Spinal Muscular Atrophy Type 4

This information sheet explains the cause, effects and management of Spinal Muscular Atrophy (SMA) Type 4 which is an adult onset form of SMA. It includes sources of further information and support. It is for individuals who have been diagnosed with SMA Type 4 and their families. It may also be useful for healthcare and other professionals.

The glossary at the end further explains the words that appear in bold.

There are several different types of adult onset SMA. This information sheet focuses on SMA Type 4 but some of the practical information on mobility, equipment, adaptations, holidays, employment and financial and emotional support may still be helpful to individuals affected by one of the other types of adult onset SMA.

SMA is a complex condition that can vary in severity - every person with SMA is different. Your medical team will be able to provide you with information and advice specific to you.

What is SMA Type 4 and how is it caused?

Usually, electrical signals from our brain are sent down our spinal cord along our nerve cells and through to our muscles. This makes it possible for us to consciously contract our muscles and make them move.

SMA affects a particular set of nerve cells called the lower motor neurons which run from the spinal cord out to our muscles. For our lower motor neurons to be healthy, we need to produce an important protein called the Survival Motor Neuron (SMN) protein. Our ability to do this is controlled by a gene called Survival Motor Neuron 1 (SMN1).

If an individual has two faulty SMN1 genes then they are only able to produce very low amounts of the SMN protein. This causes the lower motor neurons in their spinal cord to deteriorate. Messages from their spinal cord do not efficiently get through to their muscles,
which makes movement difficult. Their muscles waste due to lack of use and this is known as muscular **atrophy**.

In addition to **SMN1**, we possess a second gene that is able to produce some functional SMN protein. This gene is almost identical to **SMN1** and is called the **SMN2** gene. However, **SMN2** only makes a small fraction of functional protein (about 10%).

For more information on the inheritance of SMA and how **SMN2** is linked to the severity of an individual’s SMA please see ‘The Genetics of Spinal Muscular Atrophy’: www.smasupportuk.org.uk/the-genetics-of-sma

In SMA Type 4, the muscles that are particularly affected are those we use to walk and move our arms. In other more severe forms of childhood onset SMA, breathing and swallowing muscles are also affected. This is rarely the case with SMA Type 4.

The symptoms of SMA Type 4 can include: tired, aching muscles; a feeling of heaviness; numbness; cramp; a slight shaking of the fingers and hands. **Fatigue** is also common. Over time, increased muscle weakness can impact on daily living activities such as walking, dressing and bathing.

SMA Type 4 progresses steadily and slowly over time. Muscle weakness usually progresses gradually and, as mentioned above, SMA Type 4 rarely affects swallowing or breathing. It does not affect intelligence and life expectancy is normal.

It is important not to confuse SMA Type 4, which affects the lower **motor neurons**, and Motor Neurone Disease (MND) - also known as Amyotrophic Lateral Sclerosis (ALS) - which affects both the upper and lower motor neurons. SMA is classed as a motor neuron disease but it is not MND / ALS. Whereas MND / ALS is almost always life-threatening, SMA Type 4 is not.

**Diagnosis**

SMA Type 4 is most often diagnosed in early adulthood. (Individuals with other forms of Adult Onset SMA might not show any symptoms until later in life.)

Getting a **diagnosis** of SMA can take time. This can be because tests may not conclusively prove that an individual has SMA and therefore other **neuromuscular** conditions will need to be ruled out. Ask for the testing process to be explained to you so that you understand what tests are being carried out, for which conditions, and why.

A doctor will diagnose SMA after taking a medical history, doing a physical examination and taking a blood sample for **DNA** testing. Other tests may include an **electromyogram** (EMG) and a **muscle biopsy**. The EMG will show if the nerve supply is diminished and the biopsy will reveal any reduction in muscle cells.
Some people may also have MRI (Magnetic Resonance Imaging) and CT (Computerised Tomography) scans as well as a range of blood tests. Even with all these tests it is not always straightforward to determine the particular type of neuromuscular condition.

Confirmation of a specific diagnosis of SMA Type 4 can only be made following DNA testing.

Waiting for appointments, test results and a diagnosis can be very stressful. If you are concerned about your symptoms, have had some tests but have not been referred to a consultant neurologist, you might want to request a referral from your General Practitioner (G.P.).

Receiving a diagnosis of SMA Type 4 may have a significant impact on you and your family both physically and emotionally. You might find it helpful to seek support from a counsellor or clinical psychologist and you can ask your G.P. for a referral.

**Is SMA Type 4 hereditary?**

If an individual has one faulty copy and one healthy copy of the *SMN1* gene then they are a carrier of SMA and do not show any symptoms. If both parents are carriers then each of their children will have a 1 in 4 (25%) chance of having SMA. This is known as **autosomal recessive inheritance**. SMA Type 4 occurs because the affected individual receives two faulty *SMN1* genes, one from each parent.

For more information on ‘The Genetics of Spinal Muscular Atrophy’ please see: [www.smasupportuk.org.uk/the-genetics-of-sma](http://www.smasupportuk.org.uk/the-genetics-of-sma)

For the purpose of the diagram below, a ‘non-carrier’ means a person who does not carry the faulty gene and does not have SMA.

**Autosomal recessive inheritance: both parents are carriers**

If two carriers have a child together, the chances are as follows:

- Child does not have SMA and is not a carrier: 1 in 4 chance (25%)
- Child does not have SMA but is a carrier: 2 in 4 chance (50%)
- Child has SMA: 1 in 4 chance (25%)
Please remember that if you have a rare form of adult onset SMA, the diagram may not apply to you. If this is the case, your medical team will be able to give you information about your particular genetic situation. You can also read SMA Support UK’s leaflet ‘The Genetics of Some Rarer Forms of SMA’: [www.smasupportuk.org.uk/the-genetics-of-some-rarer-forms-of-spinal-muscular-atrophy](http://www.smasupportuk.org.uk/the-genetics-of-some-rarer-forms-of-spinal-muscular-atrophy)

If you have any queries about the genetics of your SMA please speak to your consultant.

**Genetic counselling**

If a genetic cause has been found for your SMA you should be offered a referral for genetic counselling. You can also request a referral from your General Practitioner (G.P.).

Genetic counselling takes place with a healthcare professional who has expert training in genetics. They will help you to understand how SMA is passed on and what the chances are of other people in your family being affected. Genetic counselling also provides you or other members of your family with the opportunity to discuss your choices for any future pregnancies. You will be able to go back to your genetic counsellor at a later date if you have more questions.


**Is there a treatment or cure?**

Although there is currently no cure for SMA, this does not mean that nothing can be done. Symptoms can be managed so that individuals with SMA Type 4 can achieve their maximum mobility, independence and quality of life.

If you are already on, or you have been newly prescribed, medication for your health (separate to your SMA), it can be useful to check with your consultant how these might interact with your SMA.

To help manage your SMA symptoms you may have contact with a number of different healthcare professionals. These could include a neurologist, a physiotherapist, an occupational therapist and a dietitian. You can find out more about how these people can help in the information sheet ‘Who’s Who of Professionals’: [www.smasupportuk.org.uk/whos-who-of-professionals](http://www.smasupportuk.org.uk/whos-who-of-professionals)

**Physiotherapy**
Movement and mobility can be maintained by following exercises guided by qualified practitioners. The availability of appropriate services varies greatly depending on where you live. In some areas people have access to 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 General Practitioner (G.P.) can refer you to a physiotherapist.

A thorough initial assessment is essential to take a medical history and get a picture of your joint range, muscle power and functional ability. Advice may then be given and / or a treatment programme discussed. Reassessments are necessary at regular intervals so discuss with your physiotherapist how this will be arranged.

One of the main aims of a physiotherapy programme is to maintain or improve walking (ambulation) and independence. The inability to walk, which is rare in SMA Type 4, can be due to a combination of muscle weakness and contractures. This is why it is important to do regular stretches as part of your exercise programme.

Your physiotherapist will design an individualised exercise programme that aims to help achieve the main goal of maintaining or improving your ambulation and independence. The exercise programme will have different elements to it:

- **Range of movement**

  Range of movement at a joint may be limited by tendon shortening / tightness. If this is one sided it can lead to poor posture, cause discomfort, and in later stages can limit your ability to function day-to-day. Daily stretching can help this, with the aim of preserving symmetry and flexibility. The most affected muscles are: tendon / achilles (heel), hamstrings (back of thigh), hip flexors (front of hip and thigh) and hip abductors (outside of thigh). You may also experience muscle weakness / tightness in your shoulders, arms and fingers.

  When stretching you may feel tension or pulling, but stretches should not be painful. If you do have any pain when stretching do talk to your doctor or physiotherapist.

  You may enjoy doing your stretching exercises in the bath, a swimming pool, or a hydrotherapy pool as warm water can make muscles easier to stretch. Talk to your physiotherapist or occupational therapist about doing this safely.

- **Posture and standing**

  Maintaining a good posture is important for the body / muscles to work effectively and this applies to both sitting and standing. Posture exercises often involve working on core stability (the ability of the muscles in the abdomen area to help maintain good posture and balance).

  Standing is also a very important part of a physical management programme for many individuals with neuromuscular conditions, even if you are unable to walk independently. Your physiotherapist may suggest you try a standing frame which encourages equal weight
bearing through both legs and can help prevent and reduce contractures. Straps are used to help stretch feet, knees and hips and reduce asymmetry and tilting of the pelvis, which can lead to scoliosis.

Regular standing also promotes normal bodily functions, for example kidney drainage, and reduces calcium loss in bones.

- **Muscle strength and control**

  Exercises to help maintain or improve your muscle strength and control.

- **Balance**

  Exercises to help you with your balance in order to prevent falls.

- **Fitness**

  Inactivity can lead to ‘deconditioning’ - the ‘use it or lose it’ principle. Inactivity can further limit mobility which is why fitness is important. See more detail on exercise below.

- **Respiratory function**

  SMA Type 4 does not normally cause problems with breathing or swallowing but it can be useful to learn breathing exercises and effective coughing for your general health and well-being. This will help if you have a chest infection or breathing difficulties. Your physiotherapist will be able to provide you with advice on this.

  Respiratory function can also be improved by exercising in a hydrotherapy pool or by going swimming. Swimming is a good general exercise that can help to maintain muscle condition without over-exercising or damaging them. Your physiotherapist will be able to provide you with further advice on this and also provide you with information on your nearest accessible swimming or, if there is one available, hydrotherapy pool.

**Exercise**

It is important to maintain cardiovascular fitness and stamina. Physical activity is also good for maintaining psychological health and well-being and it is important even if you have limited mobility. You might want to try swimming or adaptive sports for example. Check with your doctor and physiotherapist first what types and intensity of exercise might be suitable for you and they will also be able to tell you about local centres and organisations that you could try.
‘Exercise advice for adults with muscle-wasting conditions’ is a leaflet available from Muscular Dystrophy UK. You can request a copy by phoning 0800 652 6352 or you can download it from the website: www.musculardystrophyuk.org/app/uploads/2015/05/Exercise-advice-for-adults.pdf

‘Doing Sport Differently’ is a guide available from Disability Rights UK. You can request a copy by phoning 0207 250 8181 or you can download it from the website: www.disabilityrightsuk.org/how-we-can-help/publications/doing-life-differently-series/doing-sport-differently

**Nutrition**

A healthy diet is important for everyone. If needed, your General Practitioner (G.P.) or consultant will be able to refer you to a dietitian to provide you with advice and support on eating and nutrition.

Reduced mobility and lack of regular exercise can be a factor in some individuals becoming overweight. If this happens the extra weight can increase the stress on muscles, bones and joints, making physical activity even more difficult. A dietitian will be able to provide advice on a healthy diet that will suit individual needs.

SMA Type 4 rarely causes difficulty with chewing and swallowing but your medical team will provide you with advice and support with this if necessary.

**Pain and fatigue management**

Individuals with SMA Type 4 sometimes report experiencing pain and **fatigue**. This can happen because you can overuse some muscles in order to try and compensate for the weaker muscles. A physiotherapist may be able to help you to manage this and your General Practitioner (G.P.) might be able to prescribe you suitable medication for pain relief.

It is important to sit and lie comfortably so that your muscles and joints can relax. You might find it helpful to use pillows to support certain muscles. Your physiotherapist will be able to advise you on how to help your posture in order to aid muscle relaxation.

A hot water bottle, heat pad, or a microwaveable heat bag can also help to relax muscles and a cold pack can also be used for pain relief.

Regular exercise may help but please speak to your physiotherapist for advice on what will work best for you.

Fatigue can be managed by pacing yourself and preventing your body from becoming overly tired. Regular exercise will help you to optimise your physical capacities and to understand
your limits better. Your physiotherapist will be able to advise you on strategies to help with fatigue.

A number of individuals affected by SMA Type 4 have told SMA Support UK that they find meditation and mindfulness helpful in managing pain and fatigue. There is now a wide range of information available on these subjects both on the internet and in libraries.

**Equipment**

As muscle weakness increases it may be necessary for you to use various aids, for example a walking stick, raised toilet seat, bath hoist, handrails. Social Services departments may supply such items and your occupational therapist and / or physiotherapist will be able to advise you on equipment suitable to your individual needs.

A very small number of people who have SMA Type 4 may eventually need to use a wheelchair. When walking becomes difficult, a wheelchair can improve your quality of life by reducing fatigue and any fear of falling. Wheelchairs are provided by NHS wheelchair services or may be bought privately. Referrals for an NHS wheelchair can be made by your General Practitioner (G.P.), consultant or your local Social Services. Getting the right wheelchair is important so make sure you get a wheelchair assessment, even if you decide to buy privately. Advice on this can be provided by your physiotherapist and / or occupational therapist.

**Home adaptations**

You may need to make adaptations to your home or consider moving to a more accessible property. You will need to think ahead about long term needs and allow plenty of time for planning and implementing any changes. If you need adaptations to your home you can refer yourself, or request that your occupational therapist or physiotherapist refer you, to the local authority occupational therapy service.

**Care and Repair England** is a charity working to improve the housing and living conditions of people with disabilities and older people. They can provide advice on local Care and Repair agencies and provide information on house adaptations: [www.careandrepair-england.org.uk](http://www.careandrepair-england.org.uk) or phone 0115 950 6500.

In Scotland you can contact **Care and Repair Scotland**: [www.careandrepairscotland.co.uk](http://www.careandrepairscotland.co.uk) or phone 0141 221 9879.

In Wales contact **Care and Repair Cymru**: [www.careandrepair.org.uk](http://www.careandrepair.org.uk) or phone 02920 674 830.

In England, Northern Ireland and Wales, depending on your financial circumstances, you may be eligible to apply for a disabled facilities grant (DFG) to help with the cost of adapting your home. This grant is available both to home owners and tenants and will not affect any benefits
that you might be receiving. For more information on DFGs in England and Wales see: [www.gov.uk/disabled-facilities-grants/overview](http://www.gov.uk/disabled-facilities-grants/overview) For more information on DFGs in Northern Ireland see: [www.nihe.gov.uk](http://www.nihe.gov.uk) For information on housing adaptations if you live in Scotland please visit: [www.gov.scot/Topics/Built-Environment/Housing/access/adaptations](http://www.gov.scot/Topics/Built-Environment/Housing/access/adaptations)

**Mobility**

- **Blue Badge Scheme**

If you have a car you might want to find out if you are eligible to apply for a Blue Badge. Local authorities administer the Blue Badge system which entitles people with severe mobility problems to parking concessions. The concessions may apply to street parking and might include free use of parking meters and pay-and-display bays. Some areas might also offer exemption from toll charges but you will need to check this with your local authority. For more information and to find out how to apply for a Blue Badge visit the GOV.UK site: [www.gov.uk/blue-badge-scheme-information-council](http://www.gov.uk/blue-badge-scheme-information-council)

- **Road tax**

If you receive the enhanced mobility component of Personal Independent Payment (PIP), or the higher rate mobility component of Disability Living Allowance (DLA), you are also entitled to free road tax. For more information on this please see: [www.nhs.uk/Conditions/social-care-and-support-guide/Pages/transport-and-mobility-issues.aspx](http://www.nhs.uk/Conditions/social-care-and-support-guide/Pages/transport-and-mobility-issues.aspx)

- **Cars and adaptations**

If you might need adaptations to enable you to continue to drive safely you can arrange an assessment. Suitable adaptations can then be fitted privately or through the Motability Scheme. The following organisations provide information on getting an assessment:

  - **Driving Mobility** - give practical and independent advice to disabled drivers. They will assess an individual's ability to drive, provide advice about suitable vehicles and any adaptations that may be needed. There are centres across the United Kingdom (UK). To find one please see: [www.drivingmobility.org.uk](http://www.drivingmobility.org.uk) or phone 0800 559 3636.

  - **Motability** - provide assessments if the individual is in receipt of the enhanced rate of the mobility component of Personal Independence Payment (PIP), or the higher rate of the mobility component of Disability Living Allowance (DLA): [www.motability.co.uk](http://www.motability.co.uk) or phone 0300 456 4566.

  - In Northern Ireland, **Disability Action** can carry out assessments and provide information on the process of learning to drive and help set up appointments with Motability. For more information please see: [www.disabilityaction.org/services-and-projects/driving/](http://www.disabilityaction.org/services-and-projects/driving/)
If you are a wheelchair user your occupational therapist can provide you with advice on whether your wheelchair is suitable to drive a car from and whether any adaptations will need to be made to your chair.

If you receive the enhanced mobility component of Personal Independent Payment (PIP), or the higher rate mobility component of Disability Living Allowance (DLA), (see the section on financial support for more details) the Motability Scheme will enable you to lease a new car, powered wheelchair or scooter for three years, or five years for a Wheelchair Accessible Vehicle (WAV). For more information please see: www.motability.co.uk or phone 0300 456 4566.

If you need a wheelchair accessible vehicle (WAV) these can be bought privately or leased through the Motability Scheme. For more information on WAVs please see:

- **RICA** (Research Institute for Consumer Affairs) provide guides to purchasing a WAV which can be found at: www.rica.org.uk/content/wheelchair-accessible-vehicles-wavs or you can phone RICA on: 0207 427 2460.

- **Driving Mobility** offer information and advice about accessible vehicles for people with disabilities: www.drivingmobility.org.uk or phone: 0800 559 3636.

- **The Mobility Roadshow** is a free roadshow that has a wide range of adapted cars and mobility products to try and see. For more information see: www.mobilityroadshow.co.uk

  - **Public transport**

If you use public transport, many operators offer concessions for people with disabilities. Contact the relevant transport provider for more details. A guide to accessible public transport is available from RICA: www.rica.org.uk/content/accessible-public-transport or you can phone and request a copy on: 0207 427 2460.

If you use the train you can book free ‘Passenger Assistance’ through National Rail Enquiries www.nationalrail.co.uk You can also book through the train operating company, in person at a local station, online at: www.disabledpersons-railcard.co.uk/travel-assistance or by phoning National Rail Enquiries on: 03457 48 49 50. For information on Disabled Persons Railcards please see: www.disabledpersons-railcard.co.uk or phone 0345 605 0525.

**Holidays**

As your mobility changes, holidays may need more considered planning in order to avoid frustrations and to ensure that the travel and accommodation booked meets your needs. The
term ‘accessible’ can have many interpretations so ask specific questions about your needs before booking.

**Tourism for All UK (TFA)** is a charity website that has been developed as a one stop shop to provide information on accessible accommodation, holidays and trips in the UK and overseas. Click on the TFA Directory for a list of services such as holiday charities, attractions and accommodation, care services, short breaks, etc. They also list travel insurance companies for people with disabilities: [www.tourismforall.org.uk](http://www.tourismforall.org.uk) or phone 0845 124 9971.

**What other help is available?**

- **Neuromuscular centres**

There are two neuromuscular centres in the UK. Both are charities and both offer physiotherapy, complementary therapies, support and advice to adults who have a neuromuscular condition. The services are free to the user but you will need to be referred by your General Practitioner (G.P.) or consultant. More information is available directly from the centres:

**The Neuromuscular Centre Winsford**
The Neuromuscular Centre, Woodford Lane West, Winsford, Cheshire, CW7 4EH.
The centre covers mainly the North of England and North Wales.
01606 860 911.
[www.nmcentre.com](http://www.nmcentre.com)

**The Neuromuscular Centre Midlands**
Hereward College, Bramston Crescent, Coventry, West Midlands, CV4 9SW.
The centre covers the Midlands region.
02476 100 770.
[www.nmc-midlands.co.uk](http://www.nmc-midlands.co.uk)

- **Support services**

A diagnosis of SMA Type 4 and learning to live with the condition may have a significant impact on you and your family. It is important that you have emotional support and plenty of time to talk and ask questions. This can be with your consultant, your local General Practitioner (G.P.), a social worker, psychologist or a counsellor. You might also want information, advice and support on topics such as mobility, equipment and financial assistance. You can find out more by talking to your healthcare team, Spinal Muscular Atrophy Support UK (SMA Support UK) and the other people and agencies listed in this leaflet.

**SMA Support UK** provides information and support, by telephone and email, to individuals and families affected by SMA in the UK. Our Outreach Workers are able to visit you at home and can discuss with you the health, social, financial and care support you may be entitled to.
We can also tell you about opportunities for having contact with others who have personal experience of living with SMA. Information about these services is available on our website: www.smasupportuk.org.uk or please phone us on 01789 267 520 or email: supportservices@smasupportuk.org.uk

Muscular Dystrophy UK also provides information, support and advocacy services, including grants towards specialist equipment, for people affected by a range of neuromuscular conditions. Their website is: www.musculardystrophyuk.org or you can phone them on 0800 652 6352 or e-mail: info@musculardystrophyuk.org

Regional care advisors, and sometimes neuromuscular nurse specialists, are attached to NHS neuromuscular clinics in various regions of the UK. They provide support and information to children and adults with muscle diseases and their families. They link up with other professionals and services so that people receive the local health and social support they need. Regional care advisors’ contact details are available on Muscular Dystrophy UK’s website: www.musculardystrophyuk.org/get-the-right-care-and-support/people-and-places-to-help-you/care-advisors/

- **Employment**

If you are working, depending on your type of employment, you may be able to continue to do so following your diagnosis. It may however be necessary for some adaptations to be made at your workplace. Financial assistance is available to employers to make such provision through the Access to Work scheme, details of which are available from the website GOV.UK: www.gov.uk/access-to-work or from your local Job Centre Plus.

- **Financial support**

For those unable to continue working, Employment and Support Allowance (ESA) is available. For more information please see: www.gov.uk/employment-support-allowance/overview

If you are working but your earnings fall below a certain level you may be eligible for the disability element of Working Tax Credit. More information is available at: www.gov.uk/working-tax-credit/overview

Whether working or not, if you are aged 16-64 and a new claimant, you may be entitled to Personal Independence Payment (PIP). (PIP has replaced Disability Living Allowance - DLA. If you are under 65 and already receiving DLA you will be reassessed for PIP at some point. If you were aged 65 or over on the 8th April 2013 and already receiving DLA then this will continue.) PIP has two components - mobility and daily living. More information on PIP is available from: www.gov.uk/pip/overview

If you are aged 65 or over, and are not already claiming either PIP or DLA and you need help with your personal care, then you might be able to claim Attendance Allowance. More information on this is available from: www.gov.uk/attendance-allowance
For further information on benefits visit the GOV.UK website www.gov.uk and look at the section ‘Benefits’ and ‘Carers and Disability Benefits’. The Department of Work and Pensions (DWP) can be contacted on: 0345 608 8545.

To find out more about other financial help you may be entitled to, including help with travel and with your home, please visit the GOV.UK website: www.gov.uk/financial-help-disabled

Disability Rights UK publishes free factsheets on a range of benefits and the ‘Disability Rights Handbook’ annually. For further information visit: www.disabilityrightsuk.org or phone 0207 250 8181.

Turn2Us is a charity which helps people access money available to them through welfare benefits, grants, and other help. They can be contacted on 0808 802 2000 or through their website: www.turn2us.org.uk

Your local Citizens Advice Bureau might also be able to provide you with financial advice and help with completing benefit applications. Visit your local office or phone 03444 111 444. You can also access information via the website: www.citizensadvice.org.uk

Age UK provide a range of services for older people including financial advice and information. The age at which you can access Age UK services can vary between local branches, but generally they are for the over 60s. You can phone Age UK’s free national helpline: 0800 169 2081 or visit their website: www.ageuk.org.uk

Your neuromuscular care advisor, social worker, or local welfare rights advisor may also be able to help you with applications for financial benefits.

- Emotional support

If you feel that you would benefit from being able to talk to a professional counsellor about how you are feeling, you can ask your General Practitioner (G.P.) to refer you to a counselling service. Waiting times for an appointment vary.

Some employers offer employee assistance programmes that include access to a counselling service.

If you attend a rehabilitation centre, some offer counselling services.

Some people consider private counselling services. The following organisations can help you to find a private counsellor in your local area:

British Association for Counselling and Psychotherapy (BACP)
Provide guidance on the counselling process and how to find a suitable counsellor. 01455 883 300.
www.bacp.co.uk
Counselling Directory
Online information about different types of counselling, useful articles, events and a directory to search for qualified counsellors.
www.counselling-directory.org.uk

What does the future hold?

As mentioned earlier, SMA Type 4 usually progresses steadily and slowly over time. However, every person is different and it can be frustrating as there are no certainties about what the future holds for any individual or how quickly their mobility will decrease.

In general terms, research into SMA is ongoing and you can find out more on our research pages at: www.smasupportuk.org.uk/research

As new treatments for SMA are being developed they need to be tested in clinical trials but because SMA is a rare condition it can sometimes take years to find enough patients for a clinical trial. The UK SMA Patient Registry aims to speed up this process. It is a database of genetic and clinical information about people affected by SMA who have a confirmed mutation in the SMN1 gene. The Registry also helps specialists gain more knowledge about the condition and the number of people affected by SMA. This information helps to develop and improve worldwide standards of care for people with SMA. You can find out more by looking at their website: www.treat-nmd.org.uk/registry e-mailing: registry@treat-nmd.org.uk or phoning: 0191 241 8605.

Further Resources

SMA Support UK information:

Copies of SMA Support UK leaflets can be requested on 01789 267 520 or downloaded from the website: www.smasupportuk.org.uk/about-sma
**Glossary**

**Amino acid**
The individual building blocks of **proteins**. There are 20 different amino acids that are naturally incorporated into proteins. The specific order of the amino acids determines the structure and function of a protein.

**Amniocentesis**
The removal of a sample of **amniotic fluid** (the fluid around an unborn baby) for **prenatal testing**. Cells in the fluid can be tested for certain **genetic disorders**.

**Amniotic fluid**
The fluid surrounding a **foetus** in the womb.

**Anterior**
Front or forward.

**Anterior Horn**
The front part of the **spinal cord** where the **cell** bodies of the lower **motor neurons** are located. Long, slender projections of the motor neurons called **axons** migrate out from the anterior horn in large bundles of nerves in order to reach muscles.

**Anterior Horn Cell**
The **nerve cells** that make up the **anterior horn** of the **spinal cord**. Also known as lower **motor neurons**, these **cells** are the main cell type affected in SMA.

**Atrophy**
The wasting or shrinkage of a part of the body. SMA is called Spinal Muscular Atrophy because the lower motor neurons within the spinal cord degenerate, which leads to the wasting of skeletal muscles.

**Autosomal recessive inheritance**

When a genetic disorder is recessive, two faulty copies of a gene, one from each parent, must come together for the disease to occur. If a person has only one faulty copy, they do not usually have the symptoms of the disease, but are known as carriers because they can pass on the faulty gene to their children. A disease is autosomal when the faulty gene is found on one of the autosomes. SMA is usually an autosomal recessive condition.

**Autosome**

Any of the 22 pairs of chromosomes found in the human body that are not involved in the determination of sex. They are identical in both males and females. Each pair of autosomes (one from the father, one from the mother) contain genes for the same traits (characteristics).

**Axon**

The long, slender main projections of a nerve cell. Axons carry electrical impulses away from the cell body (where the nucleus is) to its target, such as muscles.

**Carrier**

This term relates to autosomal recessive inheritance and X-linked recessive inheritance patterns. A person who has both a faulty copy and a healthy copy of a gene is a carrier. Carriers usually have no symptoms due to the healthy copy of the gene, but they may pass on a condition to their children.

**Cell**

The basic building block of all known living organisms. Cells come in many different forms such as motor neurons (a type of nerve cell), keratinocytes (main cell type of the skin), or erythrocytes (red blood cells).

**Central nervous system (CNS)**

The central nervous system consists of the brain and the spinal cord. The CNS is connected to other tissues and organs in the body, such as skeletal muscles, by the peripheral nervous system (PNS).

**Chorionic villus sampling (CVS)**

CVS is a way to test if an unborn baby has SMA. A sample of chorionic villous cells (placental tissue) is removed using a needle. This is usually done between the eleventh and fourteenth week of a pregnancy. The cells can then be genetically tested for SMA.

**Chromosomes**

Chromosomes are compact bundles of DNA. Humans have 46 chromosomes in each cell (with a few exceptions, including sperm and egg cells). They inherit 23 from their mother and 23 from their father to make 23 pairs.
**Clinical**  
The observation and treatment of patients, rather than laboratory studies that do not directly involve patients.

**Clinical trial**  
A trial done on humans, usually to test a treatment or intervention, or to find out more about a disease.

**Contracture**  
A tightness in the connective tissue and tendons around a joint that results from weakness and inability to move a joint through its full range of motion.

**Diagnosis**  
Identifying a disease from its signs and symptoms or from its genetic cause. A clinical diagnosis is given when a doctor sees enough signs or symptoms to be confident that a person has the disease in question. In genetic disorders, a genetic diagnosis is given when a genetic test has been performed and the fault in the gene that is known to cause the disease is found. Doctors who are experts in SMA can usually diagnose the condition with a high degree of accuracy from the clinical signs and symptoms alone. However, genetic tests are usually recommended for all genetic disorders to increase certainty, to make sure any treatment is correctly targeted and to enable the family to have prenatal testing in future pregnancies if they wish.

**DNA (Deoxyribonucleic acid)**  
DNA is the molecule that contains the genetic instruction manual to build all known organisms. DNA is often compared to a set of blueprints, a recipe, or a code, since it contains the instructions needed to construct other components of cells, such as proteins.

**Electromyogram (EMG)**  
A test that assesses the electrical activity of the muscles and the nerves controlling the muscles. It is used to help diagnose neuromuscular disorders. There are two kinds of EMG: intramuscular and surface. An intramuscular EMG involves inserting a needle electrode, or a needle containing two fine-wire electrodes, through the skin into the muscle. A surface EMG involves placing an electrode on the surface of the skin.

**Embryo**  
The name given to the developmental stage from fertilised egg up until about eight weeks of pregnancy when the embryo becomes a foetus.

**Fatigue**  
Great tiredness. A common symptom of neuromuscular conditions.

**Foetus (fetus)**  
The term used for an unborn baby after the eighth week of development until birth.
Gene
A section of DNA that carries the information to produce a specific protein. Genes are the unit of heredity that are passed from one generation to the next. We usually possess two copies of each gene, one inherited from each of our parents. When genes are altered through mutation, this can affect the structure and function of the proteins that they produce, leading to disease.

Genetic counselling
Information and support provided by a genetic specialist to people who have genetic disorders in their families or are concerned about a genetically transmitted condition. Genetic counselling helps families understand things like how the condition is passed on, what the chances are of children being affected, and which other family members may be at risk of carrying the affected gene. It also helps affected teenagers / young adults to understand their future choices.

Genetic disorders
Conditions resulting from alterations to an individual’s genes. Genetic disorders can be caused by defects in one or more genes, or whole chromosomes.

Genetic testing
The examination of an individual’s genes to identify any faults that could cause a genetic disorder.

Genetics
The study of genes and inheritance.

Heredity
The passing of traits (characteristics) through the inheritance of genes from one generation to the next.

Inheritance
The process by which an individual acquires traits (characteristics) from his or her parents.

Magnetic resonance imaging (MRI)
A non-invasive body imaging procedure that uses powerful magnets and radio waves to construct pictures of the internal structures of the body.

Molecule
Two or more atoms chemically bonded together. For example, water is a molecule made up of two hydrogen atoms and one oxygen atom bonded together (H₂O).

Motor neurons
The nerve cells that connect the brain and spinal cord to skeletal muscles allowing conscious muscle contraction (movement). They act as a message delivery system: electrical signals originating in the brain are fired down the spinal cord along upper motor neurons; the electrical signals continue along lower motor neurons, which project out to skeletal muscles to control movement. Lower motor neurons are located in the anterior horn of the spinal
cord and are the main cell type affected by SMA. In SMA, low levels of the **Survival Motor Neuron (SMN) protein** cause the deterioration of lower motor neurons leading to muscle weakness and **atrophy**.

**Muscle biopsy**
Removal of a small amount of muscle tissue for analysis.

**Mutation**
A permanent change in the **DNA** sequence of a gene that can be inherited by subsequent generations. Dependent upon the type of mutation and where it occurs within the gene, it might have no effect on the **protein** produced, or it might disturb the protein’s function causing a **genetic disorder** such as SMA.

**Nerve Cells**
Also called neurons, nerve cells allow the quick transmission of electrical signals throughout the body. Different types of nerve cell make up the nervous system which functions to allow us to perceive and react to our surroundings. For example, the brain sends a signal along the nerves to tell a muscle to contract (move). Nerve cells are important for both involuntary (unconscious) functions like the beating of the heart and voluntary (conscious) functions like moving your arm.

**Neuromuscular**
Anything that relates to the nerves, muscles or the **neuromuscular junction**.
**Neuromuscular Junction (NMJ)**
The specialised connection, known as a synapse, between the lower **motor neurons** and **skeletal muscle** fibres. The NMJ allows signals from the nerves to get through to the muscles enabling them to contract (move).

**Nucleus**
The control centre of a cell that contains the **DNA** wrapped up within chromosomes.

**Peripheral nervous system (PNS)**
Consists of the nerve cell extensions found outside of the **central nervous system** (CNS). The PNS acts to connect the CNS with the muscles and internal organs. The lower **motor neuron axons** and their connections with the muscle (neuromuscular junctions) are found within the PNS.

**Physiotherapy**
Physical techniques used to promote, maintain and restore physical function of the body.

**Prenatal testing**
The **genetic testing** for diseases or conditions in a foetus or embryo. This is done by removing a sample of fluid or tissue by procedures such as amniocentesis or chorionic villus sampling (CVS).
Protein
Proteins consist of chains of amino acids arranged in very specific orders. The order of amino acids within a chain is determined by the genetic code (DNA). Different genes have the “instructions” for making different proteins. Proteins are the building blocks of our bodies and are essential for the structure, function, and regulation of cells, tissues and organs.

Rare Disease
The European Union (EU) considers diseases to be rare when they affect not more than 5 per 10,000 persons in the EU.

Recessive
Autosomal recessive describes a form of inheritance in which two faulty copies of a gene are required in order for a person to be affected by a genetic disorder. This means that a faulty copy of a gene is inherited from each parent. Survival Motor Neuron 1-associated SMA is an autosomal recessive condition. In X-linked recessive conditions, two faulty copies of the gene are needed for the genetic disorder to show in females, but only one faulty copy in males. This is because X-linked recessive conditions are caused by mutations in genes found on the X chromosome, but that are missing from the Y chromosome. Males have one X and one Y chromosome, while females have two X chromosomes.

Scoliosis
Sideways curvature of the spine.

Skeletal muscle
Consciously controlled muscle that attaches to bones allowing movement. Examples include the biceps, triceps, and thigh muscles.

Spinal
Relating to the spine.

Spinal cord
The bundle of nervous tissue within the spine. It includes nerve cells and extends out from the brain. The brain and spinal cord make up the central nervous system (CNS).

Survival Motor Neuron 1 (SMN1)
The gene that when mutated or deleted can lead to the development of SMA. For our lower motor neurons to survive and thrive we need a certain amount of the full-length SMN protein produced by the SMN1 gene.

Survival Motor Neuron 2 (SMN2)
The gene that can have an impact on the severity of SMA because it is able to produce a small amount of functional SMN protein. In people with a fault in the SMN1 gene, this can be important because the more copies of SMN2 that someone has, the more functional SMN protein they can produce. Individuals with more severe forms of SMA, for example Types 1 and 2, usually have fewer copies of the SMN2 gene than those with SMA Type 3.
**Survival Motor Neuron (SMN) gene**

A gene that produces the Survival Motor Neuron protein. Mutations in the **SMN1** gene are the cause of some forms of SMA. There are two types of SMN genes - **SMN1** and **SMN2**.

**Survival Motor Neuron (SMN) protein**

Produced from both the **SMN1** and **SMN2** genes, the SMN protein is required for the survival of lower motor neurons. If there is no SMN protein in a cell, the cell will die. Of all the different cell types, the lower motor neurons seem to be most affected by low levels of SMN protein.

**Tissue**

A collection of cells that work together to perform a common function. For example, organs are formed from multiple tissues.

**References**


