

If your child has been recently diagnosed with:

## Spinal Muscular Atrophy with Respiratory Distress 1 (SMARD1)

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Spinal Muscular Atrophy with Respiratory Distress 1 (SMARD1) is a complex condition. The impact and severity varies, and every child with SMARD1 is different. Though there's limited information about SMARD1 because it is so rare, your child's medical team will be able to discuss how it affects your child and how to manage symptoms for maximum comfort and quality of life.

### Other useful resources:

Our information sheet:  
**'The inheritance patterns of some rarer forms of SMA'**

### Includes

Symptoms, diagnosis and effects

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What causes SMARD1?

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Looking after yourself

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Support and resources

## What is SMARD1?

SMARD1 is an inherited condition that causes muscle weakness and respiratory failure, usually beginning between the ages of 6 weeks and 6 months. SMARD1 is an extremely rare disease; the number of children affected is very small. Currently, it's not possible to collect accurate numbers of those affected but this may change as knowledge of the condition increases.

## What are its symptoms and effects?

Usually, the first and most noticeable symptom of SMARD1 is breathing difficulties (respiratory distress). This is caused by the diaphragm - the large, thin sheet of muscle separating the chest from the stomach that is essential for normal breathing - becoming paralysed. Along with this, and usually progressing rapidly after breathing difficulties start, there are signs of weakness in the muscles which are furthest from the centre of the body (distal muscles) - such as in the hands and feet. This weakness then spreads to all muscles, but within 2 years the muscle weakness usually stabilises. This is sometimes described as a 'disease plateau'.

A child's walking, crawling, arm and hand movement, head and neck movement and swallowing may all be affected, but the impact on each child varies. Some children may still have some ability to move their muscles while others lose all this ability. Other early symptoms of SMARD1 can include a weak cry and misshapen feet. For some children, fatty finger pads are another symptom.

SMARD1 may also affect nerves for sensation and control of body functions (the autonomic nervous system). This can lead to symptoms such as excessive sweating, an irregular heart rate or rhythm (cardiac arrhythmia), constipation, bladder incontinence and a reduced response to pain.

Sadly, life expectancy for babies who have SMARD1 is reduced. It's difficult though to predict life expectancy for any individual child because the progression of SMARD1 varies between children, as does the use of appropriate medical interventions to support breathing.

In the UK, SMARD1 is sometimes also called:

- Distal Spinal Muscular Atrophy 1 (DSMA1) **or**
- Distal Hereditary Motor Neuropathy Type VI (dHMN6 or HMN6).

The impact of SMARD1, varies greatly between individuals and this impact may change over time.

## What causes SMARD1?

Usually, nerve cells called lower motor neurons carry electrical signals from the brain to the muscles and make it possible to move them. In SMARD1, due to the changes in genetic material, the lower motor neurons degenerate which affects the link between the brain, the spinal cord and muscles. This means these muscles can no longer be stimulated, which causes them to waste or atrophy.

The affected gene in **SMARD1** is:

- the **IGHMBP2** gene (immunoglobulin mu-binding protein 2) found on **chromosome 11q13.3**

More than 60 different mutations in the *IGHMBP2* gene have been found to cause SMARD1. Most of these affect either the amount or efficiency (functionality) of the **IGHMBP2 protein** produced by the *IGHMBP2* gene.

Although the exact mechanism is unknown, if the IGHMBP2 protein isn't working properly, or there isn't enough of it, this leads to the degeneration of the lower motor neurons.

It's thought that the amount of 'functional' protein the mutated *IGHMBP2* gene produces may be linked to the severity of an individual's SMARD1. The less functional IGHMBP2 protein a child has, the earlier they are likely to develop symptoms. However, many differences have been seen even between individuals who have identical changes in the *IGHMBP2* gene, which suggests that other factors may also have an effect.

## How is SMARD1 diagnosed?

As SMARD1 is a very rare condition, it sometimes doesn't get recognised and it can take a very long time to get a diagnosis - perhaps several months. Sometimes it's not possible to confirm the diagnosis.

It's usually diagnosed following your child being in hospital with severe breathing difficulties. If doctors think this could be caused by SMARD1, a blood sample is taken for genetic testing to help confirm the diagnosis. They may ask for further tests, such as an electromyogram (EMG) or muscle biopsy if they're still unsure.

SMARD1 is an autosomal recessive genetic disorder. For further information about the genetics of the condition and how it is inherited, please see our information sheet: **'The inheritance patterns of some rarer forms of SMA'**.

As one parent said, 'Looking for the genetic confirmation for SMARD1 can be a bit like looking for a spelling mistake in a novel'.

## Looking after your child

When your child's diagnosis is confirmed, they may already be under the care of specialists in neuromuscular conditions and respiratory care. If not, you would expect to have a referral to these specialists so that you can meet to discuss next steps. Though families are prepared to travel, infants with SMARD1 may not cope well with a long journey so where this discussion takes place will very much depend on how well your child is and where the nearest specialist centre is.

Although there's currently no treatment or cure for SMARD1, there are options to manage your child's care so that they have the best possible quality of life and are as comfortable as possible. There's a wide range of severity in SMARD1 so discussions will focus on the options most beneficial for your child.

## Breathing difficulties

At first these are likely to be of greatest concern as once a child with SMARD1 starts to show symptoms of severe breathing difficulties, they will need a lot of help to survive. This can involve the need for surgery to create an opening in their windpipe so that they can breathe through a tube rather than their mouth (a tracheostomy). This is a very big step and any family facing this option will have a full and careful discussion with their specialists as to whether this is best for their child.

## Advance care plan

Though it's a difficult discussion to have, it's important that you have the earliest possible opportunity to discuss in-depth with your medical team the range of future care options that may be available so that they can record the treatment you do or don't wish your child to have if their health deteriorates or in an emergency.

SMA UK provide free multisensory toy packs for babies in the UK diagnosed with SMARD1. Contact us and ask for one.

These are all very difficult discussions and you should have time and support to consider and talk through the options thoroughly with your child's medical team.

This plan can be reviewed and you can change your mind at any time.

## Who else may become involved in my child's care?

As your child's condition is complex and potentially life-threatening and affects you and all your family, you'll need a lot of support from a number of different health professionals and the palliative care services. Palliative care includes the management of symptoms, information and practical support, and, if wanted, the provision of short breaks from caring for your child at home. The overall aim is to achieve the best quality of life for your child and to support you, whichever medical options are decided on.

If it's possible for you to take your child home, some of the professionals may be based in your local hospital or children's hospice. As well as your child's neuromuscular and respiratory specialists, your team may include specialists in:

- physiotherapy
- orthopaedics
- occupational therapy
- speech and language therapy
- dietetics
- community nursing

All these professionals will work as a team aiming to actively support your child and family's physical, emotional and practical needs. They will be careful to respect any cultural and spiritual needs you may have.

One of the team should act as your keyworker to help co-ordinate services for your family.

You can find out more about how these people can help in SMA UK's leaflet, '**Who's Who of Professionals**'.

## Going home

If you're able to go home, you'll need a great deal of practical support. A detailed 'care package' will be set up before your child is discharged from hospital. This often takes a long time to organise and is likely cover the following topics:

- Housing
- Equipment and supplies
- Day and night-time carers / who will provide this service / hours and rotas
- Short breaks / respite care
- Procedures to cover emergencies
- Getting out and about - including play, leisure and education
- Sources of financial help
- Sources of support - including those in your local area

You should also receive information and advice about other financial, practical and emotional support available in your local area.

With your permission, your child's emergency advance care plan can be shared with all the professionals supporting your child, including ambulance services, so that everyone is aware of your wishes. You should have your own copy so that you can give it to hospital services if you are away from your home area.

## Your local children's hospice services

Your local children's hospice is there to support families, both practically and emotionally. They provide support and respite at any stage from diagnosis onwards. As well as offering nursing care, they provide a range of services that may include physiotherapy, complementary therapies, play and music therapy.

They can support you as you review and make decisions about your baby's ongoing and future care should they become very unwell. They can also offer invaluable practical and emotional support and specialist care if, sadly, you are faced with the end of your child's life. This may be within the hospice and in some areas in your own home.

You can find further information, practical ideas and links to resources at: [www.livingwithsma.org.uk](http://www.livingwithsma.org.uk)

Although not everything is relevant for SMARD1 (as primarily written for those with 5q SMA) many sections may still be helpful (discuss with your medical team if you're not sure).

It's important that you know how to access medical care in case your child has a respiratory emergency. Open access to your local children's ward can be arranged with your medical team so that, whenever needed, you can take your child straight to hospital.

Many families find hospices to be happy and positive environments where they can spend time together doing activities as a family.

## Looking after yourself and your family

The impact of a diagnosis of SMARD1 on families is enormous. It often comes as a shock and you may experience feelings such as disbelief, confusion, anger and sadness. You may find it difficult to take everything or anything in.

Everyone's different, but it's important for you and your family to have access to emotional support and to have plenty of time to talk and ask questions. This can be with members of your child's medical team, your local GP, health visitor, social worker, psychologist or a counsellor, family and friends, a spiritual leader, support groups, or online communities.

Even though you'll be focused on your child, it's important to try and look after yourself too. Things like remembering to keep your vaccinations up-to-date and getting the 'flu' jab. Perhaps ask friends and family to help out with practical jobs such as shopping and cleaning to help save you time and energy.

You may also benefit from accessing local short break services. These may be available in your home or at your children's hospice. Some local authorities also have residential short breaks provision for children with disabilities. You can ask your GP, community nurse, health visitor or social worker for more information.

## Genetic counselling

A genetic counsellor is a healthcare professional who has expert training in genetics. They will explain how SMARD1 is inherited and what the chances are of other family members also being affected. Genetic counselling also provides the opportunity to discuss options and choices for any future pregnancies.

Following your child's diagnosis, you should be offered genetic counselling. A referral to genetic services is usually made through your General Practitioner (GP), paediatrician or neurologist.

The 24 hour-a-day responsibility of caring for a child with complex medical needs can be physically, emotionally and psychologically exhausting. Everyone is different and it's OK to ask for support if, and when, you need it.

Genetic counsellors can also be contacted at a later date to discuss any further questions you may have.

## Financial support

You may be eligible for a number of financial benefits to help towards the cost of providing the extra care your child needs. This does depend on your individual circumstances. For further information visit the website: [www.gov.uk](http://www.gov.uk)

Your health visitor, community nurse, neuromuscular care advisor, family support worker, social worker or outreach worker may be able to help with applications for financial benefits.

There are also a number of charities that can assist with the cost of general household goods, specialist equipment and holidays / days out.

## Research

Though there's very limited research into SMARD1, what we do find out we report on this page: [www.smauk.org.uk/research-into-rarer-forms-of-sma](http://www.smauk.org.uk/research-into-rarer-forms-of-sma)

Patient Registries are databases of genetic and clinical information about people with a particular condition. They provide points of contact for when new treatments develop that need to be tested in clinical trials. They also help specialists gain more knowledge about the condition and the number of people affected which helps to develop and improve worldwide standards of care for people with the condition.

Currently in the UK there isn't a registry specifically for SMARD1. The UK SMA Patient Registry is mainly for people affected by SMA due to mutation in the *SMN1* gene (known as 5q SMA), however the registry does accept registrations from people affected by Spinal Muscular Atrophy with Respiratory Distress 1 (SMARD1) with mutation in the *IGHMBP2* gene.

Please contact SMA UK for more information or see:

[www.livingwithsma.org.uk/children/support](http://www.livingwithsma.org.uk/children/support)

To find out more about the UK SMA Patient Registry website: [www.treat-nmd.org.uk/registry/](http://www.treat-nmd.org.uk/registry/)

You can also contact the Registry Curator by e-mail: [registry@treat-nmd.org.uk](mailto:registry@treat-nmd.org.uk)

or phone: 0191 241 8605.

## Support and Resources

### SMA UK

**Phone:** 01789 267 520

**Email:** [supportservices@smauk.org.uk](mailto:supportservices@smauk.org.uk)

**Website:** [www.smauk.org.uk](http://www.smauk.org.uk)

We provide a free **Support & Outreach Service** for families by email, phone, text and outreach home-visiting. Our experienced team offer personalised support and information and are available to answer questions and talk things through. Though we don't give medical advice, we can discuss with you the support you and your family can access. **Multisensory toy packs** are also available free of charge for babies in the UK diagnosed with SMARD1.

You can upload a copy of your child's latest **emergency health plan** using our **EHP app** which follows the NHS guidelines for data privacy. It's free to download on Android and iOS. You can find out more at: <https://ehpapp.com>

The **Living With SMA** area of our website builds on knowledge and advice from the SMA Community and SMA UK's Support Services team, and covers a whole host of topics including equipment, homes, education, transport, leisure, holidays, financial, emotional and social support. In the **Health & Wellbeing** section of this website, we refer to the **2017 Standards of Care (SoC)** for SMA which covers **5q SMA**. If your child has been diagnosed with SMARD1, the guidance in the SoC may not be relevant for you and it's important to discuss your child's individual care and management with your medical team.

Other sections of the Living With SMA website will be relevant for you and your child and, we hope, may provide some useful information and ideas for day-to-day living. If you're at all unsure if something's relevant for you, we suggest you discuss it with a healthcare or other professional such as a physiotherapist, occupational therapist (OT), education or social worker who is supporting you and your child.

### Together for Short Lives

**Phone:** 0808 8088 100

**Website:** [www.togetherforshortlives.org.uk](http://www.togetherforshortlives.org.uk)

Provide information and support to families who have a child with a life-limiting condition.

### Contact for Families with Disabled Children

**Phone:** 0808 808 3555

**Website:** [www.contact.org.uk](http://www.contact.org.uk)

Provide information and support to families who have a child with a disability.



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## General References

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*We are grateful to the writers and reviewers who assist us in our information production. A list of who this includes may be viewed on our website: [www.smauk.org.uk/our-writers-and-reviewers-panel](http://www.smauk.org.uk/our-writers-and-reviewers-panel) or requested from [supportservices@smauk.org.uk](mailto:supportservices@smauk.org.uk)*

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**[supportservices@smauk.org.uk](mailto:supportservices@smauk.org.uk)**