**Section 661.400 Spinal Muscular Atrophy (SMA)**

a) Interpretation of Results. Although the majority of infants affected by SMA will be identified by this screening, due to genetic variabilities and variations in health status, sample quality, and timing of sample collection, not all infants affected by the disorder may be identified. As with any laboratory test, false positive and false negative results are possible. Newborn screening test results are insufficient information on which to base diagnosis or treatment.

1) SMA is indicated when an SMN1 deletion is detected in dried blood spot samples by using DNA-based methods, such as polymerase chain reaction (PCR) or other methods. Normal testing parameters will be established using accepted statistical techniques (for example, as described by the Association of Public Health Laboratories, see Section 660.20).

2) When the SMN1 deletion is found, thus indicating the possibility of SMA, the Department will recommend referral of the newborn to a designated medical specialist for appropriate definitive testing and diagnostic studies.

b) Designation of Medical Specialist. In addition to the minimum qualifications set out in Section 661.230, medical specialists designated by the Department to follow-up on a screen positive for SMA shall have certification by the American Board of Medical Genetics and Genomics in Clinical Biochemical Genetics. Certification by the American Board of Psychiatry and Neurology in Pediatric Neurology or Pediatric Neuromuscular Medicine, or certification by the Board of Pediatrics in Pediatric Pulmonology with at least one year of experience post-training in the diagnosis and treatment of spinal muscular atrophy. Medical specialists shall have the capacity to provide prompt access to a multidisciplinary care center which includes the availability of pediatric genetics, neurology, pulmonology, cardiology, orthopedics, medical nutrition therapy, physical therapy, and occupational therapy.

c) Diagnosis and Treatment. Medical management by a designated medical specialist is highly recommended to confirm the diagnosis of spinal muscular atrophy. Referral to the appropriate medical team, including pediatric neurology, is critical for treatment. Long-term follow-up is necessary to document and to assess growth and development.

(Source: Amended at 48 Ill. Reg. 5175, effective March 12, 2024)