

Appendix. Information and Documents Reviewed by the CLIAC Biochemical Genetics Workgroup

42 CFR Part 493. Clinical Laboratory Improvement Amendments (CLIA) Regulations and Interpretive Guidelines.

2001 CLIAC recommendations on genetic testing.

American Board of Bioanalysis. Board certification information.

American Board of Clinical Chemistry in Molecular Diagnostics. Board certification information.

American Board of Clinical Chemistry. Board certification information.

American Board of Forensic Toxicology. Board certification information.

American Board of Histocompatibility and Immunogenetics. Board certification information.

American Board of Medical Genetics. Board certification information.

American Board of Medical Laboratory Immunology. Board certification information.

American Board of Medical Microbiology. Board certification information.

American College of Medical Genetics (ACMG). ACMG Standards and Guidelines for Clinical Genetic Laboratories.

Association of Public Health Laboratories Policy Statement. Parental Consent in Public Health Newborn Screening Programs.

CAP Molecular Pathology Retention Periods from LAP audioconference: Inspecting the Molecular Pathology Laboratory. August 16, 2006

CAP laboratory accreditation standards relating to ensuring clinical validity of genetic tests.

Centers for Disease Control and Prevention (CDC). Good Laboratory Practices for Molecular Genetic Testing for Heritable Diseases and Conditions. Morbidity and Mortality Weekly Report Recommendations and Reports. Vol. 58, No. RR-6. June 12, 2005.

Centers for Medicare & Medicaid Services (CMS). Survey Procedures and Interpretive Guidelines for Laboratories and Laboratory Services.

CLIAC Recommendations for Good Laboratory Practices for Molecular Genetic Testing.

Clinical and Laboratory Standards Institute (CLSI) Guidelines:

- ✓ A Quality Management System Model for Healthcare (HS1-A2).
- ✓ Application of a Quality Management System Model for Laboratory Services (GP26A3E).
- ✓ Collection, Transport, Preparation, and Storage of Specimens for Molecular Methods (MM13AE).
- ✓ Defining, Establishing and Verifying Reference Intervals in the Clinical Laboratory (C28-A3).
- ✓ Evaluation of Precision Performance of Quantitative Measurement Methods (EP5-A2).
- ✓ Evaluation of the Linearity of Quantitative Measurement Procedures: A Statistical Approach (EP6-A).
- ✓ Interference Testing in Clinical Chemistry (EP7-A2).

- ✓ Method Comparison and Bias Estimation using Patient Samples (EP9-A2).
- ✓ Molecular Diagnostic Methods for Genetic Diseases (MM01A2E).
- ✓ Protocols for Determination of Limits of Detection and Limits of Quantitation (EP17-A).

College of American Pathologists (CAP) Laboratory Accreditation Checklists:

- ✓ Chemistry and Toxicology.
- ✓ Cytogenetics.
- ✓ Laboratory General.
- ✓ Molecular Pathology.

Cowan, T and Stroval, E. Management and Quality Assurance in the Biochemical Genetics Laboratory. Current Protocols in Human Genetics. Supp 59, 2008.

Food and Drug Administration (FDA) Guidance Documents:

- ✓ Draft Guidance for Industry and FDA Staff – Pharmacogenetic Tests and Genetic Tests for Heritable Markers (2006).
- ✓ Class II Special Controls Guidance Document: Newborn Screening Test Systems for Amino Acids, Free Carnitine, and Acylcarnitines Using Tandem Mass Spectrometry.

ISO 15189 Medical laboratories — Particular requirements for quality and competence. 2007.

Javaher P, Kääriäinen H, Kristoffersson U, Nippert I, Sequeiros J, Zimmern R, Schmidtke J. EuroGentest: DNA-Based Testing for Heritable Disorders in Europe. Community Genetics 2008;11:75–120.

National Board in Clinical Chemistry. Board certification information.

National Credentialing Agency for Laboratory Personnel. Certification information.

New York State Civil Rights Law. Section 79-I. Confidentiality of records of genetic tests.

New York State Clinical Laboratory Evaluation Program (CLEP) Laboratory Standards:

- ✓ General Systems.
- ✓ Genetic Testing.
- ✓ Cytogenetics.

Organization for Economic Co-operation and Development (OECD). OECD Guidelines for Quality Assurance in Molecular Genetic Testing. 2007.

Performance Evaluation and Assessment Scheme (PEAS) for Newborn Screening Systems. Developed as part of a cooperative agreement between Health Resources and Services Administration, Maternal and Child Health Bureau, Genetic Services Branch and the National Newborn Screening and Genetics Resource Center.

Rinaldo P, Hahn S, Matern D. Clinical biochemical genetics in the twenty-first century. Acta Paediatr Suppl 445:22-27, 2004.

Secretary's Advisory Committee on Genetics, Health and Society (SACGHS). U.S. System of Oversight of Genetic Tests: A Response to the Charge of the Secretary of HHS. SACGHS Report 2007.

Sequeiros, J. and Guimarães, B. EuroGenTest Report: Definitions of Genetic Testing. Third Draft. EuroGenTest. 2007.