Adult Onset SMA-Diagnosis and Care

You can also read this guide on our website at smauk.org.uk/djya where you can follow all the links we give to further information.

Who this is for

This guide is for adults who:

- have had a recent diagnosis of an adult-onset form of SMA *or*
- are wondering if this might be what is causing symptoms they are experiencing.

It is designed to be used alongside medical advice. This should always come from a member of your medical team.

1. Trying to Get a Diagnosis

Getting a diagnosis of adult onset SMA can take time. The symptoms can seem similar to the symptoms of other neuromuscular conditions. Waiting for appointments, test results and a diagnosis can be stressful and frustrating. You might want to request a referral to a consultant neurologist from your GP (General Practitioner) if you are concerned about your symptoms or have had some tests but not yet been referred. As SMA is a rare condition, many GPs will not have come across it before.

Possible tests are:

- an electromyogram (EMG) which shows if the nerve supply is diminished
- a muscle biopsy which shows if there is any reduction in muscle cells

- an MRI (Magnetic Resonance Imaging) scan which produces detailed images of the inside of the body
- a CT (Computerised Tomography) scan which is another way of producing detailed images of the inside of the body
- a range of blood tests, including a blood sample for DNA testing.

2. Different Forms of Adult Onset SMA

Even with all these tests it is not always straightforward to say exactly what neuromuscular condition an adult has. A specific diagnosis of SMA Type 4 can be made following DNA testing.

The most common form of SMA is known as 5q SMA due to its genetic cause. 5q SMA includes SMA Type 4. If you have been diagnosed with SMA Type 4, you may also want to read:

- **What is 5q Spinal Muscular Atrophy?**
- **Symptoms and Effects of SMA – Type 4**
- **The Genetics of 5q SMA**

You can find these on the About SMA page > of our website.

The impact of adult onset SMA varies greatly between individuals and may change over time – no matter what form of SMA you have.

If you have been diagnosed with:

- **Distal SMA Type 5 (DSMA V)**
- **Spinal Bulbar Muscular Atrophy (SBMA) – Kennedy's Disease**

You may want to read information in our webpages about Rarer forms of SMA >

3. First Reactions to Your Diagnosis

Finally knowing your condition - and what it isn't– can bring mixed emotions for you and your family.

Some of the most common reactions people have include:

- Relief – you now know what your condition is.
- Shock
- Anxiety about the future
- Sadness and upset
- Anger or frustration

You may have all, some or none of these reactions – there is no 'right or wrong' way to feel. People can experience different combinations of these emotions at different times. Whatever you are feeling, it is very understandable. It is important to know that as well as your partner, family and friends, there are other people you can turn to for support at any time (please see below).

There is a lot of information to take in. You will need to give yourself time to ask your clinical team all the questions you have, as many times as you need to.

The section below on **Emotional Support** talks about the importance of this and suggests sources of support.

4. Movement, Mobility and Exercise

Whatever form of SMA you have, it will be important to maintain as much movement, mobility and independence as you possibly can. An individualised exercise programme guided by a qualified practitioner can help you do this.

Unfortunately, the availability of appropriate services to support you varies greatly depending on where you live. In some areas people have access to the services of a neuromuscular centre (see the Posture, Movement & Exercise > page in our Living with SMA section). This is where specially trained physiotherapists can set up a programme for you. Where these services are not available, your GP can refer you to a neurological physiotherapist.

To set up your personal programme, a physiotherapist will need to understand your medical history. They will need to get a picture of your joint range, muscle power and what you are able to do. You may then need to be seen at regular intervals to check how you are getting on.

An exercise programme would include:

- **Stretching exercises**

It is rare for people with adult onset SMA to be unable to walk. If walking is becoming difficult this is due to a combination of muscle weakness and a reduced ability to move some joints that may become tight (contractures). The most affected muscles and joints are:

- heel (achilles tendon)
- back of thigh (hamstrings)
- front of hip and thigh (hip flexors)
- outside of thigh (hip abductors)

You may also experience muscle weakness or tightness in your shoulders, arms and fingers. If your joint tightening is on one side it can lead to poor posture or cause you discomfort. In time this may affect your ability with day-to day tasks. Daily stretching can help this.

When stretching you may feel tension or pulling; but stretches shouldn't be painful. If they are, do talk to your doctor or physiotherapist.

You may enjoy exercising in the bath, a swimming pool, or, if one is available in your area, a hydrotherapy pool. Warm water can make muscles easier to stretch. Swimming is a good general exercise that can help to maintain muscle condition without causing muscle damage or over-exercising. Talk to your physio about doing this safely and for information about your nearest facilities. Most public pools have standard disabled changing facilities. If these do not meet your needs your physiotherapist or occupational therapist (OT) may be able to advise.

- **Exercises for posture, standing, balance, muscle strength and control**

Good posture is important for the body and muscles to work effectively. This applies to both sitting and standing.

Posture exercises often involve working on the muscles in the abdomen which help maintain good posture and balance. This is often called 'core stability'. Having good balance helps prevent falls. Standing balance, posture and muscle control can be maintained through exercise, such as yoga, pilates and tai chi. A physiotherapist will be able to advise you about appropriate exercises.

Standing is also a very important part of a physical management programme for many individuals with neuromuscular conditions. This includes those who are unable to walk independently. If needed, your physiotherapist may suggest equipment, such as a standing frame, which can help with this.

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

- **Exercises to improve cardiovascular fitness, stamina and mental health**

You have probably heard the phrase 'use it or lose it'. Inactivity can further limit mobility, which is why fitness is important. Physical activity is also good for maintaining psychological health and well-being and is important even if you have limited mobility. You might want to try swimming or adaptive sports as ways to keep fit. Check with your doctor and physiotherapist what types and intensity of exercise might be suitable for you. They should also be able to tell you about local centres and organisations that you could try.

You can download Muscular Dystrophy UK's Exercise advice for adults with muscle-wasting conditions > or request a copy by phoning 0800 652 6352 .

5. Managing Any Fatigue and Pain

People with adult onset SMA sometimes report experiencing pain and fatigue. This can happen because of overusing some muscles to try and compensate for the weaker ones. If this does happen to you, a physiotherapist may be able to help you manage better. 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 physio would be able to advise you how to do this.

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

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

You can help to manage fatigue by pacing yourself and preventing your body from becoming overly tired. Where possible, breaking demanding tasks into smaller chunks can be helpful. Planning your weekly schedule can also help to ensure that you do not overdo it. If you are in paid work you may ask about the possibility of, for example, working from home on some days or working more flexible hours. Regular exercise will help you to optimise your physical ability and understand your limits better. Your physiotherapist and occupational therapist (OT) can help support you with strategies to better manage any fatigue.

Though Muscular Dystrophy UK's Fatigue Management for people with neuromuscular conditions > is not SMA specific, you may find it helpful. You can request a copy by phoning 0800 652 6352.

Some people with adult onset SMA have told us 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.

6. Diet

A healthy diet is important for everyone. If needed, your GP or consultant can 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 for why some people become overweight. Extra weight can increase the stress on muscles, bones and joints, making physical activity even more difficult. If this is happening to you, a dietitian will be able to provide advice on a healthy diet that will suit your needs.

Adult onset SMA rarely causes difficulty with chewing and swallowing, though this may happen with Kennedy's Disease. Your medical team will provide you with advice and support with this if necessary.

7. Breathing

Adult onset SMA doesn't usually cause problems with breathing. If you have any shortness of breath, chest infections, early morning headaches or fatigue, your medical team will discuss if you need any baseline lung function tests or a sleep study. Further assessment usually takes place in your local ventilation clinic.

8. Treatment

In the UK, there is currently no NHS-funded drug that treats the genetic cause of any form of adult onset SMA.

- ### Type 4

In the UK, access to NHS funded drug treatment is only possible for those who have a clinical diagnosis of SMA Type 1, 2 or 3 and who meet the eligibility criteria. No drug treatments are currently available in the UK for people who have been given the clinical classification diagnosis SMA Type 4. Discussions leading to this diagnosis should include a careful review of the history of the person's onset of symptoms.

In 2024 / 5, the National Institute of Health and Care Excellence (NICE) is reviewing whether the NHS in England will continue to fund the two drug treatments, nusinersen and risdiplam. These may be prescribed for people who have SMA Type 1, 2 or 3. SMA UK is advocating for adults diagnosed with SMA Type 4 to have access to drug treatment. You can read more about the review >.

You can find out more by talking to your healthcare team, SMA UK and the other people and organisations listed in the support and resources section below.

9. Medications for Other Health Issues

If you are already on any medications for your health that are separate from your SMA, check with your consultant how these might interact with your SMA.

10. Emotional Support

Though it may come as some relief to have a diagnosis, having adult onset SMA 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. You may find some of the following suggestions helpful:

- ## Your Clinical Team

Over time, you may be offered support from health and social care professionals. This could include:

- Your Neuromuscular Care Advisor (NMA)
- Your nurse or physiotherapist
- A counsellor or psychologist

You can read more about the different sorts of health and social care professionals who may be involved in your care in our information sheet: **Who's Who of Professionals >**

- ## SMA UK's Support Team

Our small, experienced **Community Support Team ›** offers a UK-wide service for anyone affected by SMA. We are flexible in how we can support you and can be in contact by email, phone, text or virtual meetings (such as on Zoom or Microsoft Teams). We are also able to home visit if you would find it helpful to talk something through in person. Though we do not give medical advice, we can discuss with you the support you and your family can access.

- ## Family and Friends

You may have concerns about how this diagnosis and the care you may need will affect your partner and / or immediate family. Your family and friends will want to know how best they can support you, but sometimes it can be difficult for them to know what to do or say. If you are comfortable doing so, talking with them about how you are feeling can help. There may also be practical things they might be able to do (see more below).

- ### Religious / Spiritual Support

You may have your own religious or spiritual leader or community who can give you support.

- ### The SMA Community

Adult onset SMA is very rare and people's diagnosis journey and the impact of their SMA may not be like yours. However, if you would like to talk with someone who is affected by Adult Onset SMA, you can speak to our Community Support Team to see if it is possible for them to put you in touch with someone else.

At some point, you may be interested in joining our **Adults' WhatsApp Network >** . This is a safe space to connect with others, share experiences and ask questions. This group is for those who have a diagnosis of any Type of SMA and are aged 18+.

- ### Talking Therapies and Counselling

Some people find it helpful to talk about their feelings and how they are managing day-to-day. There may be things you want express that you have not wanted to say out loud to your partner, or anyone in your family or friendship group. An independent, trained professional can therefore be an option:

Free Talking Therapy Services:

- **England:**

You can access free NHS Talking Therapies (formally known as IAPT – Improving Access to Psychological Therapies) for anxiety and depression. You can either refer yourself directly without a referral from a GP, or a GP can refer you. Either way, you need to be registered with a GP to get talking therapies on the NHS.

Find out more >

- **Scotland:**

NHS Scotland offers 'Living Life' – free phone support sessions using Cognitive Behavioural Therapy (CBT) based techniques. You can refer yourself for an assessment by phoning 0800 328 9655.

Find out more ›

- **Wales:**

You can sign up for a free 12-week course of online therapy called 'SilverCloud'. This uses methods like Cognitive Behavioural Therapy (CBT) to help people manage their problems.

Find out more ›

Northern Ireland:

Referrals for any talking therapies must go through your GP.

Find out more ›

Waiting times for an appointment with these services will vary and may be quite long.

Private counselling

If you think counselling would be helpful for you and it is an affordable option, you could consider seeing a local private counsellor. These organisations can help you find one:

- **British Association for Counselling and Psychotherapy (BACP) ›** – guidance on counselling and how to find a suitable counsellor. Phone: 01455 883 300.
- **Counselling Directory ›** – online information about different types of counselling and a directory to search for qualified counsellors.

If you are not sure who to speak to or which is the right route for you, our Community Support Team > is here to talk through your options.

11. Practical and Financial Support

If you have been used to being independent and doing things yourself, letting others take over tasks can be emotionally difficult. Help with personal care can be equally, if not more, challenging to ask for. You may find it helpful to read our page on **Personal Life – Tab 1 Care and Relationships >**.

You may be eligible for financial support and benefits. This includes if you find you need and want external care and support from a Personal Assistant (PA). There is further information about this in our **Financial and PA Support pages ›.**

You can also talk to our **Community Support Team ›** for further information and / or **contact your Local Authority >** for support or to find out more about an assessment.

12. Support and Resources

Support and advice can help you manage your symptoms and the effects of your condition. Your neurologist, GP, physiotherapist and OT will all support you.

SMA UK ›

Phone: 01789 267 520
Email: office@smauk.org.uk

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The **Living With SMA area of our website ›** gives useful information and ideas. It builds on knowledge and advice from the SMA Community and SMA UK's Support Services Team. It covers a whole host of topics, including: equipment, homes, transport, leisure, holidays, financial, and emotional and social support.

We offer a number of ways for you to **Connect with the SMA Community ›.**

We have a number of **Grant Schemes ›** that may be useful.

You can keep up to date about events, new research developments and much more by **signing up for our monthly E-news ›.**

Muscular Dystrophy UK ›

Phone: 0800 652 6352
Email: info@musculardystrophyuk.org

MDUK provide information, support and grants towards specialist equipment for people affected by a range of neuromuscular conditions.

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 ›**
  Phone: 01606 860 911
  Neuromuscular Centre, Woodford Lane West, Winsford, Cheshire, CW7 4EH.
  The centre covers mainly the North of England and North Wales.

- **Muscular Dystrophy Support Centre ›**
  Phone: 02476 100 770
  Hereward College, Bramston Crescent, Coventry, West Midlands, CV4 9SW.
  The centre covers the Midlands region.

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Information
Creator
Patient Information Forum

Version 6

Author: SMA UK Information Production Team

Last updated: June 2025

Next full review due: June 2026

Links last checked: June 2025

This publication, and its links, provides information. This is meant to support, not replace, clinical and professional care.

*Find out more about **how we produce our information** ›.*

If you have any feedback about this information, please do let us know at: information@smauk.org.uk