Clinical
Laboratory
Improvement
Advisory
Committee

Genetic Testing Subcommittee

Summary Report
May 27-28, 1998

U.S. DEPARTMENT OF HEALTH & HUMAN SERVICES

Public Health Service

CDC
CENTERS FOR DISEASE CONTROL AND PREVENTION

Genetic Testing Subcommittee

May 27 - 28, 1998

Summary

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Record of Attendance

Committee Members
Dr. Patricia Charache
Dr. Susanne Gollin
Dr. Verlin Janzen
Dr. Edward McCabe
Dr. Margaret McGovern
Dr. Toby Merlin
Dr. Wendell O'Neal
Dr. Kenneth Pass
Ms. Sharon Radford
Mr. Mark Rothstein
Dr. Morton Schwartz
Mr. Elliott Segal
Dr. Lawrence Silverman
Ms. Stephanie Smith

Ex Officio Members
Dr. Carlyn Collins, CDC
Dr. Steven Gutman, FDA
Ms. Judith Yost, HCFA

Executive Secretary
Dr. John Ridderhof (representing Dr. Edward Baker)

Centers for Disease Control and Prevention
Ms. Nancy Anderson
Dr. Rex Astles
Ms. Carol Cook
Dr. Joe Boone
Dr. Ed Holmes
Dr. John Ridderhof
Ms. Renee Ross
Ms. Elva Smith
Ms. Rhonda Whalen
Ms. Laurina Williams

Welcome and Introductory Information

The meeting was called to order by Genetic Testing Subcommittee Chair Dr. Wendell O'Neal. Dr. Carlyn Collins, Director, Division of Laboratory Systems, Public Health Practice Program Office, welcomed the Subcommittee members and described the logistics for the meeting. She explained that the pre-analytic, analytic, and post-analytic workgroups would meet individually until mid-afternoon. During this time, each workgroup would discuss the topics identified at the previous Subcommittee meeting relevant to their phase of genetic testing, to consider whether the Clinical Laboratory Improvement Amendments of 1988 (CLIA) regulations are appropriate for that phase of testing, or if revisions to the requirements are needed to address genetic testing. After completion of the individual discussions, the three workgroups would meet jointly to review the results of each workgroup's deliberations and prepare a summary report for presentation to the Clinical Laboratory Improvement Advisory Committee (CLIAC).

Presentations and Subcommittee Discussion

When the three workgroups jointly reconvened, each group presented an outline of the topics they discussed for their respective phases of genetic testing (see addenda). The Subcommittee addressed each issue, noting comments and suggestions made by the workgroup pertaining to the appropriateness of CLIA. Due to time constraints, the Subcommittee did not complete its discussion for all of the relevant topics. In preparing the summary report for CLIAC, the issues which were not adequately addressed were noted as needing further discussion by the full Subcommittee.

PRE-ANALYTIC PHASE OF GENETIC TESTING — Addendum S-1

For the following topics, the Subcommittee agreed with the workgroup that these areas are either adequately addressed by the current CLIA regulations or do not need to be addressed by CLIA for the pre-analytic phase of genetic testing, or suggested minor revisions:

- Appropriateness of tests (suggested adding "Appropriate clinical information must be provided on the request form.")
- Specimen handling/preparation
- Confidentiality
- Communication with provider community
- Ordering additional tests (by laboratory, patient, or public health agency)
- Ownership of specimen

The Subcommittee considered the following items, but determined either that additional time is needed to fully address the item, or that input pertaining to topics addressed by the other workgroups would be helpful prior to making a final decision as to appropriate CLIA coverage of that item. The issues raised during the Subcommittee discussion of each topic are:

- Informed consent - The workgroup reported that informed consent is appropriate for some genetic tests, and may be needed for re-use of patient specimens (see below). Several

Subcommittee members suggested that enforcement of informed consent is not the responsibility of the laboratory , but that the laboratory should provide limitations of the testing to the ordering physician. It was proposed that a definition for informed consent be added to the CLIA regulations, and that requisitions for genetic testing of a sensitive nature must indicate that informed consent has been obtained for that test. In obtaining informed consent, the limitations and consequences of the test results must be discussed with patients.

- Consent to re-use specimens - The workgroup proposed that excess material from patient specimens may be used for quality control, quality assurance, or test development purposes without obtaining informed consent if patient identifiers are removed, but that informed consent would be needed if identifiers are not removed. This topic was discussed extensively by the Subcommittee, with a strong view expressed by some members that patient specimens must not be re-used under any circumstances without informed consent.

- Genetic counseling - This topic is closely related to personnel issues, and was discussed in conjunction with the role and qualifications of the clinical consultant for genetic testing. Subcommittee members suggested that a board-certified genetic counselor, or an individual with a master's degree in genetic counseling, is qualified to serve as a clinical consultant for genetic testing. However, another member stated that although a genetic counselor may be appropriate to serve as a clinical consultant for the pre-analytic phase, it would not be appropriate for the analytic phase of genetic testing.

ANALYTIC PHASE OF GENETIC TESTING **Addendum S-2**

The Subcommittee was only able to complete its discussion of the personnel qualifications for laboratory director and technical supervisor in the time allotted for discussion of the analytic phase of genetic testing. In addressing these personnel categories for high complexity testing, Dr. Collins reminded the Subcommittee that the laboratory director is responsible for the overall operation and administration of the laboratory, whereas the technical supervisor is responsible for the technical and scientific oversight of the laboratory. Qualifications for laboratory director and technical supervisor were part of the summary report to the CLIAC. The proposed qualifications below include the current CLIA requirements with suggestions for revision noted in bold.

- Laboratory director - Several Subcommittee members stressed the importance of including provisions in the regulations that would permit both an M.D. and a Ph.D. Board-certified in medical genetics to direct a genetic testing laboratory. This was incorporated into the following proposed qualifications for laboratory director:

  -- Be an M.D., or D.O. with certification in clinical and/or anatomic pathology, or
  -- Be an **M.D.**, D.O. or Ph.D. and be certified in medical genetics, or
  -- Be an M.D. or D.O. and have two years directing or supervising high complexity testing, or
  -- Hold a doctorate degree in chemistry, physical, biological, or clinical laboratory sciences, be certified, and have two years of supervisory experience in high complexity testing, or

-- Be grandfathered.

- Technical Supervisor - In discussing the qualifications for this personnel category, the Subcommittee noted that the technical supervisor is responsible for the technical aspects of testing and that extensive education, training and supervisory experience in the relevant subspecialty of high complexity genetic testing are essential to ensure appropriate technical supervision of genetic testing. The following suggested qualifications for technical supervisor in genetic testing include this requirement:

  -- Be an M.D., or D.O. with certification in clinical and/or anatomic pathology and two years subspecialty training in genetic testing plus two years supervisory experience in high complexity genetic testing, or four years supervisory experience in high complexity genetic testing in the relevant subspecialty, or
  -- Be an M.D., D.O. or Ph.D. and be certified in the appropriate medical genetics specialty and have two years experience directing or supervising high complexity genetic testing in the relevant subspecialty, or
  -- Hold a doctorate degree in chemistry, physical, biological, or clinical laboratory sciences, and have four years of training or supervisory experience in high complexity genetic testing in the relevant subspecialty, or
  -- Be grandfathered.

Issues addressed by the analytic workgroup but not discussed by the full Subcommittee are:

- Personnel qualifications for general supervisor, clinical consultant, testing personnel
- Personnel responsibilities for all categories
- Contamination
- Specimen integrity
- General and specific quality control measures
- Proficiency testing
- Validation of tests
- Re-use of previously tested specimens
- Confidentiality

POST-ANALYTIC PHASE OF GENETIC TESTING **Addendum S-3**

Due to insufficient time, the Subcommittee did not address the following issues for the post-analytic phase of genetic testing that had been considered by the workgroup:

- Special reporting requirements
- Consultation to non-geneticist caregivers
- Role of the laboratory in genetic counseling
- Confidentiality
- Use of tested specimens
- Use of test data

Public Comments

The were no public comments for the Subcommittee.

Concluding Remarks

It was decided that the Genetic Testing Subcommittee would meet again prior to the next CLIAC meeting (Note: The Subcommittee meeting was subsequently scheduled for July 30 - 31, 1998). Dr. O'Neal then adjourned the Subcommittee meeting.

I certify that this summary report of the May 27 - 28, 1998, meeting of the Genetic Testing Subcommittee of the CLIAC is an accurate and correct representation of the meeting.

/S/ Wendell R. O'Neal, Ph.D.
Chairman