CLIA & Genetic Testing
Oversight
Topics for Discussion:

• Background & history of GT regulation
• What CLIA requires for GT in yellow
• Why no GT specific standards?
• CMS’ plan to enhance GT oversight w/existing authority
• Other quality & oversight efforts underway
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Background & History:

• Final CLIA regulations—1992
• NIH/DOE Task Force report—1997
• CDC Notice of Intent—2000
• CMS CLIA Final QC regulations—2003
• SACGHS recs to the Secretary HHS—2008
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General CLIA Information:

• Impetus was deaths from incorrect Pap smears
• **Intent**—ensure accurate, reliable, timely testing
• Requirements minimal; based on test complexity
  – 3 categories: waived, moderate & high
  – More complex tests have more stringent standards
• **Most GT are high complexity***
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General CLIA Information:

- Program entirely funded by user fees; not gov’t.
- Covers *all testing* on human specimens for health assessment--not just Medicare or FDA approved.
- >200,000 labs enrolled; **approx. 600-1K are GT***
- Excludes research, forensic, VA labs.
  - Research covered when patient-specific results are returned.
- Data indicates *improved performance* over time.
What CLIA Already Covers

- **Quality control (QC)** — real time check of test quality
  - Monitors the performer, test & lab’s environment
    - *PCR, tests w/extraction & 2 levels of QC/day;*
    - Daily QC w/ some specific to GT;

- Test method (analytic) validation;
- Calibration/calibration check;
- Instruments, reagents, supplies;
- Maintenance; Procedure manual;
- Test results comparison;
- Corrective actions; & Specialties.
What CLIA Already Covers

- **Proficiency testing**—accuracy measure (external QA)
  - Tests listed in regulations (83) – enroll in PT program
  - Tests not listed -- check test accuracy 2X/year
    - **Applies to GT**
- **Audit trail, confidentiality, specimen integrity & identification, complaints;**
- **Specimen collection, processing, test referral, test orders, result reporting;**
- **Facilities**----**Uni-directional workflow for GT**

What CLIA Already Covers

- **Personnel**—Required positions w/ education, experience, training & **quality** responsibilities
  - *Laboratory Director* — overall quality responsibility
  - *Clinical Consultant*
  - *Technical Supervisor*
  - *General Supervisor*
  - *Testing Personnel*

- **Competency**—annual checks of personnel performance

- **Highest qualifications apply to GT labs**
What CLIA Already Covers

• **Quality Assurance/Assessment**
  – Overall plan to monitor test systems & quality;
  – Encompasses all CLIA standards;
  – Correct problems/complaints effectively; &
  – Communicate with staff, clients.

• **Biennial surveys** look at outcomes (test results).
  – Assess *All* lab’s systems & processes to assure quality

• Menu of **enforcement actions** for noncompliance
  – Sanctioned labs’ Registry posted on CLIA web site
Why no Genetic Testing Specialty?

• Survey data *doesn’t indicate a problem*;
• GT specialty *will not*:
  – provide clinical validity;
  – solve PT/QC sample paucity;
  – address ELSI or DTC issues.
• **No widely accepted definition** of a GT
• Dynamic GT area: prescriptive standards will be outdated; lock labs into outmoded compliance.
• Disruption to existing infrastructure & specialties
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Labs with Deficiency Citations

Survey Cycles

- Labs with Std. or Cond.-Level Def.
- Labs with Cond.-Level Defic.
Is There a Comparative Advantage to a Specialty?

- Labs already covered by CLIA;
- CMS can use *existing* regs to enhance outcomes;
- Professional & accred. orgs. have standards;
- Some advisory com. recommendations published in 2003 CLIA regulations;
- Lab community isn’t unanimous;
- Rule = 3 yrs. & uses scarce CMS resources.
Using Existing CLIA Rules Effectively

What is CMS doing to strengthen GT oversight?

- Transmit specific guidance to State surveyors
- Conducted surveyor technical & procedural training
- Publish educational MMWR for labs w/ CDC
- Explore survey alternatives w/ oversight agencies
- Partner to design alternative PT/QC
- Work w/ CLIAC, CDC, FTC, NIH & FDA
Using Existing CLIA Rules Effectively

What is CMS doing to strengthen GT oversight?

• Collaborate with CLSI on professional standards
• Request FDA aid in complex test valid. reviews
• Collect data on GT laboratory performance
• Enhance CLIA web site for easy public access to lab certification info
  – Additional info on GeneTests & AMP web sites
• Monitoring DTC labs for certification
Other Ongoing Efforts Underway:

• CDC, in partnership w/ GT community estab. GeT-RM:
  – Provide materials for QC, PT;
  – Facilitate test development;
  – Determine method validation;
  – Encourage research.

• CDC’s further efforts:
  – Rare diseases; newborn screening pgm., CETT, EGAPP;

• CAP, JCAHO, NY have GT standards;
• CLSI/ACMG molecular guidance docs; more planned.
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**Items CMS Will Augment:**

- **QC**—partner to identify creative mechanisms & materials;
- **PT**— update ‘92 regulations w/ CDC to include some GT in existing specialties—if samples available;
  - Expand alternative GT PT mechanisms/approaches;
- **Personnel**— develop interpretive guidance w/ experts
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- **Issues Beyond the Scope of CLIA:**
  - Clinical validity;
  - DTC claims;
  - Informed consent;
  - Genetic counseling;
  - Tests that don’t assess health (e.g., gender); &
  - ELSI.
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SACGHS Recommendations for CMS to Sec. HHS---2008

- Include all non-waived tests in PT regulations;
  - Promote further development of GT PT;
  - Identify more effective forms of alt. assessment;
- Train surveyors in GT technology; creative surveys;
- Develop a registry of GT w/ clinical validity information;
- Register uncertified labs;
  - Take approp. enforcement actions;
- Develop professional standards;
- Monitor DTC; Hire staff;
- Cover all health-related tests.
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• **Suggested Resolutions:**
  – Work with SACGHS & private sector to:
    • Study the best mechanisms for oversight & test quality.
      – Partner w/ other oversight agencies
    • Continue to develop & follow professional standards.
  • Continue to augment lab oversight & educate labs under current CLIA.
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Next Steps for CMS:

• Heighten surveyor awareness & train---complete;
• Initiate development of updated PT regs;
• Collaborate ongoing w/ advisory groups, experts, CDC, FDA, etc.;
• Encourage CLSI to develop more GT standards;
• Educate GT laboratories;
• Expand CMS/CLIA web site.
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An Offer You Can’t Refuse!

- Assist HHS in GT oversight efforts to assure quality.
- Tell us your concerns, so we can address them using --
  - Existing CLIA infrastructure;
  - Current or updated mechanisms;
  - Your expertise;
  - Other programs/experts.
Where to Find CLIA Info:

- CMS CLIA Web site:
  - [www.cms.hhs.gov/clia](http://www.cms.hhs.gov/clia)
- CMS Central Office in Baltimore:
  - 410-786-3531
- Judy Yost’s email:
  - Judith.yost@cms.hhs.gov
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The End!
Thank You!!