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 that provides accurate information and support to anyone affected by SMA through our support line, website and outreach services.

As advocates for the SMA Community, we work in partnership with other organisations to improve access to the best care, services and treatments available within the UK to allow people living with SMA to lead the lives they want to lead.

How to contact us

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

www.smauk.org.uk

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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 ability, arm, hand, head and neck movement, 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 called '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 1.69 million carriers in the UK. Carriers do not have SMA.

- If two carriers of the 'altered' gene have a baby, there 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 are used by doctors at the time of diagnosis. They 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.

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, how severe and what impact SMA has 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 for those who have SMA Type 1, 2 or 3. These can change what motor milestones babies and children may be able to achieve and improve their general health. These drugs must be given as early as possible to maximise the potential benefits. This is why clinicians and patient groups are calling for the earliest possible introduction of newborn screening for SMA in the UK.

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