

CDC Update

Devery Howerton, Ph.D.

Director, Division of Laboratory Science and Standards

CLIAC Meeting

March 2, 2011

Atlanta, Georgia

The findings and conclusions in this report are those of the author and do not necessarily represent the official position of the Centers for Disease Control and Prevention.

Office of Surveillance, Epidemiology, and Laboratory Services

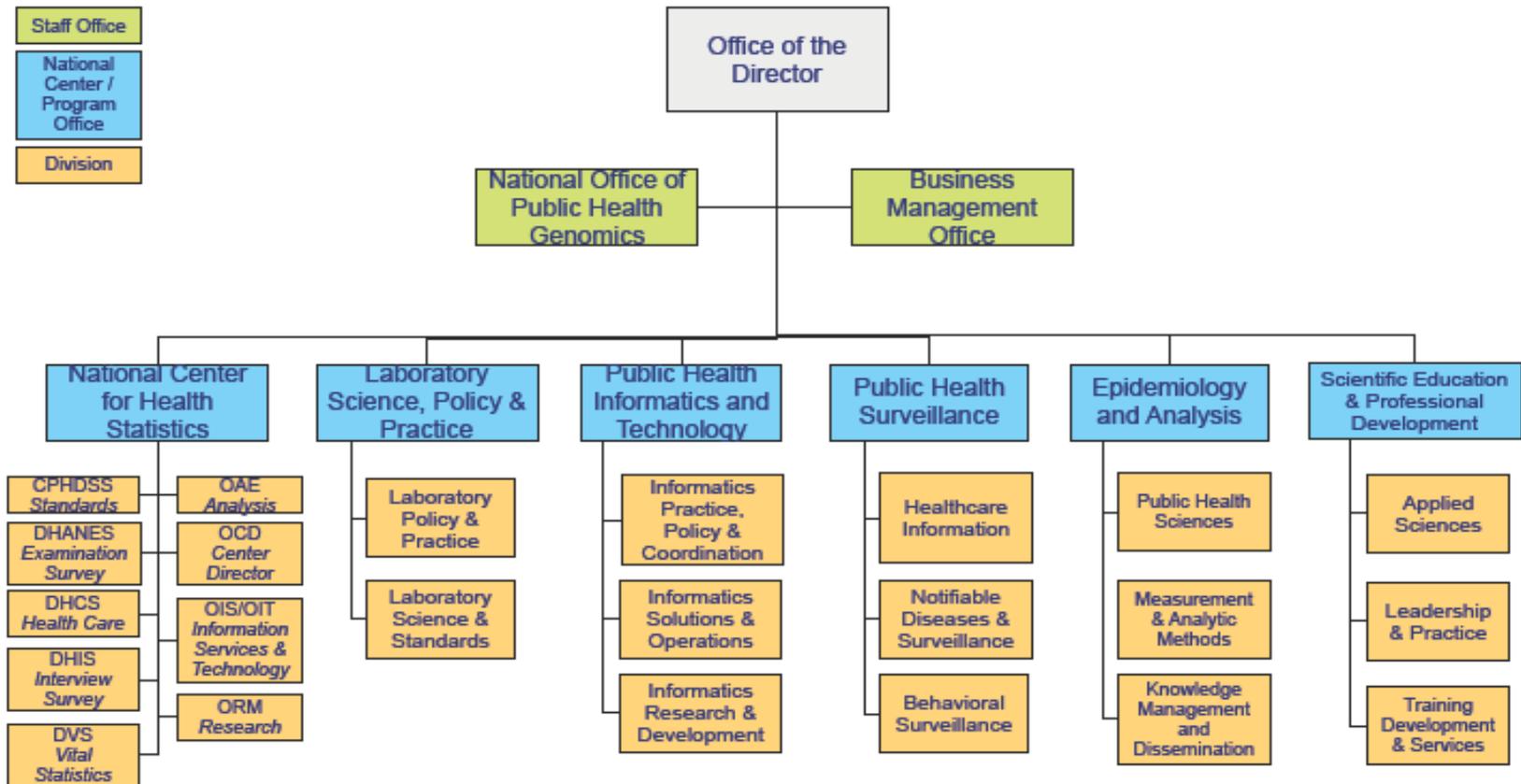
Laboratory Science, Policy and Practice Program Office



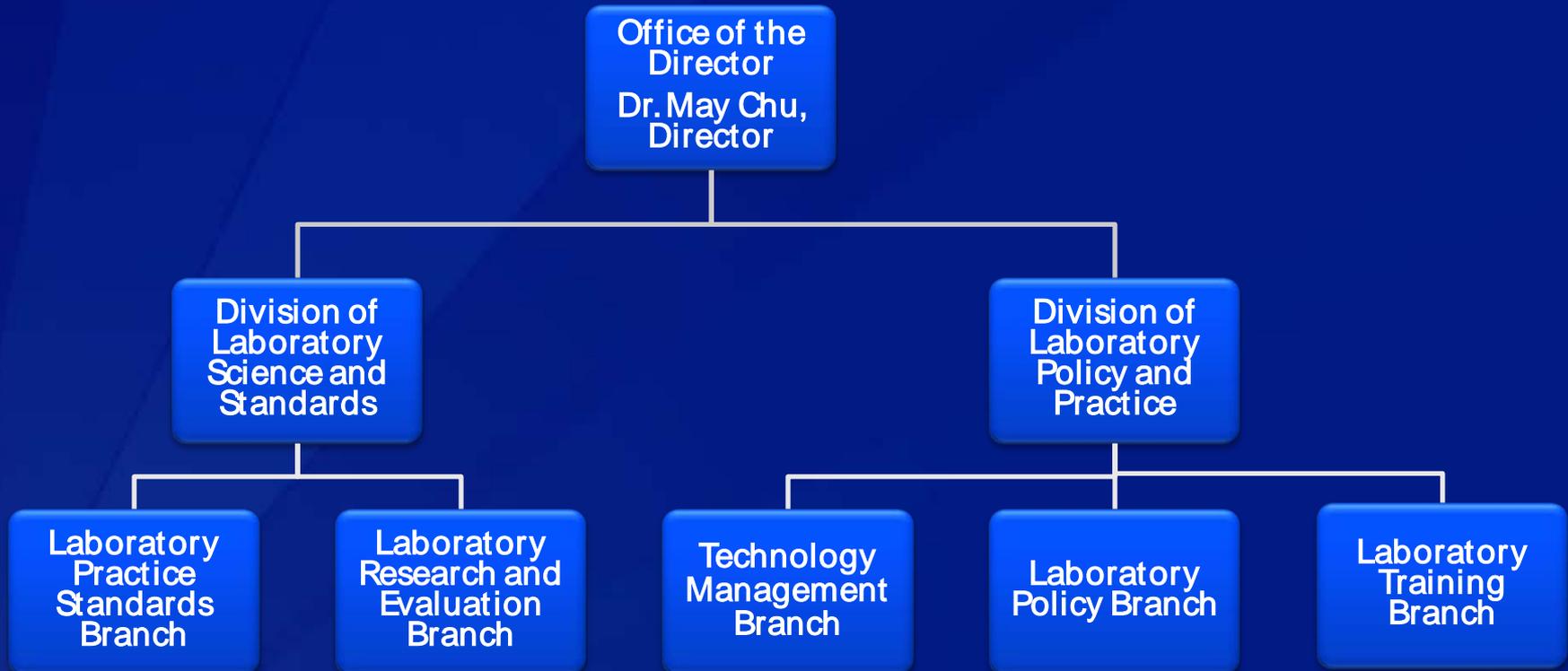
Outline

- ❑ Organizational changes
- ❑ Status of proposed rule for proficiency testing
- ❑ Update on cytology projects
- ❑ Development and promotion of good laboratory practices guidelines/products
 - Waived testing
 - Molecular genetic testing
 - Biochemical genetic testing

Office of Surveillance, Epidemiology and Laboratory Services



Laboratory Science, Policy & Practice Program Office (LSPPPPO)



Outline

- ❑ Organizational changes
- ❑ **Status of proposed rule for proficiency testing**
- ❑ Update on cytology projects
- ❑ Development and promotion of good laboratory practices guidelines/products
 - Waived testing
 - Molecular genetic testing
 - Biochemical genetic testing

Status of Proposed Rule for Proficiency Testing

- ❑ September 2010 – CLIAC provided 22 recommendations to HHS
- ❑ CLIAC recommendations addressed:
 - Analyte inclusion/prioritization
 - Criteria for acceptable performance/grading criteria
 - Microbiology PT requirements
 - PT referral
- ❑ CDC and CMS staff working together on project plan for development of the proposed PT rule

Outline

- ❑ Organizational changes
- ❑ Status of proposed rule for proficiency testing
- ❑ **Update on cytology projects**
- ❑ Development and promotion of good laboratory practices guidelines/products
 - Waived testing
 - Molecular genetic testing
 - Biochemical genetic testing

Cytology Projects

Two cooperative agreement awards funded in 2010-2011

- ❑ College of American Pathologists
 - Survey all cytology labs
 - Review current practices
 - Analyze responses and post on CAP website
 - Convene consensus conference in 2011
- ❑ Michigan Public Health Institute (MPHI)
 - Survey of Pap smear providers (clinicians, nurses)
 - Use of laboratory services, interpretation of test reports and effect on clinical decision-making
 - Partnering with MI Cancer Consortium



- ❑ Additional questions will be posted on CAP website
- ❑ Announcement for The GYN Practices consensus conference – June 4, 2011; Chicago, IL
- ❑ Workgroups
 - Monitoring Diagnostic Rates
 - Rescreening/Retrospective Review
 - Pap PT
 - Biopsy Correlations
 - Monitoring HPV testing

MPHI Cytology Survey Status

- ❑ Plan to survey 5,000 physicians and 3,000 nurses
- ❑ Looking for a 30-40% response rate
- ❑ Survey should be complete by the end of March
- ❑ Includes questions about
 - Specimen collection
 - Test ordering practices
 - Test reports
 - Consultation from laboratory
 - HPV testing practices

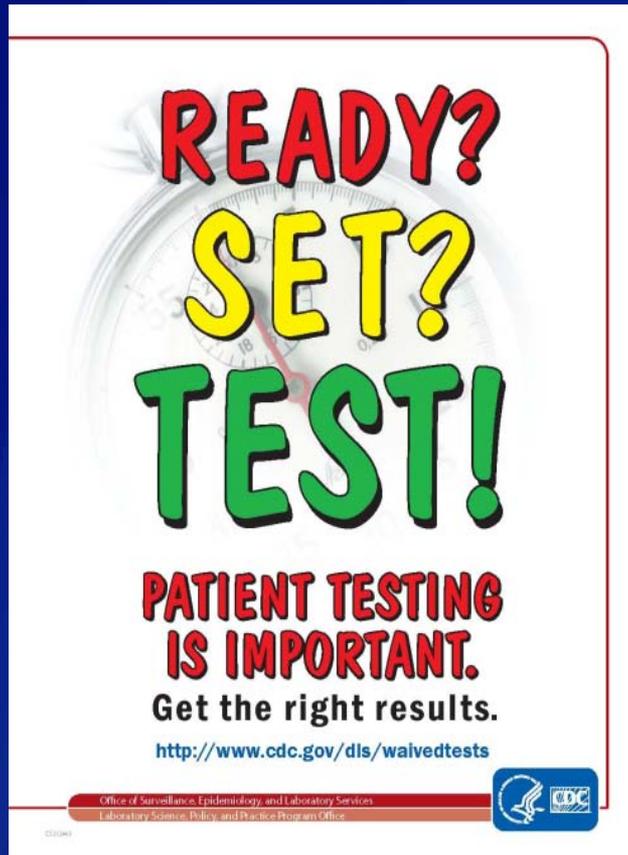
Outline

- ❑ Organizational changes
- ❑ Status of proposed rule for proficiency testing
- ❑ Update on cytology projects
- ❑ **Development and promotion of good laboratory practices guidelines/products**
 - Waived testing
 - Molecular genetic testing
 - Biochemical genetic testing

Promotion of Good Laboratory Practices for Waived Testing Sites

- ❑ Following the 2005 MMWR publication, CLIAC suggested educational materials be developed
- ❑ Poster and postcards completed in 2009
 - 700 posters distributed to date
 - 800 postcards distributed to date
- ❑ Educational booklet with job aids published in 2010
 - 100 booklets distributed to date

Good Laboratory Practices for Waived Testing Sites



PATIENT TESTING IS IMPORTANT.

Get the right results.

**READY?
SET?
TEST!**

- Have the latest instructions for ALL of your tests.
- Know how to do tests the right way.
- Know how and when to do quality control.
- Make sure you do the right test on the right patient.
- Make sure the patient has prepared for the test.
- Collect and label the sample the right way.
- Follow instructions for quality control and patient tests.
- Keep records for all patient and quality control tests.
- Follow rules for discarding test materials.
- Report all test results to the doctor.

<http://www.cdc.gov/dls/waivedtests>



Poster and postcard

Educational booklet with job aids

Future Waived Testing Products

- ❑ **Ready? Set? Test! On-line Training**
 - Recommended practices for performing waived testing
 - CE credits will be available
- ❑ **To Test or Not To Test? Booklet on Considerations for Waived Testing**
 - Issues to consider prior to performing waived testing
 - Assistance to those who want to initiate or direct testing under a CLIA Certificate of Waiver.

Promotion of Good Laboratory Practices for Molecular Genetic Testing

- ❑ **MMWR published in 2009**
 - 1,200 distributed to date; also available on-line
 - CE credits awarded to over 2,500 individuals
- ❑ **Created fact sheets for laboratory professionals, health professionals, and consumers**
 - 1,000 distributed to date
- ❑ **Published commentaries and articles**
- ❑ **Presented at numerous professional conferences**
- ❑ **Writing new CLSI guideline (MM20) on quality management for MGT (Dr. Chen)**

Molecular Genetic Testing

Top 10 Recommendations for Laboratories Performing Molecular Genetic Testing



Top 10 Recommendations for Laboratories

- 1. Provide information regarding the laboratory's molecular genetic tests to users to facilitate appropriate test selection and requests, specimen handling and submission, informed decisions, and patient care. Test information to be provided should include:**
 - Information necessary for selecting and requesting appropriate tests;
 - Information on appropriate collection, handling, transport, and submission of specimens;
 - Patient information that the laboratory needs to perform the test and report test results;
 - Implications of test results for the patient's family members, when applicable;
 - Laboratory and/or genetic consultation regarding test selection and ordering, specimen submission, result interpretation and implications; and
 - Cost of testing, whenever possible.
- 2. Ensure test requests solicit information needed for selecting appropriate test methods, determining the mutations or variants to be tested, interpreting test results, and timely reporting of test results. Information to be solicited includes:**
 - All information required by CLIA; and
 - Indications for testing, relevant clinical and laboratory information, patient race/ethnicity, family history, and pedigree when applicable.
- 3. Ensure adequate establishment and verification of analytic performance specifications before introducing any new molecular genetic test for patient testing, and document available information on clinical validity.**
- 4. Implement specific quality control (QC) practices in addition to meeting the applicable general CLIA requirements:**
 - Ensure control materials are comprehensive and resemble patient specimens when possible;
 - Include an extraction control in any test that has a nucleic acid extraction step;
 - Perform control procedures each time patient specimens are tested;
 - Ensure appropriate alternative control procedures when control materials are not available; and
 - Ensure adequate procedures to monitor unidirectional workflow for amplification procedures and prevent cross-contamination.
- 5. Participate in available proficiency testing for each molecular genetic test that the laboratory performs, and perform alternative performance assessments for those molecular genetic tests for which no proficiency testing program is available.**

Office of Surveillance, Epidemiology, and Laboratory Services
Division of Laboratory Science and Standards



CS210245

- 6. Molecular genetic test reports should provide information necessary for accurate understanding and interpretation of test results. In addition to meeting CLIA test report requirements, laboratories should:**
 - Include the recommended additional information (e.g., test method, performance specifications and limitations, result interpretation and implications for relatives); and
 - Maintain an up-to-date database of the laboratory's molecular genetic tests and provide updates to users when knowledge advancement affects performance specifications or result interpretation.
- 7. Ensure adequate retention of test reports, records, and tested specimens for quality assurance and quality assessment:**
 - Molecular genetic test reports should be retained for a minimum of 25 years after the date reported.
 - Previously tested specimens that are stable should be retained until the next proficiency testing event or the next alternative performance assessment is performed.
- 8. Ensure confidentiality of all patient information, including information regarding family members. This is especially important when patient test results are used for the assessment and care of family members.**
- 9. Comply with applicable federal, state, and local requirements regarding direct-to-consumer genetic testing:**
 - Verify that test requests are from authorized persons;
 - Release test results only to persons authorized by applicable laws and regulations to receive test results, persons responsible for using the test results, and/or the referring laboratory; and
 - Follow accepted professional guidelines.
- 10. Ensure personnel have appropriate qualifications needed to fulfill their responsibilities for the high complexity molecular genetic testing they perform:**
 - Laboratory Directors must, at a minimum, meet the CLIA requirements for laboratory directors of high complexity testing.
 - Technical Supervisors should have qualifications equivalent to the CLIA qualification requirements for clinical cytogenetics technical supervisors or current certification in molecular genetic testing.
 - Clinical Consultants must meet the minimum qualifications required by CLIA and should have relevant training, experience, or both with the testing for which they provide consultation.
 - General Supervisors must fulfill the CLIA requirements for high complexity testing and should have specific training and/or experience with the molecular genetic testing the laboratory performs.
 - Testing Personnel must meet the CLIA qualification requirements for high complexity testing and have training/competency with the molecular genetic testing they perform.

For additional information go to:
<http://www.cdc.gov/dls/moleculartesting>

September 2010



GENETIC TESTING

What you need to know



1. Do I need genetic testing?

Genetic tests are used to:

- Detect or confirm genetic diseases and inherited conditions
- Predict risk of disease for an individual and their children or relatives
- Select treatments and monitor patient's response
- Predict the outcome of disease

2. Who can help me choose the right test?

Genetic tests for medical purposes should be ordered and interpreted with the assistance of your doctor. Talk to your doctor before having a genetic test performed.

The laboratory should help you and your doctor to select the right genetic test by telling you the following:

- The genetic tests that they offer
- Explanation of how each test works and what the results will mean
- Instructions on how specimens must be collected and sent to the laboratory
- An estimated cost

3. What information will I need to give to the laboratory?

The laboratory requires certain information before performing a test. This may include:

- Race/ethnicity
- Family history
- Health information

Several state and local governments require that you or your guardian sign an agreement that says you understand and agree to have a laboratory test performed.

4. What will the results tell me?

Your laboratory results should include:

- The name of the test and how well it works
- The results of the test and what they mean
- Whether genetic counseling is indicated and where to get counseling
- Whether your family may also be affected

5. Will my results be confidential?

The laboratory must, by law, keep your personal information and test results confidential.

In instances where your results can be used to help care for a family member, the laboratory should first request permission from you to share the results.

6. How can I learn more?

Ask your doctor.

Visit www.cdc.gov/dls/moleculartesting



Office of Surveillance, Epidemiology, and Laboratory Services
Laboratory Science, Policy, and Practice Program Office

CS21629

Health Professionals and Genetic Testing

What you need to know



1. Why perform molecular genetic testing?

Molecular genetic testing may be performed to detect and confirm genetic diseases or heritable conditions, to identify increased risks for health problems, and to choose or monitor response to treatment for a condition.

2. What information should the laboratory provide to help me make an informed decision on choosing a molecular genetic test?

- the genetic tests that they offer and the purposes of these tests
- how the test works and what the test will tell you
- who will benefit from testing
- whether the test is FDA cleared or approved
- the laboratory's federal certification or accreditation status
- how to collect and send the laboratory a specimen for testing
- what advice or consultation is available from the laboratory
- an estimated cost

3. What information will the laboratory require from me?

- several state and local governments require the patient or the patient's guardian sign an agreement that says they understand and agree to have a laboratory test performed
- the patient's name or identifier, date of birth, and health information related to the test
- some tests need more information such as race and ethnicity, and family medical history

4. What should I expect the test results to include?

- the date of the report with the name and address of the laboratory
- the patient's name or identifier, date of birth and why the patient was tested
- the date and time the specimen was collected
- the name of the person that ordered the test
- the name of the test and how it works
- the results and an explanation of what they mean
- some tests may indicate a need for genetic counseling and whether the patient's relatives may be affected

5. Are molecular genetic test results confidential?

Yes. Laboratories must keep all patient information confidential, including genetic test information.

6. How long is genetic test information stored by the laboratory?

The laboratory should keep molecular genetic test results on file for at least 25 years for health care and quality assurance purposes.

For additional information go to:

<http://www.cdc.gov/dls/moleculartesting>

September 2010



Office of Surveillance, Epidemiology, and Laboratory Services
Division of Laboratory Science and Standards

CS21629

Genetic Testing Fact Sheet for
Consumers

Genetic Testing Fact Sheet for
Health Professionals

Future Molecular Genetic Testing Product

Good Laboratory Practices for Molecular Genetic Testing On-line Training –

- ❑ Will include recommended quality practices for performing molecular genetic testing
- ❑ CE credits will be available
- ❑ Targeted to laboratory professionals

Web Sites

- ❑ Waived Testing MMWR, educational booklet with job aids, and poster: www.cdc.gov/dls/waivedtests
- ❑ Molecular Genetic Testing MMWR, educational materials, and frequently asked questions: www.cdc.gov/dls/moleculartesting

Home | About CDC | Press Room | Funding | A-Z Index | Centers, Institute & Offices | Training & Employment | Contact Us

CDC Department of Health and Human Services
Centers for Disease Control and Prevention

Enter Search Term

DLS Content

- > DLS Home
- > Best Practices
- > CLIA
- > Genetics
- > Institutes
- > International
- > MASTER
- > Monitoring
- > MPEP
- > Publications
- > Quality Conferences
- > Training
- > Waived Tests

Other DLS Resources

- > Resource Links

Waived Tests

[Ready? Set? Test! Booklet](#)

[Ready? Set? Test! Poster, \[HTML Version\]](#)

[MMWR R&R Good Laboratory Practices for Waived Testing Sites](#)

Other materials being developed by CDC will be posted in the future.

Background

All facilities in the United States that perform laboratory testing on human specimens for health assessment or the diagnosis, prevention, or treatment of disease are regulated under the Clinical Laboratory Improvement Amendments of 1988 (CLIA) (1). Waived tests include test systems cleared by the FDA for home use and those tests approved for waiver under the CLIA criteria. Although CLIA requires that waived tests must be simple and have a low risk for erroneous results, this does not mean that waived tests are completely error-proof. Errors can occur anywhere in the testing process, particularly when the manufacturer's instructions are not followed and when testing personnel are not familiar with all aspects of the test



Home | About CDC | Press Room | Funding | A-Z Index | Centers, Institute & Offices | Training & Employment | Contact Us

CDC Department of Health and Human Services
Centers for Disease Control and Prevention

Enter Search Term

DLS Content

- > DLS Home
- > Best Practices
- > CLIA
- > Genetics
- > Institutes
- > International
- > MASTER
- > Monitoring
- > MPEP
- > Publications
- > Quality Conferences
- > Training
- > Waived Tests

Other DLS Resources

- > Resource Links

Molecular Genetic Testing

[MMWR R&R Good Laboratory Practices for Molecular Genetic Testing for Heritable Diseases and Conditions, June 12, 2009, Vol. 58, No. RR-6 | \[PDF Version\]](#)

- [Questions and Answers](#)
 - General
 - Laboratories
 - Healthcare Providers
 - Patients and Consumers
- [Top 10 Recommendations for Laboratories](#)
- [MMWR one pager](#)
- [Health Professional fact sheet](#)
- [Patient fact sheet](#)

Background

With the increasing use of genetic tests in healthcare services, concern has been raised regarding the adequacy of regulatory oversight and quality assurance measures in the area of molecular genetics laboratory testing. Since 1997, CDC, in collaboration with the Centers for Medicare & Medicaid Services (CMS), the Food and Drug Administration (FDA), and other federal agencies, has been working with stakeholder groups and organizations to promote the quality of genetic testing and improve the appropriate use of genetic tests in healthcare. Currently, laboratories performing molecular genetic testing are subject to the general requirements for high complexity testing under the Clinical Laboratory Improvement Amendments of 1988 (CLIA) regulations. While promoting high quality molecular genetic testing, additional voluntary guidance for good laboratory practices is needed to improve the quality of test performance and enhance the oversight of genetic testing under the current regulatory framework.



Biochemical Genetic Testing (BGT) & Newborn Screening (NBS) Good Laboratory Practice Recommendations

□ 2009: CLIAC BGT workgroup

- 13 experts representing key BGT & NBS perspectives
- Comprehensive evaluation of laboratory standards and guidelines

□ Feb. 2010 CLIAC meeting

- CLIAC review of workgroup report
- Recommendations for BGT and NBS for diagnosis and monitoring of inborn errors of metabolism (<http://wwwn.cdc.gov/cliac/default.aspx>)

□ May-Nov. 2010: Additional input to complement CLIAC recommendations

- Secretary's Advisory Committee for Genetics, Health, and Society (SACGHS)
- Secretary's Advisory Committee for Heritable Diseases in Newborns and Children (SACHDNC)
- Association of Public Health Laboratories

BGT-NBS MMWR Projected Timeline Next Steps



MMWR Guideline for BGT and NBS

□ Overall intent

- Provide good laboratory practice recommendations for genetic testing performed for screening, diagnosis, monitoring, and treatment of inherited metabolic disorders
- Complement 2009 MMWR guideline for molecular genetic testing (<http://www.cdc.gov/mmwr/pdf/rr/rr5806.pdf>)

□ MMWR recommendations will -

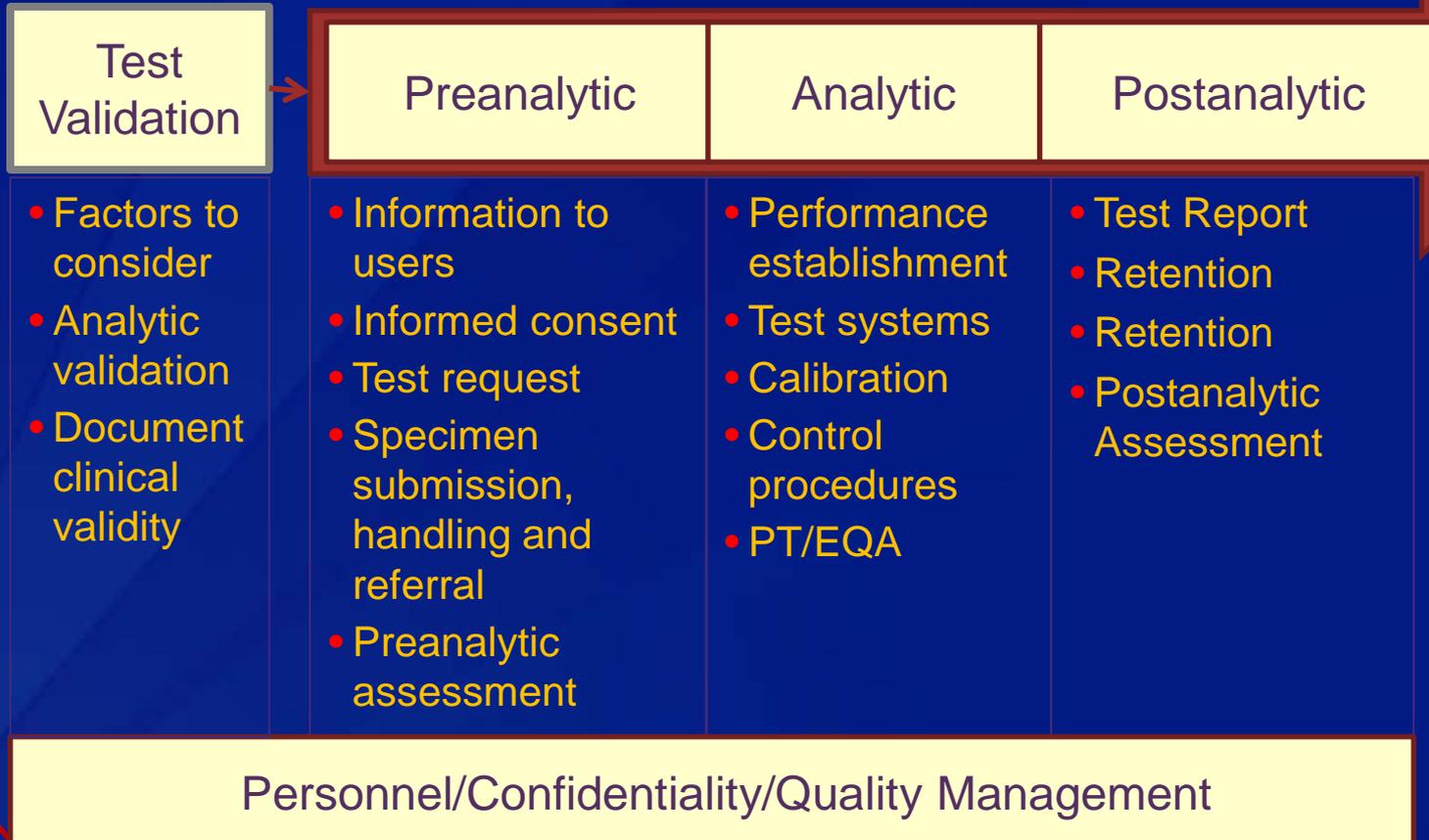
- Clarify applicable CLIA requirements
- Provide recommendations for QA practices in addition to CLIA
- Apply to testing for inherited metabolic disorders, but address BGT and NBS separately when practices are different

□ Expected outcomes -

- Improve quality of laboratory genetic services
- Enhance oversight for genetic testing under the current regulatory framework
- Improve healthcare outcomes from genetic testing

Highlights of Recommended Practices

Total Laboratory Testing Process



Summary

- ❑ PT proposed rule under development
- ❑ Cytology practice surveys underway
- ❑ CAP cytology consensus conference - June 2011
- ❑ Waived testing practice recommendations: booklet, & poster available; on-line tutorial in development
- ❑ MGT practice recommendations: fact sheets available; on-line tutorial in development
- ❑ BGT and NBS practice recommendations: MMWR in development

Questions?

Comments?

Office of Surveillance, Epidemiology, and Laboratory Services

Laboratory Science, Policy and Practice Program Office

