CDC Update
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CDC
• Progress in Genetics

• 2007 Conference - “Managing for Better Health”

• Research – “Defining Best Practices in Laboratory Medicine”
  ❖ Structure - Where are we now and where are we going?
  ❖ Process - How can we identify and implement Best Practices?
  ❖ Outcome - Can we make PT more effective?

• Workgroup – Impact of Rapid and Molecular Tests for Infectious Diseases on Public Health
Purpose: Facilitate translation of genetic tests from research to practice

Process: Collaboration - research laboratory, clinician involved in the study of the disease, a CLIA laboratory, patient advocacy group

1st Test: July 21 - Cornelia de Lange Syndrome  University of Chicago
Cost: $2400  Turn-around time: 10 weeks
Link to information: http://genes.uchicago.edu/DiagnosticTests.html

Other tests in Fall 2006:
- ATP7B sequencing for Wilson disease
- GP1BB sequencing for Bernard-Soulier syndrome

CETT Site: http://www.cettprogram.org/
Purpose: Explore CETT program for biochemical genetic tests

Concerns:
- Tests from limited number of laboratories
- Laboratories discontinuing testing services
- No process to move tests from the research to practice
- Materials for QC and PT are often not available
- Collaboration - biochemical and molecular testing laboratories
- Reimbursement and liability may restrict services, such as prenatal diagnosis
- Testing service information often not readily accessible.
- Shortage of trained personnel and training programs.
Recent Success

• Verified 14 Huntington Disease Cell Lines
• Verified Ashkenazi Jewish panel of 27 variants
• Coriell cell lines typed for Pharmacogenetic loci

Coming soon

• Fragile X

www.phppo.cdc.gov/dls/genetics/qcmaterials
Deriving Actionable Information from Testing
Focus: Genetic Testing

- Individual
- Family
- Health care provider
- Test Orderer
- "The Laboratory(ies)"
- Information Resources

Test Order
Test Report
Oversight of Genetic Testing

US Issues

• Analytical validity
• Clinical validity
• Home brew testing
• Internet (Tests to Public)
• Informed consent
• Education: Users of Lab Services
• Communication
• Use of Residual Specimen
• Balancing access and quality
Oversight of Genetic Testing

International Organization for Economic Cooperation and Development (OECD)

“Guidelines for Best Practices in Quality Assurance for Molecular Genetic Testing”

• 2002 Survey of 18 countries
• Principals and Best Practices
  ❖ General
  ❖ Quality Assurance
  ❖ PT
  ❖ Result Reporting
  ❖ Education and training of laboratory personnel

http://www.oecd.org/sti/biotechnology/qualityassurance
Purpose: To identify ways to enhance the management of laboratory practices and use of laboratory services to promote better health.

Assumptions:
- Current practices cannot cope with anticipated changes in technology, patient care options, knowledge management needs, and resource limits
- Laboratory services are essential to patient care and public health and appropriate and effective use of these services must be a high health care priority.
- Enhancements in informatics capability could allow the laboratory to play a major role in the coordination and continuity of care.

Topic Areas
Management systems, Informatics, Technology
Integrated laboratory services, Customer expectations
For Each Topic Area

Key Questions

- What are the major changes that have occurred in the past 5 years or are expected to occur in the next 5 years?
- What major changes need to occur in the next 5 years to enhance laboratory services?
- What are the expected benefits and costs of laboratory services if these changes do occur?

Workgroup Discussion

- What are the major impediments, including policy issues and barriers of effectiveness, to implementing needed changes?
- Who should be responsible for assuring that promising changes are implemented?
- What resources would be required to make these changes within institutions, regionally, and nationally?
Location of conference - Atlanta
Date and Time - ?

Next Steps - Form Steering Committee and select Workgroup Leaders

Structure of the Conference
• Opening and Closing sessions broadcast over internet.
• Workgroup sessions recorded, but not broadcast
• Conference proceedings published and posted on the CDC web-site
Defining Best Practices in Laboratory Medicine

Report on Status of Laboratory Medicine

- Scope and magnitude of the field
- Customers of laboratory services
- Factors affecting the delivery and quality of services
- Impact of regulation and accreditation on the field
- Common practices, performance measures
- Workforce trends
- Expectations for the Future
Defining Best Practices in Laboratory Medicine

Developing a Process for Identifying Best Practices

• Form an Expert Workgroup
• Define “Best Practice”
• Establish criteria for selecting candidate practices
• Solicit candidate Best Practices in target area
  ❖ POCT for Infectious Disease
• Conduct pilot test of process
• Disseminate findings
Evaluation of Proficiency Testing Services

• Form an Expert Workgroup
• Establish set of goals for PT programs
  ❖ Do US Programs meet quality improvement goals?
  ❖ Do US Programs meet regulatory goals?
  ❖ Do US Programs meet education goals?
• How could PT programs enhance their value to laboratories?
• Are PT programs keeping pace with changing technology?
• Would conformance to an international standard enhance PT program usefulness?
• Disseminate findings
Purpose: Discuss concerns raised during February 2006 CLIAC Meeting

Concerns:

• Need for confirmatory testing for some screening tests
• Specimen or cultures not available for confirmatory testing or epidemiologic surveillance
• PH epidemiology data missing for outbreak investigation and disease surveillance
• Test sites unaware of PH reporting requirements – State to State variation
• Testing service information often not readily accessible
Thank you
Questions/ Comments