



STATE OF MICHIGAN

DEPARTMENT OF HEALTH AND HUMAN SERVICES
LANSING

GRETCHEN WHITMER
GOVERNOR

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DIRECTOR

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Newborn Screening for Spinal Muscular Atrophy

Dear Practitioner(s):

Following recommendations by the Michigan Newborn Screening (NBS) Quality Assurance Advisory Committee and approval by the Legislature required by law (MCL 333.5431), all Michigan newborns will be screened for spinal muscular atrophy (SMA) beginning March 9, 2020.

SMA is an autosomal recessive neurodegenerative disorder, affecting approximately 1 in 11,000 individuals. It is caused by a defect in the survival motor neuron 1 (SMN1) gene which leads to the loss of motor neurons in the spinal cord. Individuals with SMA develop progressive muscle weakness and atrophy typically involving muscles that control mobility, breathing, and swallowing. There are four main types of SMA, varying in age of onset and clinical severity. The most severe and common form, SMA Type 1, is usually fatal by two years of age if not treated. New FDA approved therapies are available to treat SMA and have been shown to significantly modify the course of the disease when introduced early.

Most cases of SMA are due to two identical deletions in the SMN1 gene. The Michigan Department of Health and Human Services (MDHHS) Newborn Screening Laboratory will use real time polymerase chain reaction (PCR) to determine the presence or absence of these deletions. The newborn will be screened for SMA using the dried filter paper blood spots collected as part of the current newborn screening process. A normal result does not rule out the diagnosis of SMA. Approximately 5% of cases are caused by other genetic changes that will not be detectable through newborn screening.

The Newborn Screening SMA follow-up coordinating center at Michigan Medicine is responsible for the follow-up of all positive screens for this disorder. Coordinating center staff will contact primary care providers to instruct them on the next steps to take after a positive SMA screening result is received. The baby will be referred to a designated MDA clinic for confirmatory testing and clinical management. To ensure the best possible outcome, it is very important to complete the follow-up steps in a timely manner.

The SMA follow-up coordinating center in Michigan is:

- SMA Follow-up at Michigan Medicine (Ann Arbor)
Phone: 734-647-8938

MDHHS and follow-up coordinating center staff will provide prompt communication and are available to answer questions. Additional resources for providers and families are available on our website, www.michigan.gov/newbornscreening. Please do not hesitate to contact us at 517-335-4181 or newbornscreening@michigan.gov.

Sincerely,

Mary Kleyn, MSc
Manager, Newborn Screening Follow-up Program