American College of Medical Genetics and Genomics (ACMG)

TECHNICAL STANDARDS FOR CLINICAL GENETICS LABORATORIES (2021 Revision)

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Disclaimer: These ACMG Technical Standards for Clinical Genetics Laboratories are developed primarily as an educational resource for clinical laboratory geneticists to help them provide quality clinical laboratory genetic services. Adherence to these standards is voluntary and does not necessarily assure a successful medical outcome. These standards should not be considered inclusive of all proper procedures and tests or exclusive of other procedures and tests that are reasonably directed to obtaining the same results. In determining the propriety of any specific procedure or test, the clinical laboratory geneticist should apply his or her own professional judgment to the specific circumstances presented by the individual patient or specimen. Clinical laboratory geneticists are encouraged to document in the patient's record the rationale for the use of a particular procedure or test, whether or not it is in conformance with these standards. They also are advised to take notice of the date any particular technical standard was adopted, and to consider other relevant medical and scientific information that becomes available after that date. It also would be prudent to consider whether intellectual property interests may restrict the performance of certain tests and other procedures.

A: OVERVIEW

A1 Purpose of Standards

These voluntary standards have been established as an educational resource to assist clinical geneticists in providing accurate and reliable diagnostic genetic laboratory testing consistent with currently available technology and procedures in the areas of clinical cytogenetics and genomics, biochemical genetics, and molecular genetics and genomics.

A2 Minimal Criteria

These standards establish minimal criteria for clinical genetics laboratories. Many laboratories will exceed these minimal standards.

A3 Voluntary Adherence
Adherence to these technical standards is completely voluntary and does not necessarily assure a successful outcome. The standards should not be considered inclusive of all proper procedures and tests, or exclusive of other procedures and tests that are reasonably directed to obtaining the same results.

A4 Acceptable Variations
It is acknowledged that numerous acceptable variations exist in genetic testing methodologies. The accuracy and precision of all procedures should be documented in each laboratory. This should include in-house validation and/or references to appropriate published literature.

A5 Specialized Testing
Specialized testing (testing that is limited to one or a few laboratories) requires appropriate and sufficient documentation of effectiveness to justify its use.

A6 Maintenance of Standards
These standards will be reviewed and updated periodically to assure their timeliness in this rapidly developing field.