THE JENNIFER TRUST
FOR SPINAL MUSCULAR ATROPHY

Factsheet

Severe (Type I) Spinal Muscular Atrophy

What Are The Effects of SMA Type I?

This is the most severe form of SMA. Its effects are seen within the first six months of life, and can affect babies even before birth. Mothers frequently remember that their baby had become less active towards the end of pregnancy.

Within a few weeks or months of birth, babies are limp and floppy because most of their voluntary muscles are affected. This means that babies are unable to lift their heads, have difficulty rolling over and are unable to sit unsupported. Muscles used for sucking and swallowing can also be affected and may cause difficulty when feeding. These problems can, to some extent, be overcome and most babies are still able to make the most of their limited abilities (see section on treatment). What they seem unable to do physically, they appear to compensate for with facial expressions, particularly their beautiful eyes and smiling faces! Also it must be remembered that the brain is totally unaffected and these babies are often bright, alert and responsive.

However, it is usually difficulties with breathing, coughing and the baby's susceptibility to respiratory diseases and infections which make it unlikely that babies suffering from SMA Type I will live to see their first birthday. It must be remembered, though, that it is practically impossible to put a time limit on each baby's life, because they are all so very different (with differing muscles being more affected than others). Many consultants inform parents at the time of diagnosis that SMA Type I babies can live up to two years old (which is medically correct). This, however, is exceptional. The majority of babies (approximately 80%) die in the first year of life.

Loan Library

JTSMA has some equipment available on loan, which you may find of use at some point in time. Specific for Type I SMA are: -

Toy Packs

Suction Machines (portable)

Seats/Car Seats/Buggy

Combisystems: Tendercare Snugseats

These are available on loan, free of charge. Carriage is kindly donated by Securicor, usually within 48 hours. Please contact The Jennifer Trust for more details - the postal address is at the end of this document.

Toys

Even though Type I babies have only a relatively short life; their lives can very easily be surrounded by joy and happiness. This can be fulfilled fairly easily by using carefully selected toys from which they can gain a great deal of enjoyment. It can often be difficult, however, to

find toys and games that they can use and play with themselves because of their limited movements. Basically, you need toys that are light or can operate by themselves. Below is a list of suggestions and ideas as to how you may be able to give your child as fulfilling a life as possible. Not all ideas may be suitable for you or your child, but hopefully you may be able to find something listed here that benefits your child.

1. Bright bangles, pencils, pegs, feathers, straws, tissue-paper - all different colours, and light enough to hold.
2. Balloons - plenty of fun, especially helium filled with a string attached.
3. Silver bottle tops on a string - light and good to feel.
4. Bubbles - great to watch.
5. Wind up toys, e.g. musical television, clock.
6. Activity Arch with additional items hanging down, e.g. silver paper, feathers - easier to move when the child is lying underneath.
7. Mobiles and musical toys with a pull down string.
8. Videos - lots of movement and colour, especially the Walt Disney Sing-a-long videos.
9. Toys and Games that are touch sensitive.
10. Lullaby Light Show.
11. Wrist Rattles.

Babies can be propped up fairly easily in a bean bag, especially with a frame over them with a variety of toys hanging off strings. This all depends, of course, on how comfortable the baby feels in the sitting position. The standard baby seats can also be propped up at different angles and babies are often better off at a slightly elevated angle at perhaps 30° or 40°, than lying flat on their backs.

JTSMA also has a Toy Library that has many of the above-mentioned toys. They also stock a limited number of specially adapted switches that respond to a very light touch.

The benefits of water must not be forgotten either. Baths give these babies the ability to kick around a little and bath time can often be the best time. Assisted movement can also be a good game as well as being excellent therapy.

Above all, however, what these babies need most of all are people. Being so alert, they need plenty of communication. Singing games, e.g. This Little Piggy, Round and Round the Garden are long time favourites. Changes of scenery can also be very beneficial - take them into different rooms or outside (depending on the weather), which will help to stimulate them. Give them plenty of opportunity to touch, taste and smell with plenty of approval and recognition whenever they achieve anything, no matter how small it may be.

Is There Treatment For SMA?

Research into SMA is progressing, but at the present there is no known cure and no treatment that will repair the damage to the anterior horn cells or reverse the weakening of the muscles. But who knows what the future will bring?

However, PHYSICAL THERAPY can provide movements that babies are unable to make. The babies will enjoy these whilst it is also being very good for their circulation and avoids any stiffening of the joints. Respiratory exercises are very important. They help to clear the chest when babies have difficulty in coughing and also help to reduce the effect of chest infections.

HYDROTHERAPY can also be of excellent benefit to babies, as they can really enjoy the sensation of their limbs moving in warm water.

When babies have difficulty clearing phlegm from the back of their throats, a SUCTION MACHINE may be useful. This involves popping a thin, plastic tube to the back of their mouths, which rids all mucus collecting there. This usually gives great relief to the baby and although they don't always enjoy the actual process, they certainly feel the benefit afterwards.

Sometimes, a special FEEDING TUBE may also be helpful when swallowing becomes difficult. It can often become a battle to feed babies, causing extreme concern about whether they are eating enough. Tube feeding can take all the stress and worry out of feeding times, both for you and the baby. It is worth remembering that the sight of a tube in your baby's nose is a lot more distressing for you than it is for the baby.

OXYGEN may also be of help. Your GP will be able to advise you best, should the need for this course of action arise.

Advice on all these techniques can, and should, be obtained from a physiotherapist who will be attached to your local hospital, or GP's surgery. These various forms of treatment can seem a little daunting and not all babies will need them. Our babies are very special, and with care and consideration their symptoms can be controlled. They deserve a good quality of life and surely anything that makes them more comfortable is worth pursuing? Once the varying techniques have been learnt, day-to-day living can be very normal, enabling you to make the most of every moment with your baby.

WHAT HELP IS THERE?

If you live in the United Kingdom, your baby will be eligible for the DSS benefit - Disability Living Allowance (DLA). This helps towards the cost of extra care your baby may need and is quite a substantial amount each week, but cannot be backdated. The Special Rules apply to babies with severe SMA, which means there is no 'waiting or qualifying period'. Application forms are available from main post offices. The DLA Helpline is open: -

> 8.30 - 6.00 Monday - Friday, and 9.00 - 1.00 on a Saturday on 0800 882 200 or write to: - DSS, PO Box 50, Heywood, Lancashire, L10 2GF.

If you have any trouble filling in the form, do not hesitate to contact either your JTSMA Contact Family, or The Jennifer Trust who can help advise how to complete the form. The Trust can also write supporting letters when necessary.

Listed below are details of different services that may be available in your area. Health Authorities vary enormously and you may not have access to all. Ask your GP, Consultant or Health Visitor for more information.

Physiotherapists:	Community or Hospital based.
Night Nurses:	If you are on your own, or have difficulty coping during the night.
District Nurses:	May help or assist with tube feeding.
Counselling & Macmillan Nurses:	During this difficult time you may feel the need to talk your feelings through.
Hospices:	There are special children's hospices which many families use for respite care, to give them a break.
Genetic Counselling:	Your GP can make an appointment with your local genetics service. They will discuss the implications for future pregnancies. (see Pre-Natal Diagnosis Section)
JTSMA Contact Family:	If you have contacted the JTSMA they will have informed your local Contact Family about your family. Please, get in touch with them if they haven't already contacted you. They will be able to give you all the support and information you need.
Equipment and Toy Library:	JTSMA have a well stocked toy and equipment library which loans specialist toys, games and equipment suitable for babies with SMA Type I. Contact The Jennifer Trust for more details.
Family Care Officers: (Muscular Dystrophy Campaign)	They can give specialist advice, counselling and support to families who have a child with severe SMA.

Pre-Natal Diagnosis

As if being told that your baby has a terminal illness isn't bad enough, finding out that it could happen again can be just as devastating.

SMA is a recessive disorder, which means that in every pregnancy there is a 3 out of 4 chance of the baby being unaffected, with only a 1 out of 4 chance of it being affected. A pre-natal test will reveal whether SMA affects the unborn child.

Previously, in order for a pre-natal test to be carried out, DNA (extracted from blood samples) was needed from both parents, the affected child and any unaffected children (if there were any). Scientists could then use the DNA to look for 'markers' close to the gene for SMA (on chromosome 5). These markers are harmless variations in the DNA, which lie close to the site of the gene; they were then used for 'tracking' the condition through the family.

Recent advances, however, show that mutations (changes) have been identified in two closely related genes near the SMA gene. This now makes it possible to identify a missing part of the gene in affected infants. Around 95% of all infants and children with SMA, show the deletion in the one gene whereas only about 50% of the more severely affected ones show a deletion in the second gene. Once the deletion has been shown in an affected child, pre-natal diagnosis is now possible on a chorionic villi sample in any future pregnancy by direct assessment as to whether the foetus is carrying the deletion, and is therefore affected or not.

In any future pregnancy, a CVS test (chorionic villus sampling) can be carried out. This involves the removal of chorionic villi for pre-natal testing. The chorionic villi are cells situated on the wall of the uterus (womb), which form the early placenta (afterbirth). They have the same genetic make-up as the unborn baby and can be tested to detect SMA. The test is usually carried out between ten and twelve weeks of the start of pregnancy.

Because the pre-natal test reveals if the unborn baby is affected with SMA, careful thought needs to be taken about what will be done with the information derived from the test. Most couples, having made the decision to undergo pre-natal testing, have usually decided to terminate a pregnancy if their unborn baby is said to be affected with SMA.

In order for all this to take place, though, you need to ensure that your GP or Consultant refers you for genetic counselling. This is support provided by a specialist to people who have genetic conditions in their families.

What is the Jennifer Trist for Spinal Muscular Atrophy (JTSMA)?

The JTSMA is a support group run by parents of children with SMA and adults who have SMA. The main aim of the group is to provide support, information, understanding and friendship to those whose lives are affected by SMA.

A quarterly newsletter, "Holding Hands", is produced, and an annual weekend conference is held offering the opportunity to meet each other and share experiences in a relaxed atmosphere.

The Jennifer Trust is open to anyone who comes into contact with SMA. It now has contact with several hundred families, who, between them, have a wealth of experience to share. Anita Macaulay, whose daughter, Jennifer, had SMA Type I, founded the Jennifer Trust in 1985.

JTSMA has an Area Contact Network of people who provide local support and advice and are experienced in SMA and aware of the wider issues. The Trust will be able to give you the name address and phone number of your local contact family in the UK, who should be able to answer many of your questions, offer advice and just be there whenever you need them. Many areas also produce regional newsletters on a regular basis informing you of all the events taking place in your area. As well as the annual conference, there is a national Christmas Party, Regional Fun Days and Regional Lunches which all provide you with an excuse to get together. These are often invaluable meetings with other families who have similar problems. It can be very

positive to meet other families who have had the same experience as yourselves and have come through it.

What Does the Future Hold?

JTSMA and other organisations throughout the world are funding research into SMA.

Researchers are studying affected children to pinpoint the reason for degeneration of the anterior horn cells, and how this occurs. They are now investigating the anterior horn cells of unaffected people to see how damaged nerves might be repaired. It is possible that this might lead to the development of drugs that will be able to improve the regeneration of nerve cells.

Research is advancing in many areas. JTSMA will keep all members up-to-date with any developments.