

## **OTHER HEALTH CARE ISSUES RELATED TO SMA:**

**Respiratory**

**Orthopedic**

**Therapeutic**

**Nutrition**

**Genetics**

### **Respiratory**

Respiratory care is often a central focus in the medical management of SMA. Intercostal muscles (muscles supporting the chest wall) may be weak leaving the diaphragm as the primary breathing muscle. This may lead to decreased respiratory function with lung underdevelopment and difficulty in coughing and clearing secretions. This has the potential to cause recurrent chest infections.

The breathing function can be assessed by pulmonary function testing in children over 3-4 years. They are asked to blow into a tube that measures their lung capacity.

Chest physical therapy may be used to assist your child in clearing secretions. It often includes position changes that allow gravity to help mobilize secretions and keep lungs clear. Each of our lungs has several compartments that we call lobes. It is important to make sure secretions do not collect in these lobes. The individual can be placed in different positions to allow gravity to empty out each lobe/compartments of our lungs. We can this systematic change of positions postural drainage. It can often be performed at home with the supervision of a therapist.

A cough assist device may also be used at home to help children cough and clear their secretions. Children with SMA rely on their diaphragms to breathe. At night when they are lying down, the abdominal contents push up against the diaphragm and gravity is eliminated, making it harder to take deep breaths. This can cause our lungs to not get enough oxygen and cause carbon dioxide to build up. Children may experience daytime sleepiness or headaches from this. Doctors may help by giving a BIPAP machine, which helps the child breathe by giving extra bursts of air (intermittent positive inspiratory pressure). This assistance allows the child's breathing muscles to get a rest.

If a chest infection occurs, all the above supportive care can be done at the first sign of any chest problem. Antibiotics and inhalation therapy may also be needed. Sometimes hospitalization is required to best manage and care for the child

### **Orthopedic Care**

Orthopedic issues refer to the state of our bones and the associated muscles, ligaments and other soft tissue. Maintaining proper alignment and muscle balance helps us avoid problems in this area. Children with SMA, who may be unable to maintain good alignment and/or who have difficulty moving, may be at risk for certain orthopedic issues.

One of the most common concerns is spinal alignment. A curved spine affects our comfort, ability to balance ourselves in sitting and standing, our arm function, and, most importantly, can affect

our breathing. If an individual does develop a side-to-side curvature of the spine, it is referred to as scoliosis. The spine of an individual who has scoliosis may look like a "C" or an "S", instead of being straight.

Scoliosis may develop in children who are wheelchair bound due to weakness of the muscles supporting the spinal column. Scoliosis is slowly progressive in SMA, and needs to be followed periodically. Doctors will follow this with a physical examination or may ask to obtain x-rays.

There are two ways to help. The first is positional curve control. This in any means that puts the spine into better alignment. This can be done with either wheelchair modifications (positioning devices) or bracing with a soft spinal orthosis to help the child maintain a straight sitting posture. The wheelchair modifications mentioned above can range from a simple lateral support that promotes upright sitting to custom molded back support that fits your child's body exactly.

These positional curve controls may improve function, independence, comfort and quality of life. However, since they can typically not prevent progression, they should not be used if they cause discomfort.

Many surgical techniques exist to correct scoliosis. The purpose may be to correct the curve and stabilize the spine. A consultation with an orthopedic doctor is recommended.

Hip surgery is usually not needed in non-ambulatory SMA patients. Other orthopedic concerns can be due to joint contractures. Tendon release is typically not needed unless it can improve function, e.g. allow for the use of a standing device.

Range of motions exercises and positioning (e.g. night splints) may assist your child in preventing joint discomfort and malalignment.

### **Therapeutic Care**

The role of exercise in Spinal Muscular Atrophy is to assist in improving flexibility, function, independence and quality of life. There is no evidence to support traditional strength training. Therefore it is often best for children with SMA to exercise by practicing the movements and tasks they want to perform throughout the day. We call this type of practice functional exercise. It can consist of any movement or position one would like to strengthen in order to improve their activities of daily living, for example rolling, reaching, sitting and maybe even walking. These activities are dependent on age, amount of neuromuscular involvement, and developmental stage. Although exercise is important, excessive exercising may cause over-fatigue and damage. Check with your physical and occupational therapists to ensure you are performing appropriate types of exercise.

### **Nutrition**

Diet is very important and deserves careful considerations to help limit unnecessary losses of muscle mass; explanation of the importance on energy, protein, fat, and fiber requirements specifically to SMA children; frequent gastroesophageal reflux disease in SMA 1, 2, & 3; abnormal fatty acid metabolism in more severe forms of SMA; serious concerns with obesity, and management of diet during illness.

### **Genetics**

Spinal Muscular Atrophy is an autosomal recessive disease. In order for a child to be affected by SMA, both parents must be carriers of the defective gene and both must pass this gene on to

their child. Although both parents are carriers, the likelihood of passing this gene along to a child and having an affected child is 25%, or 1 in 4.

SMA is caused by a mutation in the survival motor neuron gene 1 (SMN1). This SMN1 gene is responsible for the production of a protein that is essential to the proper working of the motor neurons. People who are missing both copies of SMN1 have SMA, while carriers are missing only one copy. There is a similar copy of SMN1 called SMN2 that even people with SMA always have. The SMN2 gene differs by producing only a little SMN protein. This small amount of SMN protein is not sufficient for the normal function of the motor neuron. The lack of this protein causes the motor neurons in the spinal cord to degenerate resulting in SMA.

90-95% of all cases of SMA can be detected through a blood test to screen for SMN deletion. For carriers there is a prenatal test available that allows for prenatal testing with 98% reliability.