

REGULATION OF LABORATORY-DEVELOPED TESTING PROCEDURES

Highlights from [A primer on regulation of laboratory-developed testing procedures: A points to consider statement of the American College of Medical Genetics and Genomics \(ACMG\)](#)
Leung, Marco L. et al. (2025) [Genetics in Med](#), 27(6):101391

For more than 30 years, clinical testing has been regulated through the Clinical Laboratory Improvement Amendments (CLIA), ensuring high standards of accuracy, reliability, and patient safety while allowing laboratories to innovate. **Laboratory-developed tests (LDTs) play a critical role in patient care, especially in cancer and rare disease genetic testing.**

What Are Clinical Testing Laboratories?

- Certified healthcare facilities that provide professional testing services to aid in diagnosis, management, and treatment.
- Run by highly trained professionals, which for genetics laboratories includes laboratory directors, technologists, genetic counselors, variant scientists, and bioinformaticians.

What Are Laboratory-Developed Tests?

- Testing procedures that are designed, developed, validated, performed, and interpreted by board-certified laboratory professionals.
- Includes FDA-cleared/approved tests that must be modified and validated to meet clinical needs.

How Are Laboratories and LDTs Regulated?

- Through the CLIA regulations enforced by the Centers for Medicare and Medicaid Services (CMS).
- Includes standards for testing personnel qualifications, quality assurance, test development and performance, as well as proficiency testing.
- May also be subject to additional state regulations and national accrediting bodies.

What Clarifications to CLIA Are Needed?

- Require prompt inspections before a new laboratory performs high-complexity clinical testing.
- Establish industry-wide minimum performance standards for higher-risk assays.
- Require laboratories to submit key validation data to a publicly accessible database.
- Examine both analytical and clinical validation of LDTs.
- Review marketing materials and claims during inspections.

Why It Matters

Essential for Patients: Many genetic, rare disease, and cancer tests are only available as LDTs; over-regulation could limit access.

Built-in Oversight: CLIA already ensures rigorous quality standards for laboratories, personnel, and testing procedures.

Room for Modernization: Updating CLIA can strengthen patient protections while keeping pace with rapid scientific advances.

Balanced Regulation: Policies must protect patients without creating barriers that delay or block access to lifesaving testing services.