



Current Oversight of Genetic Testing

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Genetic Testing Oversight Issues

- Genetic revolution – New scientific, medical, social, legal, ethical concerns
- Genetic evolution – potential benefits and potential risks

Challenge:

Bring our public policies in line with the genetic revolution

Quality Testing



Access

Cost



Positive aspects of regulation/oversight

- Protection of the public – Sanction
- Level playing field – Minimum Standards
- Provide benchmarks for good practice
- Monitor attainment of goals (PT, QA, QC)



Negative aspects of regulation/oversight

- Always out of date
- Focus on process rather than outcome
- Increases costs
- May not prevent bad outcomes
- May impede new technology
- May impose rigid requirements (personnel)



Background

- CLIA enacted - 1988
- NIH/DOE Task Force Report - 1997
- CLIAC recommends changes to CLIA – 1998
- SACGT recommends increased oversight – 1999
- CDC Notice of Intent – May 2000



NIH/DOE Report: Areas of Concern

- Appropriate introduction of new genetic tests into clinical practice
- Adequate regulation of laboratory testing
- Increasing healthcare provider and patient understanding of genetics
- Maintaining access to quality testing for rare diseases

Proposed CLIA Genetics Specialty



- **Definition – What is included and excluded?**
- **General requirements**
 - **Documentation of clinical validity**
 - **Person authorized to order a genetic test**
 - **Informed consent**
 - **Confidentiality**
 - **Genetic counseling**
- **Requirements for specific testing phases**
 - **Pre-analytic phase**
 - **Analytic phase**
 - **Post-analytic phase**



CLINICAL VALIDITY

1. Lab director's role in documenting clinical validity:

- no responsibility for documentation; or
- ensures that documentation exists in literature;

or

- documents clinical validity of all tests offered

2. Should clinical validity be established before a test can be offered?

- mixed opinions on this issue



INFORMED CONSENT

1. Should CLIA require documentation?

- Is documentation an integral part of laboratory practice?
- Is CLIA the place to “police” ordering physicians?

2. Should the laboratory’s role in assuring documentation of IC include:

- documentation that an authorized person has obtained IC?
- alerting health care providers when IC is needed?
- providing IC forms to health care providers?
- documenting the adequacy of IC forms?

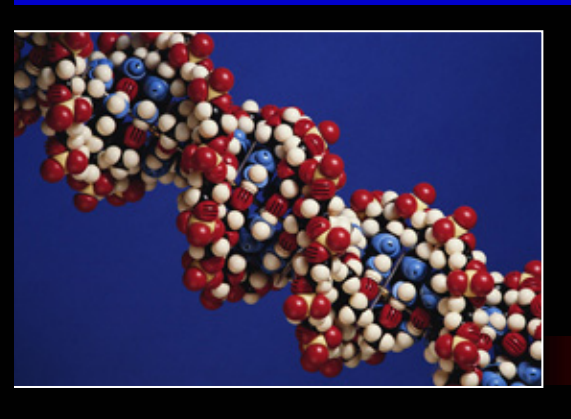


CLIAC Issues

- QA/QC/PT
- Re-use of samples
- Authorized person to order genetic tests
- Confidentiality
- Test requisition and clinical information
- Result reporting
- Record and specimen retention

Secretary's Advisory Committee on Genetic Testing - Recommendations

- Strengthen human protection in research
 - IRB review and informed consent
- Augment CLIA to address genetic testing
- Establish FDA review of all new genetic tests
- Develop information on the clinical utility of genetic tests



Current US Oversight of genetic testing

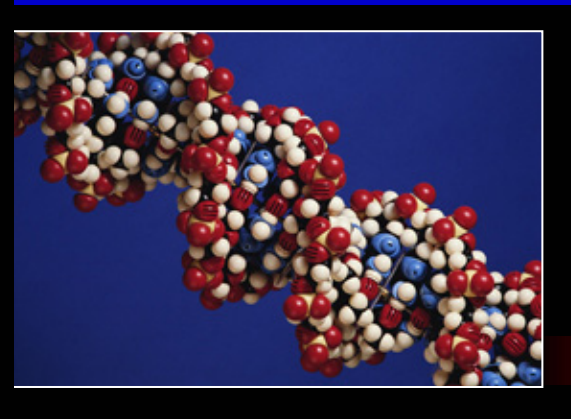
- CLIA for laboratories
- FDA for kits and devices
- IRB for patients in research
- NYS – QC, personnel, test validation, test review and approval
- Professional guidelines and standards of practice (AMP, ACMG, CAP, etc.)

International oversight of genetic testing

- UNESCO
- OECD
- European Commission
- ISO
- ILAC/WHO
- Eurogenetest

International oversight of genetic testing

- WHO
- Professional guidelines and standards of practice
- Sweden/Norway Biobanks
- Others



Secretary's Advisory Committee on Genetics, Health and Society – Additional Concerns

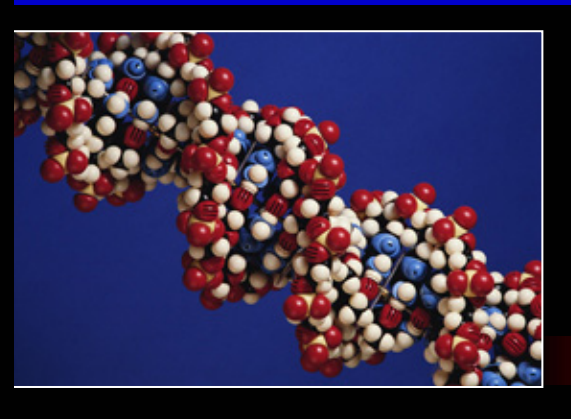
- Coverage and Reimbursement
- Large Population Studies
- Pharmacogenomics
- Direct-to-Consumer Marketing



The Genetic Testing Environment

- Rapid advances in genetic technology
- Molecular basis of both rare and common disorders
- Commercialization of testing
- Genetic testing no longer for rare diseases or conditions

Public Policy Challenges



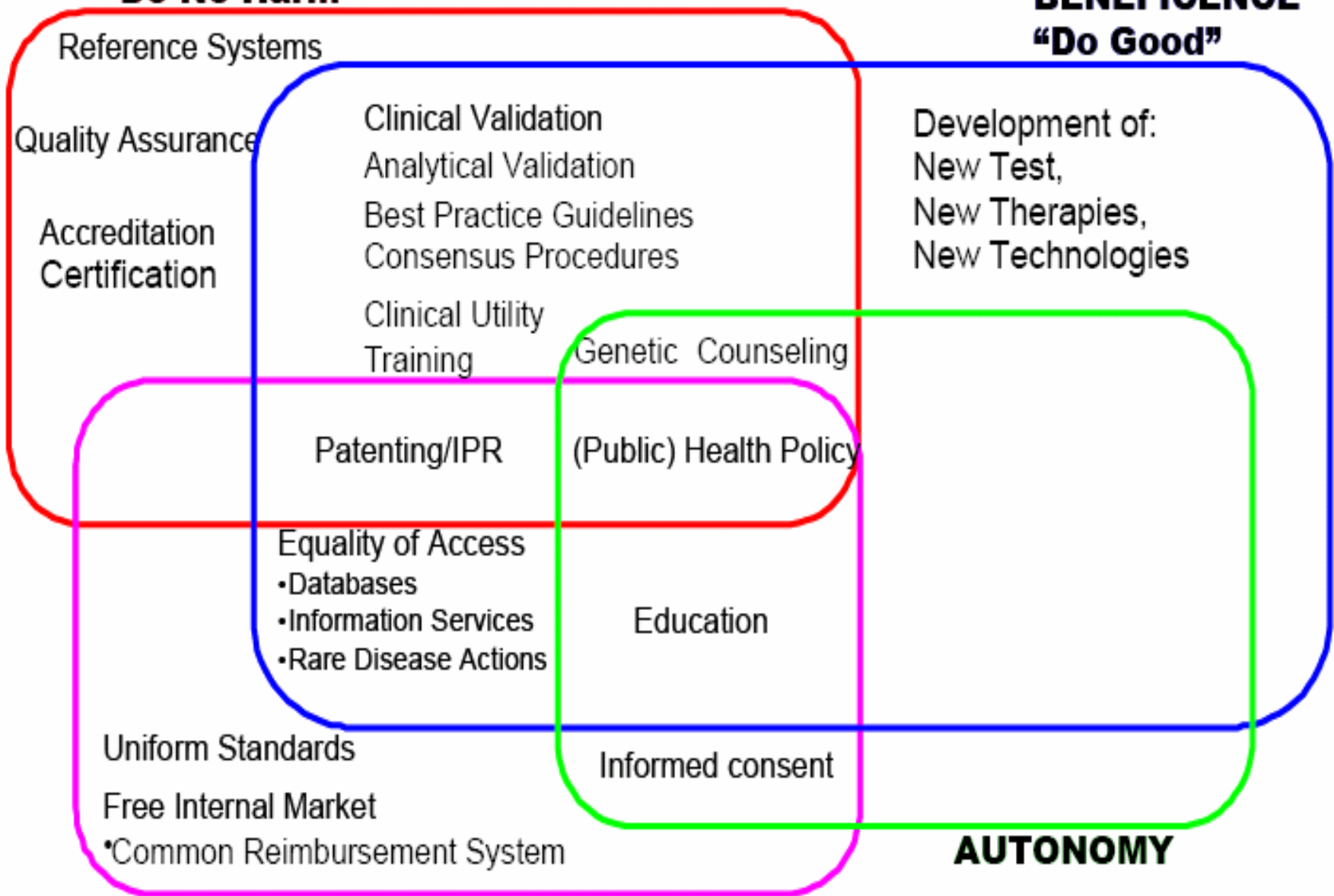
- Balancing access, costs, and quality of services
- Clarifying roles of government, professional organizations, advocacy groups in ensuring adequate oversight
- Dealing with new issues posed by genetic testing
- Obtaining data needed to guide policy decisions

NONMALEFICENCE

“Do No Harm”

BENEFICENCE

“Do Good”



JUSTICE

AUTONOMY

