KANSAS DEPARTMENT OF HEALTH AND ENVIRONMENT

NEWBORN SCREENING ACT SHEET

SCREEN FOR: Homozygous deletion exon 7

CONDITION: Spinal Muscular Atrophy (SMA)

METABOLIC DESCRIPTION: SMA is a neurodegenerative disease caused by mutations in the SMN1 gene. It is an autosomal recessive disorder affecting the motor neurons in the spinal cord and brainstem. The homozygous mutations or deletions of the SMN1 gene produces a deficiency of SMN protein, which causes degeneration of motor neurons in the spinal cord. Motor neurons are specialized nerve cells that control the muscles used for activities such as breathing, crawling, and walking. Babies affected with SMA gradually lose motor neurons which causes progressive weakness and atrophy (muscle wasting).

MEDICAL EMERGENCY - ACTION TO BE TAKEN WITHIN 24 HOURS:

- Notify the parents about the newborn screening results. Arrange for immediate referral to neuromuscular specialist (see below) to provide direction regarding recommended evaluation and treatment.
  - Genetic testing is needed to confirm diagnosis.
- Evaluate the infant for signs and symptoms of SMA, including hypotonia, areflexia, swallowing and feeding difficulties and tongue fasciculations.
- Ensure blood sample for genetic testing to confirm diagnosis is obtained.

CONFIRMATION OF DIAGNOSIS: Confirmatory testing is done by genetic testing of the baby's blood sample. The primary care physician or neurologist may order additional tests, coordinate further care and refer to genetic counseling.

CLINICAL EXPECTATIONS. Newborn screening for SMA allows for earlier diagnosis and treatment. There are treatments available that significantly modify the course of the disease, producing better outcomes. The treatment works best when given before symptoms of weakness start. The treatment increases the body's ability to produce more functional SMN protein, helping motor neuron cells to stay healthy.

Without treatment, babies with the most severe form of SMA, whose symptoms start in infancy, typically die in early child hood. Although SMA cannot be cured, it can be treated. Early intervention and treatment of SMA is important because the therapies and medication are most effective in improving children's quality of life when started within the first few weeks of life.

REPORTING: Report diagnostic result to family and Kansas NBS program.

PEDIATRIC SPECIALISTS:

Children's Mercy	Kansas City, MO	Dr. Ann Modrcin – pediatric neuromuscular rehabilitation medicine	816-302-3970
Salina Pediatric Care	Salina, KS	Dr. Britton Zuccarelli- pediatric neurologist	785-825-2273

Cotton O'Neil Clinic	Topeka, KS	Dr. Daniel Katz- pediatric neurologist	785-368-0448
Via Christi Clinic	Wichita, KS	Dr. Bassem El Nabbout- pediatric neurologist	316-796-5610
Pediatric Neurology of Wichita		Dr. Dwight Lindholm- pediatric neurologist	316-651-0033
Neurology Center of Wichita		Dr. Subhash Shah- pediatric neurologist	316-686-6866

ADDITIONAL INFORMATION:

www.curesma.org ***www.ghr.nlm.nih.gov*** ***www.babysfirsttest.org***

DISCLAIMER: These standards and guidelines were adapted from the American College of Medical Genetics ACT sheets. They are designed primarily as an educational resource for
Physicians to help them provide quality medical services. Adherence to these standards and guidelines does not necessarily ensure a successful medical outcome. These standards and guidelines should not be considered inclusive of all proper procedures and tests or exclusive of other procedures and tests that are reasonable directed to obtaining the same results. In determining the propriety of any specific procedure or test, the healthcare provider should apply his or her own professional judgment to the specific clinical circumstances presented by the individual patient or specimen. It may be prudent, however, to document in the patient's record the rationale f