Kansas Newborn Screening Program

NEWBORN SCREENING DISORDER FACT SHEET
INFORMATION FOR PARENTS

Spinal Muscular Atrophy (SMA)

NEWBORN SCREENING FINDINGS

A baby with a deletion in each copy of the SMN1 gene has a presumptive positive diagnosis of Spinal Muscular Atrophy (SMA). The baby must have confirmatory testing by genetic testing on a blood sample to rule out or confirm SMA.

ABOUT THE CONDITION

SMA is a genetic condition affecting the motor neurons in the spinal cord and brainstem. 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 (wasting).

SYMPTOMS

The features of SMA are based on the severity of the condition and the age at which symptoms begin. Hypotonia or muscle weakness are common signs that can be noticed at birth or can start later in childhood. Newborn screening cannot predict when symptoms will start. 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.

CAUSE

SMA is an autosomal recessive genetic condition due to mutations in the SMN1 gene. It occurs when both copies of the SMN1 gene have a deletion which makes it nonfunctional. This leads to a shortage of the SMN protein, which causes degeneration of motor neurons in the spinal cord.

Babies usually receive two copies of this gene, one each from the mother and the father. Because you need only one functioning SMN1 copy gene to be healthy, parents may pass down a missing or faulty SMN1 copy without knowing it. A baby born with SMA received nonfunctional SMN1 gene copies from both parents.

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 functional SMN protein. The body is then able to produce more functional SMN protein, helping motor neuron cells to stay healthy. The treatments are available through consultation with a specialist in SMA care.

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

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CONFIRMATION OF DIAGNOSIS

Confirmatory testing is done by doing genetic testing from the baby's blood sample. The primary care physician or neurologist may order additional tests, coordinate further care and refer you for genetic counseling.

REPORTING

When SMA is confirmed, the Kansas Law 65-180 through 65-183 requires reporting 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