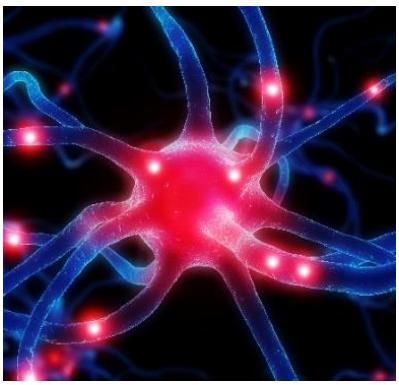
Nusinersen in the UK (marketed as SPINRAZA™)

Find out more about the treatment and what SMA Support UK is doing to help get access to it in the UK.

What does nusinersen do? How does it work?



Spinal Muscular Atrophy (SMA) affects a set of nerve cells called the lower motor neurons which run from the spinal cord out to our muscles. The lower motor neurons carry messages that make it possible for us to move the muscles we use to crawl and walk, to move our arms, hands, head, neck, and to breathe and swallow.

For our lower motor neurons to be healthy, we need to produce an important protein called the Survival Motor Neuron (*SMN*) protein. Our ability to do this is controlled by a gene called *Survival Motor Neuron 1* (*SMN1*).

Having two faulty *SMN1* genes means that a child has SMA and is only able to produce very low amounts of the SMN protein. This causes their lower motor neurons in their spinal cord to deteriorate. Messages from their spinal cord do not efficiently get through to their muscles, which makes movement difficult. Their muscles waste due to lack of use and this is known as muscular atrophy.

Another gene called *SMN2* also helps with the production of SMN protein. Children with SMA Type 1 usually have fewer copies of this gene so also miss out on this 'back-up' source of SMN protein.

Nusinersen is a highly specialised medicine that can increase the production of SMN protein by targeting the process through which it is produced by the *SMN2* gene.

Nusinersen (also known by its brand name SPINRAZA™) is produced by the pharmaceutical company Biogen.

What are the benefits of nusinersen?

In Biogen's clinical trials some individuals who had, or were likely to, develop SMA Types 1, 2 or 3 were treated with nusinersen and some of these individuals showed improvements, including:

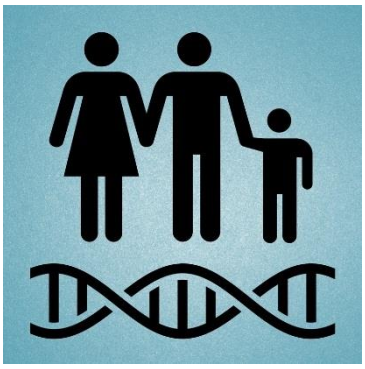
- achieving physical milestones which they would not have reached without treatment
- maintaining physical milestones which they would not have done without treatment
- surviving longer than expected considering the typical course of their condition

More follow-up trial results are being worked on and will be published in due course.

What other options are there?

Nusinersen is not a cure for SMA; it is a treatment and, at the moment, it is the only one available. There are however other clinical trials of other drugs in the pipeline.

It is important to remember that with or without nusinersen treatment, there are a range of options aimed at managing symptoms, reducing complications of muscle weakness and maintaining the best quality of life for as long as possible.



Each individual or family should have the opportunity to discuss with their medical team which treatment and management options are clinically and personally appropriate for them or their child.

Can nusinersen be accessed in the UK?

Currently the only way to access the treatment in the UK is through what is called an Expanded Access Programme (EAP). This programme is only available to children with SMA Type 1 where both the child's medical team and the child's parents/guardians agree that it will be of benefit and that the child is eligible for the treatment. Any family with a child with SMA Type 1 wanting to find out if they are eligible should ask their medical team.

Since December 2016, nusinersen has been licensed for use in the United States of America. On 1st June 2017, it was approved by the European Commission for marketing as 'Spinraza™' in Europe 'for 5q SMA'. This includes SMA Types 1, 2, and 3 where there is genetic confirmation of a fault on the *SMN1* gene found on Chromosome 5. For other types of SMA, including some forms of Adult Onset, there are other genetic causes which nusinersen cannot treat.

In the UK, the availability of the drug now depends on a recommendation by the National Institute for Health and Care Excellence (NICE), NHS England, the Scottish Medicines Consortium and other authorities in the devolved nations to decide whether to fund the drug in England, Scotland, Wales and Northern Ireland. Availability may also depend on the treatment readiness of specialist centres.

NICE National Institute for Health and Care Excellence

So what are the problems?

On 1st April 2017, NICE and NHS England jointly announced that they were pressing ahead with changes to the Highly Specialised Technologies (HST) drug funding criteria in England. The HST assessment process is specifically for considering whether NICE will recommend that the NHS fund treatments for rare diseases. The changes lowered a number of cost thresholds that a drug would need to meet to be approved. These changes threaten the possibility of nusinersen getting to the point of being funded by the NHS.

Additionally, despite the efforts of clinicians across the UK, there continue to be unacceptable delays in the roll out of the Expanded Access Programme (EAP) for SMA Type 1 and, as a result, inequitable access across the country.

What is SMA Support UK doing?

- We continue to publish up-to-date, accurate information about the treatment, including any new clinical trial results. You can find this on our website here: www.smasupportuk.org.uk/drug-treatments-proven-to-be-effective-for-sma

- We listen to questions from families and talk through their concerns. You can see many of these questions and answers in the above section of our website

- We keep the SMA community up-to-date with developments via our monthly E-newsletter and social media. Anyone can sign up to these – just go to the top of our home page

- Importantly, we are advocates for:

  - the EAP being rolled out equitably and quickly for eligible children with SMA Type 1

  - nusinersen being available via the NHS in the UK as quickly as possible for all who could benefit from the treatment – those with SMA Type 2 or 3 as well

- We are working with Muscular Dystrophy UK, the SMA Trust, Genetic Alliance UK and the family led campaign group TreatSMA on these goals

- You can join us by responding to any 'calls for action' that you read about in our updates. Letters from those affected by the condition, addressed to the right people, can have the biggest impact. When we reach the stage of giving evidence to NICE, you can respond to any calls to add your voice

This work is hugely important to us and we are giving it high priority as well as continuing to offer emotional support, practical advice and guidance to children, young people and their families, and to adults affected by any form of SMA anywhere in the UK.

Please help us carry on keeping families informed and working to make nusinersen available in the UK.

You can donate online through our website:

www.smasupportuk.org.uk/donate

Or send a cheque made payable to **SMA Support UK** at:

Spinal Muscular Atrophy Support UK
40 Cygnet Court
Timothy's Bridge Road
Stratford-upon-Avon
CV37 9NW

If you have questions about SMA, nusinersen and/or want support, please:

Phone: 01789 267 520. Mon – Thurs (9.00 am—3.30 pm), Friday (9.00 am—1.00 pm) and closed on public holidays.

Email: supportservices@smasupportuk.org.uk

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