## Memorandum – MolePath #35

To: UNC Health System Attending Physicians, Housestaff, Nursing Coordinators, Department Heads and Supervisors

From: Karen Weck, MD; Director, Molecular Genetics Laboratory

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Medical Director, McLendon Clinical Laboratories

Date: November 23, 2021

**Subject: Spinal Muscular Atrophy (SMA) Diagnostic and Carrier Testing**

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Effective December 01, 2021, the UNC Medical Center Molecular Genetics Laboratory will offer diagnostic and carrier testing for Spinal Muscular Atrophy (SMA) by molecular analysis of the *SMN1* and *SMN2* genes. Currently, this testing is sent out via Referral services.

The assay has been validated for detection of *SMN1* and *SMN2* exon 7 copy number, reported as 0, 1, 2, 3, or ≥ 4 copies. In addition, variants associated with silent carriers of SMA (*SMN1* c.*3+80T>G and *SMN1* c.*211_*212del) will be reported for carrier testing and the disease modifier variant (*SMN2* c.859G>C) will be reported for diagnostic testing.

**Specimen Requirements**
1-3 mL peripheral blood in EDTA (lavender) or ACD (yellow) tube

**Test Name in Epic**
Spinal Muscular Atrophy (SMN 1/2) DNA Assay

**Test Order (EAP) number in Epic**
LAB11165A

**Turn-around time**
10 days

**CPT code**
81329

**Questions?** Email Jessica Booker (Jessica.Booker@unchealth.unc.edu) or call the UNCH Molecular Genetics Lab at (984) 974-1825

Website, https://www.uncmedicalcenter.org/mclendon-clinical-laboratories/directory/molecular-pathology-and-genetics/