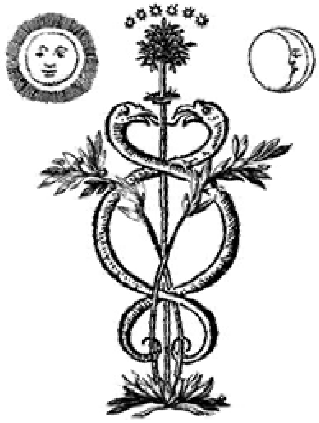

CLAIRE ALTMAN HEINE FOUNDATION, INC.

dedicated to identifying carriers of SMA

Questions about SMA

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What is SMA?

Spinal Muscular Atrophy (SMA) is a hereditary disease that destroys the nerves responsible for controlling voluntary muscle movement. Muscles that control breathing, swallowing, head and neck control, walking, and crawling are the most severely affected. There are four types of SMA, SMA Type I, II, III, IV. The determination of the type of SMA is based upon the physical milestones achieved. Symptoms of the disease range from mild (symptoms that do not appear until adulthood and are not life threatening) to severe (symptoms that are present in early infancy and lead to a severely shortened lifespan). Over 60% of those who are diagnosed with SMA are severely affected. SMA does not affect intelligence. There is no cure or treatment for SMA. Research on effective treatments and cure are underway.

What Causes SMA?

SMA is a genetic disease. In order for a child to be affected by SMA, both parents must be carriers of the abnormal gene and both must pass this abnormal gene on to their child. If both parents are carriers the likelihood of a child inheriting the disorder is 25%, or 1 in 4. If both parents are carriers of SMA the likelihood of a child who is a carrier of SMA is 50%, or 2 in 4. If both parents are carriers of SMA the likelihood of a child with who is neither a carrier of SMA nor affected with SMA is 25%, or 1 in 4.

Who is Affected by SMA?

Spinal muscular atrophy (SMA) is the number one genetic killer of children under the age of two.

SMA is the 2nd most common diseased gene, making it one of the most prevalent genetic disorders.

One in every 40 people carries the abnormal gene that causes SMA. That is 7 million Americans.

Unlike other genetic diseases, SMA has no gender or racial preferences.

One in every 6,000 babies is born with SMA; two children each day in the United States. Over 60% of children are diagnosed before 6 months of age and 90 percent of those children will die before their second birthday.

SMA is relatively common yet virtually unheard of in the general public. SMA is as common as Cystic Fibrosis in the entire population and Tay-Sachs in the Jewish population.

What are the health needs of children with SMA?

There are four types of SMA, SMA Type I, II, III, IV. The determination of the type of SMA is based upon the physical milestones achieved. It is important to note that the course of the disease may be different for each child.

Type I: Type I SMA (also called Werdnig-Hoffmann Disease) is the most severe form of SMA and the most common. Over 60% of children diagnosed with SMA are Type I's.

Usually a child with Type I is never able to lift his/her head or accomplish the normal motor skills expected early on in infancy. They generally have poor head control, and may not kick their legs as vigorously as they should, or bear weight on their legs. They do not achieve the ability to sit up unsupported. Swallowing and feeding may be difficult and are usually affected at some point, and the child may show difficulties managing their own secretions. The tongue may show atrophy, and rippling movements or fine tremors, also called fasiculations. There is weakness of the intercostal muscles (the muscles between the ribs) that help expand the chest, and the chest is often smaller than usual. The chest may appear concave (sunken in) due to the diaphragmatic (tummy) breathing. Due to this type of breathing, the lungs may not fully develop, the cough is very weak, and it may be difficult to take deep enough breaths while sleeping to maintain normal oxygen and carbon dioxide levels.

Type II: The Diagnosis of Type II SMA is almost always made before 2 years of age, with the majority of cases diagnosed by 15 months. Children with this type may sit unsupported when placed in a seated position, although they are often unable to come to a sitting position without assistance. At some point they may be able to stand. This is accomplished with the aid of assistance, bracing or standing frame. Swallowing problems are not usually characteristic of Type II, but vary from child to child. Some patients may have difficulty eating enough food by mouth to maintain their weight and grow, and a feeding tube may become necessary. Children with Type II SMA frequently have tongue fasciculations and manifest a fine tremor in the outstretched fingers. Children with Type II also have weak intercostals muscles and are diaphragmatic breathers. They have difficulty coughing and may have difficulty taking deep enough breaths while they sleep to maintain normal oxygen levels and carbon dioxide levels. Scoliosis is almost uniformly present as these children grow, resulting in need for spinal surgery or bracing at some point in their clinical course. Decreased bone density can result in an increased susceptibility to fractures.

Type III: The diagnosis of Type III, often referred to as Kugelberg-Welander or Juvenile Spinal Muscular Atrophy, is much more variable in age of onset, and children can present from around a year of age or even as late as adolescence, although diagnosis prior to age 3 years is typical. The patient with Type III can stand alone and walk, but may show difficulty with walking at some point in their clinical course. Early motor milestones are often normal. However, once they begin walking, they may fall more frequently, have difficulty in getting up from sitting on the floor or a bent over position, and may be unable to run. With Type III, a fine tremor can be seen in the outstretched fingers but tongue fasiculations are seldom seen. Feeding or swallowing difficulties in childhood are

very uncommon. Type III individuals can sometimes lose the ability to walk later in childhood, adolescence, or even adulthood, often in association with growth spurts or illness.

Type IV (Adult Onset): In the adult form, symptoms typically begin after age 35. It is rare for Spinal Muscular Atrophy to begin between the ages of 18 and 30. Adult onset SMA is much less common than the other forms. It is defined as onset of weakness after 18 years of age, and most cases reported as type IV have occurred after age 35. It is typically characterized by insidious onset and very slow progression. The bulbar muscles, those muscles used for swallowing and respiratory function, are rarely affected in Type IV.

Do all people with SMA have the same symptoms?

No. As described in the previous section some individuals have milder or more severe symptoms than others. It is not always possible to tell from a prenatal test how mild or severe a child's symptoms will be. In general, people with SMA have a severely shortened life span, many die in childhood others live into their 30's or longer. Although there is no cure or treatment for SMA, research on effective treatments and cure are underway.