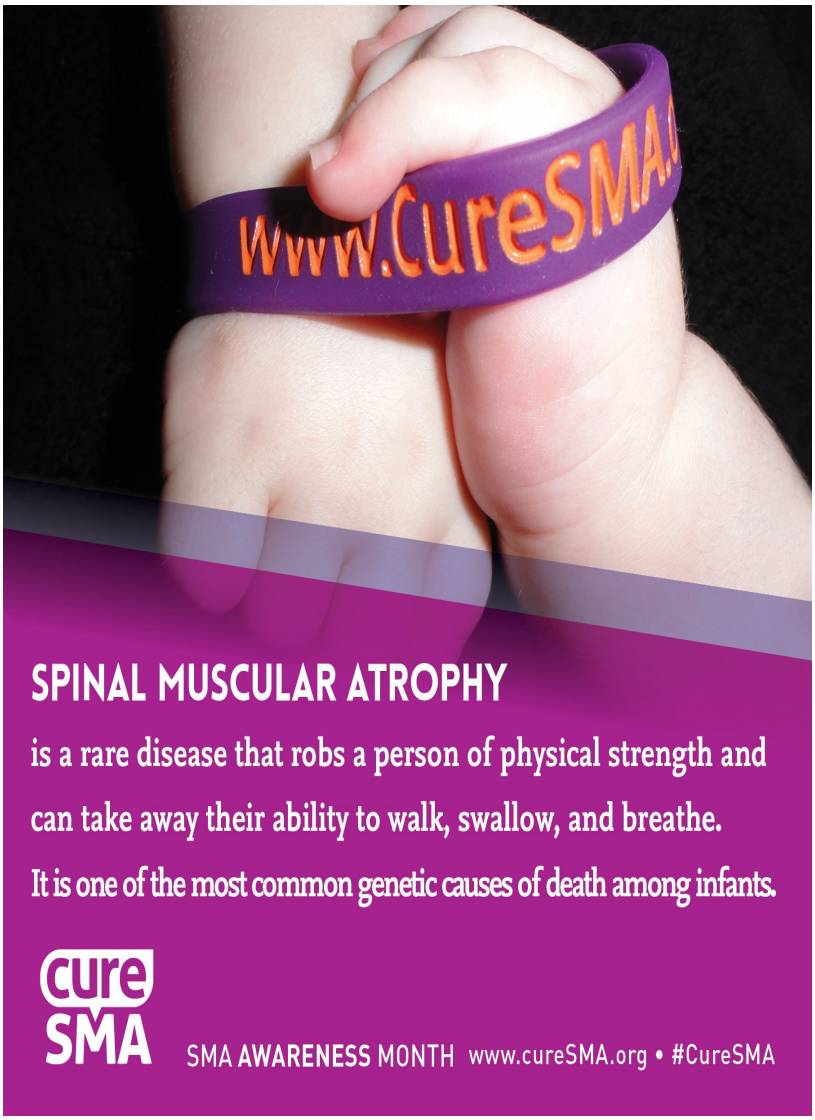
Newborn Screening Follow-up for Spinal Muscular Atrophy (SMA)

The Community Health Clinic



Spinal muscular atrophy (SMA) is a disease that robs people of physical strength by affecting the motor nerve cells in the spinal cord, taking away the ability to walk, eat, or breathe. It is the number one genetic cause of death for infants.

SMA is caused by a mutation in the survival motor neuron gene 1 (*SMN1*). In a healthy person, this gene produces a protein that is critical to the function of the nerves that control our muscles. Without it, those nerve cells cannot properly function and eventually die, leading to debilitating and sometimes fatal muscle weakness.

SMA affects approximately 1 in 11,000 births, and about 1 in every 50 Americans is a genetic carrier. SMA can affect any race or gender. There are four primary types of SMA—1, 2, 3, and 4—based on the age that symptoms begin, and highest physical milestone achieved.

In July of 2018, the Indiana State Department of Health (ISDH) started screening all Indiana newborns for Spinal Muscular Atrophy (SMA). From the beginning, The Community Health Clinic (CHC) has partnered with ISDH to help coordinate confirmatory testing and treatment for newborns affected with SMA. Indiana's screening protocol reports SMN1 absent cases only, which means a baby with a positive screen has some form of SMA. Therefore, every positive screen is considered extremely urgent; time is of the essence in all cases.

If an infant has a screen positive for SMA, the newborn screening lab notifies the CHC via fax and a phone call. The CHC's NBS follow-up coordinator discusses the case with the CHC provider and develops a plan of care. The follow up coordinator then calls the PCP to discuss results, get any updates on the baby's current status and provide instruction for confirmatory testing. If the PCP has not recently evaluated the infant, plans are made for an immediate examination either with the PCP or at the CHC.

Once a plan is in place with the PCP, the NBS follow-up coordinator calls the parents to discuss the findings on the NBS, provide a brief overview about SMA, discuss immediate actions that are needed and coordinate confirmatory testing with the family. During this conversation, the family is also informed that they will need to see a neurology specialist. The CHC follow-up coordinator advises the family of options for neurologists who can provide treatment for SMA within the region and briefly discusses treatment options.

Based on the family's preference, the CHC NBS follow-up coordinator reaches out to the neurologist's office immediately to facilitate an evaluation while confirmatory testing is processing. This early contact helps ensure infants receive life-saving interventions (such as gene therapy) in the most timely manner possible.