

**Addendum:** Technical standards and guidelines for spinal muscular atrophy testing

Joshua L. Deignan, PhD, Caroline Astbury, PhD, Andrea Behlmann, PhD, Saurav Guha, PhD, Kristin G. Monaghan, PhD, Honey V. Reddi, PhD, Bryce A. Seifert, PhD, Marwan Tayeh, PhD and Erin Wakeling, PhD; ACMG Laboratory Quality Assurance Committee

*Genetics in Medicine* (2021) 23:2462; <https://doi.org/10.1038/s41436-020-00961-6>

Addendum to: “Technical standards and guidelines for spinal muscular atrophy testing”. Thomas W. Prior, PhD, Narasimhan Nagan, PhD, Elaine A. Sugarman, MS, CGC, Sat Dev Batish, PhD and Corey Braastad, PhD; ACMG Laboratory Quality Assurance Committee *Genetics in Medicine* 13:686–694 (2011); <https://doi.org/10.1097/GIM.0b013e318220d523>, published online 13 June 2011 and *Genetics in Medicine* 18:752 (2016); <https://doi.org/10.1038/gim.2016.76>, published online 30 June 2016.

These documents were reaffirmed by the ACMG Board of Directors as of 24 August 2020 with the following addendum:

The Molecular Genetics Subcommittee members of the ACMG Laboratory Quality Assurance Committee conducted a critical review of the 2011 technical laboratory standard and on 3 July 2020 they recommended that it be reaffirmed with the following URL updates on page 688: “The coding region for SMN can be found on Ensembl ([www.ensembl.org](http://www.ensembl.org)). Proper nomenclature must be used to report any verified sequence variant (<http://varnomen.hgvs.org>).” The 2016 update was reaffirmed as written. The full ACMG Laboratory Quality Assurance Committee endorsed the recommendations to update and reaffirm these documents on 10 July 2020.