Spinal Muscular Atrophy with Respiratory Distress 1 (SMARD1)

You can also read this guide on our website at smauk.org.uk/na3g where you can follow all the links we give to further information.

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

Sadly, there are no disease-modifying drug treatments for SMARD1 which is a very rare condition. Infants and children who have SMARD1 need supportive care from a specialist clinical team.

SMARD 1 is not the same as SMA 1 or Spinal Muscular Atrophy Type 1 >. SMA Type 1 has a different genetic cause. There are disease-modifying treatments for SMA Type 1 (Nusinersen, Risdiplam and Zolgensma). These treatments do not work for conditions that have other genetic causes. Infants and children who have SMA Type 1 also need supportive care from a specialist clinical team.

Who is this for

This information sheet is for parents / carers whose child has been diagnosed with SMARD 1. We hope it will provide some answers to the many questions you will have. It covers:

- **The symptoms, effects and causes of SMARD 1**
- **Looking after your child**
- **Support for you and your child**
- **Genetic Counselling**
- **Research**

You might at some point want to read about the **personal experiences of some other families >**

1. What is SMARD1?

SMARD1 is an inherited nervous system condition that causes muscle weakness and respiratory failure. Symptoms usually start to show between the ages of 6 weeks and 6 months. It is an extremely rare disease. The number of children affected is very small.

In the UK, SMARD1 is sometimes also called:

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

2. What are its Symptoms and Effects?

The first and most noticeable symptom of SMARD1 is usually breathing difficulties (respiratory distress). These are caused by issues with the large, thin sheet of muscle separating the chest from the stomach (the diaphragm) that is essential for normal breathing becoming paralysed.

After breathing difficulties start, signs of weakness in the muscles that are furthest from the centre of the body (distal muscles) usually progresses rapidly. This weakness shows first in the hands and feet. Muscles in the legs are normally being affected before muscles in the arms. It 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. Exactly how each child is affected varies. Some children may still have some ability to move their muscles while others lose this ability completely.

Other early symptoms of SMARD1 can include a weak cry and misshapen feet. For some children, fatty finger pads are another symptom.

The impact of SMARD1 varies greatly between individuals. This impact may change over time.

SMARD1 may also affect nerves for sensation (called sensory neurons) 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 severely reduced. It is difficult though to predict life expectancy for any individual child. The progression of SMARD1 and appropriate medical interventions to support breathing varies between children.

3. 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. This affects the link between the brain, the spinal cord and muscles. The muscles can no longer be stimulated, which causes them to waste or atrophy.

The gene that is altered in SMARD1 is:

- ***IGHMBP2* gene (immunoglobulin mu-binding protein 2)**

It is found on chromosome 11q13.3[1–2]

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 is not working properly, or there isn't enough of it, the lower motor neurons degenerate.

It is thought that the amount of working or 'functional' protein the altered *IGHMBP2* gene produces may be linked to the severity of a child'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. This suggests that other factors may also have an effect.

SMARD1 is an autosomal recessive genetic disorder. For further information about the genetics of the condition and how it is inherited, please see: The Inheritance Patterns of Some Rarer Forms of SMA >

4. How is SMARD1 Diagnosed?

As SMARD1 is a very rare condition, it sometimes does not get recognised. It can take a very long time to get an accurate diagnosis – perhaps several months. Sometimes it is not possible to confirm the diagnosis.

A child is usually diagnosed if they have been 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. If they are still unsure, they may ask for further tests, such as an electromyogram (EMG) or muscle biopsy.

As one parent said:

"Looking for the genetic confirmation for SMARD1 can be a bit like looking for a spelling mistake in a novel".

5. 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. Where this discussion takes place will very much depend on how well your child is and where the nearest specialist centre is.

Although there is currently no treatment or cure for SMARD1, there are options to manage your child's care. The aim is to give them the best possible quality of life and for them to be as comfortable as possible. As there is such a wide range of severity and impact with SMARD1 and family circumstances vary greatly, discussions are very individualised.

SMA UK provides free multisensory toy packs > for babies in the UK diagnosed with SMARD1.

6. Breathing Difficulties

At first these are likely to be of greatest concern. 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. Any family facing this option will have a full and careful discussion with their specialists as to whether this is best for their child.

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

7. Advance Care Plan

Though it is a difficult discussion, it is important that you have the earliest possible opportunity to discuss the range of future care options with your medical team. They can then record the treatment you do or do not wish your child to have if their health deteriorates or in an emergency.

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

8. Who Else May Become Involved in my Child's Care?

Managing your child's complex and potentially life-threatening condition will affect you and all your family. You will 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.

All these professionals will work as a team aiming to actively support your child and family's physical, emotional and practical needs.

If it is 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

You can find out more about how these people can help on the page Who's Who of Professionals >.

9. Going Home

If you are able to go home, you will 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 to 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.

You can find further information, practical ideas and links to resources in the Living with SMA/Children > section of our website. Although not everything is relevant for SMARD1, many sections may still be helpful.

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.

It is important that you know how to access medical care in case your child has a respiratory emergency. Your medical team can organise open access to your local children's ward. This will mean that whenever needed, you can take your child straight to hospital.

10. Your Local Children's Hospice Services

Your local children's hospice is there to support families, both practically and emotionally. They provide support and breaks from caring 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 with 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.

Many families find hospices to be welcoming and positive environments where they can spend time together doing activities as a family. Details of hospice services are available from Together for Short Lives >

11. Looking After Yourself

The impact of a diagnosis of SMARD1 on families is enormous. It often comes as a huge shock. You may experience feelings of disbelief, confusion, anger and sadness. You may find it difficult to take everything or anything in. The 24 hour-a-day responsibility of caring for a child with complex medical needs that follows can be physically, emotionally and psychologically exhausting.

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

Even though you will be focused on your child, try and look after yourself too. Things like remembering to keep up to date with your own health checks and getting the 'flu' jab are important. Perhaps ask friends and family to help out with practical jobs such as shopping and cleaning, which can help save you some time and energy.

You may also find it helpful to use local short break services. These may be possible at your home or at a children's hospice. Ask your GP, community nurse, health visitor or social worker for more information.

12. Financial Support

Families living in the UK 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, see **Living With SMA – Financial Support & Benefits ›**

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 may assist you with the cost of general household goods, specialist equipment and holidays / days out.

Please contact **SMA UK's Community Support Team›** for more information.

13. 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 GP, paediatrician or neurologist.

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

14. Research

Though there is very limited into SMARD 1, what we do find out we report in our **SMARD Research page ›**

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. This helps to develop and improve worldwide standards of care for people with the condition.

Currently in the UK there is no specific Patient Registry 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 those affected by SMARD1 who have the altered *IGHMBP2* gene.

To find out more, visit the UK SMA Patient Registry website ›.

15. Support and Resources

SMA UK >

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

Our **Community Support Team >** offers a free confidential UK-wide service. We don't provide medical services or advice. But, we can give emotional support, practical advice, and guidance. We do this by email, phone, text, or virtual or face-to-face meetings.

You can ask for a free **Multisensory toy pack** > if you live in the UK and your child is under 24 months of age and has been diagnosed with SMARD1.

The **Living With SMA / Children >** area of our website builds on knowledge and advice from the SMA Community and SMA UK's Support Services team. It covers a whole host of topics that might be useful including equipment, homes, transport, emotional and social support. The Health & Wellbeing section focuses on care for those who have 5qSMA so is not relevant for families affected by SMARD1.

Children's hospices

Located throughout the UK, these also offer a wide range of services and support to eligible children and families; some also offer short breaks. Details of hospice services are available from **Together for Short Lives >**

Together for Short Lives >

- Phone: 0808 8088 100

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

Contact >

- Phone: 0808 808 3555

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

16. References

1. Pitt et al. (2003) Severe infantile neuropathy with diaphragmatic weakness and its relationship to SMARD1. Brain 126: 2682-2692.

2. **www.omim.org/entry/604320** (last accessed 16th January 2025)

General References

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Patient Information Forum

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

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