Spinal muscular atrophy (SMA) is a fairly common genetic disorder, affecting approximately one in 6,000 babies. It is estimated that one in every 40 Americans carries the gene that is responsible for SMA. SMA is an autosomal recessive disorder that leads to the degeneration, atrophy, and eventually to the death of the motor neurons in the spinal cord. It is a disease that is most frequently diagnosed in children, and is the most common fatal neuromuscular disorder in infants. There are three distinct types of spinal muscular atrophy: type I, type II, and type III, all of which are seen in children, and type IV which is diagnosed in adulthood. Type I SMA is the most severe and the most commonly diagnosed type of SMA (Lamb).

The spinal cord, specifically the anterior horn cells, is affected by spinal muscular atrophy. These cells are responsible for the voluntary movements of large muscle groups such as the arms and legs. Proximal muscles are more severely affected than the hands, fingers, and toes. SMA only damages motor neurons, so the patient’s sensory, mental, and other functions remain normal. Genetic testing is done to confirm a diagnosis, as well as other clinical assessments. A patient is typically diagnosed with SMA after an electromyography (EMG). This test measures electrical impulses as they travel from the brain to the voluntary muscles. SMA can also be diagnosed with a muscle biopsy, in which a small piece of muscle tissue is examined for degeneration (Young).

Spinal muscular atrophy is an autosomal recessive disorder, in which both parents must be carriers in order for the disease to show. If both parents carry the gene, there is a 25% chance the child will be afflicted with the disease. All three forms of SMA result from a decrease
in the Survival of Motor Neuron (SMN) protein. The SMN protein is coded by two genes is located on chromosome 5; SMN-1 and SMN-2. For an unknown reason, only SMN-1 abnormalities result in spinal muscular atrophy. Approximately 2% of all SMA cases result in new mutations to the SMN-1 gene, and are not inherited from the parents. In North America and Europe, 1 in 25,000 infant deaths is due to SMA. It is second most common autosomal recessive disorder behind cystic fibrosis. It affects populations uniformly throughout the world. There is a slight reduction in cases in Asian populations, but the reason for this is unknown (Young).

Type I SMA is also known as Werdnig-Hoffmann disease. It is first noticed either during pregnancy or within the first few months of life. Symptoms include weakness and limited movement in the limbs and trunk, impaired breathing, and feeding difficulties. While pregnant, women may notice decreased fetal movement. Children who suffer from type I SMA never are able to gain gross motor skills, and the majority of these children die before their second birthday (Lamb).

Type II spinal muscular atrophy appears slightly later in life than type I, usually diagnosed in infants ranging in age from three to 15 months. The symptoms of type II are similar, and include floppy limbs, decreased reflexes, and muscle twitches in the arms, legs, or tongue. They are able to develop more gross motor skills, such as sitting, but are unable to learn to stand or walk. Respiratory issues are common, and greatly influence the life expectancy of these children. Assistive devices are often necessary due to weakness in the chest muscles. Scoliosis is commonly found in children, and often requires surgery (Lamb).
Type III is the least severe of childhood SMA, and is also known as Kugelberg-Welander Disease. It can present itself anywhere between two and 17 years of age. The symptoms include abnormal gait and difficulty running or climbing. These can become progressively worse as the child ages, and can be further escalated by a serious injury. A broken bone, which leads to less use for a substantial length of time, can lead to quickened atrophying of the muscles and inability to regain strength (Lamb).

There is no cure for any form of SMA. There is hope that gene therapy may become possible to replace the mutated or missing gene, but this possibility is still years away. Currently, the only treatment options involve the management of symptoms (Young). Respiratory complications are the most dangerous issue facing SMA patients, and result in the majority of deaths. As time progresses, a ventilator is typically required to aid breathing. The intercostal muscles between the rib cages are typically weakened, which causes lungs to not develop properly. As a result, the diaphragms face an increased demand and are responsible almost solely for breathing. This does not allow the rib cage to expand as necessary during inhalations. Hypoventilation is common during sleep. It is often the first sign of respiratory illness seen in patients (Poutney). Coughing is essential in SMA patients to clear the lungs and throat of moisture and prevent secondary infections, such as pneumonia. Chest physiotherapy (CPT) is used to trigger and assist patients in coughing (Young).

Mallory is an 8 year old female who suffers from spinal muscular atrophy type 1. She was born on May 5, 2001. She receives home health speech, physical therapy, and occupational therapy from Pediatric Therapy Works as well as physical therapy and occupational therapy from homebound school services. She is a very bright child who thoroughly enjoys participating
in her therapies. When not under the care of her mother, she is cared for by a private nurse.

She is fully dependent for all daily living tasks. She now uses a tracheotomy for respiration, and receives nutrition through a gastric tube.

During occupational therapy treatments, the therapist works on maintaining passive range of motion in her extremities as well as limited active range of motion in her wrist. Her current therapy goals are to maintain her current range of motion, to provide as many opportunities as possible for her age level, and to provide perceptual motor and visual motor activities with adaptations as required. Splints and other equipment are also provided as needed. During a typical weekly session with Mallory, soft tissue massages and stretches are used, as well as an activity for entertainment as well as an opportunity to use active range of motion with a switch adaptive toy.
References

