THE JENNIFER TRUST
FOR SPINAL MUSCULAR ATROPHY

Factsheet

Adult Onset Spinal Muscular Atrophy

What are the Effects of Adult Onset SMA?

The effects of Adult Onset SMA depend upon the muscles affected - proximal (near the body, e.g. thighs and upper arms) or distal (extremities, e.g. hands and feet).

In the majority of cases the weakness is asymmetrical at the onset, maybe in one leg but not the other, but may progress with time to both sides. Often there is no pain, but tired aching muscles, a feeling of heaviness, numbness, tingling and cramp may be experienced. Fatigue is also common.

SMA is a disease where there is degeneration of the anterior horn cells of the spinal cord resulting in muscular weakness. It is a condition where the motor neurones do not work properly. A distinction should be made between Adult Onset SMA which affects the lower motor neurones and Motor Neurone Disease (Amyotrophic Lateral Sclerosis or ALS) which affects both the upper and lower motor neurones. People are sometimes told they have *a* motor neurone disease and assume they have ALS. Whereas ALS may prove fatal, Adult Onset SMA is not life threatening.

Prognosis of the progression of Adult Onset SMA is difficult because individuals vary enormously.

How is Adult Onset SMA Diagnosed?

The main diagnostic methods are by electromyogram (EMG) and muscle biopsy (taking a tiny sample of muscle). The EMG will show if the nerve signals are diminished, and the biopsy will reveal any reduction in muscle cells. Some people may have MRI and CT scans as well and a range of blood tests. However even with all these tests it is not straightforward to ascertain at which point in the motor unit the damage is occurring.

Is Adult Onset SMA Hereditory?

There are many different inheritance patterns to SMA, so the answer to this question is not as simple as for Type I or II SMA.

In some cases the pattern of inheritance may be autosomal recessive (i.e. both parents must carry the faulty gene) in others it may be autosomal dominant (i.e. it needs only one parent to pass on a faulty gene). Recent research has shown that a small number of individuals with Adult Onset SMA have been shown to have mutations in the Survival Motor Neuron (SMN) gene, in these cases it is believed that Adult Onset SMA is non-hereditory.

There is an X-linked form of Adult Onset SMA known as Bulbo-SMA or Kennedy's Syndrome which is only seen in males. The gene is recessive and is found on the X-chromosome. Women may carry it and pass it on to 50% of their offspring. Men who have it pass it to all of their daughters and **none** of their sons. Daughters who inherit the gene are carriers, and sons who inherit it will show the symptoms.

What Treatment is There for Adult Onset SMA?

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

Maintaining movement and mobility can be improved by following exercises guided by qualified practitioners. The availability of appropriate services varies greatly depending on where you live. A few people have the services of a neuromuscular centre where specially trained physiotherapists can set up a programme suitable for the individual. Where these services are not available your GP can recommend you to a physiotherapist; or you may find that qualified complementary therapists can aid in devising appropriate gentle exercise programmes.

The aims of an exercise programme are to:-

1. Maintain safe ambulation for as long as possible.
2. Maintain a good joint range and prevent asymmetry and scoliosis (curvature of the spine)
3. Encourage good respiratory function.

Assessments

A thorough initial assessment is essential to take a history and get a picture of joint range, muscle power and functional ability. Advice may then be given and a treatment programme discussed. Re-evaluation assessments are necessary at regular intervals.

Hands on Treatment

Stretches. Range of movement at a joint may be limited by tendon shortening or tightness. If this is one sided it can lead to postural asymmetry (lop-sidedness). Reducing this can be helped by regular stretching.

The most affected muscles are:-

Achilles Tendon (Heel),

Hamstrings (Back of thigh),

Hip Flexors (Front of hip and thigh),

Hip Abductors (Outside of thigh).

Stretches should not be painful. Tension or pulling may be felt but not pain. Arms should be regularly checked to maintain a good range of movement, particularly in the shoulders when muscle weakness prevents full elevation of the arms. The finger flexors (the muscles used for gripping) also tend to tighten as they are used more than the extensors which straighten the fingers.

Standing

Standing is a very important part of a physical management programme for many individuals with neuromuscular conditions, especially once they lose independent ambulation. Contractures, in a lot of cases, creep in and can make activities of daily living much more difficultand erode independence. For instance dressing, bathing, showering, and transfers from and to wheelchairs become affected.

Standing in a standing frame has benefits *even when independent ambulation is still possible*. It encourages equal weight bearing through both legs. Corrective straps stretch feet, knees & hips and reduce asymmetry and tilting of the pelvis which can lead to scoliosis. One of the main aims of a physiotherapy programme is to maintain independent ambulation for as long as possible. Often it is not muscle weakness but muscle contracture which causes the inability to walk.

Once an individual is dependent on a wheelchair, standing is even more important as contractures quickly appear.

Regular standing also promotes normal bodily functions such as kidney drainage & reducing calcium loss in bones. These benefits are all felt even if the individual is unable to stand completely upright.

Respiratory Function

Breathing exercises and effective coughing should be taught at an early stage. This makes it easier in the future when chest infections or breathing difficulties occur. Respiratory function can be improved with regular hydrotherapy.

Swimming is a good general exercise that can help to maintain muscle condition without over exercising or damaging the muscles. It also allows a greater degree of physical fredom than may be experienced elsewhere.

What Aids and Equipment will be Helpful?

As weakness increases it may be necessary to use various aids such as a walking stick, a raised toilet seat and a bath hoist. Social Services departments supply such items, and an Occupational Therapist should be able to advise on suitable equipment. Some people will eventually need to use a wheelchair. Wheelchairs can be provided by the NHS in the UK or may be bought privately. Referrals for an NHS wheelchair are made by a GP or Social Services.

Wheelchairs

A lightweight wheelchair, when walking gets difficult and trips out are limited by the fear of falling and being unable to manage steps etc., can improve the quality of life.

The quality of wheelchair seating is very important from the start. The key elements to good seating are, a firm base cushion which will encourage a level pelvis, a firm back canvas which will encourage an erect posture, arm rests at the correct height and foot rests at the correct level so there is a 90° angle at the hips, knees and feet.

It is important that the chair is as light as possible so it is easy to push and propel.

The position of the wheels should be adjustable, and should initially be set by an expert. Even a light weight chair can be difficult to use if the wheels are wrongly set for the length of the user's arms and their strength, weight and balance. This setting will probably need changing after a new user becomes proficient, but this is generally easy to do.

A wheelchair is not a last resort only to be considered when mobility becomes too difficult. It *is* a tool to enable the user to get the most out of life. If it is going to be necessary to have one, get it in good time so that skills can be acquired before it becomes essential to use it all the time.

How will Adult Onset SMA Affect Employment?

Depending upon the type of their employment, people with Adult Onset SMA may be able to continue working for many years. It may be necessary for some adaptations at the workplace. Money is available to employers to make such provision in the UK through the "Access to Work" scheme, details of which are available from the JTSMA Office

What Benefits are Available in the UK?

Those who are only able to work part-time or whose earnings fall below a certain level may be eligible for Disability Working Allowance. For those unable to continue working, Incapacity Benefit or Severe Disablement Allowance is available.

In addition to all these benefits there is Disability Living Allowance (DLA) which has two components - mobility and personal care. The mobility component is paid to people who have difficulty walking and in using public transport; the personal care component is paid to those who need help with such tasks as bathing and dressing. DLA application forms are available from the Department of Social Security.

What is the Jennifer Trust 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. The Jennifer Trust was founded in 1985 by Anita Macaulay, whose daughter, Jennifer, had severe SMA.

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 Other Support Services can Help?

The Muscular Dystrophy Group provides some information and assistance for those affected with SMA and their families. The MD Group has a network of Family Care Officers attached to some neuromuscular clinics who give help and advice.

General Practitioners (GPs) and hospitals should also be able to offer you help, however SMA is not so common that all GPs or hospitals know much about it. They may need to refer to specialists in other hospitals. If you are not getting as much information or help as you want, keep asking.

Where Can I Get More Information?

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