

**TECHNICAL STANDARDS FOR CLINICAL GENETICS
LABORATORIES
(2021 Revision)**

Disease-Specific Laboratory Standards and Articles Addressing Special Topics

DISEASE-SPECIFIC

Biochemical Genetics

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Strovel ET, Cowan TM, Scott AI, Wolf B. [Laboratory diagnosis of biotinidase deficiency, 2017 update: a technical standard and guideline of the American College of Medical Genetics and Genomics](#). Genet Med. 19, 1079 (2017).

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Cytogenetics

Wolff DJ, Van Dyke DL, Powell CM; Working Group of the ACMG Laboratory Quality Assurance Committee. [Laboratory guideline for Turner syndrome](#). Genet Med. 12, 52–55 (2010).

Molecular Genetics

Mao R, Krautscheid P, Graham RP, Ganguly A, Shankar S, Ferber M, and Hegde M; ACMG Laboratory Quality Assurance Committee. Genetic Testing for Inherited Colorectal Cancer and Polyposis 2021 Revision: A Technical Standard of the American College of Medical Genetics and

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Addendum: [Technical standards and guidelines for spinal muscular atrophy testing](#). Genet Med. Published October 13, 2020

Addendum: [American College of Medical Genetics and Genomics Standards and Guidelines for Clinical Genetics Laboratories, 2014 edition: technical standards and guidelines for Huntington disease](#). Genet Med. Published July 20, 2020

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Zhang S, Taylor AK, Huang X, Luo B, Spector EB, Fang P, Richards CS; ACMG Laboratory Quality Assurance Committee. [Venous thromboembolism laboratory testing \(factor V Leiden and factor II c.*97G>A\), 2018 update: a technical standard of the American College of Medical Genetics and Genomics \(ACMG\)](#). Genet Med. 20, 1489–1498 (2018).

Bean L, Bayrak-Toydemir P. [American College of Medical Genetics and Genomics Standards and Guidelines for Clinical Genetics Laboratories, 2014 edition: technical standards and guidelines for Huntington disease](#). Genet Med. 16, e2 (2014).

Hegde M, Ferber M, Mao R, Samowitz W, Ganguly A; Working Group of the American College of Medical Genetics and Genomics (ACMG) Laboratory Quality Assurance Committee. [ACMG technical standards and guidelines for genetic testing for inherited colorectal cancer \(Lynch syndrome, familial adenomatous polyposis, and MYH-associated polyposis\)](#). Genet Med. 16, 101–116 (2014).

Lyon E, Gastier Foster J, Palomaki GE, Pratt VM, Reynolds K, Sábato MF, Scott SA, Vitzka P; working group of the Molecular Genetics Subcommittee on behalf of the American College of Medical Genetics and Genomics (ACMG) Laboratory Quality Assurance Committee. [Laboratory testing of CYP2D6 alleles in relation to tamoxifen therapy](#). Genet Med. 14, 990–1000 (2012).

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Monaghan KG, Feldman GL, Palomaki GE, Spector EB; Ashkenazi Jewish Reproductive Screening

Working Group; Molecular Subcommittee of the ACMG Laboratory Quality Assurance Committee. [Technical standards and guidelines for reproductive screening in the Ashkenazi Jewish population](#). Genet Med. 10, 57–72 (2008).

SPECIAL TOPICS

Chao EC, Astbury C, Deignan, JL, Pronold M, Reddi, HV, and Jeffrey N. Weitzel, MD; ACMG Laboratory Quality Assurance Committee. [Incidental detection of acquired variants in germline genetic and genomic testing: a points to consider statement of the American College of Medical Genetics and Genomics \(ACMG\)](#). Genet Med. Published 16 April 2021.

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