Spinal Muscular Atrophy (SMA) Test

Fact Sheet for Physicians

GENERAL INFORMATION

Spinal muscular atrophy (SMA) is an autosomal recessive disease caused by mutations in the *SMN1* gene. The State of Wisconsin screens all newborns for a deletion in exon 7 of *SMN1*; this method is expected to identify about 95% of all newborns with SMA.

Typically, SMA presents at different ages. In all types, the disease results in progressive motor weakness due to motor neuron loss. Most patients will develop symptoms in childhood, with the most severe type (Type 1) affecting 40% of all patients. In these severely affected patients, death by two years of age occurs in 80% of patients who do not receive assisted ventilation and nutritional support. Other patients may develop progressive weakness, and severe motor, pulmonary and feeding disabilities.

However, there is current treatment for this disease, with evidence showing that earlier intervention improves outcomes, including less weakness, improved developmental milestones, and better long-term health outcomes. Every newborn in Wisconsin will undergo the SMA screening testing. Positive screening results are communicated to the primary care physician and one of the newborn screening program neurology consultants. Those consultants will contact you to arrange for your patient to have confirmatory blood testing done and be available to you for further clinical questions.

WHAT TO DO WHEN YOUR PATIENT HAS A SMA SCREENING POSITIVE RESULT?

Babies who have a positive SMA screening at birth urgently need an additional blood test to confirm the presence or absence of the disease, as well as severity.

Because your families may have questions about newborn screening for SMA, a fact sheet for parents is available at: www.slh.wisc.edu

If you have more questions regarding newborn screening for SMA,
email SMA@slh.wisc.edu