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Division of Public Health

## Memo

To: North Carolina Newborn Screening Stakeholders

Scott M. Shone, PhD, HCLD(ABB), Director, NC State Laboratory of Public Health

From: Kelly Kimple, MD, MPH, FAAP, Section Chief, Women's and Children's Health

Date: April 29, 2021

Re: Newborn Screening for Spinal Muscular Atrophy

On May 1, 2021, the North Carolina Newborn Screening Program will begin screening all newborn screening specimens submitted to the North Carolina State Laboratory of Public Health (NCSLPH) for Spinal Muscular Atrophy (SMA). SMA is a genetic disorder that affects the nerve cells in the spinal cord and impacts the muscles used for activities such as breathing, eating, crawling, and walking. It is the number one genetic cause of death for infants.

Newborn screening for SMA will be integrated into the standard newborn screening panel and results will be included on all NCSLPH Newborn Screening reports issued starting May 4, 2021. Reports are provided in hardcopy form and are also available via the NCSLPH Clinical and Environmental Lab Results (CELR) online portal.

Screening results that suggest elevated risk for SMA will be communicated to the newborn's health care provider by the <u>North Carolina Newborn Screening Follow-up Program</u>. The Follow-up Program will provide consultation services and recommendations for follow-up testing.

In addition to SMA, the Newborn Screening Program is working towards the implementation of screening later this calendar year for Pompe Disease, Mucopolysaccharidosis Type I (MPS-I), and X-linked Adrenoleukodystrophy (X-ALD). Additional information on the implementation of screening for these disorders will be shared later this year.

For questions about newborn screening laboratory testing for SMA, please contact the NCSLPH Newborn Screening Laboratory at 919-807-8938. For questions about newborn screening follow-up for SMA, please contact the Newborn Screening Follow-up Program at 919-218-6460.

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