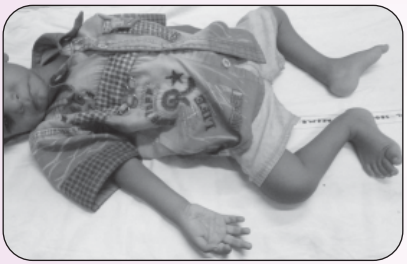
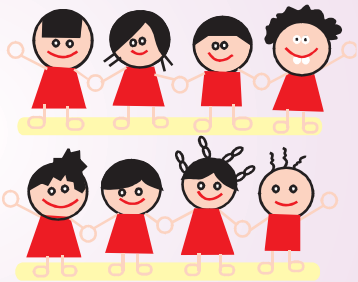
ALL YOU NEED TO KNOW ABOUT SPINAL MUSCULAR ATROPHY !

Child Neurology Division
Department of Pediatrics, AIIMS, New Delhi

Parents/Patient Education Guide





Child Neurology Division
Center of Excellence & Advanced Research on Childhood Neurodevelopmental Disorders
Department of Pediatrics
All India Institute of Medical Sciences
New Delhi India
www.pedneuroaiims.org

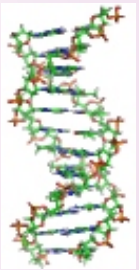
What is SMA ?

Spinal muscular atrophy (SMA) is a genetic disease that destroys the nerves responsible for controlling most of our body movements. It does not affect intelligence. It presents at a variable age depending on the type of disease (discussed later). Diagnosis requires a simple blood test. There is currently no cure but early diagnosis and initiation of supportive management prolongs the survival of these children. A lot of research is going on around the world to search for its cure.

What causes SMA ?

SMA is an inherited disorder of nerve cells which is caused by faulty genes (the units of inheritance that parents pass on to their children). This fault impairs the functioning of a specific nerve cell in the body called *anterior horn cells*. This causes weakness of muscles and lack of muscle control. This causes the children with SMA to be 'very loose' or floppy with inability to sit, stand, walk, drink or eat and even breathe.

Who gets SMA ?



Both boys and girls can get the disease. It affects one in 10,000 infants.

Understanding the genetics of SMA !

Almost all children with SMA are missing a copy of a gene called SMN1, which is important in producing a protein needed for some nerve cells to live. They do have a back-up SMN2 gene, but it does not work properly. Therefore the functioning and life of these nerve cells is affected.

If both parents are carriers of the disease (i.e they each have one normal and one abnormal copy of the gene and have no symptoms of the disease), then the risk of having a child with SMA is 25% with each pregnancy. If none of the parents are carriers then the risk of having a child with SMA is just around 1%.

What are the different types of SMA ?

There are 3 types of SMA depending on the age of presentation :

Type I SMA : It is the most severe form of the disease. It presents in the first 6 months of life. They may present

immediately after birth with difficulty in breathing and breast feeding. Many of them die soon after birth. Those who survive continue to show poor feeding, labored breathing, noisy breathing, floppiness, decreased body movements and weak cry. They may have repeated pneumonias. Most of them are not able to sit ever. They rarely cross the age of 2 years.

Type II SMA : It presents between 6 and 18 months of age. They present with floppiness, weakness of limbs and delayed motor development. They have variable degree of feeding and breathing problems. Most of these children will be able to sit one day and very few will be able to walk. They may live upto their 20s and 30s.

Type III SMA : These children present after 18 months of age. They may have delayed walking. They have difficulty in rising from sitting position and climbing stairs. They usually have waddling gait and abnormal curvature of the spine. They may have tremulousness of hands. They are usually able to walk and may have normal lifespan.

How SMA can be diagnosed ?

If clinically the doctor strongly suspects SMA, then he/she goes for genetic testing. 4 in 100 patients with SMA may have negative results with this test. Otherwise to differentiate from muscle diseases children are usually screened with a blood test called *creatine phosphokinase (CPK)*. The results of this test is variable in SMA but helps the doctor to differentiate SMA from other diseases which present in a similar fashion. This test is available in most laboratories.

If both genetic testing and serum CPK are inconclusive, then electrical stimulation test of nerve and muscle is done followed by muscle and/or nerve biopsy.

Can carriers in the family be identified?

If the parents are planning for any future pregnancy, both can be screened for the carrier status for SMA. Specialised genetic counselling is now available to all families. If both parents are carriers of the disease (i.e they each have one normal and one abnormal copy of the gene and have no symptoms of the disease), then the risk of having a child with SMA is 25% with each pregnancy. If none of the parents are carriers then the risk of having a child with SMA is just around 1%.

Is there any treatment?

Unfortunately no cure has yet been discovered. We do have ways to manage its complications. These have had a very important impact on the quality

and length of life that can be expected with this condition. Intensive research to find a cure is carrying on in many centres around the world. Trials with valproate and gabapentin have not been very encouraging.

What medical help is required ?

Immunization : The routine immunization as per the national schedule must be completed. As these children are prone to develop pneumonia, the following additional vaccines are usually recommended:

- **Pneumococcal Vaccine :**
  - If age < 2 years : PCV vaccine is recommended (protects against 7 types of a germ called pneumococcus which causes pneumonia).
  - < 6 months : 3 doses 4-8 weeks apart, booster at 15-18 months
  - 6-12 months : 2 doses 4-8 weeks apart, booster at 15-18 months
  - 12-23 months : 2 doses 4-8 weeks apart
  - 24-59 months : 1 dose
  - After 2 years of age, a single dose of PPV23 (protects against 23 types of the germ called pneumococcus which causes pneumonia) must be given. At least there should be a gap of 2 months between PCV and PPV23. The repeat dose of PPV 23 may be given after 3-5 years if the child is less than 10 years of age and after 5 years if child is aged more than 10 years of age.
  - If the child is unimmunized at 2 years against pneumococcus, then a single dose of PCV followed by PPV23 (Separated by atleast 2 months) should be given. If parents are not affording, a single dose of PPV23 is sufficient.

- Influenza vaccine should also be given yearly to protect against pneumonia caused by influenza virus.

Rehabilitation in SMA patients : Muscle weakness is the most prominent symptom of SMA. It varies from person to person, depending on disease severity. Planning for managing muscle weakness is important for helping your child achieve his or her highest level of function and independence. Physical therapists, occupational therapists, speech therapists and /or rehabilitation specialists in coordination with a pediatric neurologist are the experts who can help you design the best plan for your child. They can recommend exercises and tools or assistive devices to help your child maintain the best posture for lung function and day to day regular

activities.These can also help manage and prevent muscle tightness, back deformity, pain and bone fractures that can worsen your child's disability

Grossly your child will fall into one of these categories: nonsitter, sitter and walker. Accordingly his requirements will be different.

In a nonsitter our focus is on nutritional support, posture management, seating, muscle tightness and pain management, therapy for activities of daily living and assistive equipment, wheelchairs for mobility, limb orthotics, and developmental therapies.

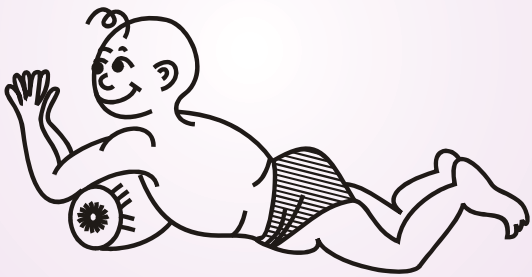
In sitters and walkers our emphasis is on wheelchair mobilty, muscle tightness management, physical therapy, occupational therapy with consideration for spine and limb orthotics and spine surgery whenever required.

Now we will discuss each component in little bit detail:

1. *Therapeutic exercises*: Here the focus is on performing and practising activities of day to day life (for example, rolling, reaching, sitting and even walking), not on traditional strength training. This should be done under the able guidance of your physio and occupational therapist.

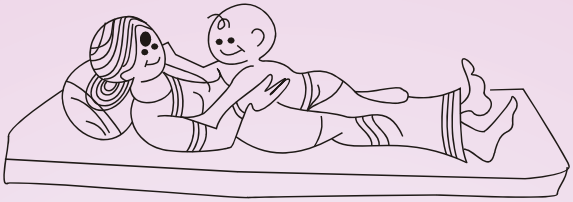
POSITION TO ACHIEVE NECK CONTROL

Bolster



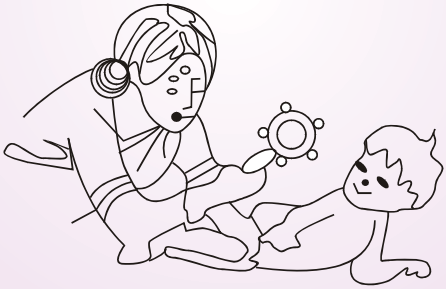
1. Make the child lie over a bolster.
2. Put his arms in front over the bolster.
3. Make the child hold his head up by showing him a toy etc.

4. In place of bolster mother can also use her lap for same.



TO ACHIEVE ROLLING AND TWISTING

1. If the child is very stiff , first help her loosen up.
2. Swing her legs back and forth .



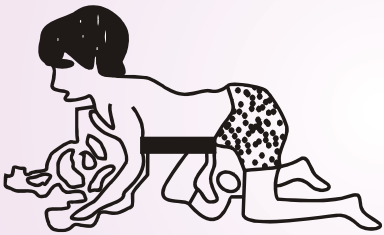
3. Then help her learn to twist her body and roll.

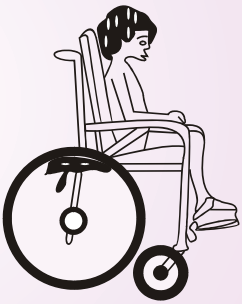
2. *Range of motion*: As children with SMA are too weak to move their joints through complete range of motion, so they have more chances of developing permanent tightness of muscles and restriction of movement at joints. Your child may be advised braces or splints to align your joint appropriately by the concerned expert.
3. *Weightbearing and mobility*: It can be achieved by the use of various assist devices like standers, walkers and orthotics(splints/braces) and wheelchairs. Orthotics are devices used to align and support the foot.

Supervised water therapy can be specially helpful in patients with SMA. Floating on the water makes your child's body light and easily manouverable.

AIDS FOR MOVING ABOUT

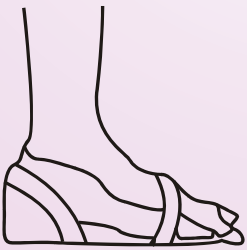
Aids for ways to get from place to place should provide corrective positions.





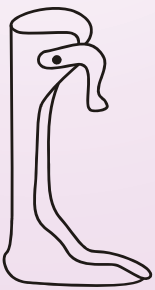
COMMONLY USED ORTHOSIS

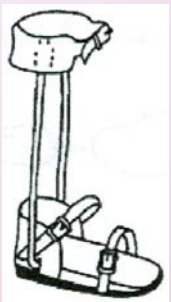
FOOT ORTHOSIS



Usually made of moulded plastic for lower leg, deformity in the foot, such as flat foot

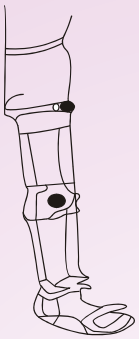
BELOW KNEE ORTHOSIS





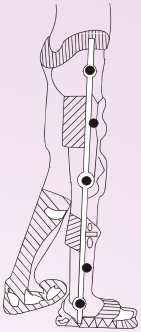
For weakness or deformities in ankle and foot

ABOVE KNEE ORTHOSIS



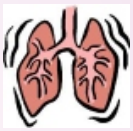
For weakness in the upper leg and knee

ABOVE KNEE ORTHOSIS WITH A HIP BAND



For severe weakness in hips and legs Possibly also for ankle and foot

4. *Orthopaedic management*: This refers to the management of your child s bones and joints by an expert. One of the glaring orthopaedic complication in patients with SMA is scoliosis which is sidewise curving of spine. This is secondary to weakness of back muscles and less mobility. Initially it is managed with postural correction and physiotherapy, in severe cases surgery may be required.



5. *Respiratory care*: This is important as patients with SMA also have weak breathing muscles. They are taught appropriate physiotherapy to help clear their secretions, sometimes assistive cough devices are also advised. They are also prone to airway infections for which appropriate medical help should be sought.

Feeding and swallowing problems : This is usually seen in Type I and II SMA. Symptoms may include prolonged meal time, fatigue with feeding, choking or coughing while feeding and repeated pneumonias. They also usually have gastro-esophageal reflux (i.e. tendency of food to move back from stomach to foodpipe or even mouth), constipation and abdominal distension. Some investigations to assess these problems may be ordered by your doctor. Referral to a dietician is essential. Some medication (taken by mouth) may be prescribed. Your child may require a nasogastric tube (tube passed through nose into the stomach) for feeding. Very rarely a surgery may be required.

Can SMA be diagnosed before birth?

Once a child with SMA has been born in a family, it is often possible to offer prenatal (meaning 'before the birth of the child') diagnosis in future pregnancies. This is normally possible when genetic studies give positive results in the child. The test is performed on a tiny piece of the developing placenta usually at about the 11th-12th week of pregnancy and studying its genetic make up to locate the faulty gene. For further details, parents should talk to a geneticist, a doctor who specializes in these issues.

What are the newest advances in the treatment of Spinal Muscular atrophy?

Recently a drug named Nusinersen has been approved to treat children and adults with spinal muscular atrophy (SMA). Nusinersen is administered via injection into CSF. It is an antisense oligonucleotide aimed at increasing concentrations of survival motor neuron (SMN) protein, which is under expressed in SMA.

Who should consider carrier testing for spinal muscular atrophy?

Those who have a family member with SMA or a family member known to be a carrier are at increased risk to be a carrier themselves. Genetic counseling is recommended in these cases to determine the likelihood of having a pregnancy or child affected with SMA. An individual without a family history of SMA can be a carrier for this condition as well. About 1 in 50 people, regardless of ethnic background, are carriers of the abnormal gene that causes SMA.

How do I know if I am a carrier for spinal muscular atrophy?

Carrier testing is available through a simple blood test. The test can detect the most common mutation that is associated with SMA and will detect approximately 90 percent of carriers. A negative carrier test will greatly reduce the likelihood of having a child affected with this condition. However there are gene changes (mutations) associated with SMA that are not included in the carrier test. In addition, in a small number of cases where a child is found to be affected, only one parent is a carrier. For both of these reasons, a negative carrier test cannot eliminate the possibility of having an affected child.

Child Neurology OPDs	**Tuesday & Friday 9 a.m. onwards**	**Room No.4, 5, 14**
Development Clinic	**Monday 2 p.m. onwards**	**Room No.5**
Neurocysticercosis Clinic	**Monday 2 p.m. onwards**	**Room No.11**
Pediatric Neurology Clinic	**Wednesday 2 p.m. onwards**	**Room No.3, 4, 5**
Autism Clinic	**Thursday 9 a.m. onwards**	**Room No.12, 13, D**
Neuromuscle Disorders Clinic	**Friday 2 p.m. onwards**	**Room No.3, 4**

For any queries please contact

Professor Sheffali Gulati
Chief, Child Neurology Division
Faculty Incharge, Center of Excellence and Advanced Research on Childhood Neurodevelopmental Disorders
Department of Pediatrics
AIIMS, New Delhi
Email- pedneuroaiims@yahoo.com, pedneuroaiims@gmail.com
Post a query on our website: www.pedneuroaiims.org