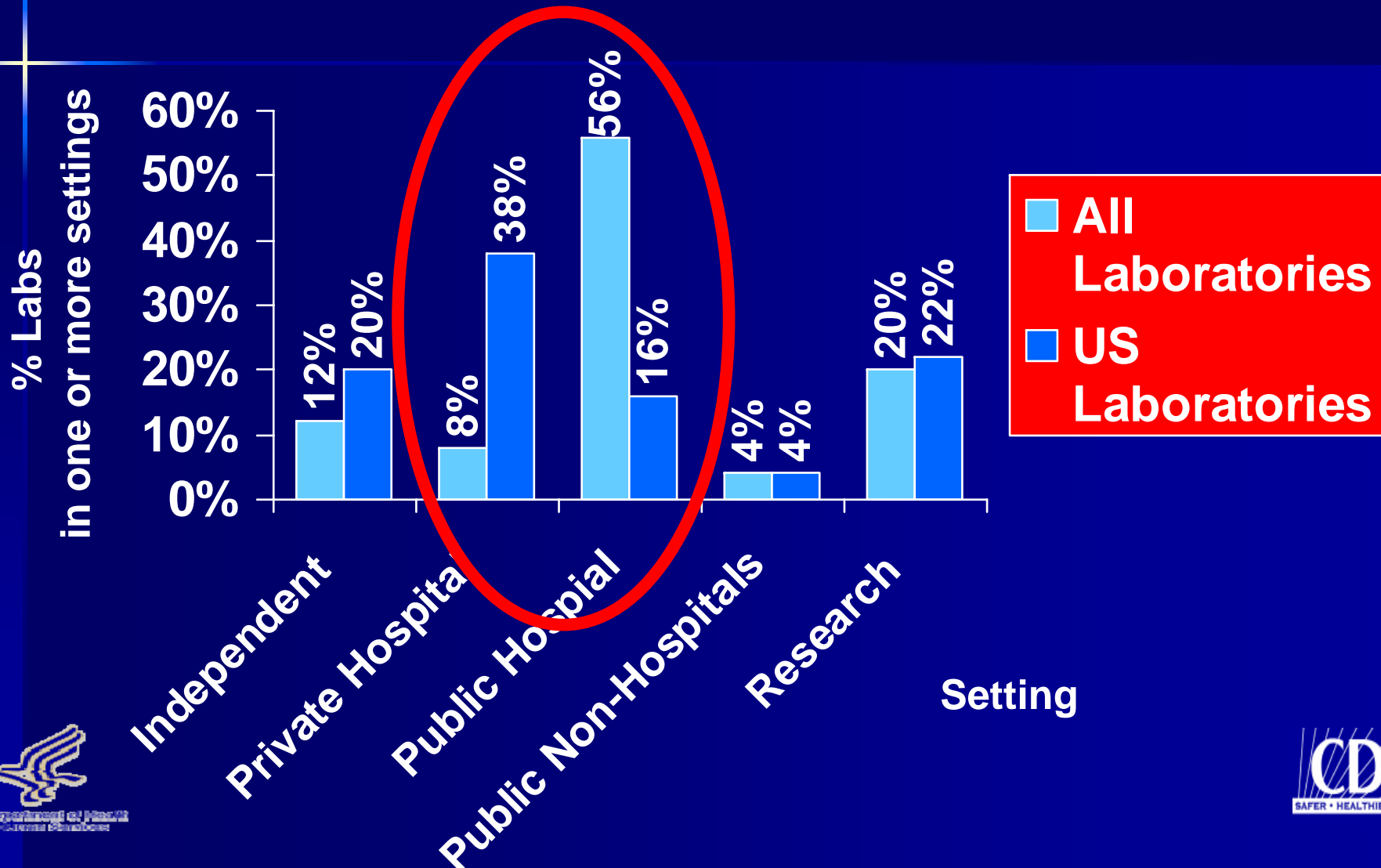
US: 226 Responses

2005

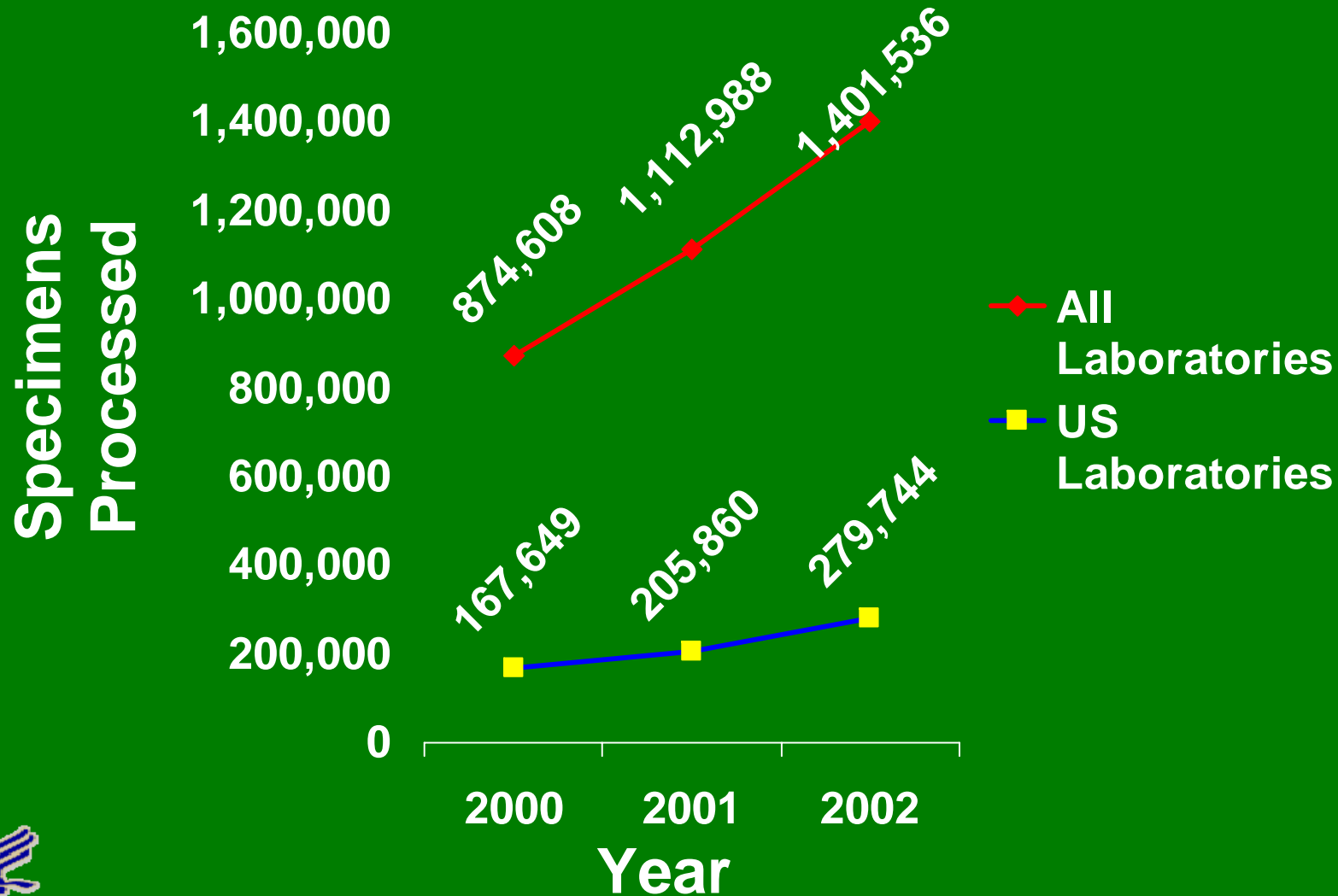
<http://www.oecd.org/dataoecd/25/12/34779945.pdf>

Laboratory Practice Settings

(from the survey)



Specimens Processed for Testing (from the survey)



Cross-Border Flow of Samples *(from the survey)*

In 2002

Number of samples sent out of country

Total: 18,000

US: 5030

Percentage of laboratories sending samples out of country

All Countries: 67% (554 out of 827)

US: 28% (63 out of 226)



Quality Assurance Index

<u>Information element</u>	<u>% Total complying</u> <u>(n=827)</u>	<u>% US complying</u> <u>(n=226)</u>
Standard operating Procedures developed	94	88
Standard operating procedures reviewed by director	82	98
Laboratory report is issued	95	96
Report is reviewed by director	84	94
Turnaround time data is collected	81	83

Reporting Quality Index

General elements

% Total complying

% US complying

Basic Elements

Two unique identifiers

100

100

Result

95

92

Added Specificity

Date of Birth

88

100

Reason for testing

89

94

Statement of limitations

78

79

Director's signature

84

94

Implications for other family members

61

64

Also Useful

Date of report

99

100

Sample collection date

86

98

Suggestion for further testing

82

79

Variables associated with higher quality indices

- **Standard Operating Procedures exist**
- **Standard operating procedures are reviewed by director**
- **Laboratory test result report is issued (written or electronic)**
- **Laboratory test result report is reviewed by director**
- **Offering of prenatal / pre-implantation testing**
- **Affiliation with a genetics unit**
- **Licensing and accreditation**
- **Participation in proficiency testing**
- **Not primarily in a research setting**
- **Maintaining data on turnaround time**
- **Lab director has an MD or PhD**
- **Lab director is certified**
- **Lab director has formal training**
- **Lab technicians have a university degree**
- **lab technicians have relevant training**

Official Recognition of Laboratories

	<u>All countries</u>	<u>US</u>
Licensed / Certified	49%	98%
Accredited	54%	84%

Barriers to Accreditation (one or more reasons given)

	All countries	US
	<u>n=362</u>	<u>n=23</u>
In process	8%	14%
Cost too high	64%	54%
Not required	46%	45%
Other	28%	14%

Guideline for Quality Assurance in Molecular Genetic Testing

OECD GUIDELINE
FOR QUALITY ASSURANCE
IN MOLECULAR GENETIC
TESTING



"Prior to form acceptance by the US and other countries of the OECD, there was a significant comment period and formal discussion"

<http://www.oecd.org/dataoecd/43/6/38839788.pdf>

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OECD Guidelines for Quality Assurance in Molecular Genetic Testing

Components

1. General principles and best practices
2. Quality assurance systems
3. Proficiency testing
4. Quality of result reporting
5. Education and training standards
6. Annotations

Organization:

Principles (1,2,3...): Primarily directed to governments

Best Practices (i, ii, iii): Primarily directed to laboratories and other entities



U.S. Department of Health
and Human Services



General principles and best practices

A.2 - Molecular genetic testing should be delivered within the framework of healthcare.

A.i - Regulatory/professional bodies should, as appropriate, review whether instruments available to manage a quality assurance framework require adaptation for molecular genetic testing

A.3 - All molecular genetic testing services should be provided and practices under a quality assurance framework

A.iii - Molecular genetic test results should be reported back to the referring health care professional to enable counseling and healthcare decision making

Quality Assurance Systems in Molecular Genetic Testing

B.1 - Governments and regulatory bodies should recognize that accreditation of medical laboratories is an effective procedure for assuring quality.

B.i. - All laboratories reporting molecular genetic testing results for clinical care purposes should be accredited or hold an equivalent recognition. (additional clarification for role of research laboratories)

B.9 - Governments should encourage international collaboration for the development and validation of molecular genetic tests.

B.viii - Laboratories should cooperative (internationally) to collect, develop, verify and make available reference materials



Proficiency Testing: Monitoring the Quality of Laboratory Performance

C.1 - The performance of laboratories offering clinical molecular genetic tests should be measured.

C.iii - Proficiency testing schemes should be structured to assess all phases of the laboratory process, including result reporting.

C.4 - Accreditation or equivalent recognition should be the basis for the international recognition of proficiency testing scheme and providers.

C.v - Laboratories should participate in a proficiency testing scheme for every test offered, when such schemes exist. When not available, they should participate in alternate methods.



Quality of Result Reporting

D.1 - All laboratories should issue molecular genetic testing results in the form of a written and/or electronic report to the referring health professional

D.i - Reports should communicate information effectively taking into account that the recipient may not be a specialist.

D.2 - Where reports are issued directly to patients, it should be encouraged that laboratories recommend consultation with an appropriate health care professional.

D.4 - The interpretation of the result should be appropriate to the individual and clinical situation and based on objective evidence.

Education and Training Standards for Laboratory Personnel

- E.2 - Standards for laboratory accreditation or other equivalent recognition should require that all molecular genetics personnel have a combination of education, training, skills, and experience that ensures their competence.**

- E.4 - Development of educational and training programmes should be encouraged where they do not exist.**

- E.vi. Comparison of education and training systems between jurisdictions should be facilitated as a means to establish equivalence.**

- E.5 - Relevant government or professional authorities should recognize medical genetics as a discipline comprising both a clinical and a laboratory specialty.**



Next Step: Implementation

OECD Perspective

- Each country charged to implement
- Follow up meeting in two years

US Perspective

- Needs: Access to quality testing, validation, and reference materials.

Eurogentests

- Road show: OECD Guidelines

International Organization for Standardization (ISO)

- Workgroup 1 (Quality and Competency of Medical Laboratories), Technical Committee 212 exploring relevance

Promoting Availability, Access, and Quality Potential Benefits to the US?

1. Encouraging development of a framework for mutual recognition
 - Quality assurance and professional competence
2. Promote options for proficiency testing or alternate assessment
3. Promote collaboration relevant to availability and access to reference materials
4. Enhance opportunities for collaboration for the development and validation of tests, particularly those for rare diseases
5. Promote importance and role of research laboratories in working with the clinical sector



Questions?



Thank You