SMA EUROPE

Care for adults living with SMA in Europe: a benchmarking report

Find out more

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Acknowledgements

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.

This benchmarking report has been made possible thanks to the invaluable contributions and support provided by several organisations and individuals, namely the Expert Advisory Group, the SMA Europe member organisations, the healthcare professionals that responded to the survey, Weber Shandwick Brussels, and Hall & Partners. Their commitment to advancing research has been instrumental, and their contributions are greatly appreciated.

During the course of this report's development, we were deeply saddened by the loss of our esteemed Expert Advisory Group team member, Clare Gray, whose invaluable contributions continue to inspire us even after her untimely passing. This report is dedicated to the her memory. Clare's remarkable commitment to advocacy was an inspiration, and her contribution leaves a lasting legacy.

Disclosure

This benchmarking report is not intended to serve as a comprehensive or scientifically exhaustive assessment of the care provision for adults living with spinal muscular atrophy (SMA) throughout Europe. For more information about the full methodology and its limitations, please refer to the Annexes.

We recognise that this benchmarking report captures only a snapshot of the landscape across 23 countries. Our findings are based on a rapid literature review, an online structured survey completed by healthcare professionals, and individual interviews with one to two patient representatives from SMA Europe member organisations.

Should you have any inquiries, requests for further information or wish to express any concerns or comments, we encourage you to reach out to SMA Europe at: secretariat@sma-europe.eu. Your input is invaluable to our ongoing efforts to enhance our understanding and advocacy in the field.

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1. Foreword from SMA Europe CEO and President

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I am honoured to welcome you to this benchmarking report, a testament to our collective commitment to drive progress and enhance the lives of those living with SMA. As the mother of a child living with SMA and President & CEO of SMA Europe, advocacy for SMA has been a personal and professional calling, and I am deeply gratified to share this significant milestone with you.

At SMA Europe, we are committed to enhancing the healthcare systems, shaping policy, and improving access to care and treatment for people living with SMA. These are not just abstract ideals but the pillars that bolster our relentless pursuit of a better tomorrow.

Today, three disease-modifying therapies have been developed – *nusinersen*, *risdiplam* and *onasemnogene abeparvovec*. But not everyone has access to them, and they are no cure for SMA. The daily objective, therefore, rests on stabilising the progression of SMA and managing its symptoms through provisioning the highest quality care. Moreover, despite recent progress in both pharmacologic and non-pharmacologic interventions, persistent gaps in treatment and care remain unmet for those living with SMA.

Acknowledging the pressing need to address the unique challenges faced by adults living with SMA, as described in the 'Why a benchmarking report' section, SMA Europe has taken a decisive step forward by establishing an adult committee. This move underscores our unwavering commitment to inclusivity and ensuring that no one within our community is left behind.

We took a major step forward in early 2023 with the launch of the OdySMA access atlas, which systematically collects real-time data to visualise and centralise knowledge around access challenges across Europe. The OdySMA initiative provides a clear platform for advocacy and action. The addition and integration of this adult care benchmarking report will serve as a valuable supplement to this important dataset.

So, what does this benchmarking report entail and why should stakeholders invest their time in its content? Within these pages, you will discover a wealth of insight into the current care landscape for adults living with SMA across Europe. You will also find calls-to-action and best practices to address the unmet needs of adults living with SMA.

Yet this report is more than just a compilation of facts and figures; it is a powerful advocacy tool, representing an opportunity to amplify our voices and advocate for the needs of adults living with SMA. This part of our community has long been underserved and deserves nothing less than comprehensive care and support.

As we embark on this journey together, I extend my heartfelt appreciation to all who have contributed to this report, whose commitment is the driving force behind our progress.

In closing, I invite you to join us in transforming the insights and policy recommendations of this benchmarking report into actions that will resonate across Europe. Together, we can create a future where all those living with SMA, regardless of age, receive the optimal care, support and recognition they rightly deserve. All together. One goal.

Dr. Nicole Gusset
President & CEO, SMA Europe

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As adults living with SMA, we view this benchmarking report as crucial because it sheds light on the state of care for adults living with SMA in Europe, highlighting key areas for improvement. We expect the report to serve as a catalyst for policy changes and actions that will enhance the quality of life of all adults living with SMA by addressing critical gaps in care provision.

The SMA Europe Adult Committee

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Foreword by MEP Stelios Kympouropoulos

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I am very glad to be writing these words about the study in hand.

One of my credos in politics is that our policies and laws should be based on sound data.

The present study does exactly that by shedding a light on the state of care for adults living with SMA in Europe.

Therefore, it makes up for a significant gap as it really puts the persons with SMA at the centre of decision-making. By being human-oriented, the study enables all of the stakeholders to consider these persons as complete individuals with rights in every aspect of life and not only as SMA patients.

At the same time, it comprehensively maps practices and divergences across EU Member States, as well as highlights practice examples and key areas for improvement.

In this respect, it is a very meaningful contribution towards enabling adults living with SMA to live and exercise their right to live independently.

But the report also comes at a very timely juncture.

On 6-9 June 2024, European citizens will go to vote and elect their representatives at the European Parliament.

Major changes will also be made at the leadership of the main EU Institutions.

Against this backdrop, I am convinced that the study in hand needs to feed into the reflection stage the European Institutions are getting into.

A reflection stage about how the next mandate's social and healthcare policies will reflect the needs of people with rare diseases and in particular, SMA patients.

And I sincerely look forward to ''participate'' in this debate towards improving care provision for adults living with SMA throughout Europe.

Stelios Kympouropoulos
Member of the European Parliament
(European People's Party, Greece)

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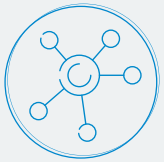
3. Calls-to-action

SMA Europe's recommendations for action

SMA Europe, with the endorsement of the Expert Advisory Group involved in this project, is calling for urgent action on 8 areas to be taken by key international and national stakeholders to collectively addressing the persistent challenges preventing adults living with SMA from living longer, healthier, more independent and fulfilling lives.

International recommendations	National recommendations		
 Developing updated European or international adult care and treatment guidelines	 Creating and/or strengthening a collaborative national network of SMA/NMD adult treatment centres	 Prioritising funding to enable accessibility to existing treatment centres	 Revisiting the support framework for persons with disabilities to ensure it is holistic and person-centric
 Reaching alignment on data collection from disease / patient registries	 Enabling access to appropriately funded and resourced multidisciplinary care for all adults living with SMA	 Addressing inequalities in access to pharmacological treatment	 Addressing insufficiencies in formal and informal assistance

These calls-to-action are further detailed in the following pages.

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Developing updated European or international adult care and treatment guidelines

We invite leading researchers and clinicians across Europe to join forces with SMA patient organisations and the SMA Patient community to define recommendations and protocols that address the specific needs of adults living with SMA. These should:

- be developed in the context of/coordinated by networks such as: European Neuromuscular centre (ENMC), European Reference Network on NMDs (EURO-NMD), or TREAT-NMD.
- cover all areas applicable across countries (e.g. multidisciplinary care, pharmacological treatment, outcome measures, training recommendations for physiotherapists and other HCPs, as well as caregivers).
- call on the national medical community to work on national chapters that should be country-specific (e.g. transition care).
- call out the current research needs and priorities for further advancing and improving diagnosis, care and treatment for adults living with SMA.

Reaching alignment on data collection from disease / patient registries

We invite relevant stakeholders across Europe – from leading researchers, to clinicians, to pharmaceutical industry, to health insurance organisation, to regulators – to, in collaboration with SMA patient organisations, develop guidance on e.g. the priority data to be collected (including treatment outcomes, clinical history and natural history data), ensuring patient-relevance of the data sets collected, how it should be collected, by whom, and what incentive systems should be put in place to incentivise participation. Discussions should focus on alignment across countries but also aim for a future EU-wide patient registry, or inter-operable registries, for all those living with SMA, including adults.

National recommendations

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Creating and/or strengthening a collaborative national network of SMA/NMD adult treatment centres

We invite leading researchers and clinicians in each country, in collaboration with local SMA patient organisations and patient communities, to develop and strengthen an adult treatment network for SMA. With the right endorsement and support from healthcare systems, such networks should play a key role in:

- developing national chapters that are to complement updated European or international adult care and treatment guidelines
- generate data on disease management/care best practices for adults living with SMA, to back up revised SoC developed on the basis of expert opinions
- encouraging sharing of expertise, case studies and information between treatment centres,
- building capacity in centres delivering adult care services,
- providing training opportunities for HCPs (inc. physiotherapists and local nurses),
- providing necessary medical training for life assistants,
- collecting and recording data on relevant registries, and
- enabling research efforts (that would not be possible in individual centres).

Enabling access to appropriately funded and resourced multidisciplinary care for all adults living with SMA

We invite health authorities to develop an action plan to implement and fund (better) access to multidisciplinary care, in line with updated European or international adult care and treatment guidelines once they are developed, regardless of where adults living with SMA are based. This could happen, depending on the organisation of the healthcare system, in the form of, e.g.:

- Creation of a hub-and-spoke model, whereby treatment centres coordinate with primary care;
- Creation of the role of a care manager/coordinator that acts as a point of contact for adults living with SMA.
- Introduction of tele-health systems, such as virtual consultations, to facilitate regular follow-up appointments between in-person consultations

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Prioritising funding to enable accessibility to existing treatment centres

We invite relevant ministries make funding available for (wheelchair-accessible) transport and facilities as well as for reimbursement of other expenses related to visits. For example, in countries where long-distance travel is often necessary, funding could also be provided for overnight stays for adults as well as for their carers. Where public transport is not accessible or not available, funding could be provided to treatment centres to charter vehicles that facilitate travel to and from the treatment centres.

Addressing inequalities in access to pharmacological treatment

We invite national medicines authorities to address current inequalities in access to pharmacological treatment, which would include eliminating any existing barriers restricting adults' access to available disease modifying therapies, such as age cut-offs, the need to have started treatment as a child, or restricting eligibility for treatment to certain SMA types.

Revisiting the support framework for persons with disabilities to ensure it is holistic and person-centric

We invite relevant ministries, including those focusing on health, social, employment and education policies, to collaborate in ensuring a more holistic and person-centric support framework for persons living with disabilities, including adults living with SMA, to ensure existing policies enable their independent living.

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Addressing insufficiencies in formal and informal assistance

We invite relevant ministries to:

- create and maintain life/personal assistant schemes that are appropriate and sufficient to address the needs of adults living with SMA. A public sector platform should be created to bring together these life/personal assistants and professional caregivers in view of providing regular training and certifications, including tailored to the disease areas and different disability types they are working on.
- recognise informal caregivers as carers and an integral part of adults living with SMA's support system, and provide them with the appropriate financial, social, and employment support, to alleviate the burden placed on them and their families.

Together, we can drive meaningful change and create a better future for all adults living with SMA. We call on you to express your support for the recommendations in this report and urge key stakeholders to turn these recommendations into actionable outcomes. Let's make a difference today.

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Key gaps identified in this report

As described in the 'Why a benchmarking report' section, with the increasing availability of innovative treatments that improve quality of life and survival rates, as well as the number of adults living with SMA being set to grow, it remains critical to provide age-appropriate, comprehensive and integrated medical care to adults living with SMA as well as strong social and support systems. This will empower adults living with SMA to achieve meaningful health outcomes and quality of life as well as to have an independent and fulfilling life.

Further explored along the 19 chapters that put the spotlight on each of the indicators analysed, the findings of our report underscore the presence of persistent gaps on how care and support are provided to adults living with SMA across the 22 European countries analysed, that demand immediate attention and targeted interventions. These gaps, some of which are described below alongside their impact on adults living with SMA, represent the foundation of the calls-to-action put forward in this report

	The challenge	The impact on adults living with SMA
Organisation of healthcare systems	Broad lack of standardised national protocols for the transition from paediatric to adult care services or, even if they exist, inconsistent implementation across SMA treatment centres.	The experience of adults living with SMA differs not only between countries but also within them, with some transitions being well-structured and smooth while others are complex and challenging, with adults becoming lost from the medical system.
	Input into existing registries is often done on a voluntary basis and the type of data collected differs from country to country.	There is no alignment between existing registries, preventing collation and comparison of data to help further understanding of e.g. the natural history or treatment benefits.
	In general, countries rely on the 2017 international standards of care or national guidelines focusing mainly on care best practices for children.	When SoC focus on best practices for children, they may overlook important considerations for adults - from hypertension, to hormonal issues, to sexual health - thereby leading to suboptimal care and a negative impact on quality of life.

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	The challenge	The impact on adults living with SMA
 **Healthcare delivery**	In many countries, even when having access to the different specialists they may need, adults with SMA lack the advantage of coordinated day-to-day care through a formal multidisciplinary care team within their treatment centre.	The quality of care is impacted as the different specialists do not communicate with each other and do not develop a joint plan for improving care and health outcomes for each individual. There is also no coordination to optimise time spent in the healthcare system.
	Shortcomings persisted in many countries, for adults trying to locate HCPs with specialist knowledge of their SMA, especially local nurses and physiotherapists.	Training for HCPs caring for adults living with SMA is extremely important to ensure they remain up to date with the latest developments and optimal disease management strategies. In its absence, adults living with SMA can experience a “postcode lottery” for accessing optimal care.
	Despite the situation changing rapidly, some countries’ eligibility criteria for access to DMTs are more restrictive than the EMA approved indication.	Restrictive criteria create persistent and significant inequalities in access to pharmacological treatment between (and sometimes within) countries, with adults in some countries having very limited access to existing treatment options.

	The challenge	The impact on adults living with SMA
 **Governmental and peer support**	Governmental support measures for persons with disabilities – including those for non-medical and medical assistive devices, social, employment, and access to education - vary widely across Europe, and sometimes within countries.	Strong support systems are key to empowering adults living with SMA, and their caregivers, to live an independent life and fulfil their personal goals. When support systems are not holistic and person-centred, they may lead to adults not being able to carry out daily activities or be professionally active, as well as to social vulnerability.
	Despite general availability of schemes for accessing life assistants, challenges persist in many countries, including inadequate financial support, restrictions on the number of hours or days permitted, and a shortage of life assistants/ professional caregivers.	The significance of life assistants lies not only in enhancing the independence of adults living with SMA, but also in relieving the burden on their families and friends. Any gaps in this support can have far-reaching effects on the physical and mental health of everyone involved.
	Regrettably, there are still countries where limited to no support is extended to informal caregivers, including family members or friends.	When existing support for informal caregivers is insufficient, a significant financial and emotional burden is placed on the adult living with SMA and their family This is especially important in countries where there are challenges in access to life assistants, who very much complement the role of the informal caregivers.

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4. Why a benchmarking report[1]

SMA has historically been considered a children's disease. Yet today, adults make up an estimated 50% of the population. And with the increasing availability of innovative treatments that improve quality of life and survival rates, those numbers are set to grow. [1] [2]

It remains crucial not to overlook the needs of those living with SMA in adulthood. [1] Yet, many essential initiatives focus predominantly on the needs of children living with SMA, leaving adults, and those transitioning into adulthood, relatively neglected. [3] [4]

The medical and patient communities see an opportunity to enhance national healthcare systems and support structures for adults living with SMA by: [1]

- Providing age-appropriate and comprehensive care that delivers the most meaningful health outcomes and quality of life for adults living with SMA.
- Establishing more integrated pathways that enable adults living with SMA to optimally manage their multifaceted healthcare needs.
- Strengthen social and financial support systems that empower adults living with SMA and their caregivers to fulfil their personal goals.

Within this context, for the first time, this report delivers a comprehensive evaluation of care and support provision for adults living with SMA in Europe, as well as individual country snapshots of performance against the different indicators. It helps identify best practice examples, as well as common areas of weakness, ultimately leading to recommendations to improve care and the overall quality of life for adults living with SMA. The report focuses not only on medical care and treatment but also on broader support provided to adults living with SMA so that they can live independently.

The ultimate objective is to provide the SMA patient community with an evidence-based resource that can be used to engage EU and national policy-makers, increase awareness of the care challenges faced by adults living with SMA, and to call for policy change.

[1] Benchmarking is defined as the process of measuring products, services, and/or processes provided/ undertaken by different organizations/stakeholders, helping make a comparison between them.

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5. Short description of methodology

SMA Europe's membership extends over 25 countries and is made up of 27 patient organisations. This benchmarking study analyses 23 of these[2], and follows a mixed data collection approach, as follows:

- The process began with a **rapid literature review**[3], making use of publicly available sources of information and data specific to each country.
- Simultaneously, an **online structured survey**[4] was conducted, targeting clinical experts who treat adults living with SMA in the respective countries.
- Finally, **semi-structured phone interviews**[5] took place with a local patient organisation in each country.

After completing the data collection phase, the obtained data was compiled and consolidated into a comprehensive report.
Throughout the study, to ensure input from the SMA community, an Expert Advisory Group (EAG) was created by Roche and SMA Europe, bringing together a representative group of adults living with SMA as well as healthcare professionals involved in their multidisciplinary care, covering the various regions of Europe where the research was being carried out.

Categorised under three overarching themes, the results were mapped against 19 indicators outlined in the next page. These were defined by SMA Europe, Roche and the EAG.
For more information about the process, please refer to the detailed methodology.

Limitations

The study's methodology has limitations that warrant consideration. These include: a relatively limited literature review, given the choice to opt for a rapid literature review instead of a systematic literature review, potential disparities in survey responses from healthcare professionals within the same country; and the limitation of conducting only one semi-structured interview per country (most of which were in English) with patient representatives.

A fuller analysis of these limitations is available in the Annex section of this report and addressing them in future iterations would enhance the study's credibility and contribute to a more robust body of knowledge in the field.

Map of member organisations that are part of SMA Europe

[2] Excluding Turkey and Cyprus - which were not covered because they became SMA member organisations after the project began.
[3] Rapid reviews are a form of knowledge synthesis in which components of the systematic review process are simplified or omitted to produce information in a timely manner.
[4] Structured surveys are a systematic and organised method of data collection used to gather information from a sample of individuals or entities in a standardised and consistent manner consisting in a set of predetermined questions, standardisations of questions and response options, and quantitative data which can be analysed statistically.
[5] Structured surveys are a systematic and organised method of data collection used to gather information from a sample of individuals or entities in a standardised and consistent manner consisting in a set of predetermined questions, standardisations of questions and response options, and quantitative data which can be analysed statistically.

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6. Spotlight on the 19 indicators

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a. Area 1: Organisation of healthcare systems

Looks at how a country organises healthcare and implements internal procedures to deliver healthcare to the individuals living with SMA to promote, restore or maintain health.

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

b. Area 2: Healthcare delivery

Looks at the most tangible aspects of the health system as experienced by adults living with SMA.

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)

c. Area 3: Governmental and peer support

Looks at the access to different types of support that are provided by the government, beyond medical care, to foster independent living of adults with SMA., as well as patient organisation 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
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a. Area 1: Organisation of healthcare systems

Transition from paediatric to adult care services

Key findings

- Standardised national protocols for the transition from paediatric to adult care services are lacking in the great majority of countries. Where they do exist, their implementation varies from centre to centre.
- Most often, transition protocols are established by each SMA treatment centre, and they may not always exist.
- In a significant number of countries, there is a need for a reassessment of re-eligibility to access pharmacological treatments that is specific to the transition period. This occurs in addition to the reassessment, which is done on an ongoing basis for all individuals living with SMA.

Situation analysis[6]

The landscape of transition care for individuals living with SMA across Europe varies greatly. Their experiences differ not only between countries but also within them. Some transitions are well-structured and smooth, while others are complex and challenging, often leaving the patient feeling disconnected and unsupported.

Transition protocols to guide the transition to adult care are non-existent across five countries: Ireland, North Macedonia, Russia, Serbia, and Ukraine. While a few countries like Denmark, Finland, France, Hungary, Poland, Switzerland, and the Netherlands do have national protocols, with the exception of the Netherlands their implementation across SMA treatment centres appears inconsistent, according to the majority of surveyed healthcare professionals (HCPs). In France, the national diagnostic and care protocol for SMA includes recommendations on transition care but establishes that each centre has its own transition care protocol to ensure a smooth transition. Meanwhile, the 11 remaining countries assessed took a more decentralised approach, whereby transition protocols are implemented on a centre-by-centre basis. In both instances, adults living with SMA can have a completely different experience, depending on the centre they attend.

In some countries, such as the Czech Republic, North Macedonia, and Ukraine, individuals living with SMA must transfer to a different centre when transitioning. This means they start being followed by an entirely different care team. In contrast, in countries like Denmark, Germany, Italy, and the Netherlands, where paediatric and adult care services are typically organised together, or coexist in the same SMA treatment centre, transitions are generally smoother.

Patient advocacy for improving the care transition has been identified in various countries. For instance, in Greece, MDA Hellas has initiated the establishment of integrated units within hospitals, to facilitate the transition of individuals living with SMA and other neuromuscular diseases (NMDs) from paediatric to adult care services. In Serbia, the National Organisation for Rare Diseases has advocated for collaboration between paediatric and adult doctors to provide joint care for individuals living with SMA aged 16 to 18.

Typically, when transitioning, there is no need for a reassessment of eligibility for access to pharmacological treatment. Instead, ongoing assessments to monitor the use and effectiveness of disease modifying therapies (DMTs) are required, regardless of whether the individual living with SMA is transitioning or not. However, according to the majority of surveyed HCPs, this assessment, with different levels of administrative burden to the adults living with SMA, appears to be necessary in nearly half of the countries, such as Denmark, France, Greece, Hungary, North Macedonia, Romania, Russia, Spain, Switzerland and the UK. This may make the transition more difficult and stressful for adults living with SMA, even more so if there is an

[6] The observations and conclusions rely largely on the perspective of surveyed HCPs across all countries, which often varied. Regarding the reassessment for re-eligibility for access to pharmacological treatment, this variability might mean that HCPs mentioning the need for reassessment were referring to the ongoing need rather than a need specific to the transition time. Additionally, specific requirements may vary among healthcare centres and regions where they are based.

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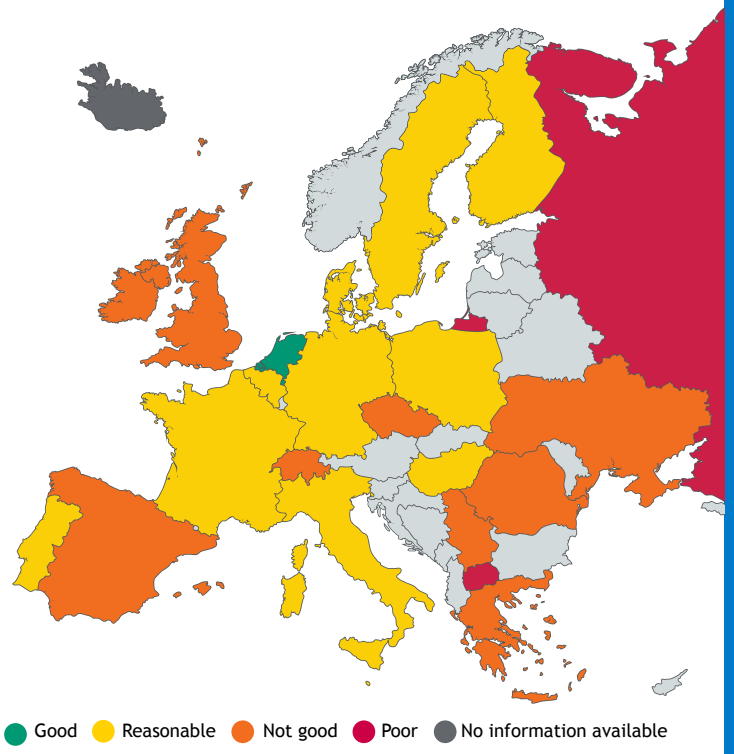
Best practices

In Poland, the Ombudsman for Patients' Rights, in collaboration with a group of medical and patient community experts, introduced a comprehensive framework to facilitate the smooth transition from paediatric to adult care services for individuals living with rare diseases. This framework is built on three fundamental principles: strategically placing the patient within the healthcare system, understanding the unique aspects of adolescent conditions and associated developmental transitions, and ensuring effective patient education.

In Denmark, at university hospitals, a formal transition process is in place, in which a paediatric neurologist typically attends the first adult care meeting.

European map



A recap of sub-indicators analysed under this indicator

- **Existence of transition care protocols/policies**

The scoring system assesses the existence and implementation of transition care protocols and policies within the healthcare system, ranging from a high score of four, signifying well-implemented national protocols, to a low score of one, indicating the absence of any transition protocols, whether national or centre-specific.

- **Need for requalification for access to pharmacological treatment**

The scoring system assesses the need for those living with SMA, specifically when transitioning from paediatric to adult care services, to go through a reassessment procedure to be able to continue accessing pharmacological treatment as well as the level of administrative burden placed on the individual. A high score of four, signifies the lack of need for those living with SMA to undergo such a procedure, while lower scores imply a need and a greater administrative burden.

Navigation in the healthcare system

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Key findings

- In half of the countries covered in the report, adults living with SMA can easily navigate the healthcare system, rarely encountering any barriers.
- In the great majority of countries covered, there is usually no care manager responsible for following the care of adults living with SMA in the healthcare system.
- In general, there is more support available to access neurologists and other specialists, compared to local HCPs such as physiotherapists.

Situation analysis

The findings of the report show a significant variation in the ease of navigating the healthcare system. In almost three quarters of the countries, adults living with SMA can easily navigate the healthcare system without significant barriers, or only face one gatekeeper to care, such as a general practitioner (GP), from whom they must receive a formal referral.

Additionally, a coordinating role for care, such as a care manager, exists only in Switzerland and France, and occasionally in Germany and the UK, in specific centres. The existence of a care manager – a contact person who support adults living with SMA in organising interdisciplinary consultations, taking over administrative/organisational tasks, and accessing government support measures – greatly supports adults living with SMA in navigating the healthcare system, organising their care and living independently. The existence or lack of this role doesn't, however, exclude the existence of someone (e.g. neurologist) who coordinates multidisciplinary care.

Roughly one-third of countries have country-wide formal pathways for referrals to an SMA or NMD specialist, or a general neurologist, meaning that once an SMA diagnosis has been documented, people living with SMA are referred to a SMA specialist/SMA treatment centre by the referring GP, family doctor or general clinic. Centralised formal pathways prevent adults living with SMA from having to spend a significant amount of their time understanding who to contact, and reduces uncertainty about the next steps in the care pathway.

The administrative burden varies between countries. In about a quarter of the countries, HCPs are not involved in scheduling specialist appointments as part of the referral process, meaning that adults living with SMA either need to initiate contact with other specialists without a formal referral or are required to obtain a referral document before initiating contact. In a small number of countries, adults living with SMA are almost completely unsupported in navigating the healthcare system. For example, in Italy and Serbia, unless adults living with SMA are referred to nearby specialist SMA centres after a diagnosis has been documented, they may have to search for specialists and initiate appointments themselves.
Regarding referrals by GPs or neurologists to other specialist HCPs, in nearly half of the countries covered by the report, formal processes for these referrals only exist within centres, meaning that adults living with SMA may face administrative hurdles if they need to consult with a specialist outside of the referring centre. Only in Portugal and Finland is there a nationally available formal pathway that enables adults living with SMA to access specialist consultations even if the specialist is not affiliated with the healthcare centre that made the referral.

When adults living with SMA have access to specialists without any significant barriers, this usually also applies to access to local HCPs, such as physiotherapists or local nurses. However, in some countries there are more barriers to accessing local HCPs compared with other specialists. For example, in Portugal, although there are formal referral pathways for adults living with SMA to access specialists, accessing local HCPs cost-free requires a referral document, and appointments might need to be pursued independently by adults living with SMA through private facilities. Administrative hurdles to care, such as physiotherapy, can have a negative impact on adults living with SMA, who may have to seek alternatives at a higher cost or rely on informal caregivers to provide this care instead.

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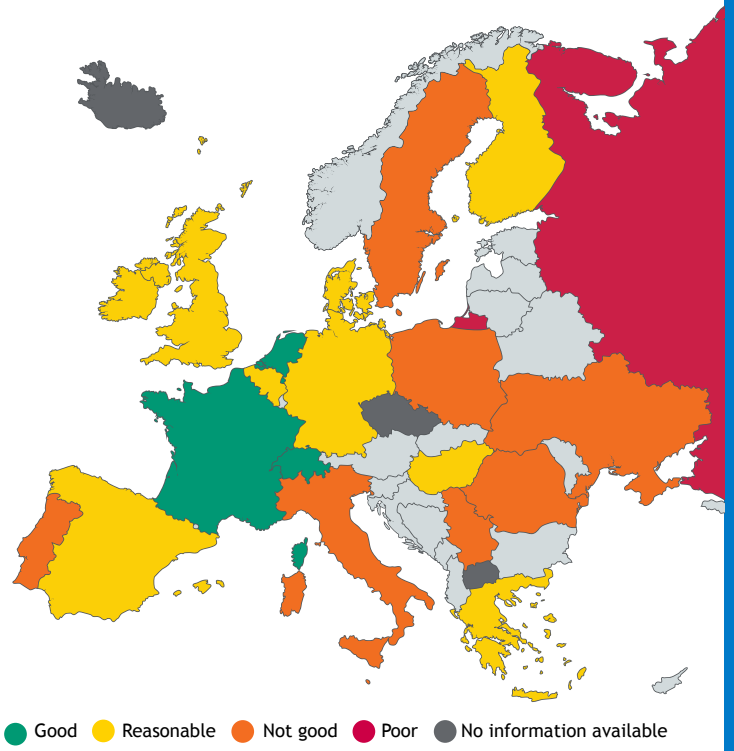
The country where adults living with SMA experience the greatest burden in navigating the healthcare system is Russia, where adults living with SMA have easy access only to local policlinics and general outpatient medical centres. Finding an SMA-specialised medical facility with access to a specialised neurologist is left completely to the individual.

Best practices

In the Netherlands, when a person is diagnosed with SMA or another NMD, they are automatically referred to the Netherlands SMA Centre for treatment and to a NMD reference centre for day-to-day care.

In Switzerland, a case care manager who manages referrals to specialists, is assigned to adults living with SMA during their first appointment in a centre. Adults living with SMA also have the flexibility to provide their own preferred contacts for referrals to specialists.

European map



A recap of sub-indicators analysed under this indicator

- **Ease of navigating the healthcare system**

This indicator assesses how easy it is for adults living with SMA to access the care they require. Higher scores indicate fewer barriers to referrals and scheduling appointments, while lower scores suggest significant barriers to navigation of the healthcare system.

- **Existence of care manager**

This indicator evaluates whether there is a designated care coordinator for adults living with SMA. Higher scores indicate the presence of such a coordinator, while a lower score suggests the absence of a care manager.

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Access to SMA treatment centres

Key findings

- In the majority of countries, half or more of the SMA treatment centres cater to adults living with SMA, with a relatively even geographical distribution of NMD or SMA-specific treatment centres.
- However, challenges arise in few countries where adults living with SMA may have to rely on general neurology services or less specialised care. These challenges include a very limited number of NMD clinics offering adult care, the need to travel considerable distances to access a treatment centre, or the non-existence of NMD clinics altogether.

Situation analysis

In almost half of the countries under examination, the great majority of, or all existing NMD clinics/SMA treatment centres exclusively focus on adults or offer comprehensive care for both children and adults living with SMA. This is particularly evident in countries like Belgium, Finland, Hungary, Switzerland, the Netherlands, and Ukraine, where adult care services are universally available in all NMD treatment centres.

Despite the considerable variation in the number of NMD treatment centres globally, even in countries where only half of these centres cater to adults with SMA, a reasonably well-distributed network of these facilities often exists. For instance, in Poland, approximately half of the centres provide care to adults, covering 13 (and soon 14) out of 16 provinces.

However, challenges arise in certain countries, such as North Macedonia, where adults living with SMA have significantly fewer options as only one of the two NMD treatment centres offers adult care. Geographical distribution poses another consideration, with some regions presenting obstacles for adults living with SMA who may need to travel considerable distances to access an NMD treatment centre, making access to SMA-specialised care challenging. Notable examples include the southern regions of Italy and the eastern part of Portugal, as well as the islands of both countries.

In countries like Finland, Ireland, Portugal and Romania, where NMD or SMA-specific treatment centres may be absent in certain regions, a well-distributed network of general neurology clinics compensates by providing care for adults living with SMA. Conversely, adults living with SMA in Greece, Hungary, Ukraine, and Russia depend more heavily on less specialised care, such as primary care centres/ GPs. NMD reference centres (if they exist) are predominantly located in large cities, making accessibility a challenge. This may lead to adults living with SMA falling out of specialist care all together.

The distribution of care varies, with slightly over a quarter of the countries reporting that the majority of adults living with SMA receive care in specialised reference centres. In others, a larger portion of the community may rely on general neurology practices or less specialised care, a variation often attributed to the geographic distribution of reference centres. For example, healthcare professionals estimate that in Portugal, a significant percentage of adults living with SMA, ranging from 15% to 50%, are not under the care of a reference centre. Meanwhile, in Finland, the number of adults living with SMA receiving treatment in an NMD reference centre is minimal, given that there is only one such centre in the country, so adults are mostly followed in neurological departments of university hospitals.

The average wait time for a first appointment with an SMA or NMD specialist following diagnosis varies significantly across the countries in the report. Waiting times can range from less than one month in North Macedonia and Ukraine to as long as six months in countries like Denmark, Germany, Poland, the Czech Republic, and Romania. In some instances, the lengthier wait is attributed to adults living with SMA choosing one clinic over another for their initial consultation. For example, adults in Serbia commonly opt for a consultation in Belgrade, even if it involves a longer wait than a consultation in another reference centre.

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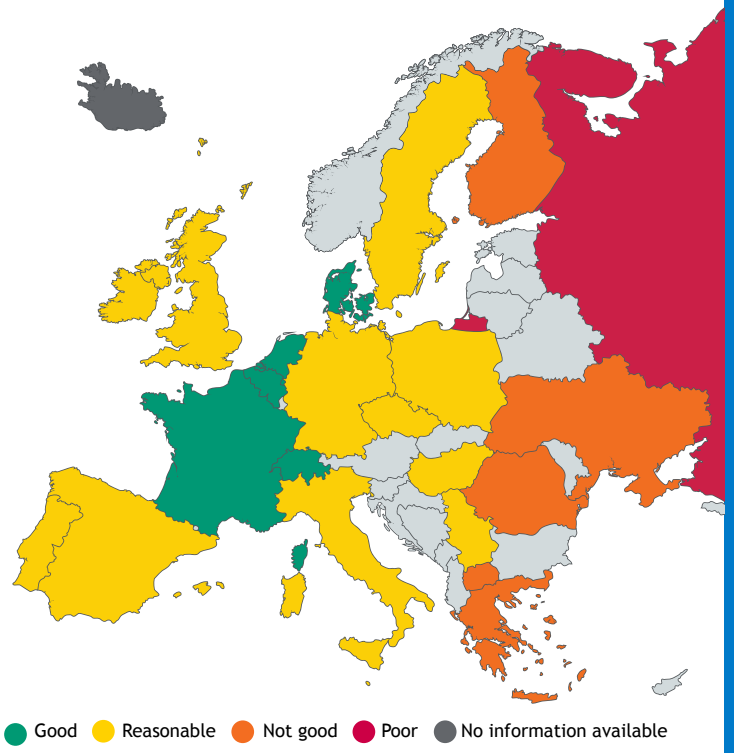
Best practices

In Serbia, there are four SMA treatment centres dedicated to providing care for adults living with SMA, located in Novi Sad, Belgrade, Niš, and Kragujevac, ensuring an equitable geographic distribution. These centres typically offer short waiting times of one to two months for an initial appointment with a specialist after diagnosis. It's worth noting that some adults living with SMA may opt for a consultation in Belgrade instead of their nearest centre, potentially extending their waiting period. However, there are plans in Serbia to designate the Kragujevac Clinical Centre as the specialist centre for rare diseases in the country. Notably, the city administration has already invested in SMA training for physiotherapists, demonstrating a commitment to enhancing specialised care.

In Spain, six out of seven NMD clinics or NMD-specialised divisions within hospitals cater to adults living with SMA. These clinics are situated in four out of Spain's 19 regions, demonstrating a fairly balanced distribution across the country, despite four (out of seven) clinics being located in Catalunya. There are short waiting times of one to two months for an initial appointment with a specialist following diagnosis.

European map



A recap of sub-indicators analysed under this indicator

- **Percentage of treatment centres treating adults**

The scoring system assesses the extent to which existing treatment centres offer care & treatment services for adults. Higher scores reflect a greater prevalence of adult care services across the centres, while lower scores indicate a limited offering of these services.

- **Geographic distribution of adult treatment centres**

The scoring system evaluates how evenly these centres are spread across a geographic area. Higher scores indicate a more even distribution, while lower scores suggest adults living with SMA may have to rely on less specialised centres for their day-to-day care.

- **Percentage of adults that are followed in SMA or NMD reference centres/clinics**

The scoring system evaluates the extent to which adults receive care at these specialised centres. Higher scores reflect a greater proportion of adults being treated in these centres, while lower scores indicate that adults living with SMA are more likely to be treated in non-specialised clinics.

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Network of treatment centres

Key findings

- Formal networks of SMA or NMD treatment centres exist in half of the countries analysed in this report.
- Collaboration between treatment centres, whether through a formal network or otherwise, occurs in the majority of the countries.
- In countries where there is no collaboration between treatment centres, this is usually due to the fact that there are few or no specific SMA treatment centres.

Situation analysis

In many countries, the establishment of a formal collaborative network of treatment centres for SMA is lacking.

In countries where formal networks do exist, these networks may engage in a variety of activities or have a very specific focus. For example, the network of treatment centres in the Czech Republic and Poland places a strong emphasis on activities such as clinical cases, training of HCPs, collecting data for the existing registry, and supporting transition care. In contrast, Spain's network focuses mainly the collection of data for the existing registry. In countries such as Ukraine, a network has been recently formed and, therefore, is still in its preliminary stages.

Formal networks not only collaborate on individual treatment and care for adults living with SMA but also promote research and international collaboration. Such comprehensive networks can be found in countries like Switzerland, the Netherlands, Germany, Italy, France and the UK.

In countries such as Denmark, Finland, Greece, Portugal, Serbia, and Sweden, where formal networks are absent, informal collaboration takes place on various activities. These include the development of clinical guidelines, data collection and aggregation, clinical trial facilitation, discussions about clinical cases, and referring adults living with SMA to specialists.

However, in a small number of countries, there is little to no collaboration between treatment centres. For example, North Macedonia has only one SMA treatment centre, which explains the absence of collaboration. Russia lacks SMA treatment centres altogether. In Ireland, Romania, and Belgium, despite having multiple SMA treatment centres, there are no notable networks or informal collaborations for activities such as discussing clinical cases or defining standards of care. Belgium does have some collaboration, primarily through the Neuro Muscular Disease Registry (BNMDR).

Collaboration between treatment centres, whether through a formal network or otherwise, is important for the standardisation of activities such as knowledge sharing between HCPs, discussion of clinical cases, enabling transition care and referrals between centres, and collection of data through existing registries, among others. The non-existence of collaboration between centres is a missed opportunity for continuously improving care for those living with SMA across the country.

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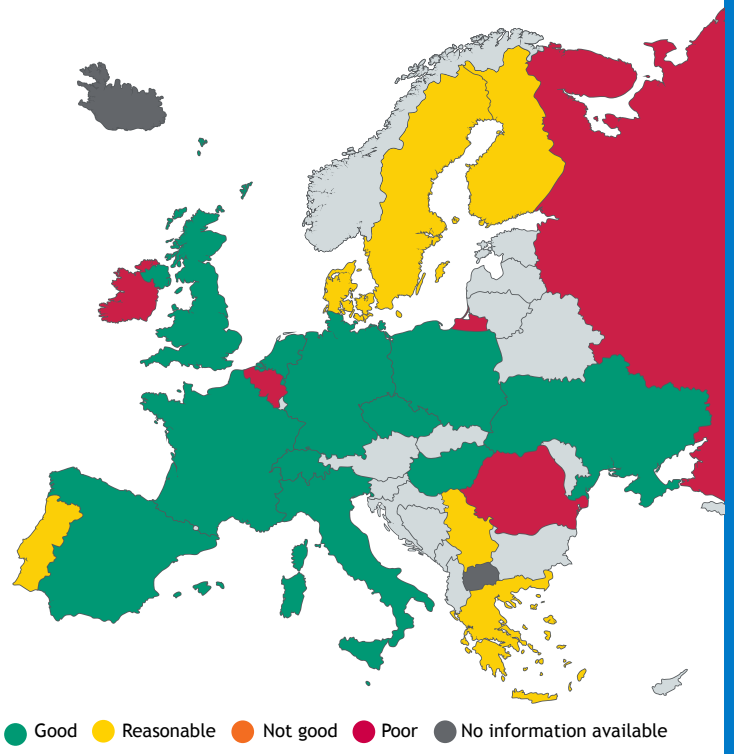
Best practices

In Italy, a collaborative network of seven centres spread throughout the country - known as the NeuroMuscular Omnicentre (NEMO) clinical centres network - has been established by hospitals and other organisations that provide funding for research and care related to NMDs. Its primary goal is to provide support to individuals living with NMDs, both in terms of clinical care and support. Additionally, there is also a wider network of sites that are affiliated with the Italian Myology Association (AIM), which also follows a multidisciplinary approach and is more widely distributed within the national territory.

In Germany, the Society for Neuromuscular Disease (DGM) has established a nationwide network of NMD centres with the aim of facilitating collaborative and multidisciplinary treatment and diagnosis, as well as streamlining the referral process between various medical disciplines. Additionally, these centres engage in research collaborations, with five of them participating in a pilot project known as “patient guides”. This project assists staff in coordinating care, organising multidisciplinary consultations, and providing information to individuals living with NMDs.

European map



A recap of sub-indicators analysed under this indicator

- **Existence of collaborative network of treatment centres**

The scoring system assesses the degree of formalised collaboration among these centres. Higher scores reflect the existence of a more established and purposeful network, while lower scores indicate a lack of formal or informal collaboration mechanisms.

Patient registry

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Key findings

- A patient registry for collecting data on adults living with SMA either exists or is in the process of being established in most countries covered in the report.
- In most cases, data is entered on a voluntary basis, either by HCPs and/or adults living with SMA themselves. In some countries, HCPs are required to input data for those adults receiving a DMT.
- About three quarters of the countries covered in this report had an adult registry supported by the majority of/all SMA treatment centres.

Situation analysis

In the countries covered in this report, the establishment and management of SMA-specific or NMD registers vary. Some registries are managed by medical societies in collaboration with healthcare centres (e.g. Czech Republic), others by healthcare centres/hospitals alone (e.g. France, the Netherlands), while others are overseen by patient organisations (e.g. Russia, and Ukraine) or national authorities/agencies (e.g. Greece, Hungary, North Macedonia, Portugal). In some instances, registries are a result of multi-stakeholder collaboration (e.g. Belgium). In some countries, such as Germany, Italy, Poland, Spain and Sweden, different complementary registries exist, managed by different stakeholders.

The mandatory requirement for HCPs to input data into these registries is primarily applicable when adults living with SMA are receiving DMT and monitoring of treatment outcomes is needed to ensure continuous access to it. In certain countries like Germany, Hungary, Spain, Sweden, and the Netherlands, adults living with SMA also input their own data. However, in Russia, Ukraine and the UK, only adults living with SMA, not HCPs, are contributing to the existing SMA registry. While the existence of registries collecting data inputted exclusively by adults living with SMA are beneficial for mapping demographics, they may not capture outcome measures scoring and progression of the disease, making them less useful for clinical research or post-marketing evaluation purposes.

Most countries have either established or are in the process of creating a registry. However, even in cases where these registries exist, they may face challenges in collecting sufficient data. For instance, in the Czech Republic, registries encounter obstacles due to insufficient participation from NMD reference centres/clinics.

In countries where data on adults living with SMA is only collected by a non-NMD-specific registry, it may not gather enough useful information. An example is the registry in Denmark, which, despite collecting some data on adults living with SMA, including on examinations, diagnoses, and treatments, does not monitor the clinical history or pharmacological treatment outcomes.

Administrative barriers can also arise, such as registries ceasing to collect new data, such as in Romania and Finland, including due to the administrative burden on HCPs.

These registries, whether specific to SMA or NMD, serve as valuable sources of epidemiological data for clinical research and post-marketing follow-up, benefiting the adult SMA community. Successful examples include the TREAT NMD Registry of SMA Patients in Poland, which has facilitated the establishment of SMA treatment centres, feasibility studies, and assessments of treatment outcomes. Furthermore, registries can promote international collaboration in research and advance SMA care. An example of such an internationally collaborative registry is the registry in France.

In conclusion, while most countries have either established or are developing a registry, in more than half of them, this process operates on a voluntary basis. Combined with other challenges related to registry management and the type of data being collected, there is still progress to be made before the collected data can fully benefit the broader adult SMA community.

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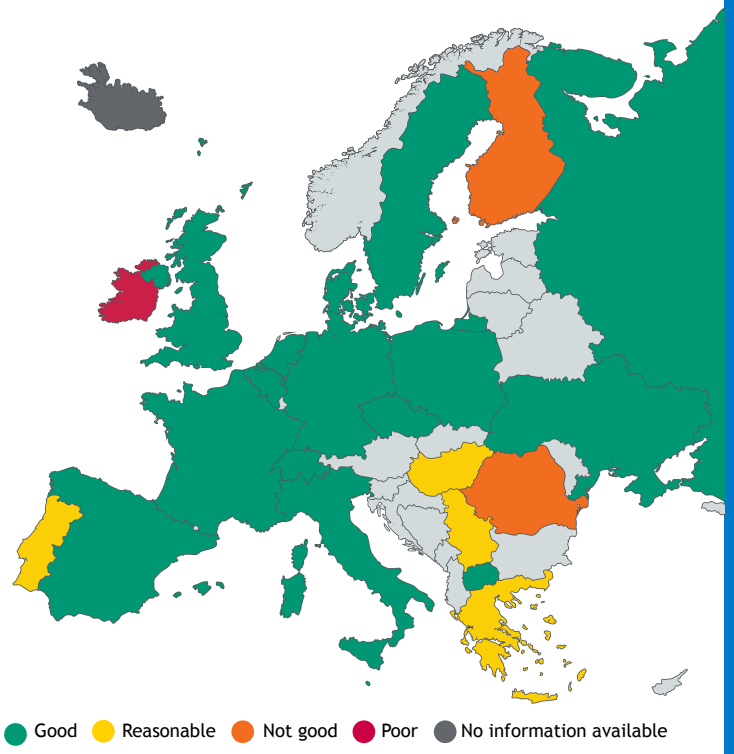
Best practices

Sweden's Neuroregister, managed by the university hospital Karolinska sjukhuset, includes a sub-registry for NMDs where both HCPs and adults living with SMA can input data and updates. There is also a compulsory registry dedicated to recording the outcomes of pharmacological treatment.

A new SMA registry has been initiated in France, with its establishment dating back to 2020. Nearly all SMA treatment centres are already actively participating and requiring their HCPs to input information. The registry compiles data on treatments, consultations, and follow-ups within reference centres.

European map



A recap of sub-indicators analysed under this indicator

- **Existence of patient registries collecting data on adults**

The scoring system evaluates the presence and functionality of registries specifically collecting data on adults living with SMA. Higher scores indicate the existence and active data collection, while lower scores reflect the non-functionality or even absence of such registries.

- **Percentage of treatment centres that participate in an existing registry**

The scoring system assesses the level of engagement by treatment centres in contributing data. Higher scores indicate a greater percentage of centres participating, while lower scores suggest limited involvement or non-participation in the existing registry.

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Standards of care (SoC) & treatment guidelines

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Key findings

- Only one country has in place national SoC for diagnosis and management of SMA that addresses specific needs in adulthood, while all others still rely on international or national standards, which focus on best practices for children living with SMA.
- Close to half of the countries lack guidelines for pharmacological treatment, while most others are in the process of developing those or already have them.
- In approximately half of the countries, the use of outcomes measures in standard clinical practice is done on a voluntary basis. In the other half, however, their use is mandatory for continuous access to DMTs.

Situation analysis

Adults living with SMA can face unique health challenges requiring solutions that pediatric approaches cannot necessarily address, including excessive fatigue, kidney stones, osteoporosis, acidosis, hypertension, weight management, sexual health and hormonal issues. Hence, when SoC focus on best practices for children, they may overlook important considerations for adults, therefore leading to suboptimal care and a negative impact on quality of life.

Across all countries examined within the report, only France has put in place SoC that address the specific needs of adults living with SMA. In others, such as Germany, Netherlands, Russia and Ukraine, stakeholders are currently working on new or revised national SoC for the diagnosis and management of SMA, which may focus on care for adults living with SMA. Yet, a significant number of countries still rely on the international SoC or national SoC focusing on children living with SMA, which fail to address the specific needs of adults living with SMA and do not cover pharmacological treatment with DMTs.

Close to half of the countries lack established guidelines on the use of DMTs and have yet to begin the process of creating them. While approximately a third of countries are in the process of developing guidelines encompassing all available DMTs for adults, only few – including France, Germany, Greece, Hungary, and Sweden – have completed this procedure thus far. Other nations, including Spain, might have outdated or incomplete treatment guidelines that do not cover the entire spectrum of DMTs available to adults living with SMA in the respective countries.

The use of outcome measure tools is important in the follow-up of adults living with SMA, to better monitor and track the progression of SMA, including whether adults are being treated with DMTs. However, the use of outcome measures in clinical practice varies across the surveyed countries. It's mandatory in approximately half of them, often linked to eligibility for reimbursement of DMTs. In all other instances, the use of outcome measures is still voluntary, but they are commonly used in clinical practice, with the exception of Russia, where they are not commonly used unless mandated by the local government for assessing access to treatment.

Adults living with SMA can face unique health challenges that require solutions pediatric approaches cannot necessarily address, including excessive fatigue, kidney stones, osteoporosis, acidosis, hypertension, weight management, sexual health and hormonal issues. Hence, when SoC focus on best practices for children, they may overlook important considerations for adults therefore leading to suboptimal care and a negative impact on quality of life.

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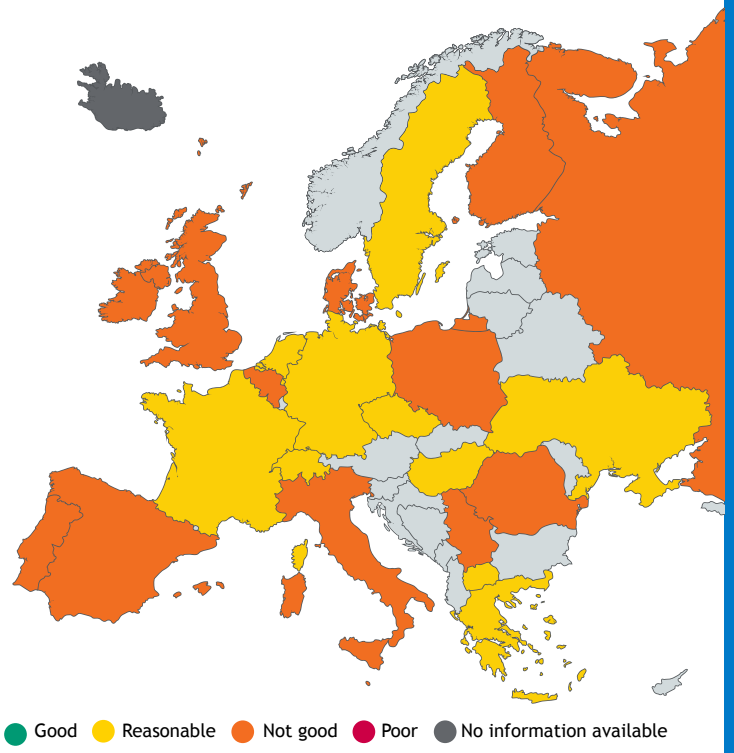
Best practices

Ukraine's Ministry of Health is in the process of developing SoC that will cover diagnosis, management and pharmacological treatment of SMA, including for adults. This will reportedly give all adults living with SMA the right to demand pharmacological treatment in any centre according to the protocol.

Greece has developed and adopted national SoC for management of SMA, including for adults, which are accessible to all on the Ministry of Health website.

European map



A recap of sub-indicators analysed under this indicator

- **Existence of SoC for diagnosis and management of SMA in adults**

The scoring system assesses the presence and relevance of these guidelines. Higher scores indicate the existence of comprehensive SoC specifically tailored for adults living with SMA, while lower scores suggest either underdevelopment, outdatedness, or reliance on international guidelines.

- **Existence of DMTs treatment guidelines**

The scoring system assesses whether these guidelines are in place and up to date. Higher scores indicate comprehensive and current DMTs treatment guidelines, while lower scores suggest the absence of such guidelines or their outdatedness.

- **Use of outcomes measures used in standard clinical practice**

The scoring system assesses the degree to which these measures are integrated into routine healthcare practices. Higher scores indicate a more structured and mandatory use, while lower scores suggest voluntary or no usage of outcome measures in standard clinical practice.

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b. Area 2: Healthcare delivery

Multidisciplinary care

Key findings

- SMA or NMD treatment centres have multidisciplinary teams (MDTs) who meet formally to coordinate adults living with SMA's care in approximately half of the countries in the report.
- Where MDTs exist, they are usually coordinated either by the lead neurologist, or alternatively by the case manager or GP.
- In approximately half of the countries, adults living with SMA have access to the relevant specialists but usually do not benefit from coordinated care via a formal MDT in their treatment centre.

Situation analysis

In the context of SMA care, the presence of MDTs varies across countries, with fewer than half of them incorporating such teams within their SMA-specific or NMD treatment centres. These teams, when present, typically encompass a spectrum of specialists, which could include neurologists, cardiologists, respirologists/pulmonologists, physiotherapists, orthopaedists, and sometimes additional professionals such as nutritionists, occupational therapists, counsellors, and social workers, among others.

The organisational structure of MDTs differs among regions, too. In some countries like Belgium, France, Germany, Greece, Italy, and Switzerland, specialists are usually centralised in one centre. France adopts a model where specialists are based in one centre but collaborate occasionally with external experts. Elsewhere, MDTs operate across various centres, engaging in formal or informal discussions, as required. On the other hand, Ireland follows a unique approach where most specialists are centralised within one hospital, with exceptions of GPs, physiotherapists, and occupational therapists.

The coordination of MDTs also varies. In Belgium and Germany, for instance, coordination is often led by the lead neurologist, while in Switzerland and France, it may be managed by the GP. In Ireland, hospital administration takes charge.

Remarkably, despite many countries providing access to relevant specialists, adults living with SMA in almost half of the countries lack the advantage of coordinated care through a formal MDT within their treatment centres. In Finland, Hungary, North Macedonia, Poland, Portugal, Spain, Serbia, Russia, Sweden, Ukraine and the UK, for example, various specialists are available, but the general absence of a structured MDT collaboration, or its existence only in few treatment centres, results in more ad-hoc to no coordination. This impacts quality of care as the different specialists do not communicate with each other and do not develop a joint plan for improving care and health outcomes for each individual.

It is worth noting some of the many initiatives by patient organisations aimed at enhancing interdisciplinary care. In Serbia, SMA Serbia collaborates with the Health Insurance Fund to establish a dedicated centre for rare diseases, offering access to MDTs. While in Russia, the SMA Families Foundation has initiated the SMA Clinic project, funded by the Presidential Grants Fund and the Family Clinic Network, allowing adults living with SMA to receive care through remote consultations with a coordinator and routine consultations in Moscow with an interdisciplinary team. These endeavours underscore the importance of collaborative efforts in improving the healthcare landscape for those living with SMA.

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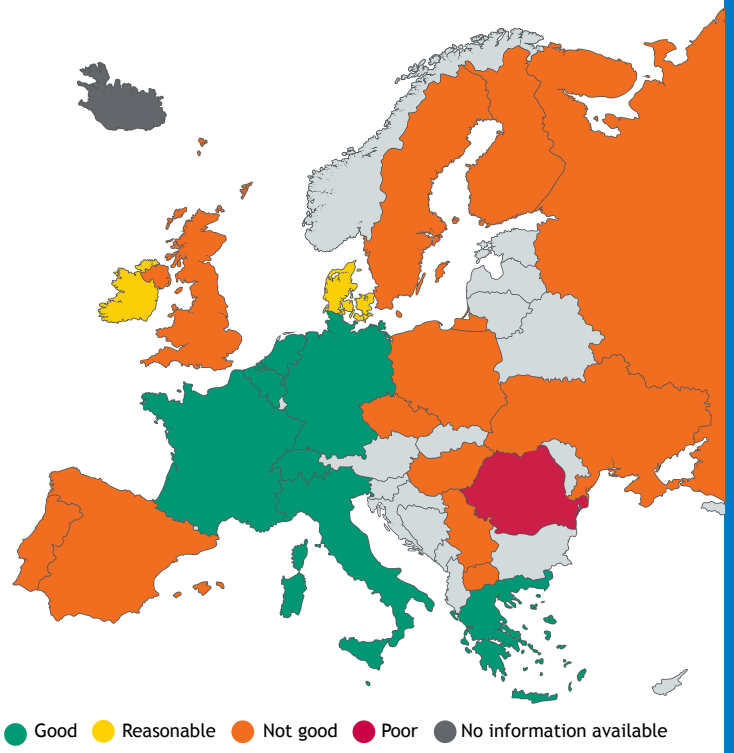
Best practices

In Belgium, MDTs are present in all NMD reference centres, overseen by a neurologist, to offer coordinated multidisciplinary care for adults living with SMA. Although the composition of these teams may differ across centres, they typically involve neurologists, cardiologists, gastroenterologists, geneticists, nutritionists/dieticians, nurses, occupational therapists, physiotherapists, psychologists, orthopaedics, social workers and speech therapists. On occasion, additional specialists like dermatologists, gynaecologists or pulmonologists may also be part of the team.

In France, all SMA treatment centres within the FILNEMUS network are equipped with an MDT to provide comprehensive support for adults living with SMA. While specialists may occasionally be located in different centres, the teams maintain formal meetings to coordinate care. The team, supervised by the designated GP acting as the “attending ”physician”, typically consists of neurologists, cardiologists, pulmonologists, physiotherapists, and/or orthopaedic specialists. Upon request, nutritionists and psychologists may also be included in the team.

European map



A recap of sub-indicators analysed under this indicator

- **Availability and access to MDTs for adults living with SMA**

The scoring system assesses the presence and collaborative nature of these teams in patient care. Higher scores indicate the availability of MDTs and collaboration among healthcare providers, while lower scores suggest the lack of collaboration among HCPs or even lack of access to HCPs.

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Shared decision-making

Key findings

- Only about half of the countries in the report have effectively implemented shared decision-making across all HCPs and clinics working with adults living with SMA. In the remaining countries, this practice is either inconsistent or not part of standard clinical procedures, being offered only upon request.
- In most countries covered in the report, there are existing resources and tools to empower adults to make informed decisions about their health. However, in some of these countries, these resources may not be easily accessible. Moreover, there are still a few countries where either the information provided is inadequate or simply unavailable.
- Despite shared decision-making being integrated into standard clinical practices in certain countries, healthcare providers sometimes encounter challenges involving adults living with SMA in this process.

Situation analysis

The report studies the implementation of shared decision-making among HCPs attending to adults living with SMA. Only about half of countries have fully integrated shared decision-making into their standard clinical practice for this demographic, as outlined in the report's findings. Within these regions, well-informed adults living with SMA benefit from receiving essential treatment and care information either directly from their HCPs or through specialised patient organisations.

However, even in countries where shared decision-making is considered standard, limitations exist in specific domains. For instance, regions imposing strict constraints on pharmacological treatments (see indicator 11 on access to EMA-approved DMTs) for adults may hinder active participation in the decision-making process. In France, the ultimate decision concerning DMTs lies with a commission following a neurologist's request, thereby limiting the autonomy of adults living with SMA in determining their treatment options.

Disparities in implementation across countries may arise from various factors, including differences in practices among HCPs or barriers that hinder the active involvement of adults living with SMA. Difficulties in accessing crucial information necessary for informed decision-making pose a significant challenge in some regions, impacting their engagement in the process.

Despite these challenges, instances of shared decision-making occur, even when HCPs lack comprehensive information to share with adults living with SMA. Many patient organisations play a crucial role by providing extensive resources, including peer support groups, empowering individuals living with SMA to make informed decisions. Moreover, in countries like Hungary, social media groups initiated by individuals living with SMA and their families serve as valuable platforms for sharing information and tools within their community.

However, in countries such as Finland, heavy reliance on international information sources and online connections like social media fall short in enabling active participation in shared decision-making among adults living with SMA.

The report highlights that only a very small number of countries, including Serbia, entirely lack shared decision-making as part of their standard clinical practice. Even in these regions, if an adult living with SMA expresses the desire to engage in decision-making, this is usually made possible.

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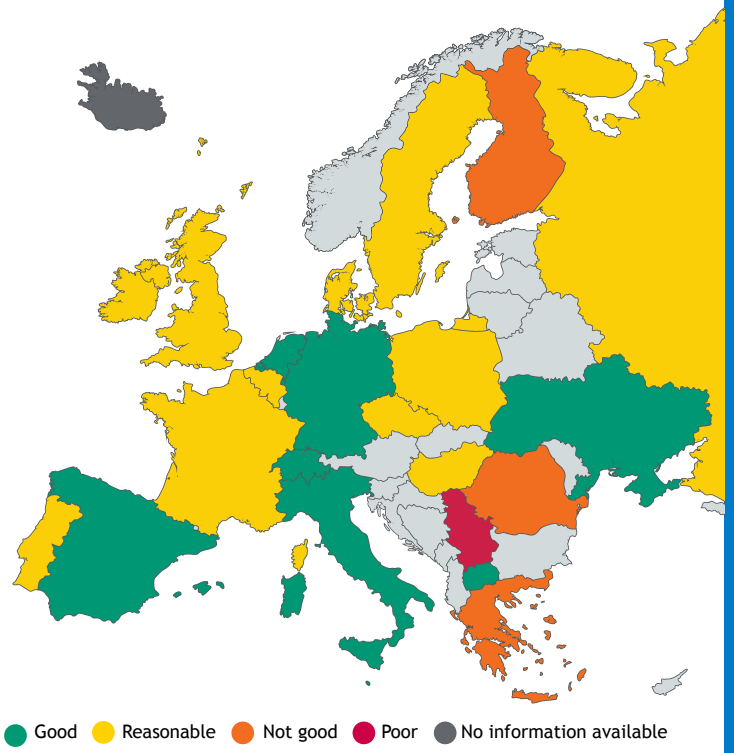
Best practices

In Germany, shared decision-making is a standard part of clinical practice for HCPs working with adults living with SMA. It is a legal requirement for HCPs to offer adequate information regarding pharmacological treatment options, and adults living with SMA must sign a consent form to confirm their understanding.

In Switzerland, adults living with SMA play a significant role in decision-making concerning their care and treatment. SMA Schweiz acknowledges that while the abundance of information accessible to these individuals might at times feel overwhelming and contain occasional gaps, it remains readily available, allowing them to make informed decisions regarding their health.

European map



A recap of sub-indicators analysed under this indicator

- **Implementation of shared decision-making in standard clinical practice**

The scoring system assesses the level of integration of shared decision-making into routine healthcare. Higher scores reflect greater incorporation of shared decision-making as a standard practice, while lower scores indicate varying levels of implementation, or its entire absence from standard clinical practice.

- **Existence of information/tools to empower adults to take informed decisions about their health**

The scoring system for the existence of information and tools to empower adults in making informed health decisions assesses the existence and accessibility of available resources. Higher scores indicate more comprehensive and appropriate resources easily available for a broader range of adults living with SMA, while lower scores reflect limited availability of information and tools for this purpose.

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Continuity of care

Key findings

- In slightly over half of the countries detailed in the report, follow-up appointments are mandatory for adults living with SMA undergoing DMT. In the remaining countries, follow-up appointments are completely voluntary.
- In the great majority of countries, healthcare centers have established procedures to encourage ongoing engagement of adults living with SMA in healthcare services, including automatic appointment scheduling.

Situation analysis

The report presents a wide spectrum of requirements concerning regular follow-up appointments for adults living with SMA across various countries. It paints a challenging picture for those not eligible for DMTs, who may be left disengaged from the healthcare system.

Meanwhile, slightly over half of the covered countries have established follow-up requirements for adults living with SMA undergoing DMT, while these appointments remain voluntary for others. Despite these variations, the great majority of countries' healthcare centres, with the exception of Russia and Ukraine, actively encourage ongoing engagement for adults living with SMA, employing strategies such as automatic scheduling, appointment reminders, and assigning a designated healthcare provider as a case manager to assist in navigating the healthcare system.

For those undergoing DMT, the frequency of required follow-up appointments varies significantly among countries. While some nations opt for yearly assessments to evaluate outcome measures, others require appointments as frequently as every three or four months.

Challenges in attending these appointments are prevalent in some countries, primarily due to geographical distance and travel difficulties. Solutions being implemented include the provision of ambulance services for transportation and facilitating specialists' remote attendance via video calls during appointments between individuals living with SMA and local non-specialist healthcare providers.

A notable observation from the report is the leniency in requirements for adults living with SMA who do not receive pharmacological treatment. This discrepancy in follow-up requirements can significantly impact disease progression tracking and hinder general data collection that holds potential benefits for the broader SMA community.

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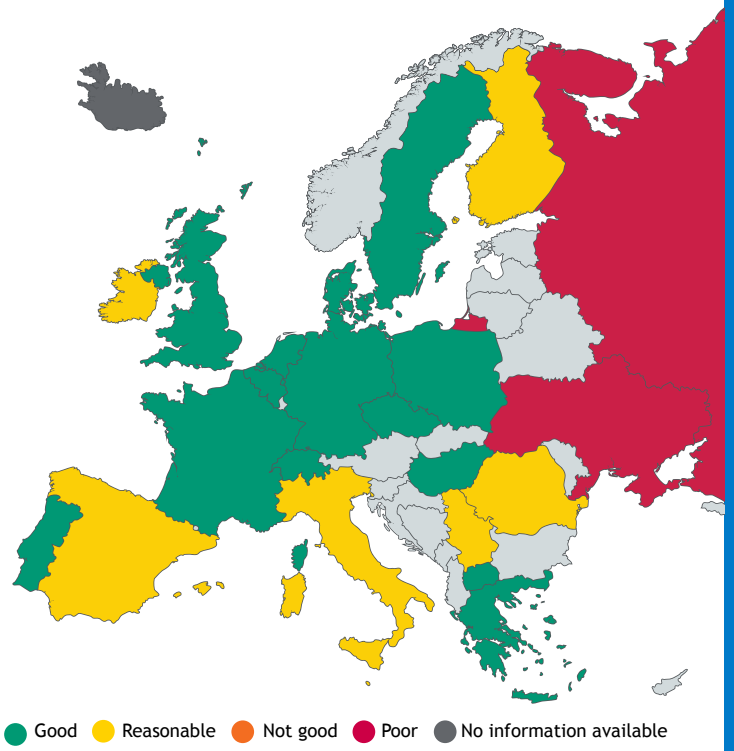
Best practices

In France, regular follow-ups are obligatory for all adults living with SMA, irrespective of their involvement in DMT. Protocols are implemented to foster ongoing participation in healthcare services, including automated appointment scheduling. Addressing transportation and accessibility barriers in challenging terrains like mountainous regions or Corsica—where no SMA treatment centres exist—France has initiated the deployment of 'telemedicine.' This enables remote consultation with a specialist through a GP.

In Switzerland, most centres require follow-ups for all adults living with SMA. In some instances, this requirement is also established by the insurer of the adult living with SMA. However, the frequency of these mandatory follow-ups might be influenced by capacity constraints within the centres.

European map



A recap of sub-indicators analysed under this indicator

- **Mandatory regular follow-ups**

The scoring system evaluates the extent to which routine check-ups are obligatory for adults living with SMA. Higher scores indicate a stronger requirement for adults living with SMA receiving DMTs to attend regular follow-ups, while lower scores a voluntary approach determined by the patient.

- **Processes in place to motivate adults living with SMA to continue engaged with healthcare services**

The scoring system evaluates the presence or absence of strategies and initiatives designed to encourage ongoing patient participation in healthcare. Higher scores indicate the existence of such processes, while lower scores reflect their absence.

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Care team & caregivers training

Key findings

- Most countries offer formal SMA training for HCPs provided by treatment centres, medical societies, or continuous medical education from SMA centres of excellence or pharmaceutical companies, among others.
- Shortcomings persisted, notably in the care provided to adults living with SMA by some HCPs, particularly local nurses and physiotherapists in certain regions.
- Although just over half of the countries covered in the report offer either formal or informal opportunities for life assistants to access SMA training, nearly half provide no such options.
- In the majority of countries, there are no SMA training opportunities for informal caregivers.

Situation analysis

Research findings revealed a landscape where accessibility to SMA training and education for HCPs varied widely across countries. In nearly three-quarters of the countries examined, formal SMA training sessions or continuous medical education were readily available for HCPs.

Over half of the countries offered formal SMA training through key NMD or SMA-specific treatment centres or medical societies, conducted either in-person or online. Many HCPs also supplemented this training with continuous education sponsored by pharmaceutical companies, often included as part of specialised university training or one-off sessions dedicated to SMA.

On the other hand, in approximately a third of the countries, only continuous medical education was available. These educational resources, predominantly provided by SMA centres of excellence or pharmaceutical companies, aimed to keep HCPs abreast of the latest SMA knowledge. Yet, despite these efforts, shortcomings persisted, notably in the care provided to adults living with SMA by some HCPs, particularly local nurses and physiotherapists in certain regions. Access to education on SMA varied among different specialisations, medical districts, or treatment centres, contributing to inconsistent knowledge levels among HCPs.

A small fraction of countries lack specific SMA training programmes for HCPs altogether, meaning they are reliant on their university studies only. The disparity widened when considering training programmes for life assistants or informal caregivers. Nearly half of the countries surveyed lacked any SMA training for life assistants, while even fewer offered such programmes for informal caregivers.

In regions where training opportunities existed for life assistants, they were often part of broader disability training initiatives or provided by non-governmental organisations or third-party service providers. However, the availability and comprehensiveness of such training varied considerably, with some countries providing only partial demonstrations on equipment use, like ventilators, at hospitals.

Informal caregivers faced even scarcer opportunities for SMA training, with formal programmes available only in a couple of countries, predominantly facilitated by care insurance funds or patient organisations. While informal training initiatives existed in some areas, they typically focused on specific needs like ventilator operation, and their accessibility remained limited. Despite mentions of training being provided by adults living with SMA or their families, the lack of formal instructional programmes for those in need was a concerning trend highlighted by several patient organisations.

Training for HCPs caring for adults living with SMA is extremely important to ensure they remain up to date with latest developments and optimal disease management strategies. The lack of such training may lead to suboptimal care that does not fit the needs of adults. Furthermore, lack of training opportunities on SMA for life assistants and informal caregivers will further increase the burden of SMA on adults, who will have to bear the burden of providing such training to their caregivers based on their individual experience and may not be always able to do so.

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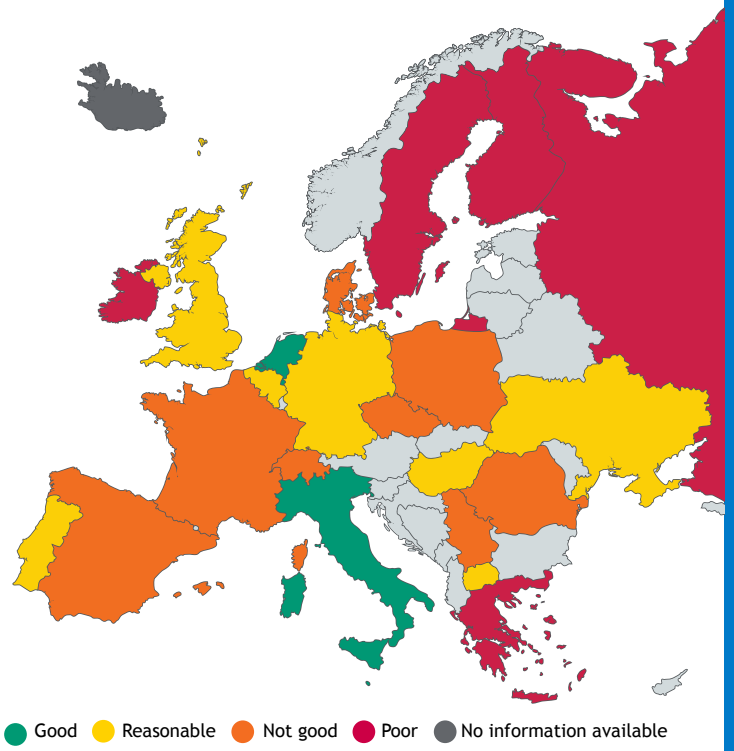
Best practices

In Italy, NMD-specialist 'NEMO' clinics offer formal training opportunities for HCPs nationwide. Simultaneously, the patient organisation Famiglie SMA conducts specialised physiotherapy training for HCPs in hospitals, focusing on SMA care. They also organise training sessions for informal caregivers, encompassing various aspects such as mobility, healthcare, psychological challenges, patients' rights, and daily life. The organisation has also developed a handbook specifically tailored to caregivers of individuals living with SMA.

In Germany, HCPs have access to training and continuous medical education offered by SMA centres of excellence, NMD clinics, or medical societies. Mandatory training for life assistants is overseen by their employers to ensure compliance. Informal caregivers also have training options available through care insurance funds.

European map



A recap of sub-indicators analysed under this indicator

- **Existence of SMA training programmes for HCPs**

The scoring system for the existence of SMA training programmes for HCPs evaluates the availability and depth of training initiatives. Higher scores indicate the presence of formal and continuous training programmes, while lower scores reflect either one-time training during university or specialist training, or the complete absence of SMA-related training programmes for HCPs.

- **Existence of SMA training programmes for life assistants**

The scoring system for the existence of SMA training programmes for life assistants assesses the availability and accessibility of training opportunities. Higher scores indicate the presence of accessible training programmes, while lower scores reflect difficulties in access or the absence of such opportunities for life assistants.

- **Existence of SMA training programmes for informal caregivers**

The scoring system for SMA training programmes for informal caregivers evaluates the presence and ease of access to these training opportunities. Higher scores suggest the existence of accessible programmes, while lower scores indicate challenges in access or the absence of such opportunities for informal caregivers.

Access to EMA-approved disease modifying therapies (DMTs)

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Key findings

- Adults living with SMA in France, Germany, Italy, Spain, Greece, Poland, Portugal, Romania and the UK have access to both *nusinersen* and *risdiplam*, according to the European Medicines Agency (EMA) label.
- In countries like Denmark, Finland, Ireland, Sweden, the Netherlands, and Switzerland, age restrictions are in place for *nusinersen* and/or *risdiplam*. These are typically set at 18 or 25 years old, significantly limiting access for many adults living with SMA.
- Adults living with SMA in North Macedonia, Serbia, and Ukraine do not have access to *nusinersen*.

Situation analysis

At the EU level, the European Medicines Agency (EMA) has approved two DMTs for the treatment of adults living with SMA: *nusinersen* (SPINRAZA®) and *risdiplam* (EVRYSDI®▼).

nusinersen received EMA approval in May 2017 for the treatment of 5q Spinal Muscular Atrophy. [5] As of November 2023, this medication is available, as per the label, to all adults living with SMA in 11 out of the 22 countries researched, including France, Germany, Greece, Italy, Poland, Portugal, Romania, Serbia, Spain, the Netherlands and the UK.

In eight other countries, additional restrictions on adult access exist, including age-specific cut-offs (e.g. Denmark and Switzerland), the need to have started treatment before individuals turned 18 years old (e.g. Finland, Ireland, and Sweden), the need for ventilatory support (e.g. Belgium). In some countries, access is granted on a case-by-case basis, such is the case in Hungary and Russia.

nusinersen is unavailable to adults living with SMA in North Macedonia, Serbia, and Ukraine.

risdiplam gained EMA approval in March 2021 for the treatment of 5q SMA, intended for individuals living with SMA type 1, type 2, or type 3, or those who have 1-4 copies of the SMN2 gene. [6]

As of November 2023, *risdiplam* is available to all adults living with SMA, as per the label, in 12 out of the 22 countries researched, including France, Germany, Greece, Italy, North Macedonia, Poland, Portugal, Romania, Serbia, Spain, Switzerland and the UK.

For *risdiplam*, restrictions are in place for nine of the countries included in the study, including age-specific cut-offs (e.g. Denmark and the Netherlands), the need to have started treatment before individuals turned 18 years old (e.g. Finland, Ireland, and Sweden), the need for ventilatory support (e.g. Belgium, Ukraine), or certain types of SMA (e.g. Czech Republic, Ukraine). In Russia and Serbia, reimbursement is considered on a case-by-case basis. In Hungary, health technology assessment (HTA) processes are ongoing.

Despite the rapidly changing situation, the landscape above shows the existence of persistent and significant inequalities in access to DMTs across Europe, with eligibility criteria for access to *risdiplam* and *nusinersen* often making access to these two DMTs more restrictive than the EMA approved indication.

SMA EUROPE

Risdiplam (EVRYSDI®▼) 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.

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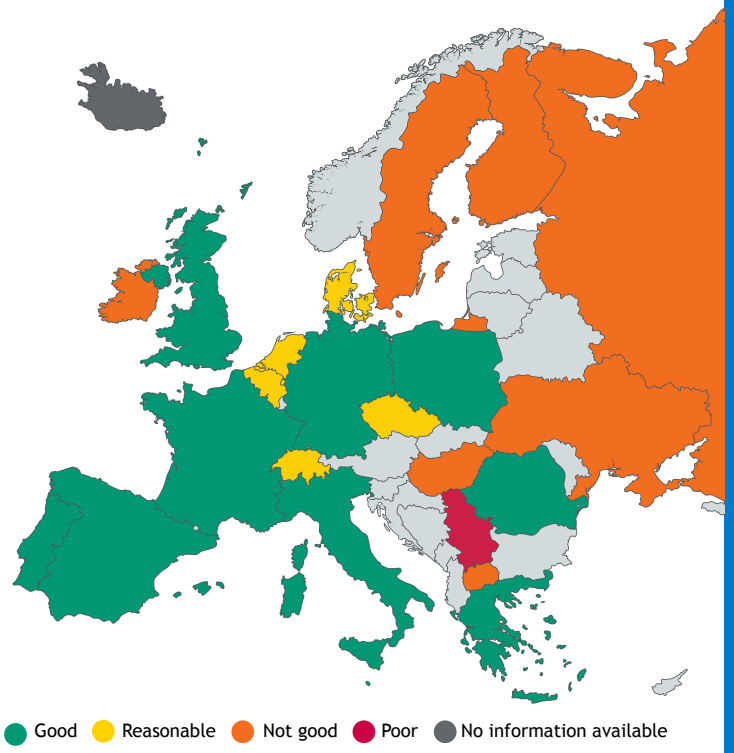
A recap of sub-indicators analysed under this indicator

- **Access to *nusinersen***
- **Access to *risdiplam***

The scoring system assesses the availability and reimbursement eligibility for these treatments among adults living with SMA. High scores indicate access for adults according to the EMA label. Lower scores reflect more limited availability for adults living with SMA, eligibility only in exceptional cases, or no availability at all.

European map



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Access to unauthorised disease modifying therapies (DMTs)

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Key findings

- The majority of the identified pharma-sponsored clinical trials took place in France, Germany, Italy, Spain, Belgium, the Netherlands, Poland and the United Kingdom between 2018 and 2022. Adults living in other countries had little or no access to pharma sponsored clinical trials during this period.
- During the same period, Early Access Programmes (EAPs) with specific entry criteria have been delivered in a majority of countries, providing access to DMTs for a proportion of adults living with SMA.
- In two countries, adults living with SMA had access neither to clinical trials nor to EAPs.

Situation analysis

Adults living with SMA have avenues for accessing investigational DMTs that are being tested but not yet approved. This can be done through participation in clinical trials, studies performed to investigate the safety and efficacy of a medicine. [7] Adults may also be able to access EAPs, also known as Compassionate Use Programmes (CUPs) at EU level. At the EU level, CUPs operate under strict conditions, allowing experimental products to be provided to patient groups living with diseases that do not have satisfactory authorised therapies, especially when they are not eligible for clinical trials. [8] These programmes often provide access to medicines that either have Phase II or III clinical studies complete or that have already have been approved in another country globally (more clarity on risk/benefit profile).

Between 2018 and 2022, a total of 12 pharma-sponsored clinical trials were identified in which adults living with SMA were eligible for participation. These trials included,

- Biogen sponsored: ONWARD [9], DEVOTEE [10], SHINE [11] and ASCEND [12] trials for *nusinersen*.
- Roche sponsored SUNFISH [13] and JEWELFISH [14] trials for *risdiplam*, as well as the MANATEE [15] trial, of RO7204239 in combination with *risdiplam*, and a study of olesoxime [16].
- Other industry sponsored studies include RESILIENT [17] for *taldefgrobep alfa* by Biohaven Pharmaceuticals, as well as TOPAZ [18] and SAPPHIRE [19] for *apitegromab* by Scholar Rock. Additionally, Catalyst Pharmaceuticals led a clinical trial for *amifampridine phosphate* [20].

Certain countries were more active in hosting pharma-sponsored clinical trials, particularly EU4, Belgium, the Netherlands, Poland and the UK. Unfortunately, adults living with SMA in Denmark, Finland, North Macedonia, Romania, Russia, and Ukraine had no pharma-sponsored clinical trials recruiting them during this period. The remaining countries saw only a limited number of clinical trials, typically one or two.

In the same timeframe, only one pharma EAP/CUP was identified in which some adults living with SMA were eligible for participation, specifically for *risdiplam* organised by Roche. In most of the countries included in this analysis, adults living with SMA type 1 and 2 were eligible to access this EAP, with the exception of Serbia. This EAP does not seem to have been rolled out in Denmark, or Finland.

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A recap of sub-indicators analysed under this indicator

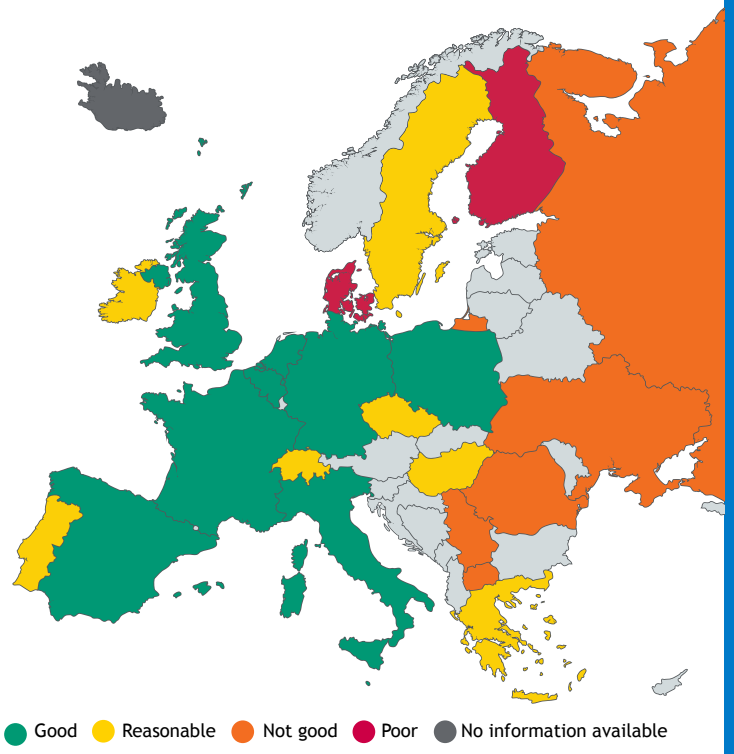
- **Availability of pharma-sponsored clinical trials for adults living with SMA (2018-2022)**

This indicator assessed countries on a scale ranging from high priority to non-priority market. The score reflects the number of pharma-sponsored clinical trials conducted in the period from 2018 to 2022, with higher scores indicating a higher number of trials for which adults living with SMA were eligible to during this period of time. These trials were identified through information available on ClinicalTrials.gov and the European Union Clinical Trials Register.

- **Availability of pharma-sponsored EAPs for adults living with SMA (2018-2022)**

This indicator assessed countries on a scale ranging from the availability of EAPs with eligibility for some adults, to no available EAPs, to the country's legislation not allowing for the roll out of EAPs.

European map



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c. Area 3: Governmental and peer support

HCP consultations reimbursement

Key findings

- In the vast majority of countries, adults living with SMA are not required to pay for consultations with pulmonologists, nutritionists, orthopaedists/orthopaedic surgeons, and physiotherapists.
- However, in a few countries, adults living with SMA are subject to co-payments, often with an annual cap that grants free consultations thereafter.
- Despite reimbursement, challenges have been highlighted concerning access to certain specialists, particularly physiotherapists. This obstacle sometimes leads adults living with SMA to seek private care and cover the costs out of pocket.

Situation analysis

In the vast majority of countries, adults living with SMA do not have to pay for consultations with specialists like pulmonologists, nutritionists, orthopaedic surgeons, and physiotherapists, which are covered under the national healthcare system/national insurance, just like any other citizens. However, in Ireland, adults living with SMA, classified as persons with disabilities, are exempt from co-payments; otherwise, they would be required to contribute. Conversely, in Russia, physiotherapy doesn't seem to be available or reimbursed.

Belgium partially reimburses consultations with these specialists through the federal government, with Belgian citizens, including adults living with SMA, having a co-payment. In Greece the same applies for physiotherapy, with the remaining three specialties fully reimbursed. Finland and Sweden also require co-payments per visit, but with an annual cap, which can limit healthcare spending for adults living with SMA. In Finland, however, physiotherapy is an exception, being fully reimbursed if the adult living with SMA receives a physiotherapy recommendation from a doctor. In Belgium, those officially recognised as disabled receive increased financial support to cover their medical appointments with specialists.

Regardless of full or partial reimbursement, challenges in accessing certain specialists, especially physiotherapists, were noted. Some countries faced long waiting lists (Czech Republic, Hungary, and Poland), shortages of physiotherapists with SMA expertise (Czech Republic, Portugal, Ukraine), limited reimbursed consultations (France, Germany, Romania, the UK), reimbursement only for the evaluation of motor scales (Spain), and regional variations in reimbursement limits (Italy). Consequently, adults living with SMA seeking physiotherapy may resort to private care, bearing high out-of-pocket costs.

Portugal's national patient organisations stepped in to address this challenge by offering rehabilitation services either in their facilities or at home, albeit for a symbolic cost.

Other access challenges surfaced too. Romania reported a scarcity of pulmonologists and inconsistent availability of nutritionists. In Ukraine, despite full reimbursement, a shortage of scoliosis surgery led adults living with SMA to seek treatment abroad, in locations such as Poland.

Beyond consultation reimbursement, other factors warrant attention, such as reimbursement limitations and access challenges (like specialist shortages, lack of SMA expertise, and healthcare system capacity). These issues force adults living with SMA to seek private or overseas care, incurring full expenses out of pocket. This contributes to the overall burden of SMA on affected individuals and their families, adding financial strain.

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Best practices

In Portugal, the national healthcare system collaborates with private healthcare centres to offer free physiotherapy services. Moreover, the national patient organisation provides a rehabilitation service to adults living with SMA, either at their facilities or home, for a nominal fee. This organisation receives public funding from the social security system to subsidise payments for various professionals working with them, including physiotherapists.

A recap of sub-indicators analysed under this indicator

- **Financial support for consultations with nutritionist**
- **Financial support for consultations with pulmonologist**
- **Financial support for consultations with physiotherapist**
- **Financial support for consultations with scoliosis surgery specialist**

The scoring for HCP consultations reimbursement assesses the extent to which consultations with nutritionists, pulmonologists, physiotherapists and scoliosis surgery specialists are reimbursed for adults living with SMA. It ranges from full reimbursement to no reimbursement.

European map

SMA EUROPE

Assistive equipment and devices support

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Key findings

- In the majority of the countries, despite existing challenges, support is available for purchasing, renting and/or repairing medical and non-medical assistive devices, including those designed for mobility aid.
- In the majority of countries, there are support measures available for employers to modify the workplace.
- With the exception of a few countries, authorities do not usually provide support for purchasing or renting accessible housing options.

Situation analysis

In most countries, assistance is available for purchasing, renting, or repairing medical and non-medical assistive devices, including those for mobility. This aid comes in various forms, from individual subsidies or grants to directly providing new or loaned equipment. However, in many countries, adults living with SMA may need to make a personal contribution, especially for more complex or expensive devices. While some nations like Denmark, Finland, Ireland, Portugal, and Serbia offer these devices free of charge (no co-pay), most require some form of personal contribution. In Sweden, apart from the local support measures determined by individual municipalities, adults living with SMA can receive monthly compensation. This allowance can be used at their discretion, including for this particular purpose.

Some countries like Belgium, Denmark, Finland, Italy, Sweden and the UK offer support for purchasing cars and/or modifying existing ones for persons with disabilities.

There were various challenges identified regarding access to assistive devices, potentially leading to significant out-of-pocket expenses for adults living with SMA. In certain countries like Greece, Hungary, North Macedonia, Serbia, Russia, and Ukraine, financial support covers only a limited range of assistive devices. For instance, in Ukraine, reimbursed devices exclude home adaptation equipment (e.g., lifts to the second floor) and medical devices (e.g., ventilation equipment, pulse oximeters). In Russia, medical equipment is solely provided to those under palliative care, in theory excluding individuals receiving DMTs (while this doesn't happen in practice). Access to electric wheelchairs was also highlighted as problematic in countries such as the Czech Republic. Similarly, in some other countries, including the Czech Republic, obtaining technology-based assistive devices (e.g., voice dictation devices) can be challenging. Moreover, in countries such as Russia, while access to non-medical equipment is available, the process is complex and time-consuming, often resulting in equipment that doesn't adequately meet the needs of individuals living with SMA (e.g. a wheelchair that lacks the proper head and back support). Finland faces challenges due a lack of rules governing the provision of assistive devices, resulting in varying practices, including decisions on who qualifies for such devices, among healthcare areas.

To alleviate the financial burden on adults living with SMA, patient organisations in various countries are intensifying their efforts to ensure timely access to necessary devices. For instance, in Hungary, charity organisations collect second-hand equipment from other countries, while North Macedonia's SMA Europe member organisation operates an equipment bank offering free rentals of rehabilitation and medical aids.

Apart from a select few, including Denmark, Finland, France, Ireland, Poland, Sweden, Switzerland, the Netherlands and the UK, most authorities do not financially support access to accessible housing options for persons with disabilities on an infrastructure level. Where support exists, it's tailored to individual disability needs. Some countries, like Hungary, offer limited home modification aid, often confined to specific regions. Others, like Romania, ease interest on loans taken for home modifications. Despite legislation often requiring accessibility standards, finding accessible housing remains a challenge in countries like Portugal.

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In many countries, employers can receive tax benefits, incentives, subsidies, and cost reimbursements for workplace adaptations. However, this support is not available in certain countries, such as Greece and Hungary and is granted on a case-by-case basis in Romania.

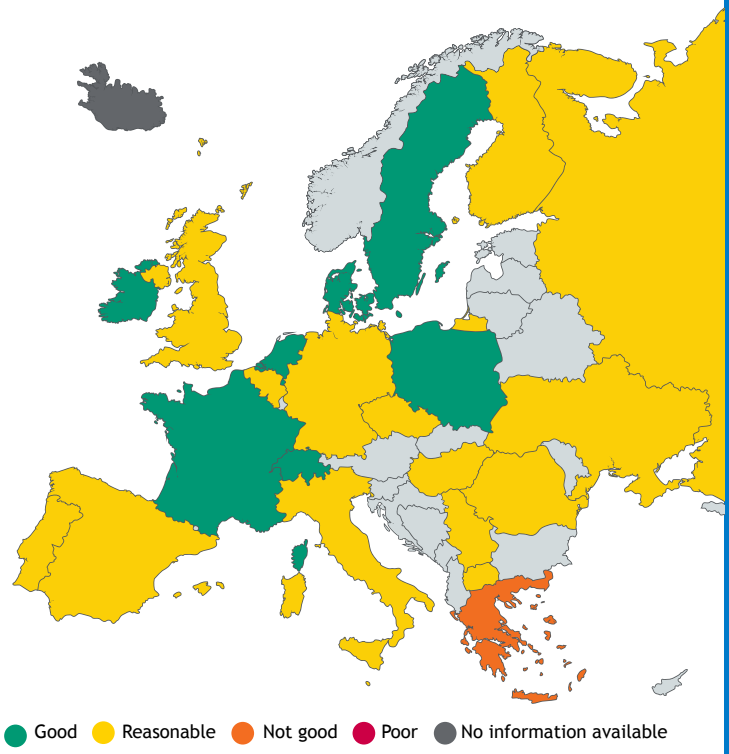
Regional variability in some of the support measures described above was identified in some countries, including Belgium, Finland, Spain, Ukraine and the United Kingdom, making the experience of those living with SMA variable depending on where they live in the country.

Best practices

Persons with disabilities are entitled to comprehensive support in obtaining medical and non-medical assistive devices that enhance their daily lives, both at home and in the workplace, including mobility aids. The National Institute for Rehabilitation (INR) oversees this programme which is funded by the Ministries of Health, Solidarity, Social Security and Education. All adults living with SMA can potentially receive financial assistance for such devices if prescribed by MDTs.

Adults living with SMA or other disabilities can apply to their local authority for a Housing Adaptation Grant for Older People and People with Disabilities. This grant covers home adaptations, such as wheelchair accessibility, home extensions, lower-level bathrooms, or stair lifts.

European map



A recap of sub-indicators analysed under this indicator

- **Availability of financial support for medical equipment and materials**
- **Availability of financial support for non-medical assistive devices**
- **Availability of financial support for workplace modifications**
- **Availability of financial support for mobility aid**
- **Availability of financial support for renting/buying accessible housing options**

The assessment considers the availability of support for persons with disabilities. A high score indicates that some form of support (independent of its quality or challenges associated) is generally available for individuals with disabilities at the national level. Lower scores indicate that there is regional variability in the support measures or that support is determined on a case-by-case basis. The lowest score implies that there is no support available for persons with disabilities.

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Social, education and employment support

Key findings

- Adults living with SMA often have access to social, employment and education support measures available for persons with disabilities.
- However, support measures vary widely across Europe and are more efficient in some countries compared to others in addressing the needs of adults living with SMA.

Situation analysis

Across Europe, adults living with SMA have access to social support, catering to their unique living requirements. These provisions stem from certified disabilities or social vulnerability, acknowledging conditions that might impede full-time employment or necessitate reduced working hours.

Different measures were identified across countries. Financial support ranges from disability-specific allowances like disability or permanent sickness grants, part-time work supplements, invalidity pensions, and ad-hoc grants, free parking certificates, public transport discounts, and tax discounts (e.g. house, mobility). Social vulnerability-related financial support measures include financial guarantees in case of reduced earning capacity, rent support and housing allowances, health insurance coverage, long-term sickness leave, child benefits, tax allowances and tax reductions, and integration assistance. Finally, non-financial assistance covers elements such as architectural accessibility, social care services, counselling, rehabilitation, occupational activities, and transport aid.

However, the uniformity of these measures varies. In countries like Germany, support isn't in general specifically designated for persons with disabilities, while regional discrepancies can also exist, as observed in Spain. In countries such as North Macedonia, social support does not seem to be provided for persons with disabilities.

Similarly, when social support is provided, this does not mean the support is always appropriate or sufficient. Disability allowances, aimed at compensating for an inability to work, often fall short in meeting basic living expenses, a concern raised in countries like Hungary and Ukraine.

Generally speaking, adults living with SMA can access support measures for finding and retaining employment due to their physical disabilities. Some countries implement rules mandating a certain employment percentage for individuals with disabilities. In other cases, compensation is available for employers to motivate them to hire persons with disabilities. This includes tax relief and concessions, wage subsidies for the private sector, compensation for sick pay, and subsidies for internships. Other measures were also identified as direct support for persons with disabilities, namely preferential access to jobs in the public sector, protected workplace schemes, vocational training and re-training, job application assistance, job-matching assistance, self-employment schemes. In some countries subsidies can be provided for hiring and training someone who can assist an employee with a disability in the workplace.

However, regional variations in the support provided persist within countries. This is evident in Belgium, Italy and Spain. In Germany, Hungary, Greece, and Switzerland, employment support for persons with disabilities is either very limited, lacking or at least not designed for this group.

Access to education is another crucial element. Financial assistance such as education subsidies, fee exemptions, and specialised allowances, along with non-financial aids like modified exams (e.g. oral instead of written), extended timelines, infrastructure adjustments, transportation means, supportive aids, and study assistance, ensure educational opportunities. Yet, issues arise in certain countries, including Belgium, Germany, Serbia, and Russia, where infrastructure adaptations in universities don't adequately cater to the needs of individuals living with SMA, particularly older ones. In countries such as Hungary, Italy, North Macedonia, Romania, Russia, Serbia and Ukraine, specific support does not seem to be provided for persons with disabilities.

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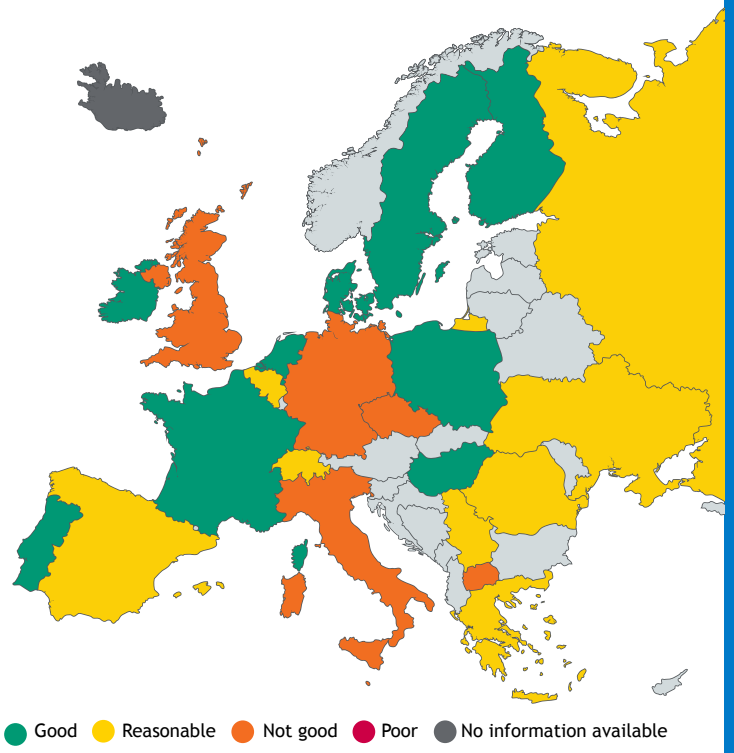
Best practices

In Poland, the State Fund for Rehabilitation of Persons with Disabilities funds several types of training and vocational courses: (1) training aimed at persons with disabilities to acquire job-related skills, undergo retraining, or enhance qualifications; (2) subsidies for three to 12-month job internships; (3) grants for employing individuals to assist employees with disabilities; (4) training for these assistants; and (5) financial aid to support persons with disabilities to start their own businesses.

Further Education and Training (FET) courses are accessible for individuals receiving disability-related social welfare payments. These courses are typically free, and participants might qualify for a training allowance, meal allowance, accommodation allowance, and/or a travel allowance. Individuals with disabilities, receiving specific payments from the Department of Social Protection, can enrol in second- or third-level education courses and receive a Back to Education Allowance.

If adults living with SMA are considered unable to work, they may be eligible for a disability compensation benefit of approximately 800 EUR per month. In cases of part-time employment, the state can supplement the salary to reach 100%.

European map



A recap of sub-indicators analysed under this indicator

- **Availability of social support schemes for adults living with SMA**
- **Availability of employment schemes for adults living with SMA**
- **Availability of access to education schemes for adults living with SMA**

The assessment considers the availability of support for persons with disabilities. A high score indicates that some form of support (independent of its quality or challenges associated) is generally available for individuals with disabilities at the national level. Lower scores indicate that there is regional variability in the support measures or that support is determined on a case-by-case basis. The lowest score implies that there is no support available for persons with disabilities.

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Life assistants/professional caregivers

Key findings

- In the majority of countries, adults living with SMA have the option to access financial assistance in the form of budgets, allowances, grants, or service vouchers. This support enables them to hire life assistants, promoting greater independence in their lives.
- However, challenges persist in certain countries, including inadequate financial support, restrictions on the number of hours or days available per month or year, and a shortage of professional caregivers or life assistants.
- In approximately half of the countries, adults living with SMA have the freedom to choose their own life assistants or receive assistance from relevant organisations in the selection and pairing process.

Situation analysis

A life assistant, also known as a personal assistant, fulfils a crucial role in the lives of adults living with SMA. These dedicated individuals offer not just in-home care but also essential assistance with day-to-day activities and routines, such as aiding with personal hygiene, meal preparation, facilitating outings, and providing mobility assistance. These assistants play a vital role in enhancing the independence and quality of life of adults living with SMA.

In most of the countries covered in the study, adults living with SMA have access to various forms of financial support. This support takes the shape of budgets, allowances, grants, or service vouchers, which empower them to hire life assistants. In some nations, such as Denmark, France, Germany, Poland, Portugal, Spain, Sweden, Switzerland and the Netherlands, the government fully covers the associated costs, ensuring that financial barriers do not hinder access to assistance. In others, such as Greece and Russia, pilot programmes are ongoing so access to a life assistant is still limited.

The extent of the financial support typically varies based on factors like the individual's level of disability or the specific assistance required. This model is employed by around half the countries included in the study.

Even when financial support is available, in certain cases, it falls short of covering the different needs of these individuals. This challenge has been highlighted by various SMA Europe member organisations, including those based in Belgium, Czech Republic, North Macedonia, and Poland. The inadequacy can stem from the low financial support provided or limitations on the number of hours or days of assistance available per month or year. Additionally, in countries such as Belgium, Italy, Spain, and the UK, the level of support may differ from region to region. This variation can lead to disparities. In contrast, some countries offer very limited governmental support to adults living with SMA. This is the case of Hungary, where adults living with SMA are entitled to just half an hour of home care per day.

The recruitment of life assistants varies from one country to another. In North Macedonia and Serbia, recruitment is typically managed by agencies or specific entities, with minimal input from the adults living with SMA in the selection process. In Ukraine and the UK, local authorities are typically responsible for recruitment, and in Ukraine, adults living with SMA can select a life assistant from a list maintained by these authorities. In Belgium and Romania, the choice of the service provider lies with adults living with SMA, who oversee the entire process. In countries such as the Czech Republic, Denmark, Finland, France, Germany, Ireland, Italy, Portugal, Spain, Sweden, Switzerland and the Netherlands, adults living with SMA have the freedom to select their life assistant independently or receive support with the selection and pairing process.

However, the freedom to choose a life assistant is limited by the shortage of professionals identified in many countries, with reasons ranging from compensation, training, complexity and burden. In the Czech Republic, Finland, France, Hungary, North Macedonia, Serbia and the UK, the availability of professional caregivers or life assistants may be limited. For example, in Hungary, the shortage is attributed to professionals seeking better salaries and working conditions abroad. In North Macedonia, the process of becoming a life assistant is seen as complex, potentially dissuading candidates. In Serbia, the scarcity of applicants is linked to low compensation rates.

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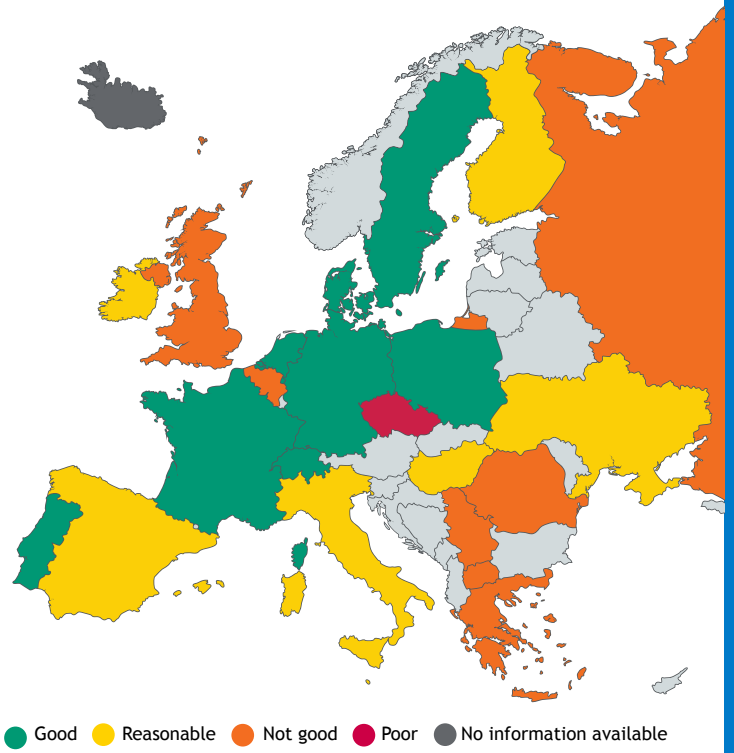
The significance of life assistants lies not only in enhancing the independence of adults living with SMA but also in relieving the burden on their families and friends, who would otherwise need to provide support. Any gaps in this support can have far-reaching effects on the physical and mental health of everyone involved.

Best practices

Adults living with SMA are eligible for compensation from the Social Insurance Agency if they require life assistance for an average of over 20 hours per week to manage various essential needs. The compensation rate falls in the range of 324.50 to 363.44 SEK, equivalent to approximately 29 to 32.5 EUR per hour. Adults living with SMA have the choice of either directly hiring their life assistants or enlisting the services of an organiser, which may be a municipality or a private organisation. For those who require a life assistant for less than 20 hours per week, municipalities are responsible for arranging support services.

In 2017, the Portuguese government initiated the MAVI pilot project (Independent Living Support Model) with the aim of providing life assistance to individuals with disabilities, thereby reducing institutionalisation and dependence on family support. This programme is accessible to adults living with SMA who meet the specified eligibility criteria. As of October 2022, there were 27 active operations within the project, offering support to more than 750 individuals each month, with a budget allocation of 35 million EUR. Building on the success of the pilot project, a permanent support model for independent living is planned for implementation as part of the National Strategy for the Inclusion of People with Disabilities 2021-2025 (ENIPD). Eligible adults living with SMA have the option to select their own life assistant, or they can rely on Independent Life Support Centres (CAVI) for assistance with the selection process. All life assistants undergo specific training to meet the established eligibility criteria.

European map



A recap of sub-indicators analysed under this indicator

- **Financial support for hiring life assistant(s)/professional caregiver(s)**

This indicator is rated across four levels, each signifying a different level of available assistance. At the highest level, individuals can hire one or more life assistants with full reimbursement, incurring no personal costs. Subsequent levels provide partial financial support for hiring life assistants or support provided only in exceptional circumstances. The lowest level indicates a complete absence of financial support for hiring life assistants or professional caregivers.

- **Ability for adults to select their life assistant(s)/professional caregiver(s)**

The highest level of this assessment allows adults living with SMA the freedom to choose whether they want to directly hire their life assistant(s) or opt for assistance from organisations, giving them a voice in the selection process. On the other hand, the lowest level indicates that adults living with SMA rely on third-party organisations that handle the hiring process, without granting them any influence over the selection of their life assistant(s).

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Informal caregivers

Key findings

- Informal caregivers of adults living with SMA receive a wide range of support from national and local authorities, and the nature of this support varies across European regions. It includes monthly full-time care allowances, temporary replacement income, recognition of care hours for pension insurance, extraordinary leave, and paid time off.
- Regrettably, there are still countries where no support is extended to informal caregivers, which not only impacts their well-being but also affects adults living with SMA.
- In many countries, informal caregivers can work as life assistants, provided they meet the relevant eligibility criteria. However, in some nations, this option is not permitted.

Situation analysis

Informal caregivers, individuals who provide unpaid care and assistance to family members or friends living with SMA, receive differing types of support from national and local authorities across Europe. The nature of this support varies not only by country but, in some cases, even within different regions of the same country. These caregivers can receive financial support, which may encompass compensation to make up for income lost due to missed work. For example, in the Czech Republic, Hungary, Poland, Ireland, and Spain, caregivers are eligible for a monthly full-time care allowance, which precludes them from holding another concurrent job. However, in some instances, such as Hungary and Spain, this allowance falls significantly below the minimum wage, rendering it insufficient to cover living expenses.

In several countries, such as Belgium (specific regions), Finland, France, Ireland, North Macedonia, Romania, Russia, and Sweden, caregivers can receive a complementary monthly care allowance while retaining their primary employment. In Portugal, however, to be eligible, the caregiver's household income needs be under a threshold that is lower than the country's minimum wage. Eligibility conditions and the criteria for this support, as well as its budget allocation, may vary among municipalities, and sometimes may be insufficient (e.g. Romania). Belgium and Denmark provide a replacement income for caregivers who need to temporarily interrupt their employment to care for an adult living with SMA. Nevertheless, the income in Belgium is reported to be inadequate to cover living expenses and the duration of its provision is very limited in time.

The Netherlands offers caregivers remuneration for the precise number of informal care hours they provide. Some countries provide additional forms of financial support, such as health insurance for carers (Czech Republic), recognition of caring hours as part of pension insurance (Czech Republic, Germany and the UK), and contributions to accident and unemployment funds based on the number of care hours provided (Germany).

In specific cases, caregivers can adjust their weekly working hours if they are employed, as seen in the Czech Republic. Italy permits caregivers to take extraordinary leave and paid time off from work. Other types of support, such as household benefits, fuel discounts, and assistance with rent payments, are offered in certain countries, as is the case in Ireland.

Unfortunately, informal caregivers in Greece, Serbia and Ukraine do not receive any financial or employment support.

In some countries, informal caregivers can also be hired as life assistants, making them eligible for the support available to this group, with potential eligibility criteria or restrictions in some cases. Conversely, in other countries like Hungary, North Macedonia, Poland, and Portugal, informal caregivers cannot be hired as life assistants by family members, or such hiring is allowed only in exceptional circumstances, as seen in Spain. In a few countries, such as Belgium (some regions), Denmark, Switzerland and the UK, eligibility criteria restricts the family members who can be hired as life assistants. The inability to employ informal caregivers as life assistants is not necessarily negative, as it is generally done to ensure that adults living with SMA are not overly dependent on them.

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Regional variations in support measures for informal caregivers were identified in countries like Belgium and Italy, where the type of support may differ based on the caregiver's place of residence.

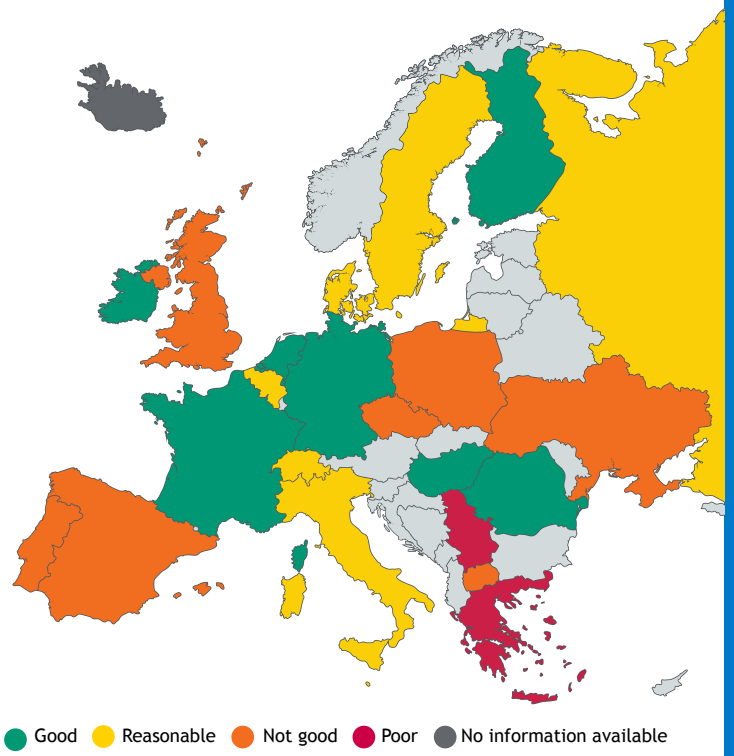
When existing support for informal caregivers is not adequate and sufficient a significant burden is placed on the adult living with SMA and their family, both from a financial perspective (due to missed time at work or the inability to work) as well as on everyone's well-being. This is especially important in countries where there are challenges in access to life assistants, who very much complement the role of the informal caregivers.

Best practices

Family members, close relatives, or friends who act as informal caregivers can access financial assistance. The amount of support varies based on the level of care and assistance needed, commencing at 439.70 EUR per month. To be eligible for this financial aid, the caregiver must establish a contractual agreement with the municipality. The municipality may also provide compensation for extra assistance from a personal assistant, up to a maximum of 40 hours per week, in addition to the allowance granted to the informal caregiver.

Individuals providing informal care to a partner, family member, or friend living with SMA can potentially receive compensation from their personal budget. To qualify for this support, they must have a care agreement in place and will be compensated at the hourly rate designated for Informal Personal Budget (PGB) care providers, which stands at 22.98 EUR per hour as of 2023.

European map



A recap of sub-indicators analysed under this indicator

- **Availability of financial/employment support for informal caregivers**

The assessment considers the availability of financial and/or employment support for informal caregivers. At the highest level, all informal caregivers are entitled to this support. Conversely, the lowest level indicates that there is no support available for informal caregivers in terms of finance and employment.

- **Possibility to hire family members as life assistants**

The assessment evaluates the possibility of hiring family members as life assistants. A high score signifies that any family member can be employed as a life assistant. In contrast, the lowest score indicates that family members cannot be employed as life assistants.

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Rare disease policies and public funding for Patient Organisations

Key findings

- Rare disease actions plans exist or are in development for around half of the countries. However, established actions plans do not specifically prioritise NMDs or SMA in most of the cases.
- From a general perspective, with the exception of some countries, public/governmental funding is not provided for SMA Europe member organisations to enable their activities at the national level.

Situation analysis

In a few countries, including Denmark, Finland, Germany, Poland, Romania, Serbia, Switzerland, Ukraine and the UK, a rare disease action plan (whether individual or integrated in more overarching plans) is currently (or at least until the end of 2022) being implemented. From a general perspective, no measures were identified that directly benefit the NMD and SMA communities. In Ukraine, on the contrary, the current action plan prioritised the establishment of reference centres, including for rare NMDs, which are and will continue benefiting adults living with SMA. In Poland, following advocacy efforts driven by the neurological diseases community, recognising the increasing burden of neurological diseases and inadequate human resources, the government has recently designated neurology as a priority area.

In some countries, such as the Czech Republic, France, Hungary, North Macedonia, and Serbia, a rare disease action plan is currently under development. It is uncertain if any measures will directly or indirectly benefit the NMD and SMA communities. However, in countries such as France and Serbia, it was noted that SMA Europe member organisations are directly involved in the development of these plans, ensuring the SMA community has their interests considered in the development process. In countries such as North Macedonia, the annual rare disease plan usually specifically focuses on pharmacological treatments.

In the remaining countries, a rare diseases action plan was in place in the past but is currently either out of date, never existed, or information about its existence could not be found.

Despite the non-existence of a rare disease action plan, other types of supporting measures were found in countries such as Spain and Sweden. In Spain, a national commission, made up of members of all regions and the Spanish Healthcare Ministry, directs money for improving the care of patients with rare diseases, whose investment focus is decided in each region. In Sweden, the National Board of Health and Welfare funds the Swedish National Agency for Rare Diseases, which seeks to promote collaboration and enhance knowledge in the field of rare health conditions nationwide.

From a general perspective, public/governmental funding is usually not provided for SMA Europe member organisations to enable their activities at the national level. However, there are some exceptions.

In countries such as Belgium, the Czech Republic, Poland, Portugal, and Russia, public funding is provided to SMA Europe member organisations for specific projects. Projects funded across these countries included free loan services for technical and mobility aids, mappings of SMA SoC in NMD centres, personal assistance programmes, and payment of professional services provided through national associations (e.g. physiotherapy, occupational therapy, psychology, legal support), among others. In countries such as Hungary, public funding is provided to the Patient Organisation that participated in this study (non-SMA Europe member) in the form of ongoing grants. Receiving such funding helps the organisations in rolling out key activities that will support adults living with SMA in having a better quality of life and living more independently. When such funding is not provided, organisations must rely on other types of funding sources - from individual donations, to fundraising activities, to pharmaceutical sponsorships - which in some cases may be challenging.

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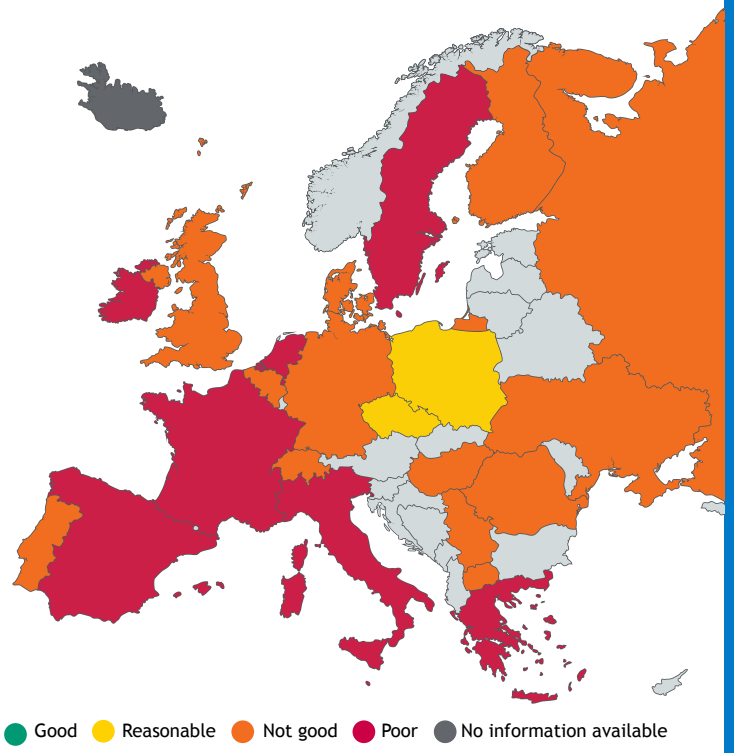
Best practices

SMÁci is currently working on the project 'Improving the care of patients with SMA in medical facilities and expanding the services of the patient organisation.' This project is financed by the EEA Funds for the period 2014-2021. The goal is to map the situation in NMD centres through an anonymous survey and subsequently enhance and standardise the level of care in individual facilities. In addition, their website, which serves as an important initial source of information on SMA, has recently been upgraded thanks to a donation from the Office of the Government of the Czech Republic

France's 4th National Plan for Rare Diseases (PNMR4 is in preparation and the French Muscular Dystrophy Association (AFM-Téléthon) is involved in a few working groups. The Rare Diseases Alliance (AFM is a member) is also leading representation of individuals living with rare diseases in all discussions.

European map



A recap of sub-indicators analysed under this indicator

- **Existence of rare diseases political action plan that addresses NMDs/SMA**

The assessment evaluates the presence of a rare diseases action plan and whether it prioritises actions that directly impact NMDs and SMA. The highest level denotes an action plan that prioritises SMA or NMDs. In contrast, the lowest level signifies an outdated or non-existent plan.

- **Provision of public funding for SMA Europe member organisations**

The evaluation of public funding provision for SMA Europe member organisations highlights the presence or absence of such funding as well as the type of funding. The highest score indicates the existence of public funding in the form of a general grant, while the lowest score signifies the complete absence of public financial support for these patient groups.

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Patient Organisations' support

Key findings

- Almost all countries covered by the report have at least one national patient group supporting adults living with SMA.
- The national SMA or NMD patient group(s) in the vast majority of countries have supporting adults as one of their main priorities.
- National patient groups most commonly facilitate peer-to-peer support, support adults in accessing healthcare services or navigating care pathways, and disseminate information about SMA.

Situation analysis

Overall, countries in the report scored well in patient organisation support for adults living with SMA. In almost all countries, at least one national patient group supporting adults living with SMA exists, with a small number of countries having two or three. In the remaining countries, adults living with SMA can receive support from a national NMD patient group.

In the vast majority of countries covered by the report, the national patient group has supporting adults living with SMA among its main priorities, as opposed to, for example, focusing mainly on supporting under-18s living with SMA. Meanwhile, the few countries that do focus primarily on paediatric SMA also support adults through certain specific initiatives or, in the case of one country (Ireland), provide more ad-hoc support to adults.
In countries where adults have the support of one or more SMA patient groups, they may benefit from initiatives such as support in navigating the healthcare system and care pathways, facilitation of peer-to-peer support networks or other opportunities to share experiences, and dissemination of information about living with SMA.

Some groups directly support adults living with SMA in accessing DMTs and other care, for example by connecting them with specialists or attending medical appointments with adults living with SMA and their families. Others have programmes such as connecting adults living with SMA with life assistants, or to providing them with mobility aids if they cannot otherwise access these.

A small number of groups provide certain healthcare services to adults living with SMA, such as physiotherapy and counselling. In some cases, patient groups also facilitate clinical trials involving adults living with SMA in their country. Others work directly with HCPs, such as physiotherapists, to provide SMA-specific trainings.

A number of patient groups with adults living with SMA as a main priority are also involved in advocating for improved care for adults, greater access to treatment and participation in national and international congresses on NMDs and SMA. Those involved in international dialogues also connect with researchers in other countries or participate in networks to share data.

Even where adults living with SMA rely on the support of a patient group representing adults living with NMDs (including SMA), the majority of these groups have SMA-focused initiatives. This is in addition to general support such as guidance on care pathways, education and career counselling, peer-to-peer support, and more.

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A recap of sub-indicators analysed under this indicator

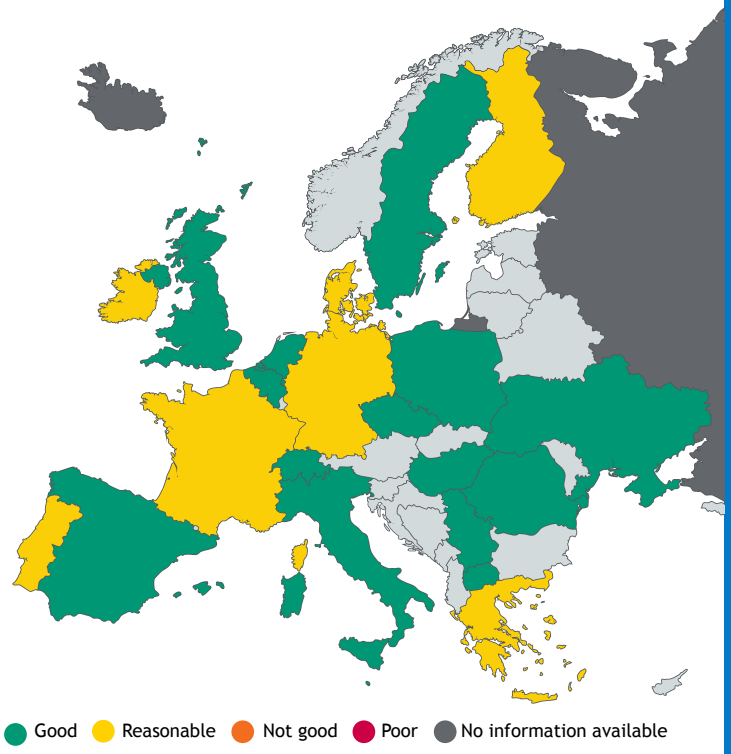
- **Existence of patient groups/networks supporting adults living with SMA**

The scoring for the existence of patient groups and networks supporting adults living with SMA is based on the availability of SMA-specific support from these available nation-wide. The highest score indicates that support is provided by at least one national SMA-specific patient group, while the lowest score reflects the absence of support for adults by patient groups or networks.

- **Level of support provided to adults by existing patient groups/networks**

The scoring for the level of support provided to adults by existing patient groups and networks supporting adults living with SMA is based on the level of adult-specific support provided. The highest score indicates that either the adult SMA population specifically, or both paediatric and adult SMA populations, are among the main priorities of the organisation. In contrast, the lowest score reflects the absence of support provided to adults by patient groups or networks.

European map



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This benchmarking report has been made possible thanks to the invaluable contributions and support provided by several organisations and individuals.

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.

Special recognition is extended to the members of the Expert Advisory Group: Bart Bartels, Stefan Bos, Yasemin Erbas, Jakub Gietka, the late Clare Gray, Anna Kostera-Pruszczyk, Pascal Laforet, Robert Muní Lofra, Valeria Sansone, Juan Vázquez, John Vissing, and Maggie Walter; whose multidisciplinary expertise and valuable guidance have been key in supporting the research project and reviewing the report. They have ensured that unbiased perspectives were driving the outcomes.

Simultaneously, appreciation is extended to the member organisations involved as well as to the Hungarian Association of Muscular Dystrophy Patients for their contribution in participating in the interviews and validating the insights gathered. By disseminating the report's findings, especially with national and local policy- and decision-makers, they will not only contribute to the success of this report, but also ensure that the research serves its intended purpose effectively as an advocacy tool.

Thanks also goes to the healthcare professionals that took part in the survey, whose insights illustrated the realities of care in Europe for adults living with SMA. Without their knowledge, this report would not have come to fruition.

Appreciation also extends to Hall & Partners, the Market Research Agency, and their field partners for managing the survey process, ranging from the questionnaire development to reaching out to local HCPs and monitoring responses.

Lastly, appreciation goes to the project team, namely to the SMA Europe representatives, Laura Gumbert, Nicole Gusset, and Véronique Van Assche, as well as the Roche team involved for driving this project; and to Weber Shandwick Brussels, especially Sofia Brandao, Daniela Negri, and Karen Schober for coordinating, compiling, and managing the project from start to finish.

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NSMA

RehabiliteringsCenter for Muskelsvind

CSMA foundation
Children with Spinal Muscular Atrophy

fundacja SMA

Famiglie SMA
Genitori per la Ricerca sull'Atrofia Muscolare Spinale

MDA ΕΛΛΑΣ
ΣΩΜΑΤΕΙΟ ΦΡΟΝΤΙΔΑΣ ΑΤΟΜΩΝ ΜΕ ΝΕΥΡΟΜΥΪΚΕΣ ΠΑΘΗΣΕΙΣ

SPIERZIEKTEN NEDERLAND

Spinal Muscular Atrophy uk
SUPPORTING • INFORMING • ADVOCATING

SMA FINLAND

DGM
Deutsche Gesellschaft für Muskelkranke e.V.

Smáci

SMAIreland

СМА Семьи

AFMTÉLÉTHON
INNOVER POUR GUERIR

SMA SRBIJA

ASOCIATIA SMACARE

STOP SMA

ABMM ASBL
Logistics - Prêt de matériel

apn

SMA Schweiz
informieren. vernetzen. fordern.

SPIERZIEKTEN VLAANDEREN

SMA EUROPE

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a. What is SMA

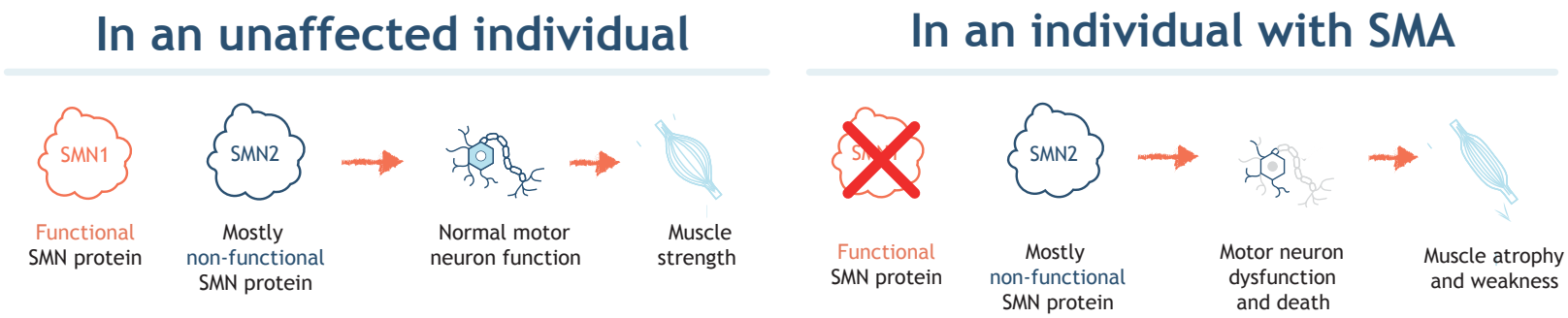
Spinal muscular atrophy (SMA) is a neuromuscular disease that affects one in 6,000-10,000 live births. Because it is characterised by the degeneration of important motor nerves in the spinal cord, people living with SMA experience progressive weakness, atrophy and paralysis of muscles. Untreated, this can result in loss of movement and even death. [21] [22] [23]

SMA is characterised by a wide spectrum of how severely children and adults are affected. The symptoms vary from person to person. SMA may affect daily activities such as breathing, eating, hugging, grabbing, nodding, sitting and walking. [21] [23] [24]

About the cause

SMA is a genetic disease caused by a mutation in the survival motor neuron gene 1 (SMN1). In a healthy person, who carries at least one or two copies of SMN1, this gene produces a protein, the SMN protein, that is critical to the function of the nerves that control muscles. Without the protein, those nerve cells (motoneurons) cannot properly function and eventually die, leading to debilitating and sometimes fatal muscle weakness. Someone develops SMA when they have inherited two faulty copies of the gene — one from each parent. [21] [23] [24] [25]

A second gene also has a role in producing almost the same protein - the survival motor neuron gene 2 (SMN2) - which serves as a backup for the SMN1 gene. The number of SMN2 genes can vary from person to person, and those with more SMN2 copies usually have a less severe form of SMA than those with fewer copies. However, the number of copies of SMN2 does not reliably predict the severity of SMA symptoms an individual will have. [21] [23] [24]



Production of survival motor neuron protein and its link with SMA

In addition, there are other forms of SMA caused by mutations in genes other than the SMN1 gene. [26] However, these won't be in focus of this report.

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Classification of SMA

Historically, SMA was classified into five severity grades (Type 0, 1, 2, 3 and 4) based on age of onset and motor function achieved. [21] [22] [26] For a more in-depth explanation of each SMA classification type, please visit SMA Europe's website.

This classification of SMA is a subject of disagreement between the patient and the medical communities, as individuals living with SMA often do not identify with these predefined categories. SMA exhibits a broad spectrum of severity, with symptoms and their impact differing from one individual to another. Natural history studies confirm the heterogeneity of disease progression, irrespective of the specific type. Further, following the approval of existing DMTs and the implementation of newborn screening for SMA, individuals living with SMA are getting treatment early in life, often even prior to the onset of symptoms, and therefore gaining more physical milestones and abilities than history would predict. [21] [22] [26]

This has led to a more holistic perspective for categorising the spectrum of disease, considering factors such as: 1) the number of SMN2 copies, 2) the age of symptom onset, 3) the maximum and current motor function achieved (e.g. non-sitter, sitter and walker), 4) the impact and severity of symptoms, and 5) the age at first treatment. [21] [26]

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b. Glossary

Disease modifying therapies (DMT)
A category of pharmacological treatments (medicines) designed to alter the course of the disease by targeting the underlying mechanisms responsible for the disease's progression, rather than just alleviating symptoms.

European Medicines Agency (EMA)
Regulatory agency in the European Union responsible for the evaluation, supervision, and regulation of medicinal products.

General practitioner (GP)
A medical doctor who provides primary healthcare services to patients and is often the first point of contact for individuals seeking medical care.

Healthcare professional (HCP)
An individual involved in delivering medical services or healthcare to patients, such as doctors, nurses, pharmacists, and other professionals that provide medical care.

Life assistant
A professional who provides one-to-one assistance for individuals in a home or office setting, enhancing the quality of life, promoting well-being, and facilitating the management of health-related tasks.

Multidisciplinary care
A healthcare approach in which professionals from multiple different disciplines or fields collaborate as a team to provide comprehensive and holistic care to those they serve.

Neuromuscular Disease (NMD)
A group of disorders that that all involve injury or dysfunction of peripheral nerves or muscles.

Neuromuscular Disease (NMD) clinics
A specialised medical facility or department within a hospital or healthcare institution that focuses on the diagnosis, treatment, and management of neuromuscular diseases.

Referral pathways
Structured and standardised processes within healthcare systems that outline the steps and actions required to provide effective and coordinated care for patients

Shared decision making
An approach to healthcare in which patients and healthcare providers collaborate as equal partners to make decisions about the patient's medical treatment or care plan.

SMA treatment centre
A specialised healthcare facility or department within a hospital or medical institution dedicated to the diagnosis, treatment, and care of individuals with SMA.

Standards of care (SoC)
The established guidelines and principles that healthcare professionals adhere to when providing medical treatment and services to individuals, ensuring that they receive safe, effective, and consistent care.

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c. Detailed methodology

Project Team

The project is a collaborative effort between SMA Europe and F. Hoffmann-La Roche Ltd, driven by their shared vision to improving care in the field of SMA. Several members of these two organisations formed the project team. They contributed as subject matter experts, reviewing every step of the study, and working in close collaboration with Weber Shandwick, an independent consultancy that oversaw the coordination, compilation and management of the project. In addition an Expert Advisory Group (EAG) ensured input from the patient and medical communities throughout the study.

Objectives

This benchmarking study was designed to measure and compare how care for adults living with SMA is provided in 23 countries across Europe. This complements the atlas of SMA Europe's "OdySMA initiative". In this atlas, data on access to diagnosis, medicines and care is collected systematically to create visualisations that illustrate access pathways across Europe, providing a comprehensive and organised representation of information related to SMA, to ensure that no one is left behind.

This project was undertaken with three primary objectives:

- To collect quantitative and qualitative data demonstrating the gaps in care for adults living with SMA and the discrepancies between countries;
- To identify patterns and best practices in care for adults living with SMA across Europe; and
- To recommend policy actions and other solutions that can improve the care and overall quality of life of adults living with SMA.

The Expert Advisory Group

As explained above, an Expert Advisory Group (EAG) was created to bring together a representative group of healthcare professionals involved in the multidisciplinary care of adults living with SMA as well as adults living with the disease, covering the various regions of Europe where the research was being carried out.

The role of the EAG was to advise on:

- the benchmarking indicators to be used in this study;
- research methodology;
- the process for resolving discrepancies within the data;
- the content of the report, including recommended stakeholder actions.

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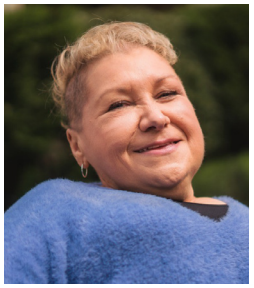
The final composition of the EAG is presented below, and included clinical experts such as neurologists, physiotherapists, and psychologists, as well as adults living with SMA.

Bart Bartels
Associate Professor, Paediatric Physical Therapist, Netherlands SMA Center, UMC Utrecht
Netherlands

Stefan Bos
Adult living with SMA & Member, Adult Committee, SMA Europe & Member, Vereniging Spierziekten Nederland
Netherlands

Yasemin Erbas
Assistant Professor in Psychology, University of Tilburg & Vice President, SMA Europe
Belgium/Netherlands

Jakub Gietka
Adult living with SMA & Member, Adult Committee, SMA Europe & SPAG Member, EURORDIS & Founder, Fundacja Celując w Przyszłość
Poland



Clare Gray
Adult living with SMA & Member, Adult Committee, SMA Europe & Trustee, SMA UK
United Kingdom

Anna Kostera-Pruszczyk
Head of the Department of Neurology, Medical University of Warsaw, ERN EURO NMD
Poland

Pascal Laforet
NMD Specialist, Neurology Department, Raymond-Poincaré Hospital
France

Robert Muni Lofra
Consultant Physiotherapist and Team lead for the Physiotherapy team, John Walton Muscular Dystrophy Research Centre
United Kingdom

Valeria Sansone
Scientific Director, Nemo Clinical Research Center, NEMO Center in Milan
Italy

Juan Vázquez
Coordinator of the Motor Neuron Diseases Unit, Hospital de la Fe
Spain

John Vissing
Director of the Copenhagen Neuromuscular Center, National Hospital, Rigshospitalet
Denmark

Maggie Walter
Associate Professor for Neurology, Nutritional Medicine, Friedrich-Baur-Institute, Department of Neurology, LMU Klinikum, Ludwig-Maximilians-University Munich
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The project phases

To accomplish the main objectives set out above, a structured research methodology was devised, comprising three distinct phases:

Phase 1

Reaching consensus on the data collection methodology and the benchmarking indicators that were used to clearly and objectively assess how care was provided for adults living with SMA in each of the 23 countries.

Phase 2

Data collection and analysis, which included:

- Rapid literature review[7] to identify publicly available sources of information/data for each country;
- Online structured survey[8] targeted at clinical experts treating adults living with SMA across the different countries;
- Semi-structured phone interviews[9] with SMA Europe national members in each country (in a few cases non-members were also interviewed).

Phase 3

Development of Benchmarking Report:

- Consolidated results were summarised in this report, which highlights key gaps, best practices and recommendations for solutions and policy changes that can improve the care and, therefore, the overall quality of life of adults living with SMA.
- The report is accompanied by individual country short reports illustrating the performance of each country against the different indicators.

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[6] Rapid reviews are a form of knowledge synthesis in which components of the systematic review process are simplified or omitted to produce information in a timely manner.
[7] Structured surveys are a systematic and organised method of data collection used to gather information from a sample of individuals or entities in a standardised and consistent manner consisting in a set of predetermined questions, standardisations of questions and response options, and quantitative data which can be analysed statistically.
[8] Semi-structured interviews combine both structured and unstructured elements so that while there is a predetermined set of questions and topics to guide the interview, there is also flexibility for the interviewer to explore additional areas of interest and adapt the conversation based on the participant's responses.

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Spotlight on Phase 1 – Defining data collection methodology

The data collection methodology was agreed upon by SMA Europe, Roche, Weber Shandwick and the EAG.

23 countries are covered by the report, namely:

- **EU Countries:** Belgium, Czech Republic, Denmark, Finland, France, Germany, Greece, Hungary, Ireland, Italy, Netherlands, Poland, Portugal, Romania, Spain, Sweden
- **Other European Countries:** Iceland, Switzerland, North Macedonia, Russia, Serbia, Ukraine, United Kingdom

Today, SMA Europe's membership extends to 25 countries. Turkey and Cyprus are not covered by this report because they became members after the project began.

Categorised under three overarching themes, the 19 indicators (outlined below), guided the data collection.

Organisation of healthcare systems: Looks at how a country organises healthcare and implements internal procedures to deliver healthcare to the individuals living with SMA to promote, restore or maintain health.

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: Looks at the most tangible aspects of the health system as experienced by adults living with SMA.

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: Looks at the access to different types of support that are provided by the government, beyond medical care, to foster independent living of adults with SMA, as well as patient organisation 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

List of overarching areas and indicators analysed in this benchmarking report

These areas were defined by the EAG, SMA Europe, and Roche. More specifically, the selection process involved research conducted by Weber Shandwick, one-on-one discussions with the EAG members, three review rounds with SMA Europe and Roche, and a live meeting with the EAG before finalisation.

As part of this process, a set of sub-indicators were also identified to aid the data collection, analysis and assessment. These are detailed in the full scoring system section.

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Spotlight on Phase 2 - Data Collection

Comprehensive data collection and analysis were carried out through the form of a literature review, an online structured survey with clinical experts, and semi-structured interviews with SMA Europe members in each of the respective participating countries.

Scan this QR code to consult the methodological framework used for this rapid literature review

Rapid literature review

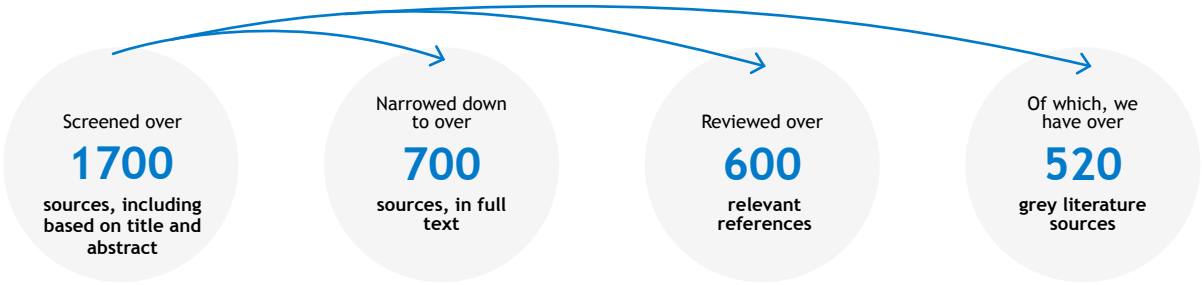
A rapid literature review was undertaken to gather quantitative and qualitative data concerning care of adults living with SMA in Europe, with a focus on comparing the provision of care in the countries in scope. The review encompassed the examination of 1700 references (as shown in the graphic below), such as:

- Scientific and academic articles, namely original articles (e.g., original research papers) and review articles (e.g., systematic reviews, literature reviews, meta-analyses), as well as open-access publications. However, the review excluded methods or methodologies, clinical case studies, clinical trial papers, notes and news, book reviews, perspective/opinion articles, and paid-access publications.
- Information sourced from relevant national organisations (grey literature), which included governmental bodies, health authorities, medical societies, and patient organisations.

Inclusion criteria for the literature review involved publications:

- Focus on the 23 countries in scope.
- Issued either in English or in any of their respective official languages.
- Issued over the past five years.
- Extracted from PubMed and the Cochrane Library. Additionally, the references of relevant articles were checked to identify other potentially useful articles. As mentioned above, if information was not available through these databases, relevant national websites were consulted for data, including grey literature.

One reviewer was designated for each country to screen articles and grey literature, and to extract, synthesise, and report the relevant data for that country in English (with the limitation of quality assessment). In the end, reviewers across all countries:



Number of sources screened, reviewed, and used as relevant references in the rapid literature review

A separate document was generated for each country, and the rapid review findings were reported in English, adhering to a standardised template. The data was presented using narrative synthesis, meaning that words and text, in a narrative style, were used to summarise and explain the findings of the rapid review.

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Online structured survey

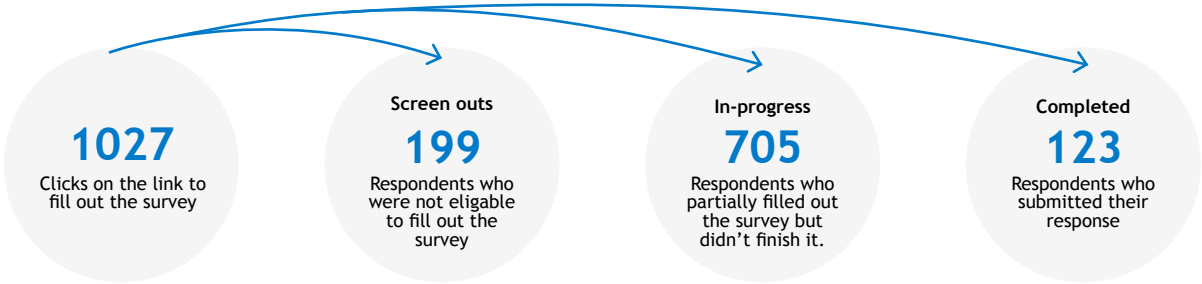
In parallel with the rapid literature review, an online structured quantitative survey was carried out with healthcare professionals treating adults living with SMA across the 23 countries. The objective was to gain a deeper understanding on how adults living with SMA were managed in their respective countries, with a focus on indicators across the areas: healthcare systems organisation and healthcare delivery.

To be eligible to participate in the survey, HCPs had to:

- be directly involved in the care of adults with SMA (e.g. neurologists and physiotherapists),
- have ≥two years of experience,
- have seen ≥five patients over the last two years;

The survey, which took on average 16 minutes to complete, was conducted by Hall and Partners, an independent market research agency. Over the course of approximately 10 weeks, that the survey was open, including a fielding period between January and March 2023, they logged:

Scan this QR code to consult the final questionnaire used for this online structured survey



Number of screen outs, in-progress and completed responses in the online structured survey

The final sample included 123 healthcare professionals, among which there were 116 neurologists, six physiotherapists and one specialist nurse.

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It is worth noting that, for each country, a target number of healthcare professionals was established based on the number of treating centres in the countries relative to the size of the country. This target varied between two and 15, given the rare nature of this disease and the focus on adults living with SMA only. The survey was distributed in English with the exception of the following countries, where the survey was translated into the local languages, based on recommendations from our market research agency, to maximise recruitment: Czech Republic, Hungary, North Macedonia, Romania, Russia and Ukraine.

Below you can find a full breakdown of targets and actual survey completions for each of the countries covered by the report:

			Breakdown of responding HCPs		
	Target completions	Survey completions	Neurologist	Physiotherapist	Nurse (IRE)
Belgium	5	3	3		
Czech Republic	5	3	3		
Denmark	5	5	5		
Finland	2	2	2		
France	15	10	10		
Germany	15	15	15		
Greece	2	2	2		
Hungary	5	5	4	1	
Iceland	2	0			
Ireland	2	1			1
Italy	15	16	16		
Netherlands	5	5	4	1	
North Macedonia	2	1	1		
Poland	10	9	9	1	
Portugal	5	5	5		
Romania	5	5	5		
Russia	5	2	1	1	
Serbia	2	1	1		
Switzerland	5	5	5		
UK	10	10	10		
Ukraine	5	3	1	2	
Spain	15	11	11		
Sweden	5	3	3		

Number of target responses vs online structured survey completions for each country

The target was not reached for Belgium, Czech Republic, Ireland, North Macedonia, Russia, Serbia, Sweden and Ukraine. No responses were received for Iceland. For more information on reasons why we may not have reached this target, please refer to the limitations section.

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Semi-structured phone interviews

The final data collection source came from phone interviews which were held with SMA Europe members in each of the 23 countries in scope. The interviews were conducted with a representative from the national patient organisation who were best placed to give an overview of the challenges facing adults living with SMA in their respective countries.

All interviews were conducted in English, with the exception of Portugal and Serbia, where the interviews were carried out in the local language.

In general, the interviews lasted for an hour and were led by Roche, with the support of Weber Shandwick. Topics covered during these interviews encompassed almost the full range of indicators across all three main overarching areas - organisation of the healthcare system, delivery of healthcare and governmental and peer support.

Scan this QR code to consult the final interview guide used to guide these semi-structured phone interviews

Given the semi-structured nature of the interviews and the time available for the interview, we were (1) not able to cover all topics across all countries, and (2) the list of topics discussed differed from interview to interview. To address this challenge, all interviewed patient groups were given the opportunity to review the individual country short reports that accompany this report, therefore being able to provide any additional pertinent information or feedback on the insights collected.

It is also important to note that:

- No interviews took place with SMA Europe members based in Iceland and Spain. In Iceland, we were unable to engage with the local patient group despite our best efforts. In Spain, patient representatives declined to participate in the interview, explaining that they lacked a comprehensive understanding of how the health system competencies and resources are managed by each autonomous community (as it is not a centralised system). Nevertheless, this decision underscores the commitment of FundAME to provide well-informed and competent responses, as they want to ensure the highest level of expertise in addressing the questions.
- For Hungary, the patient group interviewed was the Hungarian Association of Muscular Dystrophy Patients (a non-SMA Europe member).

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Spotlight on Phase 3 – Data analysis and development of Benchmarking Report

Following the data collection phase, Weber Shandwick independently conducted the data analysis, using a structured scoring system developed in collaboration with the project team (see below for more information about its development).

Scoring system development

To facilitate the analysis and comprehensive assessment of the collected data, a well-structured scoring system was devised.

The first step was to agree with the project team on the benchmarking methodology that would be used to compare countries across the final list of indicators and sub-indicators. After consideration, the project team decided to move forward with a colour-coded, four-point scale, where a score of one denoted the worst-case scenario, and a score of four indicated the best-case scenario. The number zero denoted a lack of information, making it impossible to assign a score.

The full breakdown of the four-point scale to be attributed to each indicator and sub-indicator was as follows:

- All indicators would follow a standardised four-point scale (4: good; 3: reasonable; 2: poor; 1: bad). The final score of an indicator would be derived by calculating the average score of the sub-indicators that constituted it, not taking into account those sub-indicators assigned with the number zero (no information available). Averages would always be rounded down.
- Each sub-indicator would follow its own tailored four-point scale, meaning that four specific scenarios would have to be determined for each sub-indicator.

The four specific situations that would allow us to assign scores in a tailored manner to the unique scenarios of each sub-indicator were developed and validated with the project team.

Full scoring system

See below the final version of the scoring system:

Benchmarking indicators	Sub-indicators	Sub-indicators scoring explanation
1. Transition from paediatric to adult care services	Existence of transition care protocols/policies	4. National protocols/policies exist and are well implemented throughout the country 3. National protocols/policies exist and are well implemented in some treatment centres 2. Each treatment centre implements its own transition protocols/policies 1. There are no transition policies/protocols
	Need for requalification for access to pharmacological treatment	4. Not needed 3. Needed, but without administrative burden for the patient 2. Needed and with administrative burden for the patient
2. Navigation in the healthcare system	Ease of navigating the healthcare system	4. Adults living with SMA easily navigate the healthcare system 3. Gatekeepers to care exist, but once overcome, care is easily accessed 2. Adults living with SMA face some challenges navigating the healthcare system 1. Adults living with SMA face significant challenges in navigating the healthcare system
	Existence of care manager	4. A care manager usually exists in SMA treatment centres 3. A care manager exists in few SMA treatment centres 2. A care manager does not exist

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3. Access to SMA treatment centres	Percentage of treatment centres treating adults	4. All or almost all (>85% and ≤100%) 3. More than half (>50% and ≤85%) 2. Half or less than half (>15% and ≤50%) 1. None or almost none (≥0% and ≤15%)
	Geographic distribution of adult treatment centres	4. There is an even distribution of SMA/NMD centres 3. There is an even distribution of general neurology clinics that offer care for adults with SMA 2. There is an even distribution only of less specialist care in other settings (e.g. primary care) 1. Uneven distribution of treatment centres
	Percentage of adults that are followed in SMA or NMD reference centres/ clinics	4. All or almost all (>85% and ≤100%) 3. More than half (>50% and ≤85%) 2. Half or less than half (>15% and ≤50%) 1. None or almost none (≥0% and ≤15%)
4. Network of treatment centres	Existence of collaborative network of treatment centres	4. A formal network exists with a shared purpose 3. No formal network exists but treatment centres usually collaborate with each other 1. No network or collaboration mechanism exists
5. Patient registry	Existence of patient registries collecting data on adults	4. At least one registry exists that collects data on adults with SMA 3. Is being created and will collect data on adults with SMA 2. Existed in the past/is currently not functional 1. There aren't any patient registries
	Percentage of treatment centres that participate in an existing registry	4. The majority (>50%) 3. Limited number (>25% and ≤50%) 1. None or almost none (≥0% and ≤25%)
6. Standards of care (SoC) & treatment guidelines	Existence of SoC for diagnosis and management of adult SMA	4. SMA SoC exist that cover the specific needs of adults 3. SMA SoC are under development/revision 2. SMA SoC that exist do not cover the specific needs of adults 1. SMA SoC do not exist
	Existence of DMTs treatment guidelines	4. Exist and provide treatment guidelines for all available DMTs 3. Do not exist but are under development/are outdated but being revised 2. Exist but are outdated OR do not cover all available DMTs 1. Do not exist
	Use of outcomes measures used in standard clinical practice	4. Mandatory use in standard clinical practice for eligibility to reimbursement 3. Used in standard clinical practice on a voluntary basis 1. Not used in standard clinical practice
7. Multidisciplinary care	Availability and access to multidisciplinary care teams (MDTs) for adults with SMA	4. MDTs are (in general) available; HCPs are usually based in the same treatment centre and they meet to discuss care 3. MDTs are (in general) available; HCPs are usually based in different treatment centres and they meet to discuss care 2. MDTs are (in general) not available, but patients have access to different specialists they may need 1. MDTs are (in general) not available and patients do not have access to the different specialists they may need

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8. Shared decision-making	Implementation of shared decision-making in standard clinical practice	4. Integral part of standard clinical practice 3. Implemented inconsistently and may vary from HCP to HCP 2. Not part of standard clinical practice but HCPs engage adults upon request 1. Not part of standard clinical practice
	Existence of information/ tools to empower adults to take informed decisions about their health	4. Existing information/tools are appropriate and easily accessible online 3. Existing information/tools are appropriate but not easily accessible online 2. Existing information/tools are not appropriate 1. Information/tools are not available for adults 0. Information is not available
9. Continuity of care	Mandatory regular follow-ups	4. Mandatory for at least a specific group of adults living with SMA (e.g. those that are under disease modifying therapies) 3. Mandatory for a specific group of adults living with SMA (e.g. those that are under disease modifying therapies) but follow-up is suboptimal 2. Entirely voluntary and up to the patient to fulfil 1. Entirely voluntary and up to the patient to fulfil, and patients are in general not being followed-up
	Processes in place to motivate adults with SMA to continue engaged with healthcare services	4. Processes/strategies are in place 1. Processes/strategies are not in place
10. Care team & caregivers training	Existence of SMA training programs for HCPs	4. Formal SMA trainings are delivered in key treatment centres or via medical societies 3. Continuous medical education is available 2. A one-off training is done at university, as part of specialist training 1. No training programs cover SMA
	Existence of SMA training programs for life assistants	4. Available and easily accessed 3. Available but access is difficult 2. Informal training opportunities are available, access might be limited 1. No training opportunities are available
	Existence of SMA training programs for informal caregivers	
11. Access to EMA-approved disease modifying therapies (DMTs)	Access to *nusinersen*	4. Available and fully reimbursed for adults living with SMA according to the EMA label 3. Restrictions apply making access more restrictive than EMA label for adults living with SMA more 2. Adults are not yet eligible for reimbursement but HTA processes are currently ongoing OR adults are eligible for reimbursement on a case by case/exceptional basis 1. Not available or adults are not eligible for reimbursement
	Access to *risdiplam*	
12. Access to unauthorised disease modifying therapies (DMTs)	Availability of pharma sponsored clinical trials for adults with SMA (2018-2022)	4. High priority market (>5 clinical trials available in 2018-2022) 3. Medium priority market (3-5 clinical trials available in 2018-2022) 2. Low priority market (1-2 clinical trials available in 2018-2022) 1. Non-priority market (0 clinical trials available in 2018-2022)
	Availability of pharma sponsored Early Access Programs (EAP) for adults with SMA (2018-2022)	4. EAPs have been rolled out for which at least some adults were eligible 3. EAPs have been rolled out but adults were not eligible 2. No EAPs have been rolled out 1. Country's legislation does not allow for the roll out of EAPs

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13. HCP consultations reimbursiment	Financial support for consultations with nutritionist	4. Fully reimbursed for all adults 3. Partially reimbursed for all adults 2. Fully or partially reimbursed but only for some adults 1. Not reimbursed for adults
	Financial support for consultations with pulmonologist	
	Financial support for consultations with physiotherapist	
	Financial support for consultations with scoliosis surgery specialist	
14. Assistive equipment and devices support	Availability of financial support for medical equipment and materials	4. Some type of support is available for persons with disabilities at national level 3. Some type of support is available for persons with disabilities, but only in some regions/ with regional variability 2. Some type of support is available for persons with disabilities, but determined on a case-by-case basis 1. There is no support available for persons with disabilities
	Availability of financial support for non-medical assistive devices	
	Availability of financial support for workplace modifications	
	Availability of financial support for mobility aid	
	Availability of financial support for renting/buying accessible housing options	
15. Social, education and employment support	Availability of social support schemes for adults with SMA	
	Availability of employment schemes for adults with SMA	
	Availability of access to education schemes for adults with SMA	
16. Life assistants/ professional caregivers	Financial support for hiring life assistant(s)/ professional caregiver(s)	4. One or more life assistants can be hired at no cost (full reimbursement) 3. One or more life assistants can be hired with partial financial support (partial reimbursement) 2. A life assistant can be hired (full or partial reimbursement) in exceptional circumstances 1. There is no financial support for hiring a life assistant
	Ability for adults to select their life assistant(s)/ professional caregiver(s)	4. Adults living with SMA can choose whether to directly hire their life assistant(s) or to partner with organisations that facilitate this for them 3. Adults living with SMA need to rely on third party organisations that facilitate this for them, having a say on who their life assistant(s) will be

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		2. Adults living with SMA need to directly hire their life assistant(s), and are in charge of the whole process 1. Adults living with SMA need to rely on third party organisations that facilitate this for them, not having a say on who their life assistant(s) will be
17. Informal caregivers	Availability of financial/ employment support for informal caregivers	4. All informal caregivers are entitled to financial/employment support 3. Only some informal caregivers are entitled to financial/employment support 2. Informal caregivers are not entitled to financial/employment support, but to other support programs 1. There is no support for informal caregivers
	Possibility to hire family members as life assistants	4. Any family members can be employed as life assistants 3. Only some family members can be employed as life assistants 2. Family members can only be employed as life assistants in exceptional circumstances 1. Family members cannot be employed as life assistants
18. Rare disease policies and public funding for Patient Organisations	Existence of rare diseases political action plan that addresses NMDs/SMA	4. Currently ongoing and prioritises SMA or NMDs 3. Currently ongoing/just finished (in 2022) but does/did not prioritise SMA or NMDs 2. Currently being developed 1. Currently out of date (finished by 2021) or non-existent
	Provision of public funding for SMA Europe member organisation	4. Public funding is provided on the basis of ongoing grants 3. Public funding is provided for specific projects 1. No public funding is provided
19. Patient Organisations' Support	Existence of patient groups/networks supporting adults living with SMA	4. Support is provided by at least one national SMA patient group 3. Support is provided by at least one national NMDs patient group 2. Support is only provided by regional or local SMA/NMDs patient groups/networks 1. No support is provided to adults by patient groups/networks
	Level of support provided to adults by existing patient groups/networks	4. Pediatric and adult SMA are the main priorities of the organisation 3. Some support is provided to adults but the pediatric population is the main priority 2. Only ad hoc support is provided to adults 1. No support is provided to adults

Data analysis

For each country, Weber Shandwick looked at the sub-indicator data available for each of the three data collection methods and scored the country against the scoring criteria, developing a summary of the situation for that given country across each of the sub-indicators. Based on the sub-indicators' scoring, scorings were attributed to the indicators, taking the average scores awarded to the different sub-indicators that constituted it. Where discrepancies arose, an adjudication process was followed, as agreed with the EAG and the project team.

Consolidated results were summarised and have been included in this current benchmarking report, which highlights gaps, best practices and recommendations for stakeholder actions and other solutions that can improve the care and, therefore, the overall quality of life of adults living with SMA. The report is accompanied by individual country short reports illustrating the performance of each country regarding different indicators.

The individual country short reports were reviewed by representatives from the member organisations of SMA Europe and the Roche affiliate in the given country, as well as the EAG member for that country (if applicable). The benchmarking report was reviewed and validated by the project team. Because we were not able to engage participants in Iceland for either the HCP survey, or the phone interviews, we were unable to include Iceland in the final report.

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Limitations

Limitations specific to the data collection methods

1) Rapid literature review

A rapid literature review was chosen over a systematic review due to its expediency, which reduced the timeframe and resources required for the exercise. The limitations were that it may have potentially resulted in overlooked relevant studies or limited exposure to varying perspectives. A more comprehensive and exhaustive literature review would have offered a broader foundation for the research, strengthening its theoretical framework and supporting a more comprehensive analysis of the subject matter.

Even so, it was interesting to verify that the majority of the relevant references identified came from grey literature rather than academic or scientific articles. This insight might be useful in future replications of this benchmarking report.

2) Online structured survey

During the analysis of the survey results, significant disparities were frequently observed between responses from HCPs within the same country. This variance may point to regional differences, lack of awareness of the availability of items assessed under the indicators, inter-hospital variability or different interpretations of the questions. Due to the strict inclusion criteria, all participants were actively treating adults living with SMA and, as such, the disparity of responses within a singular country is a finding in itself. Future iterations of this benchmarking report should endeavour to be powered to uncover regional variations in more detail. Furthermore, in most of the countries the survey was rolled out in English, which could potentially lead to misinterpretations of the survey questions and possible responses.

Unfortunately, the target number of desired completions was not reached for a number of countries: Belgium, Czech Republic, Ireland, North Macedonia, Russia, Serbia, Sweden and Ukraine. This may have been due to various reasons:

- The limited number of specialist HCPs in some countries;
- Difficulties experienced by the market research agency in identifying the relevant HCPs;
- HCPs' unwillingness to participate in market research;
- The ongoing conflict between Russia and Ukraine.

3) Semi-structured phone interviews

We recognise that conducting only one semi-structured interview per country, involving one to two patient representatives from the local patient organisation with the aim of obtaining an overview of their members' experiences, may introduce bias and limit the overall perspective of the country's landscape. Additionally, similarly to the HCPs survey, it's important to acknowledge that the interviews were mostly conducted in English, a second language for many participants. This may potentially have led to discrepancies, misinterpretations, or missed nuances in the responses. For future iterations of the report, it is recommended to address this limitation by conducting interviews in the local language, and/or conducting more than one interview per country, and/or increasing the number of representatives to be interviewed.

Conclusion

This analysis represents the first time a study of this nature has been conducted in the area of SMA. Furthermore, the methodology employed in this study exhibits limitations that merit consideration, which have already been described throughout this section.

Addressing some of these limitations in future iterations of the methodology will enhance the study's robustness, enrich its findings, and contribute to a more robust body of knowledge in the field.

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