SMA EUROPE

Care for adults living with spinal muscular atrophy in the United Kingdom

The project is a collaborative effort and shared outcome of a partnership between SMA Europe and F. Hoffmann-La Roche Ltd, driven by their shared vision to improving care in the field of SMA. This report has been made possible with funding provided by Roche as part of this partnership.

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About the benchmarking report on SMA adult care

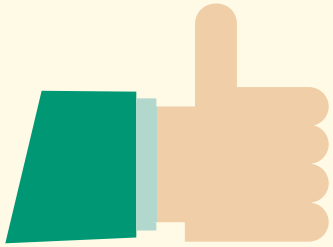
This short report complements the benchmarking report on SMA adult care published in 2024 to assess how care is provided for adults with spinal muscular atrophy (SMA) living in 22 European countries, as well as the OdySMA project, an SMA access atlas. The objective of this report is to provide an evidence-based resource for the SMA patient community to engage EU and national policymakers and increase awareness about the care challenges faced by adults living with SMA. It also features calls-to-action for policy modifications at both national and EU level, with a view to enhance care and improve the overall quality of life for adults living with SMA.

For more information explore the SMA Adult Care Benchmarking Report

Visit OdySMA website to find out more about the project

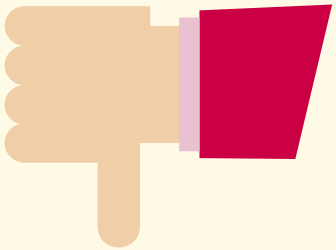
What it means to live as an adult with SMA in the United Kingdom

SOME GOOD NEWS



- A patient registry, which is co-sponsored by SMA UK, has been created, and collects data on diagnosis, overall condition, quality of life, other conditions, and participation in clinical trials.
- Both *nusinersen* and *risdiplam* are available to almost all adults living with SMA at no cost.
- Consultations with specialists such as nutritionists, pulmonologists and physiotherapists are available at no cost, though waiting times for appointments may be long and access varies geographically.

SOME BAD NEWS



- Despite the UK having a strong framework for care provision, there is a serious shortage of personal assistants (PAs) and variation in how much funding is granted to hire a PA, meaning that many adults living with SMA do not receive the care they are entitled to.
- Adults living with SMA are often subject to long waits for medical equipment. Even once the equipment is provided, they may have to wait several months for PAs to receive training on equipment use.
- In general, multidisciplinary teams (MDTs) are not available for adults living with SMA. This means there is a lack of multidisciplinary review and care coordination, often resulting in the uncoordinated and inefficient scheduling of appointments and a lack of access to some specialists.

Spinal Muscular Atrophy uk

SUPPORTING • INFORMING • ADVOCATING

“

We’re aware of adults who should be in receipt of care packages 24/7, but their care packages are never fully staffed, leaving informal carers having to step in and cover even when they are physically struggling to do so.

Representative from SMA UK

Overview of how the United Kingdom performed across the 19 indicators

Good | Reasonable | Not good | Poor | No information available

Below is a summary of how the United Kingdom scored across all indicators in this benchmarking exercise.

For more information about the scoring system please consult the main report, namely Annex C.

Healthcare systems organisation

1 Transition from paediatric to adult care services
2 Navigation in the healthcare system
3 Access to SMA treatment centres
4 Network of treatment centres
5 Patient registry
6 Standards of care (SoC) & treatment guidelines

Healthcare delivery

7 Multidisciplinary care
8 Shared decision-making
9 Continuity of care
10 Care team & caregivers training
11 Access to EMA-approved disease modifying therapies (DMTs)
12 Access to unauthorised disease modifying therapies (DMTs)

Governmental and peer support

13 HCP consultations reimbursement
14 Assistive equipment and devices support
15 Social, education and employment support
16 Life assistants/ professional caregivers
17 Informal caregivers
18 Rare disease policies and public funding for Patient Organisations
19 Patient Organisations' support

Deep dive into the 19 indicators and how the United Kingdom compares with other countries

SMA EUROPE

Healthcare systems organisation

1. Transition from paediatric to adult care services

There are no national transition care protocols for the transition from paediatric to adult SMA care in the UK. According to the healthcare professionals (HCPs) surveyed, although some protocols exist, they tend to be centre-specific, and their implementation varies [1]. SMA UK observed the existence of transition support roles in certain centres. Additionally, adults living with SMA who are seen in centres participating in the Adult SMA REACH network benefit from the network's facilitation of assessment data transfer in the transition from paediatric to adult care to minimise the administrative burden. To aid in the transition process, the organisation Alstrom UK has also developed an interactive website, T-KASH (Transition - Knowledge and Skills in Healthcare), targeting HCPs, but there is no indication of how many HCPs are aware of or use this resource [2].

70% of HCPs surveyed noted that there are re-eligibility requirements for adults living with SMA who have transitioned from paediatric care to ensure continued access to pharmacological treatments. However, with regard to the administrative burden associated with these requirements, 40% of the HCPs suggest this does not fall on the adult living with SMA and 20% said only some burden falls on the adult living with SMA, suggesting that it may be an administrative process that does not necessarily involve the adult living with SMA [1].

2. Navigation in the healthcare system

In case of a new diagnosis or if an adult living with SMA relocates, they would typically receive a referral to a neuromuscular centre through their general practitioner (GP). From there, they can be referred to other specialists or services. While GPs can also refer individuals to local HCPs, they usually do not initiate these referrals proactively.

In some specialised clinics, a neurologist may assume the role of care manager - contact person who support adults living with SMA in organising interdisciplinary consultations, taking over administrative/organisational tasks, and accessing government support measures – for adults living with SMA [3]. However, SMA UK observed that, in many cases, there is a lack of coordination and even if the centre has a range of specialists available, referrals to these specialists are not made consistently. For example, while some HCPs take the initiative to refer adults living with SMA to other specialists, such as dieticians, others only refer following a specific request from the adult living with SMA [2].

Healthcare systems organisation

3. Access to SMA treatment centres

22 out of 37 Regional Neuromuscular Centres (RNMCs), and one centre visited by a neuromuscular consultant twice a year, provide specialist care for adults living with SMA. Of the 23, one is in Northern Ireland, two in Wales, five in Scotland and 15 in England, meaning that some regions are not as well covered as others, but overall, there is a good coverage [4]. Some adults living with SMA are referred for care in general neurology clinics, which are more evenly distributed across the regions, but may not have specialist knowledge of SMA [2]. This was corroborated by the HCP survey results, with 50% of HCPs answering that fewer than half of all adults living with SMA in the UK are being monitored in SMA or neuromuscular disease (NMD) reference centres or specialist clinics.

The majority of the HCPs indicated an average waiting period of two to four months for an adult living with SMA's first appointment with an SMA specialist neurologist [1]. SMA UK has highlighted that in some cases, referral may initially be to a general neurologist and that the waiting period for this initial appointment can extend up to one year, followed by a further wait of several months for an appointment with an SMA specialist neurologist in an NMD clinic [2].

4. Network of treatment centres

A clinical network, Adult SMA REACH, has existed since 2020 with the objective of developing, implementing, and understanding the impact of standards of care, as well as implementing protocols for assessment of the impact of new treatments on the natural history of adult SMA. The network is coordinated by the Newcastle upon Tyne Hospitals NHS Foundation Trust. As of 2023, 20 National Health Service (NHS) organisations (such as regional or local Trusts and Boards) have begun participating in the network [5]. 90% of the HCPs surveyed were not aware of a formal network of SMA treatment centres, which may be due to this being relatively new or not yet involving all centres where adults living with SMA receive care [1].

Healthcare systems organisation

5. Patient registry

The UK SMA Patient Registry, co-sponsored by the patient organisation SMA UK and managed by the Institute of Genetic Medicine at Newcastle University, gathers medical and quality of life information from individuals living with SMA via regular patient-completed questionnaires [6] [7]. Notably, the NMD clinics are not involved in the registry's data collection [8].The data collected includes genetic reports, NHS/PPS numbers, diagnosis and conditions, family medical history, specific conditions such as scoliosis, patient-reported outcome measures (PROMs), hospitalisation details, and participation in clinical trials.

The UK SMA Patient Registry works closely with the Adult SMA REACH research project and network mentioned in the previous indicator, as well as with its paediatric counterpart SMA REACH UK, with the goal of systematising and harmonising data collection and integrating patient-reported data with clinical data for analysis.

6. Standards of care (SoC) & treatment guidelines

Adherence to the 2017 International Standards of Care for SMA is standard practice. However, a 2022 study which surveyed people living with SMA (73% of whom were adults) indicates some variance in the implementation of these standards across centres, such as access to different HCP specialists and interventions. Most survey respondents (85%) were located in England (35 out of 48 counties), but there was also representation from Scotland, Wales and Northern Ireland [9]. This discrepancy in implementation was echoed in responses from HCPs, with 40% stating that they are implemented only in some centres, while 30% said they are rarely implemented at all, suggesting that there may be significant differences between treatment centres where HCPs work [1].

There are currently no national guidelines for DMTs, however clinics in the UK use outcome measurement tools to assess the treatment efficacy for adults living with SMA receiving *risdiplam* or *nusinersen*. These tools gauge nutritional status, scoliosis, fractures, ventilation, and motor function. Specifically for adults receiving *nusinersen*, the focus of these tools revolves around measuring the impact on motor function [10] [11]. More than 50% of the HCPs identified several outcome measures which are in use, including the Hammersmith Functional Motor Scale-Expanded (HFMSE), the SMA Independence Scale Upper Limb Module (SMAIS-ULM) via caregiver reports, the Assessment of Caregiver Experience in Neuromuscular Disease (ACEND), the Six Minute Walk Test (6MWT), and the SMA-FRS scale [1].

Healthcare delivery

7. Multidisciplinary care

While some SMA treatment centres have MDTs dedicated to adults living with SMA, this is not universally available, with 20% of HCPs saying there are no MDTs available for adults living with SMA [1]. Additionally, a study examining access to standards of care for people living with SMA, in which 73% of the participants were adults, revealed that half of the subjects reported a lack of access to multidisciplinary care [9]. SMA UK confirmed this deficiency, noting a frequent absence of multidisciplinary reviews and a general lack of coordination of care, resulting in inefficient scheduling across multiple days or with long waits between appointments, impacting adults living with SMA's quality of life, time, and independence, and sometimes negatively affecting their engagement with the healthcare services.

In instances where MDTs are established, they are generally coordinated by an adult neurologist. SMA UK specified that these usually include NMD specialist neurologists, physiotherapists, occupational therapists or care advisors, speech and language therapists, and dieticians [2]. Additionally, HCPs who were aware of existing MDTs noted that they typically also include psychologists [1].

8. Shared decision-making

50% of the surveyed HCPs indicated that shared decision-making is an essential aspect of standard clinical practice involving all adults living with SMA. However, others noted its inconsistent implementation or reported limited engagement from adults living with SMA [1]. SMA UK shared a similar perspective, highlighting challenges stemming from infrequent appointments (which typically occur every six-12 months) and poor communication among specialists. The intermittent appointments and issues in communication often hinder the active involvement of adults living with SMA in decision-making processes. SMA UK pointed out that individuals who have longer-term relationships with a single HCP find it easier to engage in shared decision-making [2].

SMA UK has compiled a repository of SMA-related resources frequently accessed by HCPs to educate adults living with SMA. Nevertheless, they observed that many HCPs are unaware of the available resources. While adults living with SMA can access this information online, it may be perceived as overwhelming for some individuals [2].

9. Continuity of care

HCPs noted that regular follow-up appointments are deemed mandatory solely for adults living with SMA undergoing pharmacological treatment [1]. However, the frequency of follow-up varies across the four countries. For instance, adults living with SMA in Scotland are requested to complete surveys every six months, in addition to their regular appointments [2]. 50% of the surveyed HCPs mentioned the use of reminders which are sent to adults living with SMA before their specialist appointments [1]. In contrast, SMA UK stated that such reminder processes are generally not established. However, if medical appointments are missed, adults living with SMA might be warned that they will be discharged back to their GP [2].

Healthcare delivery

10. Care team & caregivers training

Some HCPs mentioned the existence of SMA training programmes, although not all were aware of them. These programmes include online continuous medical education provided by SMA centres of excellence, continuous medical education offered by pharmaceutical companies, and occasional training sessions at universities. However, 30% of HCPs stated that no such training is available [1]. Additionally, SMA UK highlighted that certain HCPs, like nurses, lack familiarity with the complexities of SMA [2].

Personal assistants (PAs) typically have access to training sessions focusing on operating medical equipment. Nonetheless, SMA UK pointed out that the wait for this training can often extend for several months, meaning that adults living with SMA may wait for months to be provided with the assistance they require [2]. It remains unclear whether there are any specific SMA-focused training programmes available for informal caregivers.

11. Access to EMA-approved disease modifying therapies (DMTs)

There are currently two Managed Access Agreements (MAA) based on recommendations of the National Institute for Health and Care Excellence (NICE) in place in England and followed in Wales and Northern Ireland. From 2019 for an initial period of five years, *nusinersen* is available in these countries at no cost for adults diagnosed with SMA types 1, 2 or 3 [12]. From 2021 until the results of the FIREFISH and SUNFISH trials are published, *risdiplam*▼ is available at no cost for adults of all ages living with SMA types 1, 2 and 3 or 1-4 copy numbers [13]. Access beyond 2024 will be dependent upon a reassessment.

In Scotland, *nusinersen* has been available at no cost to adults living with SMA types 2 and 3 in Scotland via the "ultra-orphan medicines pathway" since 2019 [14] and *risdiplam* has been available at no cost to adults living with SMA types 1, 2 and 3 or with 1-4 copies under the same pathway since 2022 [15].

12. Access to unauthorised disease modifying therapies (DMTs)

Over the past five years, seven clinical trials conducted by international pharmaceutical companies were open to adults living with SMA in the UK. These trials included SHINE [16] and DEVOTE [17] sponsored by Biogen for *nusinersen*, SUNFISH [18] and JEWELFISH [19] sponsored by Roche for *risdiplam*, another trial sponsored by Roche for *olesoxime* [20], RESILIENT sponsored by Biohaven for *taldefgrobep* [21], and SAPPHIRE sponsored by Scholar Rock for *apitegromab* [22].

Risdiplam was also made available in the UK through the Early Access to Medicines Scheme (EAMS) in 2020, including for adults living with SMA [23].

Risdiplam has been approved by the European Medicines Agency (EMA) for the treatment of 5q spinal muscular atrophy (SMA) in patients with a clinical diagnosis of SMA Type 1, Type 2 or Type 3 or with one to four SMN2 copies.
▼This compound is subject to additional monitoring. This will allow quick identification of new safety information. Healthcare professionals are asked to report any suspected adverse reactions via their national reporting system. See section 4.8 of the SmPC for details on how to report adverse reactions.

Governmental and peer support

13. HCP consultations reimbursement

The NHS offers free consultations with specialists, including nutritionists, pulmonologists and physiotherapists, if these are deemed necessary [24] [25].

14. Assistive equipment and devices support

Occupational therapists can arrange the provision of medical equipment for adults living with SMA through the NHS or local authorities at no cost [26]. However, in practice, delays in obtaining medical equipment are common, followed by further delays in training PAs to use it. Furthermore, the equipment provided varies between regions. For example, cough assist machines are provided in some regions but not others, meaning that adults living with SMA do not have equal access to medical equipment. Beyond what is provided by the NHS or local authorities, SMA UK assists individuals requiring additional funding by suggesting charities that offer grants [2].

With regards to home adaptations, persons living with disabilities are entitled to local authority funding [24] and Disabled Facilities Grants for adapting living situations for specific needs, such as installing ramps for wheelchair users [27]. However, these provisions vary regionally, with local councils in Scotland reimbursing essential equipment or adaptations up to 1,500 GBP (roughly 1746 EUR), while adults are required to contribute towards equipment and adaptations in Wales, and local trusts decide on a case-by-case basis in Northern Ireland [28]. According to SMA UK, even when non-medical equipment is provided, waiting times can be long, and housing adaptation grants are provided on a case-by-case basis, meaning that applications are sometimes rejected by the local authority [2]. Long waiting times can also mean that when the equipment is eventually provided, the results of assessments are no longer accurate as the individual's needs have changed.

The Personal Wheelchair Budget (PWB) funded by the NHS offers adults the choice of selecting the most suitable wheelchair for their needs. The PWB lasts for five years or longer unless there's a significant change in the individual's clinical condition [24]. However, SMA UK noted that the budget is insufficient in some cases, leading some adults living with SMA to use personal funds or seek charity funding. Furthermore, in some regions there are no longer routine service checks of wheelchairs, meaning that adults living with SMA have to seek maintenance themselves [2].

For workplace modifications, the Government's Access to Work grant covers special equipment or support services for individuals with disabilities in office settings to aid in daily tasks [29], however SMA UK noted that the waiting time for an assessment is up to three months. A mobility allowance of up to 71 GBP (roughly 82 EUR) per week is also provided to adults with disabilities via the Personal Independence Payment (PIP), mentioned below, and the Charitable Motability Scheme allows for the leasing of adapted cars, powered wheelchairs, or scooters using this allowance [30] [31].

Governmental and peer support

15. Social, education and employment support

Universal Credit is financial aid provided to individuals on a low income, unemployed, or unable to work [32]. Individuals with disabilities preventing them from working or preparing for work are eligible for an additional amount of Universal Credit [33] as well as the New Style Employment and Support Allowance (ESA) [34]. While assessment is only required once for persons qualified by their GP as having a 'limited capacity for work-related activity' and 'unlikely to ever be able to move into work', some adults living with SMA may be qualified as having 'limited capability for work', which requires them to periodically undergo burdensome re-assessments [35]. In addition to Universal Credit, adults with disabilities are eligible for a PIP – or Adult Disability Payment in Scotland – which includes a mobility component (mentioned above) and a daily living component of between 68.10 GBP (roughly 79.27 EUR) and 101.75 GBP (roughly 118.45 EUR) per week if they require support in carrying out everyday activities [30] [36]. However, as this is not a substantial sum, many adults living with SMA are supported by family members who take on the role of informal caregiver instead [2].

The Government's Access to Work grant, mentioned above, can finance necessary support for the workplace and transportation [37]. Employers are also obligated to make reasonable adjustments to ensure that persons living with disabilities are not significantly disadvantaged [38].

Individuals with disabilities have access to funding for education and an additional disability allowance via Student Finance England, Student Finance Wales, Student Awards Agency for Scotland or Student Finance NI [39]. The amount and eligibility criteria for this funding varies across the four countries. In England, an education, health, and care plan (EHCP) is also available from childhood up to the age of 25, identifying educational, health, and social needs and outlining additional support to address these needs [40], meaning that adults can access supported education until the age of 25. However, Wales, Scotland and Northern Ireland's equivalent support plans only extend to college or sixth form, which typically end at the age of 18.

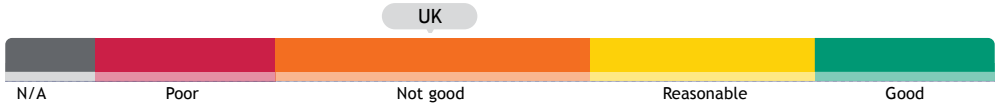
Governmental and peer support

16. Life assistants/ professional caregivers

In England, Wales and Northern Ireland, if an HCP determines that an adult living with SMA requires primary healthcare, the NHS is obliged to fund a healthcare package known as NHS Continuing Care. This package may encompass care provided in the individual's home or within the community [24]. In cases where the local authority is unable to offer a care package, some persons living with disabilities qualify for 'direct payments,' enabling them to hire a PA directly [41]. Individuals with long-term illnesses may also be eligible for a 'personal health budget' through the NHS, allowing them to manage their healthcare, treatment, equipment, and personal care [42].

In Scotland, Free Personal Care is provided by the local authority following an assessment of the adult living with SMA's needs by the authority's social work department. This can take the form of a direct payment from the local authority to the individual to cover the required support, payment by the local authority itself for a service provided by a professional caregiver of the individual's choice, or selection and payment of the professional caregiver by the local authority [43].

While the four countries have programmes in place to provide financial support for adults living with SMA to access PAs, SMA UK notes that it is a "postcode lottery" regarding the amount of funding granted to adults living with SMA to hire a PA. SMA UK also highlight that in practice, there is a significant shortage of government-appointed carers due to lack of budget and low salaries. This means that even when adults living with SMA qualify for 24/7 care, agencies struggle to provide enough carers to meet demand. Furthermore, as mentioned above, even when adults living with SMA are allocated a PA, they may have to wait up to nine months for the PA's medical equipment training [2].



17. Informal caregivers

The Carer's Allowance has specific criteria that must be met, such as providing care for an adult with a disability for a minimum of 35 hours per week and earning no more than 132 GBP (roughly 153.60 EUR) per week [44]. There is also a Carer Premium that can supplement other social benefits [45]. However, SMA UK notes that in practice, many informal carers may not qualify for financial support, and the assessment process can often be unclear [2]. In addition to these allowances, informal caregivers are entitled to Carer's Credit, which fills gaps in their National Insurance record that could otherwise affect their eligibility for the State Pension [46].

Adults living with SMA have the option to hire family members as PAs, but regulations stipulate that they cannot reside in the same household. While there may be some exceptions to this rule on a case-by-case basis [47], SMA UK note that in many cases family members are not granted any social benefits for informal care, despite this often being an adult living with SMA's only option, given the shortage of PAs [2].

Governmental and peer support

18. Rare disease policies and public funding for Patient Organisations

The England Rare Diseases Action Plan of 2022 [48], the Scotland Rare Disease Action Plan of 2022 [49], the Northern Ireland Rare Disease Action Plan of 2022/2023 [50], and the Wales Rare Disease Action Plan 2022-2026 [51], which implement the 2021 UK Rare Diseases Framework, do not include any specific actions related to SMA or NMDs as the Framework applies to rare diseases as a whole. However, the plans acknowledge the managed access agreements (MAAs) in England and the Ultra-Orphan Pathway in Scotland which facilitate access to DMTs. While partnerships with patient organisations are mentioned in the annual plans, SMA Europe member organisations are notably not allocated public funding.

19. Patient Organisations' support

SMA UK collaborates closely with adults living with SMA to assist them in navigating life with the disease. They advocate for their access to treatments and essential services while aiding in securing grants for equipment purchases. Their website offers comprehensive resources catering to adults living with SMA and their families, fostering a platform for discussions, as well as sharing experiences and knowledge [2].

Muscular Dystrophy UK provides support to adults living with SMA and various other NMDs. Their services include offering information, providing advice, granting assistance for equipment purchases, facilitating group meetings, and organising conferences [2].

Conclusion

SMA Europe, with the endorsement of SMA UK, calls on policymakers, healthcare systems, and the medical community to take coordinated action, in collaboration with the SMA patient community, to empower adults living with SMA in the UK to achieve meaningful health outcomes and quality of life as well as to have an independent and fulfilling life. We invite key stakeholders to, among other actions:

Revisit the personal assistant scheme to ensure that it is appropriate and sufficient to address the needs of adults living with SMA, without the significant regional variation which results in a ‘postcode lottery’ in access to funding and support.

Address challenges in access to assistive and medical devices and improve training for personal assistants on the use of those, to better support adults living with SMA in their daily activities.

Develop an action plan to improve access to multidisciplinary care, including coordination among different specialists/medical disciplines.

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