CLIA & Genetic Testing Oversight
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CLIA & Genetic Testing Oversight

Topics for Discussion:

• Background & history of GT NPRM.
• What CLIA already requires for GT.
• Why no GT specific standards?
• CMS’ plan to enhance GT laboratory oversight.
• Other quality & oversight efforts.
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Background & History:
• Final CLIA regulations—1992.
• CDC NOI—2000.
• Revised CLIAC recs to HHS—2001.
• CMS CLIA Final QC regulations—2003.
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.
• Covers *all testing* on human specimens for health purposes--not just Medicare or FDA approved.
• 200,000 labs enrolled; **approx. 600 are GT***.
• Excludes research, forensic, VA, labs.
  – Research is covered when 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.
  
  - **Daily QC w/ some specific to GT;**
    - *PCR, tests w/extraction & 2 levels of QC/day;*

- **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** — long-term outcome accuracy measure.
  - Tests listed in regulations – 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.

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

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

• **Biennial surveys** look at outcomes (test results).

• Menu of **enforcement actions** for noncompliance.
Why no Genetic Testing Specialty?

- Survey data doesn’t indicate a problem;
- GT specialty will not provide clinical validity;
- GT specialty will not solve PT/QC paucity;
- GT specialty will not address ELSI or DTC issues;
- No widely accepted definition of a GT;
- Disruption to existing infrastructure & specialties;
- In the dynamic GT area, prescriptive standards will be outdated; lock labs into outmoded compliance.
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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 will use *existing* regs to enhance outcomes;
- Professional standards exist;
- Several advisory com. recommendations published in 2003 CLIA regulations;
- Only a few organizations want a specialty;
- Affects approximately 600 entities; and
- Admin. 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.
• Retain experts to conduct surveyor technical training.
• Publish educational materials for labs w/ CDC.
• Explore survey alternatives w/ oversight agencies.
• Design alternative PT/QC mechanisms.
Using Existing CLIA Rules Effectively

What is CMS doing to strengthen GT oversight?

- Work with CLIAC, CDC, FTC, NIH & FDA.
- Collaborate with CLSI on professional standards.
- Request FDA/CDC aid in complex test validation reviews.
- Collect data on GT laboratory performance.
- Enhance CLIA web site for easy public access to lab certification info.
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Other Ongoing CDC 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 has molecular guidance docs & more planned.
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Rebuttal to Hudson Study Findings:

- PT enrollment isn’t required for GT, since there are few modules/samples available; so lack of enrollment has no impact.
  - Labs still subject to accuracy checks twice/yr.->1000tests;
  - PT required for only 83 tests in regs now.
- CLIA data confirms there are NO labs without a specialty.
  - CLIA fees based on the annual volume & types of tests.
  - Since there is no GT specialty, assume question was misunderstood.
- Most “potential” errors noted were not in test analysis, but pre or post test already covered by CLIA; documented in several publications & doesn’t require a specialty.
- No. of CLIA specialties have actually been reduced over time.
  - Instead QC was enhanced for ALL tests; technology improved.
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Items we can consider in upcoming regs:

- **QC**— augment QC, if mechanisms & materials are identified;

- **PT**— intersperse GT PT in existing specialties—if available; or
  - Expand 1236 –unregulated analytes w/ alternative GT PT mechanisms:

- **Personnel**— expand cytogenetic TS to include GT?
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• **Issues Beyond the Scope of CLIA:**
  – Clinical validity;
  – DTC;
  – Informed consent;
  – Genetic counseling;
  – Tests that don’t assess health (e.g., gender); and
  – Ethical, Legal, Social Issues.

• **Suggested Resolution:**
  – Work with SACGHS to:
  – Pass legislation that covers all GT issues in a separate statute instead of CLIA; or
  – Continue to develop & follow professional standards.
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Next Steps for CMS:

• Heighten surveyor awareness & train;
• Collect GT lab performance data;
• Collaborate ongoing with advisory groups, experts, CDC, FDA, etc.;
• Develop GT standards with CLSI; and
• Educate GT laboratories; expand web site.
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An Offer You Can’t Refuse!

– Assist HHS in GT oversight efforts;
– Tell us your concerns, so we can address them using --
  • Existing CLIA infrastructure;
  • Current or updated mechanisms;
  • Your expertise;
  • Other programs/experts.

CLIA

CMS/ Centers for Medicare & Medicaid Services
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Where to Find CLIA Info:

– CMS CLIA Web site:
  • 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!!