Zolgensma®▼ (onasemnogene abeparvovec) receives NICE final draft guidance in presymptomatic babies up to 12 months with spinal muscular atrophy (SMA)

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- Infants identified with SMA through genetic testing will be able to be routinely treated presymptomatically following final recommendation
- It is imperative to diagnose SMA and begin treatment as early as possible to help halt irreversible motor neuron loss and disease progression.[1]
- In the UK, there is no national newborn screening (NBS) programme for SMA. There is an urgency to introduce SMA as part of NBS to provide a nationwide solution to identify these patients from birth before symptoms manifest.[2]

London, UK, 16th March 2023 – Novartis Gene Therapies has confirmed that the National Institute for Health and Care Excellence (NICE) is recommending the use of Zolgensma (onasemnogene abeparvovec) as an option in babies with presymptomatic 5q spinal muscular atrophy (SMA) with a bi-allelic mutation in the *SMN1* gene and up to three copies of the *SMN2* gene. This recommendation will allow routine access to onasemnogene abeparvovec, which is designed to address the genetic root cause of SMA to help halt disease progression, before the onset of SMA symptoms.[3] Treatment at this early presymptomatic stage of the disease is critical to ensure babies with SMA have the best chance for optimal outcomes.[1]

Imran Kausar, General Manager at Novartis Gene Therapies UK, commented, *"Infants with SMA experience irreversible loss of motor neurons, substantially affecting their survival and impairing their quality of life. Zolgensma will be the first treatment to be routinely commissioned for presymptomatic babies in England, and as it is imperative to diagnose SMA and begin treatment as early as possible, we welcome the decision by NICE for this recommendation."*

In the UK, there is no national newborn screening programme for SMA. Currently, only infants who have a sibling history of SMA or have had family screening are identified presymptomatically. Babies without a family history are only diagnosed if symptoms are identified, which can take up to six months.[4] There is an urgency for change to ensure the next cohort of babies have a chance at the best treatment outcomes. Newborn screening could provide a nationwide solution to identify these patients from birth.[2]

Prof. Laurent Servais, Professor of Paediatric Neuromuscular Disease at the University of Oxford commented, *"I have seen too many families devastated by this disease, but we now have treatment options. However, waiting until the onset of symptoms is still too late. Every day we delay in finding and treating these infants, we could be responsible for a child spending their life in a wheelchair. Diagnosis of SMA through newborn screening is imperative to help detect the disease and treat it presymptomatically. Then we have more potential to transform the lives of infants with SMA and their families."*

Newborn screening for SMA has been shown to be a more cost-effective use of NHS resources, based on a

recent cost analysis using UK assumptions which evaluated the cost- effectiveness of NBS followed by treatment against a treatment pathway without NBS.[2] The results showed that NBS provides a cost saving of £62,191,531 over the lifetime of a newborn cohort, based on approximately 56 babies identified with SMA per year in England.[2] In addition, there were improved health outcomes from NBS with an estimated gain of 529 quality-adjusted life years (QALYs) over the lifetime of a newborn cohort identified per year. One QALY is equal to one year of life in perfect health.[2]

On 7th July 2021, following a landmark deal, onasemnogene abeparvovec was made available on the NHS recommended for patients with clinical diagnosis of SMA Type 1 up to 12 months of age as part of a managed access agreement.[5] Onasemnogene abeparvovec was granted conditional access for presymptomatic patients, pending results of the Phase III SPR1NT clinical trial. Following the completion of the trial and exit from the managed access agreement, NICE began the partial review of onasemnogene abeparvovec for treating SMA in presymptomatic babies. The NICE committee has concluded that onasemnogene abeparvovec is effective in treating presymptomatic SMA, resulting in their draft final guidance for it to be routinely commissioned for the treatment of presymptomatic babies.

Product information is available at: Summary of Product Information for Zolgensma.

Notes to Editors
The expected publication date of final guidance is Wednesday 19th April 2023.

About SMA
SMA is the leading genetic cause of infant death.[8,9] If left untreated, SMA Type 1 leads to death or the need for permanent ventilation by the age of two in more than 90% of cases.[6,7] SMA is a rare, genetic neuromuscular disease caused by a lack of a functional *SMN1* gene, resulting in the rapid and irreversible loss of motor neurons, affecting muscle functions, including breathing, swallowing and basic movement.[3] It is imperative to diagnose SMA and begin treatment, including proactive supportive care, as early as possible to halt irreversible motor neuron loss and disease progression.[10] This is especially critical in SMA Type 1, where motor neuron degeneration starts before birth and escalates quickly. Loss of motor neurons cannot be reversed, so SMA patients with symptoms at the time of treatment will likely require some supportive respiratory, nutritional and/or musculoskeletal care to maximise functional abilities.[11]

About Zolgensma®▼ (onasemnogene abeparvovec)
Zolgensma® (onasemnogene abeparvovec) is the only gene therapy for spinal muscular atrophy (SMA) and the only SMA treatment designed to directly address the genetic root cause of the disease by replacing the function of the missing or non-working SMN gene to help halt disease progression through sustained SMN protein expression with a single, one- time IV infusion. Zolgensma was approved in May 2019 by the US Food and Drug Administration and represents the first approved therapeutic in Novartis Gene Therapies' innovative proprietary platform born to treat rare monogenic diseases using gene therapy.[12] In addition to the United States, Zolgensma has been approved in over 46 countries. To date, more than 3,000 patients have been treated with Zolgensma worldwide across clinical trials, managed access programmes, and in the commercial setting.[13]

▼ This medicinal product is subject to additional monitoring. This will allow quick identification of new safety information. Healthcare professionals are asked to report any suspected adverse reactions. Reporting forms and information can be found at www.mhra.gov.uk/yellowcard.

About Novartis Gene Therapies
Novartis Gene Therapies is reimagining medicine to transform the lives of people living with rare genetic

diseases. Utilising cutting-edge technology, we are working to turn promising gene therapies into proven treatments. We are powered by an extensive manufacturing footprint, in capacity and expertise, enabling us to bring gene therapy to patients around the world at quality and scale.

Contact

UK Press office telephone number: +44 20 8481 8125
E-mail: NovartisGTUKPressOffice@Havas.com

Louise Strong, Corporate Communications, EMEA
Email: louise.strong@novartis.com
Telephone number: +44 7717 355336

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References

1. Govoni A, et al. Mol Neurobiol. 2018;55(8):6307–18.
2. Weildlich D, et al. Cost Effectiveness of Newborn Screening for Spinal Muscular Atrophy in England and Wales. medRxiv. 2023;28(5):715. doi: https://doi.org/10.1101/2023.02.09.23285715. *PREPRINT*.
3. Novartis, Summary of Product Characteristics (SmPC): Onasemnogene abeparvovec. 2023. Available at: https://www.medicines.org.uk/emc/product/11572/smpc/print#PRODUCTINFO. Date accessed: March 2023.
4. Finkel RS, et al. Observational study of spinal muscular atrophy type I and implications for clinical trials. Neurology. 2014;83:810–7.
5. NICE. Onasemnogene abeparvovec for treating spinal muscular atrophy. Highly specialised technologies guidance. 2021. Available at: https ://www.nice.org.uk/guidance/hst15/chapter/1-Recommendations. Date accessed: March 2023.
6. Landfeldt E, et al. Eur J Paediatr Neurol. 2019;23(3):347-56.
7. Kolb SJ, et al. Ann Neurol. 2017;82:883–91.
8. Anderton RS and Mastaglia FL., Expert Rev Neurother. 2015;15:895–908.
9. National Organization for Rare Disorders (NORD). Spinal Muscular Atrophy. Available at: http://rarediseases.org/rarediseases/spinal-muscular-atrophy/. Date accessed: March 2023.
10. Soler-Botija C, et al. Brain. 2002;125:1624–34.
11. Wang CH, et al. J Child Neurol. 2007;22:1027–49.
12. Novartis, Data on file.
13. Novartis, Data on file.

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