Spinal Muscular Atrophy Support UK
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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.

You may find it helpful to have a copy of our 'Glossary of terms' which you can download via the website at www.smasupportuk-glossary-of-terms

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.

More information on adult onset SMA, including SMA Type 4, is available from SMA Support UK's route map for adult onset SMA: www.routemapforsma.org.uk

SMA is a complex condition that can vary in severity - every person with SMA is different. You can always ask your medical team to provide you with information and advice specific to you.

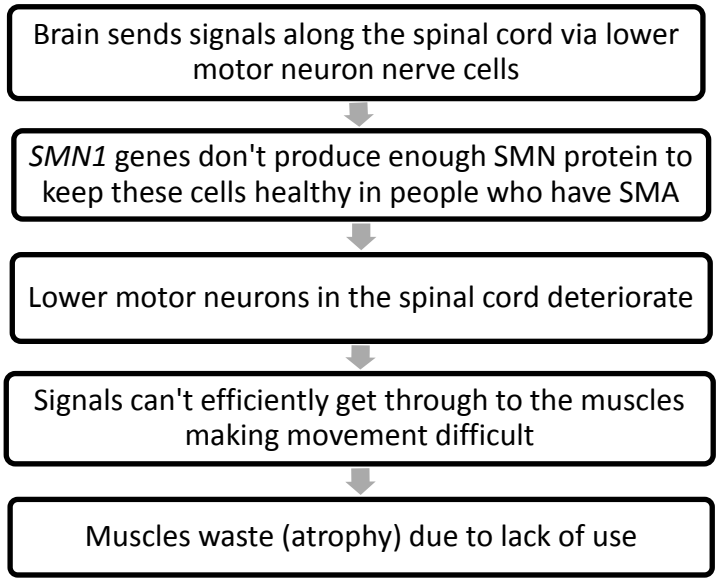
What causes SMA Type 4?

• The *SMN1* gene

SMA affects the nerve cells called lower motor neurons[1], which reside in the spinal cord and project out to muscles. These lower motor neurons carry electrical signals from the brain to move the muscles used for crawling and walking. These signals also control movement of arms, hands, head and neck as well as breathing and swallowing. For these lower motor neurons to be healthy, our *Survival Motor Neuron 1* genes (*SMN1* genes[1]) must produce enough Survival Motor Neuron (SMN) protein.

Most people have two copies of the *SMN1* gene. People with SMA Type 4 have two faulty copies of the *SMN1* gene, which means they are unable to produce enough SMN protein to have healthy lower motor neurons[2]. This causes their lower motor neurons in the spinal cord to deteriorate. This restricts the delivery of signals from the brain to their muscles, making movement difficult. Their muscles then waste due to lack of use; this is known as muscular atrophy.

In summary:



The *SMN1* gene is on the fifth chromosome in the region labelled 'q'. This is why SMA Type 4 and the other main types of SMA (childhood onset SMA Types 1, 2 and 3) are often referred to as **'5q SMA'.**

- ## The *SMN2* gene[1]

A second gene also has a role in producing SMN protein. This is the *Survival Motor Neuron 2* gene (*SMN2*), sometimes referred to as the SMA "back-up gene".

However, most of the SMN protein produced by *SMN2* lacks a key building block that is usually produced by *SMN1*. This means that *SMN2* cannot fully make up for the faulty *SMN1* gene in people with SMA.

The number of *SMN2* genes can vary greatly from person to person, from 0 – 8 copies. The severity of SMA has been linked to how much SMN protein a person makes[3-5]; individuals with more *SMN2* copies typically have a less severe form of SMA than those with fewer copies.

For more information on the inheritance of SMA please see 'The Genetics of Spinal Muscular Atrophy': www.smasupportuk.org.uk/the-genetics-of-sma

What are the effects of SMA Type 4?

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[2].

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 (GP).

Once you have your diagnosis, you may have contact with a number of different healthcare professionals to manage your SMA symptoms. 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

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.

Is SMA Type 4 hereditary?

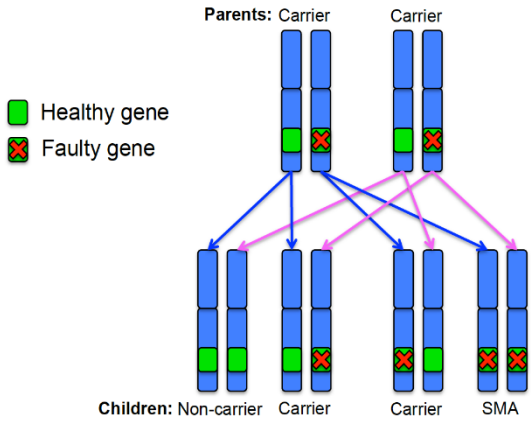
If an individual has one faulty copy and one healthy copy of the *SMN1* gene then they are a carrier of SMA and do not show any symptoms. If a couple are both carriers and they have children, then each of their children will have a 1 in 4 (25%) chance of having SMA. This is known as autosomal recessive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

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 GP.

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.

For information on '**Future Options in Pregnancy'** please see:
www.smasupportuk.org.uk/future-options-in-pregnancy

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 GP 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[6]. 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 (curvature of the spine).

Regular standing also promotes normal bodily functions, for example kidney drainage, and reduces calcium loss in bones[7].

- 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: https://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 GP 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 GP 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.

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 GP, 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 or phone 0115 950 6500.

In Scotland you can contact **Care and Repair Scotland**: www.careandrepairscotland.co.uk or phone 0141 221 9879.

In Wales contact **Care and Repair Cymru**: www.careandrepair.org.uk or phone 0300 111 3333.

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 For more information on DFGs in Northern Ireland see: 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

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

- 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

- 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 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 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/

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 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 GP 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

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

- 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 GP, 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, it may 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. If you are finding it increasingly difficult at work you may wish to talk to your consultant, physiotherapist, occupational therapist (and occupational health if your employer has this), about the support you need.

- 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: 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 678 1174 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

Receiving a diagnosis of SMA Type 4 may have a significant impact on you and your family both physically and emotionally. 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 GP 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

Is there a treatment or cure?

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

- ## Nusinersen/ Spinraza™

The first (and currently, the only) potentially available drug treatment for SMA is called nusinersen. Essentially, the drug is designed to modify the product of the *SMN2* gene to produce more functional SMN protein.

In collaboration with researchers, nusinersen was developed by Ionis Pharmaceuticals and Biogen Idec, which have run clinical trials with infants and children affected by SMA Types 1, 2 or 3. There have not yet been any clinical trials of nusinersen with anyone with SMA Type 4. On June 1st, 2017, the European Commission approved nusinersen for marketing under its brand name Spinraza™ as a treatment for those with **5q SMA**[8]. This is a broad term, that includes SMA Types 1, 2, 3 and 4.

Currently in the UK, nusinersen is only available if the medical team and family agree that an infant with SMA Type 1 is eligible and may potentially benefit from the treatment. This is possible in Scotland via the NHS and in the rest of the UK via what is called an Expanded Access Programme (EAP).

To find out more about how nusinersen works and the clinical trial results, please explore this section of our website: www.smasupportuk.org.uk/nusinersen Here, you will also find out how the UK drug approval system works, what stage nusinersen has reached, and the collaborative efforts being made to widen access to the treatment.

- ## Research and further developments

There is a considerable amount of research into SMA taking place around the world. This research will not only improve our understanding of the condition, but will also help to develop effective treatments.

One area of extensive research is the genetics of SMA and the underlying mechanisms that lead to damage of the nerve cells. The UK is a significant contributor to this, with several UK centres involved in clinical trials and international collaborations. This has led to encouraging breakthroughs in developing treatments.

SMA Support UK's website notifies the SMA community about the latest developments with drug treatments, the science behind them, and what clinical trials and other research is going on: www.smasupportuk.org.uk/research We alert people to new postings via our social media and monthly E-news. You can sign up for mailings here: www.smasupportuk.org.uk/sign-up-for-mailings

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

The adult onset SMA route map can be found at: www.routemapforsma.org.uk

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References

1. Lefebvre, S., Bürglen, L., Reboullet, S., Clermont, O., Burlet, P., Viollet, L., Benichou, B., Cruaud, C., Millasseau, P., Zeviani, M., Le Paslier, D., Frézal, J., Cohen, D., Weissenbach, J., Munnich, A. and Melki, J. (1995) 'Identification and characterization of a spinal muscular atrophy-determining gene', *Cell*, 80(1), pp. 155-165.

2. Lunn, M.R. and Wang, C.H. (2008) 'Spinal muscular atrophy', *The Lancet*, 371(9630), pp. 2120-2133.

3. Lefebvre S, Burlet P, Liu Q, Bertrandy S, Clermont O, Munnich A, Dreyfuss G, Melki J (1997). Correlation between severity and SMN protein level in spinal muscular atrophy. *Nat Genet* 16: 265–269.

4. Mailman MD, Heinz JW, Papp AC, Snyder PJ, Sedra MS, Wirth B, Burghes AHM, Prior TW (2002). Molecular analysis of spinal muscular atrophy and modification of the phenotype by SMN2. *Genet Med* 4: 20–26.

5. Eugenio Mercuri, et al., Diagnosis and management of spinal muscular atrophy: Part 1 Recommendations for diagnosis, rehabilitation, orthopedic and nutritional care. Neuromuscular Disorders (2017), doi: 10.1016/j.nmd.2017.11.005 www.sciencedirect.com/science/article/pii/S0960896617312841?via%3Dihub

6. Skalsky, A.J. and McDonald, C.M. (2012) 'Prevention and management of limb contractures in neuromuscular diseases', *Physical Medicine and Rehabilitation Clinics of North America,* 23(3), pp. 675-687.

7. Alekna, V., Tamulaitiene, M., Sinevicius, T. and Juocevicius, A. (2008) 'Effect of weight-bearing activities on bone mineral density in spinal cord injured patients during the period of the first two years', *Spinal Cord,* 46(11), pp. 727-732. 5.

8. The European Medicines Agency Spinraza Assessment Report. 21st April 2017: www.ema.europa.eu/docs/en_GB/document_library/EPAR_-_Public_assessment_report/human/004312/WC500229706.pdf

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