Kansas Newborn Screening Program

NEWBORN SCREENING DISORDER FACT SHEET
INFORMATION FOR HEALTH PROFESSIONALS

Spinal Muscular Atrophy (SMA)

NEWBORN SCREENING FINDINGS

A baby with a deletion in each copy of the SMN1 gene has a presumptive positive screening result for Spinal Muscular Atrophy (SMA). The baby must have confirmatory testing via genetic testing on an additional blood sample to rule out or confirm SMA. Newborn screening cannot determine the severity of SMA. The child should be referred to a specialist for urgent evaluation, confirmatory testing and discussion of treatment options.

ABOUT THE CONDITION

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).

SYMPTOMS

The clinical presentation of SMA are based on the severity of the condition and the age at which symptoms begin. The most severe form is the most common form affecting 60% of infants born with SMA and symptom onset before 6 months of age. Hypotonia and/or muscle weakness and atrophy are common signs that can be noticed at birth or can start later in childhood. SMA is a lifelong condition and if left untreated can cause swallowing, feeding and breathing difficulties, delayed motor milestones, loss of skills like sitting and crawling and a shortened lifespan.

TREATMENT

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.

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.

FORM CONTINUES ON BACK

REPORTING

If SMA is confirmed, Kansas Law 65-180 through 65-183 requires reporting findings back to KDHE by a physician. Financial assistance for SMA clinic services and treatments may be available to the family upon application to the Special Health Care Needs (SHCN) program. A SHCN application will be sent to the baby's address. Parents or physicians can call SHCN at (785) 296-1313 for more information.

RESOURCES

https://www.babysfirsttest.org/
https://www.curesma.org
https://www.ghr.nlm.nih.gov