Frequently Asked Questions

1. **If no members of my family are affected by SMA, can I still have a child with the condition?**
   Yes. Due to the way the disease is inherited, it is possible for the defective gene to be present in the family without anyone showing symptoms. Hence, you may not be aware of a family history.

2. **Since SMA is a genetic condition, is it possible to have more than one child with the condition?**
   Yes. When both parents are carriers of the gene change, there is a 25% risk with each pregnancy of having a child with SMA.

3. **If I am a carrier, can I develop SMA?**
   No. Carriers do not show any symptoms of the condition as they have one functioning copy of the gene which is sufficient for the production of the required amounts of protein.

4. **Would a child with SMA be able to attend school?**
   Yes. Children with milder forms of SMA are highly encouraged to attend school. The condition does not affect the mental development of patients.

5. **Can people with SMA achieve a normal lifespan?**
   Some forms of SMA shorten lifespan. The average lifespan of an SMA patient depends on the age of onset of the condition. The later the age of onset the higher the likelihood of achieving normal lifespan. Patients are advised to consult their physician to obtain specific information regarding this.

6. **What are the major challenges faced by people with SMA?**
   Weakness of the respiratory muscles is potentially the most serious issue faced by SMA patients. Ventilatory assistance is usually needed to facilitate breathing. Swallowing and chewing muscles may also be affected in some types of SMA. Those facing this difficulty would often require feeding aids.

References

Facts About Spinal Muscular Atrophy. Muscular Dystrophy Association. 2003

Tsao, B. & Armon C. Spinal Muscular Atrophy. [online] Available from: http://emedicine.medscape.com/article/1181436-overview [Accessed May 2010]

Understanding SMA. Families of Spinal Muscular Atrophy. [online] Available from: http://www.fsma.org/FSMACommunity/UnderstandingSMA/ [Accessed May 2010]

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Rare Disorders Series

Spinal Muscular Atrophy (SMA)

What is Spinal Muscular Atrophy (SMA)?

Spinal Muscular Atrophy (SMA) is a group of genetic disorders characterized by muscle wasting and loss of motor function.

It is caused by changes (mutations) in the *SMN1* gene.

Genes are located on chromosomes and contain 'recipes' to make proteins.

The proteins made by the *SMN1* gene are essential to the motor neurons. Absent or defective proteins cause the nerve cells in the spinal cord which are responsible for muscle contraction to become unhealthy. This leads to muscle weakness and wasting.

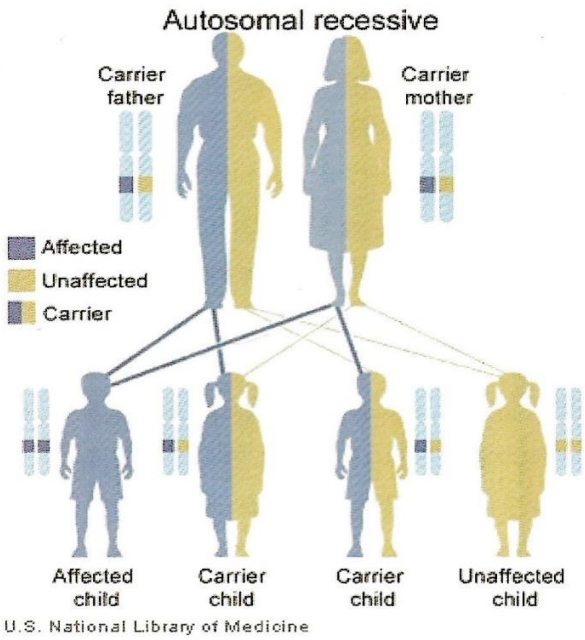
SMA is divided into four types according to age of onset and severity of symptoms.

- SMA type I: Infant onset
- SMA type II: Onset between 6 and 18 months
- SMA type III: Onset may be as early as the toddler years or as late as adolescence
- SMA type IV: Onset in mid thirties

SMA follows an autosomal recessive inheritance pattern. This means that an individual would need two copies of the changed gene to have the disease.

Inheritance of the mutation happens by chance and is not due to anything either parent did or did not do before or during pregnancy.

Patients are advised to consult a geneticist to obtain further information on disease progression.



Inheritance pattern of Spinal Muscular Atrophy

Signs and symptoms

Type I:
- Limited movement
- Difficulty in feeding, swallowing and breathing

Type II:
- Failure to crawl or walk
- Able to sit without support but not walk independently

Type III:
- Able to stand and walk, but may have difficulties standing up from a sitting position

Type IV:
- Difficulty in walking observed in mid thirties

Respiratory problems are also seen in patients with SMA types I, II and III. Its severity varies according to the type of SMA.

Testing

Genetic counselling by qualified personnel is advised if there is any family history of the condition and before any genetic testing is done.

Genetic testing is available to confirm the diagnosis of SMA if there is suspicion of the disease based on physical observation.

If genetic testing yields a negative result but symptoms typical of SMA are observed, further tests such as muscle biopsy and electromyography may be necessary.

Treatment and management

There is currently no known cure for SMA.

Treatments are given to manage symptoms and prevent complications.

Breathing aids are usually required for those with SMA type I or II. Regular chest massage and breathing exercises are also recommended to improve lung function.

Those facing difficulty in swallowing and feeding may need feeding tubes to ensure that they receive adequate nutrition.

Scoliosis (curvature of the spine) is a concern for many patients. Surgery may be required to manage this depending on the degree of the scoliosis.

Standing and mobility aids such as standing frames, braces and wheelchairs help to increase independence.