

Evidence Discussion (PN 8.2)

Spinal Muscular Atrophy

- Spinal Muscular Atrophy (SMA) is a genetic/hereditary disorder. Molecular genetic testing is the standard tool for diagnosis of Spinal Muscular Atrophy (SMA). MRI is NOT supported for diagnosis of SMA unless other diseases are being considered.^{1,2}

References

1. Prior TW, Leach ME, Finanger E. Spinal Muscular Atrophy. In: Adam MP, Everman DB, Mirzaa GM, et al., eds. GeneReviews® [Internet]. 2000 Feb 24; [Updated 2020 Dec 3]. Seattle (WA). University of Washington, Seattle. 1993-2022.
2. Oskoui M, Servais L. Spinal Muscular Atrophy. *Continuum (Minneap Minn)*. 2023;29(5):1564-1584. doi:10.1212/CON.0000000000001338