Iowa Newborn Screening Program (INSP)

Spinal Muscular Atrophy (SMA) Fact Sheet for Healthcare Providers

What is Spinal Muscular Atrophy (SMA)?

Spinal Muscular Atrophy (SMA) is an inherited autosomal recessive condition that results in progressive loss of the anterior horn cells (lower motor neurons) in the brainstem and spinal cord. Loss of these motor neurons leads to progressive skeletal muscle weakness and complications of this weakness.

If SMA is not treated, what problems occur?

The most common form of SMA has symptom onset within the first few months of life, but the disease can present at any age. Features of this disorder include hypotonia, progressive weakness, muscle atrophy, loss of reflexes and fasciculations that are most obvious in the tongue. Infants may have delayed motor development or regression of motor skills. If babies with infantile onset SMA are not treated early in the disease, worsening weakness can quickly lead to failure to thrive and progressive respiratory muscle weakness. Without treatment, the infantile form of SMA typically leads to death in the first two years of life. Untreated later-onset forms of SMA lead to physical disability due to progressive muscle weakness. The most likely type of SMA will be predicted with confirmatory genetic testing and physical findings. This will also guide urgency in the treatment plan.

How to handle an abnormal newborn screen for SMA?

Individuals with SMA require prompt evaluation by a pediatric neuromuscular specialist to confirm the diagnosis, for determination of disease subtype and formulation of a treatment plan. There are medications that can mitigate loss of anterior horn cells. Individuals with SMA will require lifelong management and follow up by a neuromuscular team.

How common is SMA?

The incidence of SMA in the USA is 1 affected individual per 6,000-10,000 live births.

Does SMA go by any other names?

SMA is divided into subtypes by severity, with type 0 being the most severe and type IV being the mildest. The severe, early infantile-onset form of SMA (type I) is the most common form and is also called Werdnig-Hoffmann disease. SMA type II was historically called chronic or intermediate SMA and sometimes Dubowitz disease, while SMA type III was historically called juvenile SMA or Kugelberg-Welander disease.

Where can I find more information?

Genetics Home Reference: https://ghr.nlm.nih.gov/condition/spinal-muscular-atrophy

Gene reviews: https://www.ncbi.nlm.nih.gov/books/NBK1352/

The Iowa Newborn Screening Program is administered by the Iowa Department of Public Health through the Center for Congenital and Inherited Disorders, in collaboration with the State Hygienic Laboratory at the University of Iowa, and the University of Iowa Stead Family Children's Hospital.