Living now with SMA

Though drug treatments have had a huge impact, they are not a cure for children, young people and adults who may still:

- Have breathing and swallowing difficulties
- Have difficulties keeping up with daily activities and need specialist equipment, e.g powered wheelchairs, to get around
- Need significant care and support

Rarer Forms of SMA

There are other very rare forms of SMA which have different genetic causes and inheritance patterns.

Who are SMA UK?

Spinal Muscular Atrophy UK is a national charity. We provide accurate information and support to anyone affected by SMA. You can reach us via our support line, website, and outreach services.

We advocate for the SMA Community. We work with other groups to improve access to the best care, services, and treatments in the UK. Our goal is to help people with SMA live the lives they want.

How to contact us

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Phone: 01789 267520
• Mon – Thurs (8.30am – 4pm)
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• Closed on public holidays.

Email: office@smauk.org.uk

Sign up for mailings:
www.smauk.org.uk/about/newsletter-sign-up/

www.smauk.org.uk

Trusted Information Creator
Patient Information Forum

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About Spinal Muscular Atrophy

Key Information

Spinal Muscular Atrophy uk
SUPPORTING • INFORMING • ADVOCATING

What is Spinal Muscular Atrophy? (SMA)

Spinal Muscular Atrophy is a rare, neuromuscular condition. It causes progressive muscle wasting and weakness. It may affect crawling and walking and movement of the arms, hands, head and neck. It may also affect breathing and swallowing. How severely people are affected, and in what way, varies greatly. There are different forms of SMA with different genetic causes. The most common form is '5q SMA'. This is passed from parents to their children through an 'altered' version of the Survival Motor Neuron 1 (SMN1) gene.

- Approximately 1 in 40 of us carry this 'altered' gene – that is around 1.7 million carriers in the UK. Carriers do not have SMA.

- If two carriers of the 'altered' gene have a babythere is a 1 in 4 chance in every pregnancy that their baby will have SMA.

How many people have SMA?

- Every month in the UK, 4 babies are born with 5q SMA. Worldwide, between 1 and 2 children, young people and adults in every 100,000 have 5q SMA.

Types of 5q SMA

These describe the age symptoms start to show and the motor milestones (e.g. the ability to sit, stand and walk) that a child or adult would be expected to achieve. They are used by doctors at the time of diagnosis.

SMA Type	Age symptoms usually begin	Motor milestones
Type 1	0-6 months	Unable to sit or roll independently
Type 2	7 - 18 months	Able to sit but not walk independently
Type 3	18 months - 18 years	Able to walk though may lose this ability over time
Type 4	18 years +	Mild walking difficulties

However, SMA's severity and impact varies from person to person, both within and between 'Types'. Each child and adult is affected differently.

Drug Treatments

There is no cure for SMA, but there are now three NHS-funded drug treatments. They are not suitable for everyone but most people in the UK who have SMA Type 1, 2 or 3 can receive one of them. These drugs can change what motor milestones babies and children may be able to achieve. They can improve their general health. The treatments work best if started before there is any muscle weakness, or when this is minimal. It is therefore important for treatment to be started as soon as possible. This is why clinicians and patient groups want newborn screening for SMA to start in the UK as soon as possible.

For adults living with SMA, drug treatment that can stabilise the condition later in life may also make a positive difference – for example, helping with fatigue or preventing the loss of the ability to use a finger to control a powerchair or laptop.

For more information about SMA, Please visit: smauk.org.uk/support-information