Spinal Muscular Atrophy (SMA)

Spinal muscular atrophy (SMA) is a group of inherited conditions that affect the motor neurons of the spinal cord. Motor neurons are specialized nerve cells that control the muscles used for activities such as breathing, crawling, and walking. In people affected by SMA, the loss of motor neurons leads to progressive muscle weakness and atrophy. There are four primary forms of SMA which are classified based on the severity of the condition and the age at which symptoms begin. The symptoms and long-term outlook of each form vary widely. In general, forms of SMA with an earlier age of onset are more severe and have a greater impact on motor function. The more severe types of SMA can affect muscles involved in feeding, swallowing and breathing. The age of onset and signs of spinal muscular atrophy (SMA) differ depending on the form. The long-term outlook for children and adults with spinal muscular atrophy (SMA) varies significantly based on the form, the symptoms present, and the response to treatment.

Incidence: It is estimated that one in every 6,000 to 10,000 babies worldwide is born with SMA.; however, an exact rate of occurrence cannot be determined until more states are screening newborns for the disease.

Diagnosis: Diagnosis is made by genetic testing which should be performed under the care of an experience pediatric neurologist who has the ability to initiate treatment as soon as results are known. Time is critical. There is a measurable difference in the outcomes of infants starting treatment at 0-3 weeks versus 4-6 weeks of age.

Treatment: Treatment includes Spinraza which requires multiple infusions into spinal fluid. A gene therapy is available as well, and Spinraza may also be used until AveXis (gene therapy and one time treatment) can be approved.

Individuals with 4 copies of SMN2 – plan is to defer treatment until the first reliable sign of SMA arises. Preliminary evidence shows biochemical changes will signal degenerative process well before onset of symptomatic weakness

False Positives: False positive results are rarely identified in newborn screening for SMA.

Resources:
Cure SMA Medical Provider Information Kit