SPINAL MUSCULAR ATROPHY- SMA
Information for Families

What is SMA?
Spinal Muscular Atrophy (SMA) is an inherited rare disease where there is progressive loss of lower motor nerve cells in the brainstem and spinal cord that control essential voluntary muscle activity such as walking, sitting, breathing and even swallowing. So there is weakness and muscle atrophy as a result of this neuromuscular disease. There are an estimated 3 to 4 lakhs affected children in India, although this may be an under prediction as it is from an extrapolation of western data and we have more consanguinity.

What are the types of SMA?
Children with SMA can be placed into different types, depending on when affected children start showing signs of the disease.

SMA Type 1: This is a devastating disease, which is usually seen by the first six months of life. Children have hypotonia (they are soft and floppy) and muscle weakness that gradually worsens; affecting the legs more than the hands as well as swallowing, although the face is often spared. Babies typically have a weak cry with breathing difficulties. Life expectancy without treatment is usually less than 2 years. It is difficult to accurately predict life span, as it is dependant on the extent of breathing difficulty the child has.

SMA Type 2: Affected children with this condition start showing symptoms between the ages of 7 and 18 months. They are unable to stand without support and are called “Sitters”. Weakness in their chest wall muscles makes it difficult for them to cough effectively, thus making them vulnerable to having repeated chest infections. This is a serious condition that can shorten life span, but with improvement in standards of care, affected children live longer and have more fulfilling lives.

SMA Type 3: This can further be divided into Type 3a and Type 3b based on whether the disease manifests before or after 3 years of age. Affected children have weakness and problems with mobility. They are described as “sitters” and “walkers”. They don’t usually have breathing problems and can live long fulfilling lives.

SMA Type 4: This is an adult and non-life threatening form, where the onset of the disease occurs later in life. There is some weakness in the legs and hands.

What causes SMA?
SMA occurs due to damage and loss of the lower motor neurons. This damage most commonly is because of defects in the SMN1 gene that is responsible for the production of an important protein called the Survival Motor Neuron or SMN protein. If there is insufficient amount of SMN protein, the lower motor neuron progressively degenerates, causing progressive weakness. The lower motor neurons carry signals from the brain to the muscle and the lack of transmission of these signals causes muscles to waste and atrophy.

How is SMA inherited?
SMA is a genetic disorder caused most commonly by a complete absence of both the copies of the SMN1 gene. Affected children have some copies of the SMN2 gene, which is similar in structure but doesn’t efficiently produce SMN protein. Parents of affected children are often carriers of the defective gene, which means that they have only one normal copy of SMN1 gene. This in turn means that they have a 1 in 4 chance of having an affected child with each pregnancy.

Is there any treatment for SMA?
Currently there is no available cure for SMA in India. This doesn’t mean that nothing can be done. A range of options are available for managing symptoms and complications and thus improving quality of life. Internationally agreed Standards of Care in SMA outlines these options and emphasises on multidisciplinary nature of the management of these affected children.

Possibilities for cure:
The genetics of SMA offers a unique opportunity for developing therapeutics as affected children with SMA have no copy of SMN1 gene but still have a backup gene: the SMN2 gene, which produces SMN protein, although very inefficiently. So SMN2 and production of SMN protein is the target of developing therapeutics.

A lot of research is being done all over the world to cure SMA. Currently the first and so far, potentially the only available drug for the treatment of SMA received FDA approval in December 2016 and is called:

Nusinersen/Spinraza™

This drug is designed to modify the production of SMN protein by targeting SMN2 gene. This is currently unavailable in India.

Some resources:

1. TREAT-NMD website: http://www.treat-nmd.eu/sma/overview/
3. A list of patient support groups for SMA all over the world are available in this website: http://www.treat-nmd.eu/sma/patient-organizations/