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ADDENDUM

Addendum: Technical standards and guidelines: Molecular genetic testing for ultra-rare disorders



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Addendum to: “Technical standards and guidelines: molecular genetic testing for ultra-rare disorders.” Anne Maddalena, PhD, Sherri Bale, PhD, Soma Das, PhD, Wayne Grody, MD, PhD and Sue Richards, PhD; ACMG Laboratory Quality Assurance Committee. *Genetics in Medicine* 7:571-583; <https://doi.org/10.1097/01.GIM.0000182738.95726.ca>; published online 01 October 2005. Revised by the Laboratory Quality Assurance Committee in 2009 and reaffirmed in August 2016.

ARTICLE INFO

This document was retired by the American College of Medical Genetics and Genomics (ACMG) Board of Directors as of May 20, 2019 with the following addendum.

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At their meeting on April 10, 2018, the Molecular Genetics Subcommittee of the ACMG Laboratory Quality Assurance Committee voted to retire the “Technical standards and guidelines: molecular genetic testing for ultra-rare disorders,” which was originally published in 2005, revised in 2009, and reaffirmed in 2016. This recommendation was endorsed by the full Laboratory Quality Assurance Committee during their meeting the following day. It was the belief of the Molecular Genetics Subcommittee members that with the advent of next-generation sequencing, a separate document addressing testing for ultra-rare disorders was no longer required. However, portions of the original document were believed to hold continuing relevance for clinical testing laboratories. These sections have been updated and are now

incorporated into the 2021 edition of the ACMG Technical Standards for Clinical Genetics Laboratories, Section G: Clinical Molecular Genetics, Sub-section G19: Testing for Ultra-Rare Disorders.¹

Conflict of Interest

The authors declare no conflicts of interest.

Reference

1. ACMG Technical Standards and Guidelines for Clinical Genetics Laboratories. Section G: clinical molecular genetics: sub-section G19: testing for ultra-rare disorders. Published 1998. Most recent update 2021. Accessed October 7, 2021. <https://www.acmg.net/PDFLibrary/ACMG%20Technical%20Lab%20Standards%20Section%20G.pdf>.

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