SMA EUROPE

European Patient Experience Survey on SMA Medicines, Access, and Treatment Journeys

One Goal Series

Together We Prioritise

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The SMA treatment journey at a glance

The SMA treatment journey is changing, but unmet needs remain

STAGE 1 · DIAGNOSIS
Unequal starting points

15%
Moved country to access care

58%
Had no treatments or trials available at diagnosis

Where you live still determines your starting point

STAGE 2 · TREATMENT DECISION-MAKING
Hopeful decisions, but limited choice

31%
Had a choice between SMA medicines

94%
Hopeful

56%
Anxious

Decisions are made under uncertainty, and emotion

STAGE 3 · STARTING TREATMENT
A moment of risk and opportunity

85%
Saw treatment start as an **opportunity**

41%
Saw treatment start as a **risk**

55%
Expected **improvement**

34%
Expected **stabilisation**

Treatment begins with both optimism and caution

STAGE 4 · LIVING WITH TREATMENT

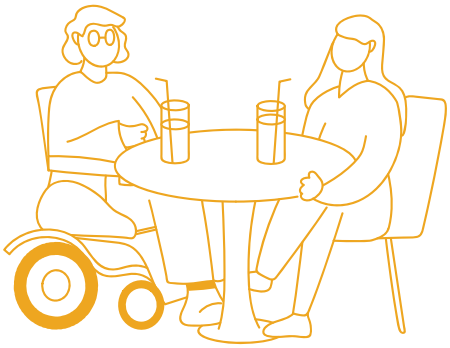
Improvement and stabilisation dominate

67%
Endurance improved

61%
Strength improved

53%
Breathing more stable

Treatment brings real-world gains, but not in every domain



STAGE 5 · SWITCHING AND INTERRUPTING TREATMENT

Looking for the right fit and experiences of interrupting treatment

13%
Switched due to lack of improvement

19%
Experienced treatment **interruption**

47%
Did not know the **potential consequences**

Continuity matters. Interrupting treatment has consequences

STAGE 6 · LOOKING AHEAD

Clear call for next-generation therapies

82%
Think there is a need for **new medicines in SMA**

89%
Want **muscle-targeting** therapies

The community needs multisystem, add-on, and future therapies

Acknowledgements

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Finally, we thank every person living with SMA and every family member or caregiver who took part in this survey. Your lived experience and generosity in sharing your stories make this work possible and meaningful. This study was supported by the Cure SMA Industry Collaboration, SMA Europe, and Tilburg University.

SMA Europe

SMA Europe is a non-profit umbrella organisation of spinal muscular atrophy (SMA) patient organisations from across Europe. Together, through greater understanding, we work towards creating a better world for all those living with SMA.

United, at European level and in collaboration with all stakeholders, we achieve our vision of creating a better world for all those living with SMA. This is our reason for being, above all else. It infuses everything we do and is what we invite others to help us achieve.

SMA Europe works to bring effective treatments and optimal care to everyone affected by SMA.

Research

We accelerate breakthrough research in SMA through our biennial call for research proposals and our scientific congresses.

Advocacy

We use evidence-based advocacy and represent the voice of patients in decisions that affect them.

Awareness and outreach

We campaign to raise awareness for SMA and promote the interests of our community.

We always put the voice of those affected by SMA at the heart of everything we do by:

Empowering our member organisations to advocate and campaign for them at a national level; and

Influencing healthcare, pharmaceutical, and regulatory decision-makers to involve them at an international level.

Only through true representation of the SMA community – the unique wants, experiences and aspirations of the people behind the condition – will their voices be heard and needs be met.

The EUPESMA Survey Series

Our European Patient Experience Survey (EUPESMA) Series is designed to map the experiences and expectations of people living with SMA and to ascertain their needs and wants. With this evidence, SMA Europe advocates for equitable access to optimal treatment and care in Europe.

Why do we need Pan-European patient experience surveys?

Access to treatment and care solutions remains fragmented and unequal. **Community-generated data gives a credible, collective voice to advocate** for fairer policies and equal access for all.

By gathering data directly from individuals and families living with SMA, **surveys translate lived experience** into reliable evidence that reflects everyday realities.

People living with SMA have diverse experiences and needs. Community surveys ensure that every voice is heard, because each single one of them is equally important.

Europe-wide results show where care and treatment work well and where gaps remain, helping to **understand expectations, design solutions** that respond to real needs, and **generate tangible impact.**

What is Patient Experience Data?

Patient Experience Data (PED) refers to information that reflects the experiences, needs and priorities of people living with SMA and their families, including their daily lives, challenges, expectations, and treatment experiences. PED has the power to capture the needs of the community and what matters in real life, beyond clinical measures.

This data helps:

- make care more responsive
- guide research priorities
- identify inequalities
- inform decisions in health systems and policy

EUPESMA is Europe's largest source of patient experience data for SMA.

SMA Europe uses this data to inform medicine development companies, clinical stakeholders, and regulators like the European Medicines Agency (EMA) on disease impact, autonomy, and expectations of current therapeutic development.

In 2025, the EMA has issued a reflection paper on PED, setting a first stone towards the systematic inclusion of this type of data in medicine development and marketing authorisation applications.

Strong of its longstanding experience with PED and with the contributions of its members, SMA Europe has shared its feedback on this draft text with the EMA to steer the conversation on how to include PED in regulatory decision while protecting the interests of people living with SMA.

EUPESMA 2025 Survey on SMA and Treatment Experiences

The European Patient Experience Survey on SMA (EUPESMA) is SMA Europe's flagship effort to gather patient experience data directly from individuals and families affected by spinal muscular atrophy across Europe and beyond.

People living with SMA and their caretakers have a central role in designing, analysing, and interpreting EUPESMA surveys to maximise representation of what matters to the SMA community.

This cross-European survey **investigates the realities and expectations of SMA patients and caregivers** regarding treatment decision-making in the current therapeutic landscape. It explores experiences with switching, bridging, and add-on therapies, with a focus on aligning care with patients' individual goals and improving shared decision-making.

How to read this report

This report describes the experiences and perceptions shared directly by people living with SMA and their families across Europe. It reflects patient and family voices, not medical evaluations or treatment comparisons.

- All findings represent what respondents chose to share.
- The report does not recommend one treatment over another.
- It does not replace clinical advice.
- It highlights themes, challenges, and priorities based on real-world experience.

The goal is to support better understanding, more equitable care, and more informed decision-making, for the community, clinicians, researchers, and policymakers.

The study aims to:

- **Map real-world treatment experiences** and preferences across Europe
- **Identify factors** influencing therapeutic choices in an evolving treatment landscape
- **Support patient-centred care and advocacy** efforts regarding access, continuity, and personalisation of treatment methods

The study was approved by **Tilburg University Research Ethics' Committee.**

The survey was developed through a **community-led, participatory research approach** and guided by focus groups with expert members of SMA Europe's TC, interviews with healthcare professionals, and a literature review.

The community survey **was translated into 18 languages** and distributed through SMA Europe's networks and member organisations across Europe.

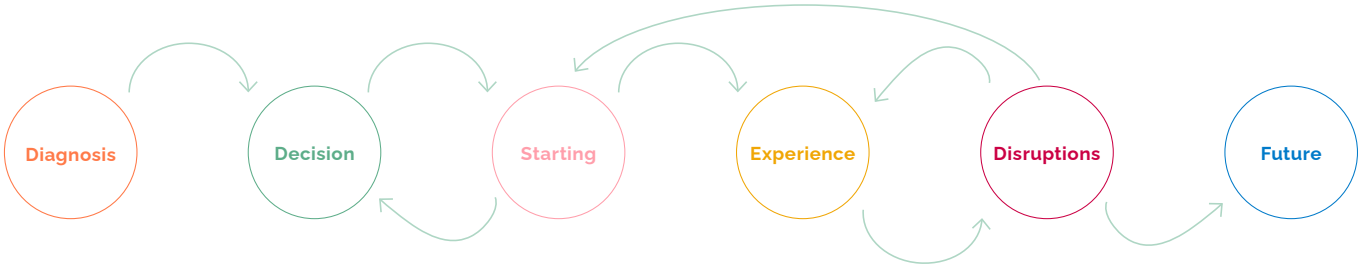
[1] **Methodological note:** All responses to our survey questions were optional and participants could skip any question. The data and percentages reported in this document reflect the proportion of people who responded to each question.

The SMA patient journey - Understanding the treatment experiences shared in EUPESMA 2025

What do we mean by 'patient journey'?

The 'patient journey' describes the series of experiences, decisions, challenges, and changes a person goes through from the moment of first symptoms or diagnosis to their daily life with SMA and their outlook on their future.

In the context of this report, the patient journey includes:



Diagnosis and early information:
What was available at the time of diagnosis, what families were told, and how inequalities emerged from the start.

Deciding whether and how to start treatment
Expectations, emotions, information needs, and the support families received at this pivotal moment.

Living with treatment over time
Real-life effects on daily life, symptoms, functioning, and wellbeing — including stabilisation and improvements.

Switching between treatments
Why people switch, how switches are experienced, and what families hope to achieve.

Disruptions in treatment
The reasons behind stopping or interrupting treatment, the consequences, and the uncertainties families face.

Looking ahead
What the community wants from SMA medicines in the future and what motivates participation in clinical research.

This journey is rarely linear. People move back and forth between stages as their needs, circumstances, and treatment options evolve.

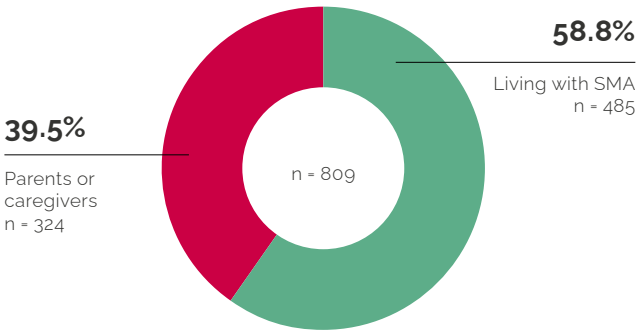
By organising the report around the patient journey, we aim to reflect real life as closely as possible, honouring the voices of people living with SMA and highlighting the points where support, information, and equity matter most.

Who participated in this survey?

The EUPESMA 2025 Survey collected responses from **826 individuals from 41 countries**, representing one of the most diverse and comprehensive community-driven datasets on SMA treatment experiences in Europe. Respondents included people living with SMA themselves as well as parents and caregivers, reflecting a broad range of lived realities across the lifespan.

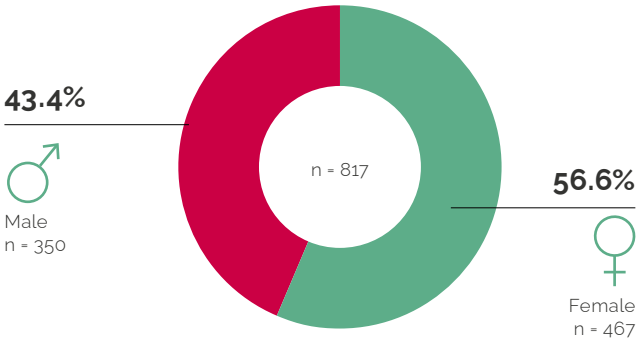
Responses were collected in early 2025, capturing current realities in the rapidly evolving treatment landscape.

Profile of respondents



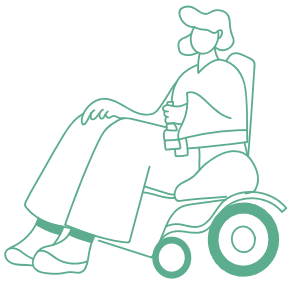
A small number of responses were submitted by other family members or representatives

Sex assigned at birth

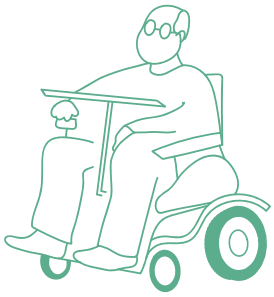


Age

Minimum age
3 months



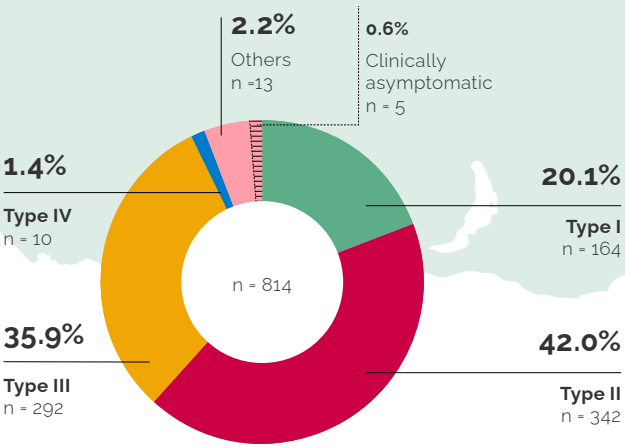
Average age
28 years



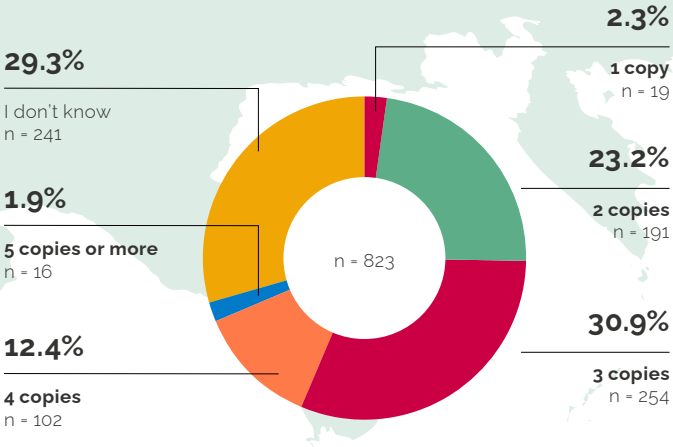
Maximum age
81 years

DISEASE SEVERITY

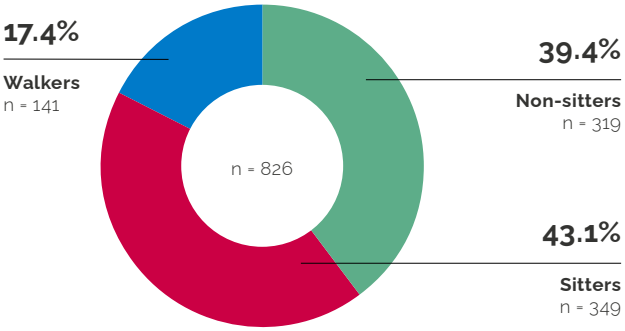
SMA types[1]



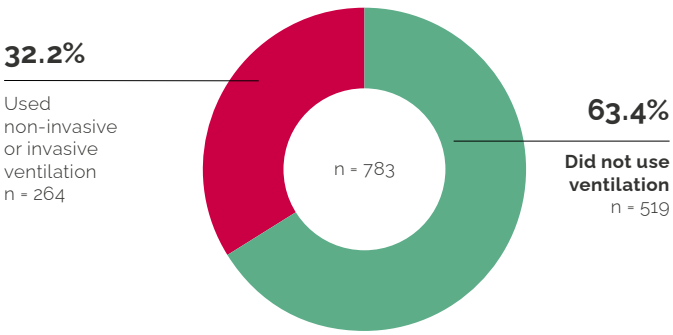
SMN2 copies



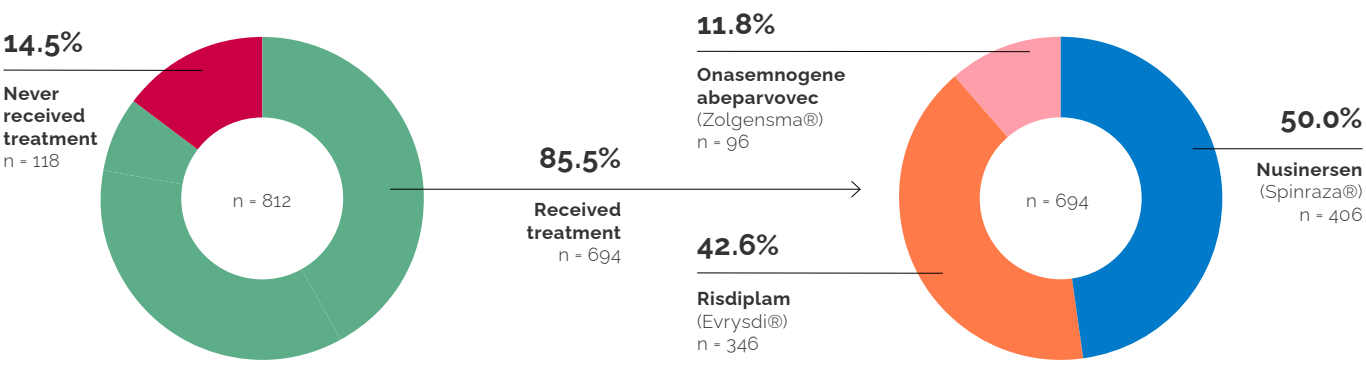
Functional status



Respiratory status



Treatment context



Some of the respondents switched medicines or received a different treatment before or after the single-dose therapy (cf. p. 49)..

[1] SMA is a heterogeneous disease that impacts individuals in different ways and to different degrees. Traditionally, SMA has been categorised in types, based on symptom onset and the motor functions achieved by an individual. However, this categorisation fails to account for the significant variability in both the symptoms and the progression of the disease among individuals living with SMA within each type. SMA Europe is working to develop an updated description of SMA that reflects the lived reality of a condition that spans multiple domains and presents differently for every individual.

STAGE 1

At diagnosis: what was available?

The moment of diagnosis is presented here as the starting point of the SMA treatment journey. At this stage, respondents reported different levels of information and access, depending on when and where the diagnosis took place. For many respondents, diagnosis occurred during periods of limited or unequal access to SMA medicines and clinical trials.

The development of disease-modifying therapies (DMTs) for SMA

Following the identification of the *SMN1* gene in 1995, efforts focused on developing DMTs. In 2011, the first clinical trial began for nusinersen (Spinraza®), an intrathecal medicine that enhances SMN protein production. Spinraza® became the first medicine approved in the European Union for SMA in 2017. Meanwhile, a gene therapy delivering a functional copy of the *SMN1* gene (onasemnogene abeparvovec, Zolgensma®) was developed and underwent clinical testing, receiving approval in 2019. Finally, in 2021, another DMTs, risdiplam (Evrysdi®), an SMN protein enhancer administered orally, was approved.

Availability of treatment options at diagnosis

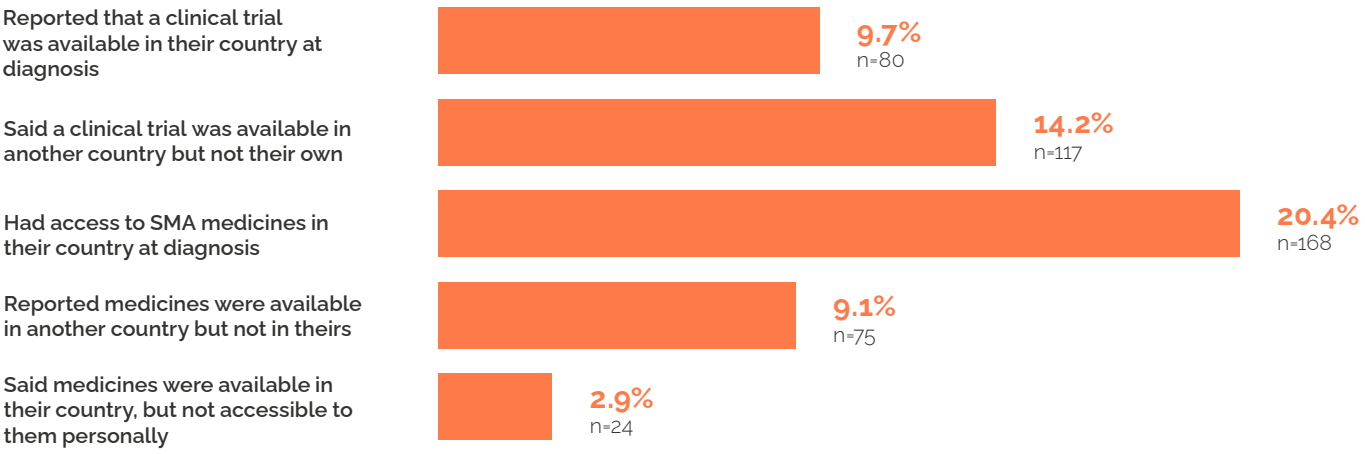
Experiences varied considerably across respondents (n = 824).

42.0% DMTs or clinical trials **were available** at diagnosis.
n = 346

58.0% **No SMA medicines or clinical trials were available** when they or their child were diagnosed.
n = 478

National versus cross-border access

Even when therapies or trials existed at the time, they were not always available in the respondent's own country (n = 824).



Cross-border mobility for SMA care

Access to SMA care varies widely across Europe. For some families, this means that the expertise, treatment options, or care structures they need are not available in their home country. As a result, a number of respondents described moving, temporarily or permanently, to another country to receive SMA-related care. Access barriers at home push some families to seek care elsewhere.

n = 823

15.0% **Moved country** to access care
n = 124

Not starting treatment at all

A subset of respondents (14.5%, n=118) reported never having started any approved SMA medicine. Reported reasons included both considerations related to availability and access and personal motivation.

Availability and access-related reasons

52.8%
Said treatment was **available but not accessible** to them
n = 57

15.7%
Said treatment was **available but not reimbursed**
n = 17

13.0%
Said treatment **was not available in their country**
n = 14

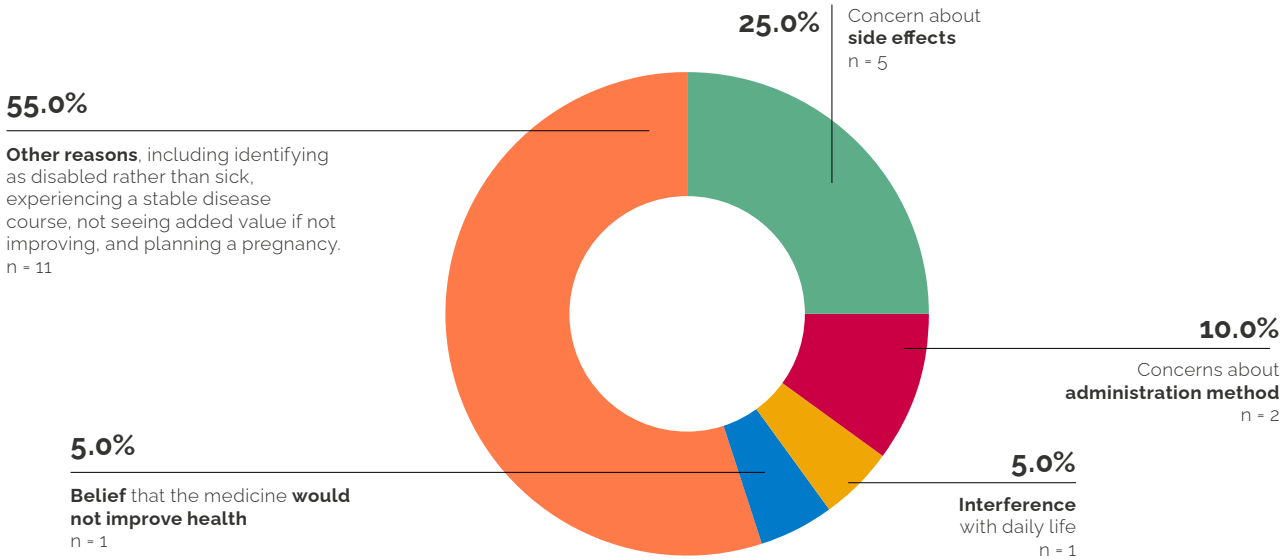
Personal reasons

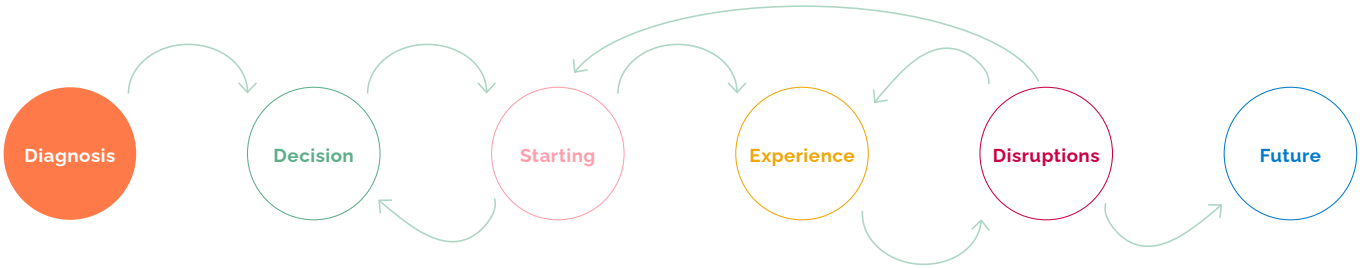
18.5%
Chose not to start treatment
n = 20

This shows that for a subset of families the treatment journey does not begin when a personal decision to start treatment is made, but rather, only when eligibility, reimbursement, and system-level barriers allow it to begin.

Among those who lacked access despite availability, common barriers included not meeting clinical, genetic, age, or performance criteria. 33.3% of the 57 individuals who answered this question identified age was the relevant criterion. Indeed, adult age is still an exclusion criterion in many European countries.

For those who chose not to initiate treatment (n=20), the reasons included:





What this stage of the journey shows

At the very start of the SMA treatment journey, people's experiences diverge dramatically. Depending on where they live, when they were diagnosed, and how eligibility criteria were defined, families encounter very different realities:

Some found themselves without any treatment options.

Others had to **fight for access** or navigate restrictive criteria.

Some even **moved countries** in search of care.

Understanding access gaps: How EUPESMA and OdySMA work together

SMA Europe works to identify and systematically address gaps in access to medicines responsible for inequities in SMA care in Europe.

In Europe, individual countries make decisions regarding which medicines will be made available, for whom they will be accessible, and whether they will be reimbursed by national health systems or insurances.

Access criteria establish which parts of the population will have access to treatment.

In SMA, these criteria may include:

- Genetic information (e.g. number of SMN2 copies)
- Clinical information (e.g. SMA type)
- Age
- Use of ventilation
- Motor function performance

These criteria directly impact opportunities to access care for each person living with SMA: not all individuals living in a country may have the same opportunities to access SMA medicines.

OdySMA: Identifying system-level access gaps

SMA Europe's OdySMA platform collects system-level information on access across Europe to understand how individual countries structure access to SMA medicines and care. This includes:

- Reimbursement status of each SMA medicine
- National eligibility criteria (e.g. genetic, clinical and functional restrictions)
- Cross-country comparisons of regulatory and policy frameworks
- Status of newborn screening
- Health economics factors and system parameters (e.g. availability of treatment centres, patient organisations)

Access to SMA treatments across Europe is shaped by national rules, eligibility criteria, reimbursement decisions, and the availability of specialised care. Much of this information exists only at the country level, in formats that are not easily comparable. As a result, it can be difficult to understand how access differs across Europe or to identify where system-level barriers may be limiting treatment availability.

OdySMA was created to address this gap. It brings together country-specific information on reimbursement, eligibility criteria, service structures, and regulatory pathways into a single, comparable European overview. This makes it possible to see patterns and differences that are otherwise hidden and helps identify where structural obstacles may exist.

More info about OdySMA

EUPESMA: Identifying patient-level access gaps

EUPESMA shows how individuals living with SMA in Europe are directly affected by barriers to access to SMA treatments, revealing inequities that begin at diagnosis and continue throughout the treatment journey. Many respondents reported that medicines or clinical trials were not available when they were diagnosed, or that therapies were available in their country but not accessible to them due to eligibility rules, reimbursement restrictions, or clinical criteria. Some even moved across borders to receive care.

These findings highlight patient-level access gaps, that is, differences in opportunities to start or continue treatment that individuals and families experience. EUPESMA captures what families experience firsthand, including:

- Whether treatments or trials were available at diagnosis
- Whether they could access them in practice
- Reasons for delayed or denied access
- System-level barriers that caused treatment interruptions
- Emotional and practical consequences of unequal access

This dataset reflects the lived reality of treatment access 'on the ground', showing the real-life impact of system-level access gaps on the lives of individuals who live with SMA.

More info about EUPESMA

Integrating patient- and system-level data on access barriers

EUPESMA and OdySMA complement each other to provide a full picture of how structural access barriers impact individual lives.

OdySMA describes how access is organised and EUPESMA reflects what this means in practice: whether treatments or trials were available at diagnosis, whether people could access them, why delays or interruptions occurred, and how these situations affected daily life, expectations, and decision-making.

Together, OdySMA and EUPESMA provide a fuller picture of access in Europe:

- **OdySMA shows *how access is structured* at the system level.**
- **EUPESMA shows *how access is experienced* by individuals and families.**

Combined, they reveal where system barriers exist and how they affect real lives.

OdySMA's Real-life Stories further integrates this evidence with personal accounts and testimonies that reflect these learnings and complete the picture: from health systems, to the community, to individuals.

STAGE 2

Treatment decision-making

Treatment decision-making involves both the choice to start treatment and of which treatment to start. In the stage of the journey, people living with SMA and their families seek professional expertise as well available evidence and consider many factors in making their choice. Many, however, face constraints with eligibility and access criteria.

Understanding SMA treatments

What are SMA DMTs?

DMTs increase or replace functional SMN protein, whose production is affected in individuals living with SMA, and can modify the course of SMA.

When this survey was fielded, three DMTs where approved in Europe:

- **Nusinersen (Spinraza®)** – an *SMN2* splicing modifier, delivered via intrathecal injection every 4 months.
- **Risdiplam (Evrysdi®)** – an *SMN2* splicing modifier, an oral medication to be taken daily.
- **Onasemnogene abeparvovec (Zolgensma®)** – an *SMN1* gene therapy, delivered in a single intravenous infusion, once in an individual's life course.

All three target the root cause of the disease by increasing SMN protein levels, but differ in mechanism, delivery, and administration frequency. In particular, *SMN2* splicing modifiers are meant to be taken continuously over the life course of an individual living with SMA.

At present, we do not have clear evidence on whether specific treatments are more beneficial than others for specific outcomes.

Additional non-DMT therapies

Other therapies for SMA are currently under investigation. These medicines do not directly address SMN deficiency but aim to improve clinical outcomes by targeting its downstream effects, including those on muscle and the neuromuscular junction.

For further information, please visit:

Opportunity to choose between different medicines

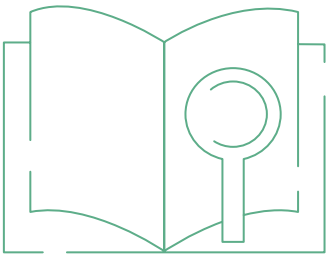
When initiating treatment, most respondents did not have the opportunity to choose which medicine they would start (n = 642):

30.7% Had the opportunity to choose between different DMTs
n = 197

69.3% Had no opportunity to choose
n = 445

Information availability and clarity

Most people felt sufficiently **informed** to make a decision:



85.5%

On whether to start a DMT
n = 554

87.8%

On which DMT to start
n = 173

The majority felt sufficiently **supported** in making the decision:

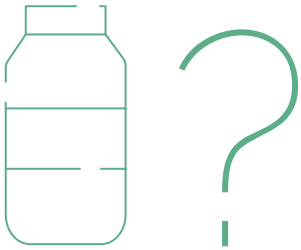
80.7%

On whether to start a DMT
n = 522

85.6%

On which DMT to start
n = 166

However, information gaps remained significant:



27.5%

Agreed or strongly agreed that it was difficult to decide whether to start a medicine due to insufficient safety and efficacy data.
n = 644

This reflects the reality of a rapidly evolving therapeutic landscape in which **long-term evidence is still emerging** and suggest that respondents had differing views about the amount and clarity of available evidence at the time of decision-making.

Sources of advice during decision-making

Respondents consulted a **range of sources** when making treatment decisions (n = 653).

85.5%
Neurologists
n=560

44.3%
Other people living with SMA
n=289

38.4%
Internet
n=251

29.6%
Scientific publications
n=193

28.8%
Patient organisations
n=188

21.4%
Social media
n=140

17.5%
Family members
n=144

15.0%
Geneticists
n=98

12.9%
EMA/FDA/ national medicines agency
n=84

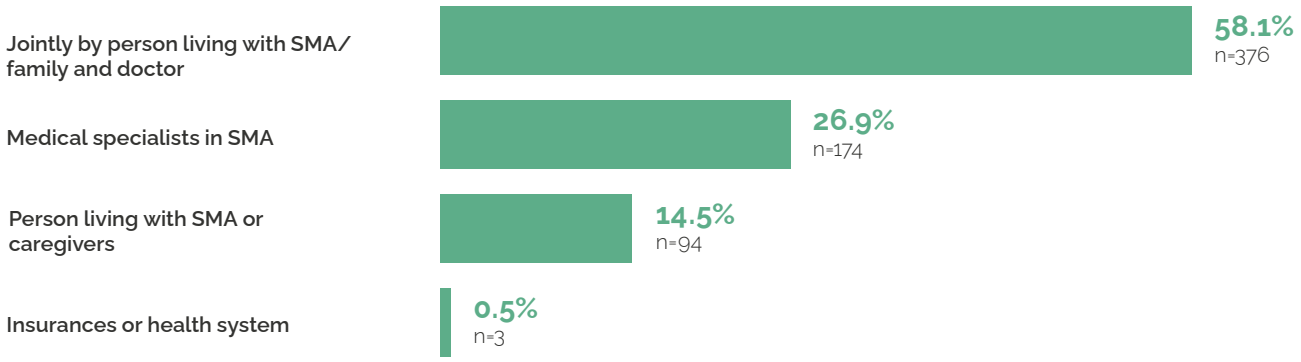
10.0%
Friends
n=65

9.8%
General practitioners
n=64

Preferences for who should make the final decision

Most respondents emphasised the **importance of shared decision-making** (n=647).

They expressed that final decisions about medicines should be made by:



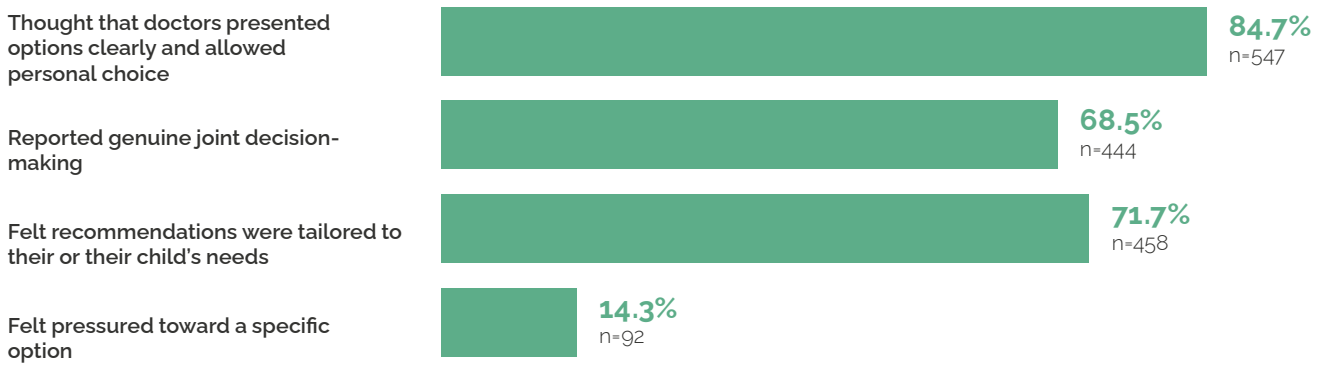
Current status of shared decision-making: Healthcare professional perspectives

In parallel to the EUPESMA survey presented here, targeting people living with SMA, their family or caretakers, SMA Europe fielded a survey on the use of SMA treatments among SMA healthcare professionals in Europe. Participants (n=122) included clinicians from various specialties as well physiotherapists, speech and language therapists, psychologists, occupational therapists, and nurses from 27 countries in Europe.

Healthcare professionals (n=102) reported that, in their professional experience, the final decision between SMA medicines is actually made by:

Experiences of shared decision-making

Experiences during decision-making reflected a preference for **collaboration** (n=646):

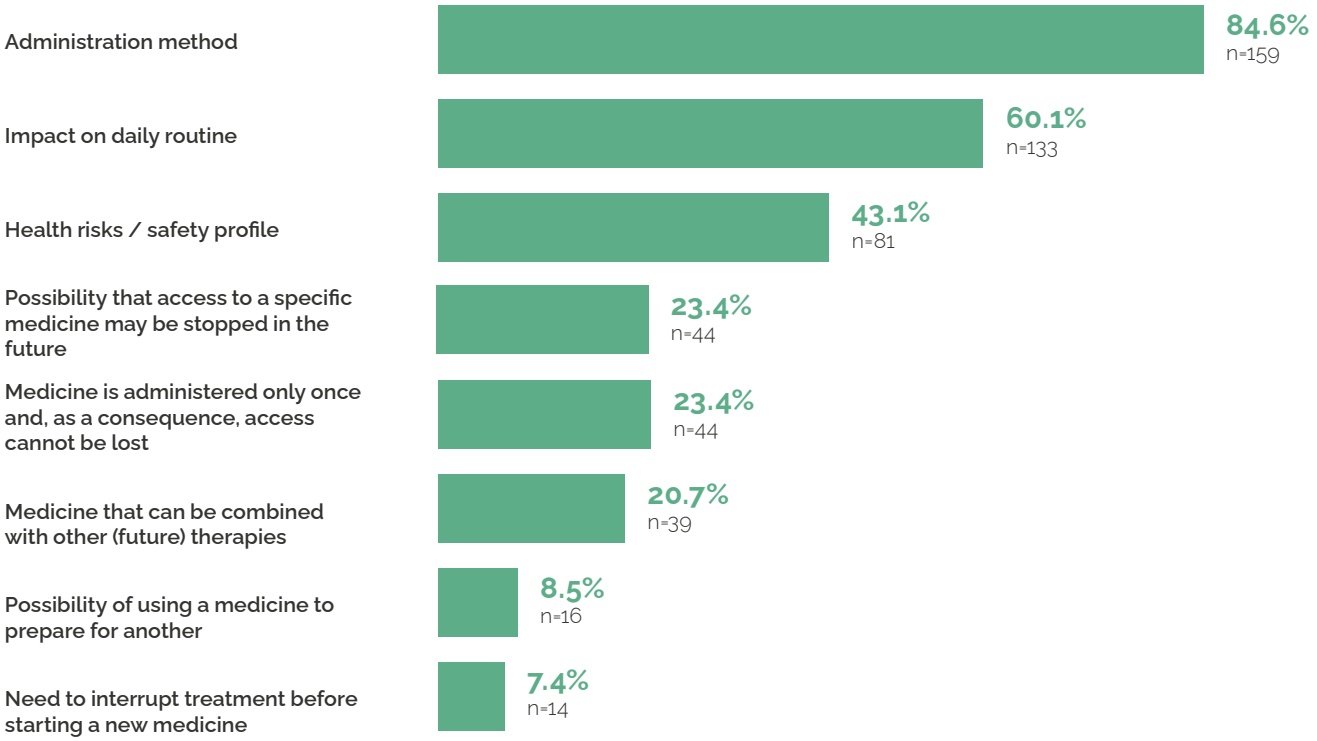


This suggests that while some respondents experienced pressure or uncertainty, the majority felt involved and respected in the process.

Factors that influenced treatment decisions

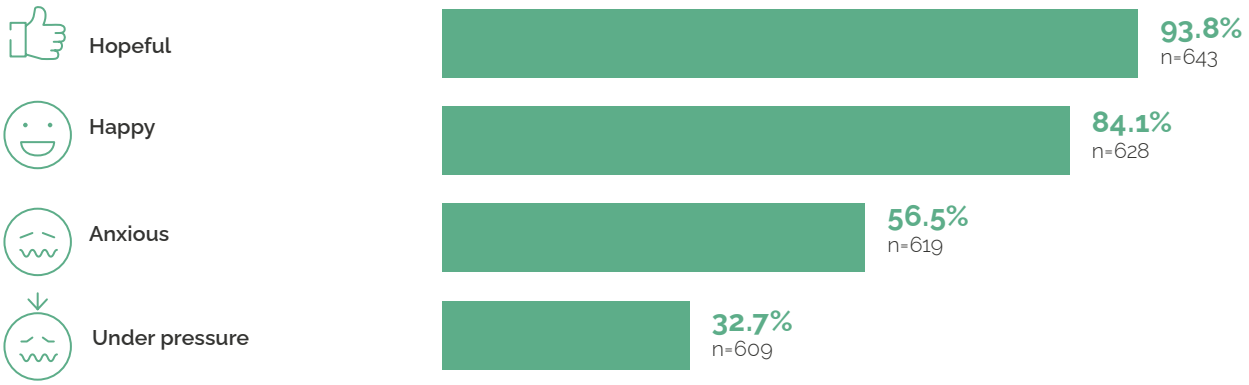
Only 30.7% (n=197) **of respondents had the opportunity to decide which medicine to start** (total n=642).

When asked which factors shaped their decision between different medicines, they (n=188) reported:



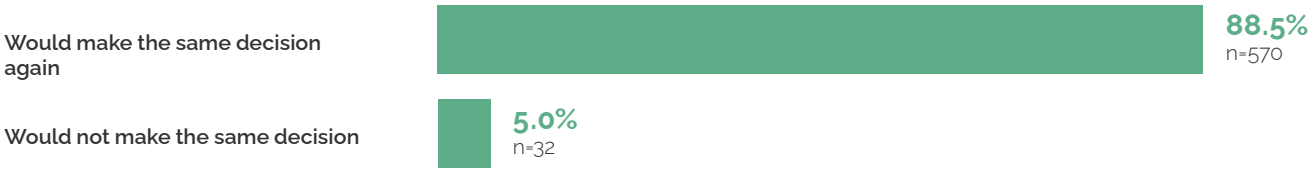
Emotional landscape of the decision-making phase

When making a decision about starting a medicine for SMA, respondents were feeling:

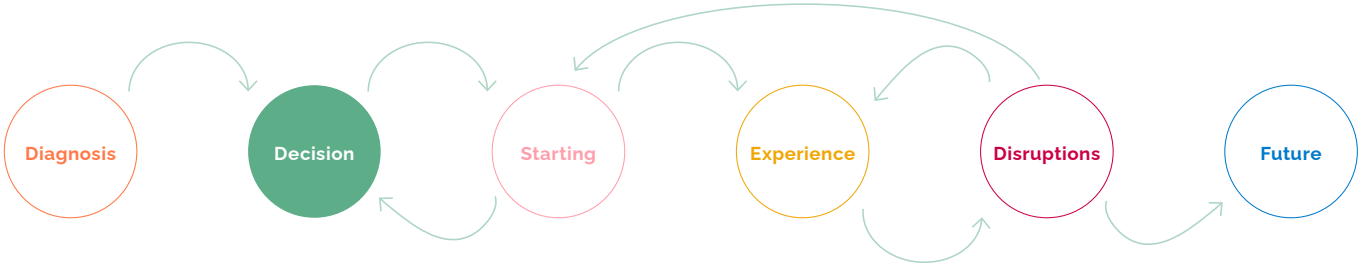


Looking back: would they make the same decision again?

Reflecting on their first decision on which medicine to start (n=646):



This suggests that, despite challenges, most families stand by the decision they made.



What this stage of the journey shows

Treatment decision-making is shaped by:

Limited choice and eligibility constraints.

Variable evidence and lingering uncertainties.

Reliance on neurologists and peer experience.

Practical factors such as administration and daily life.

Strong preference for shared decision-making.

High emotional tension, but overwhelmingly high retrospective confidence.

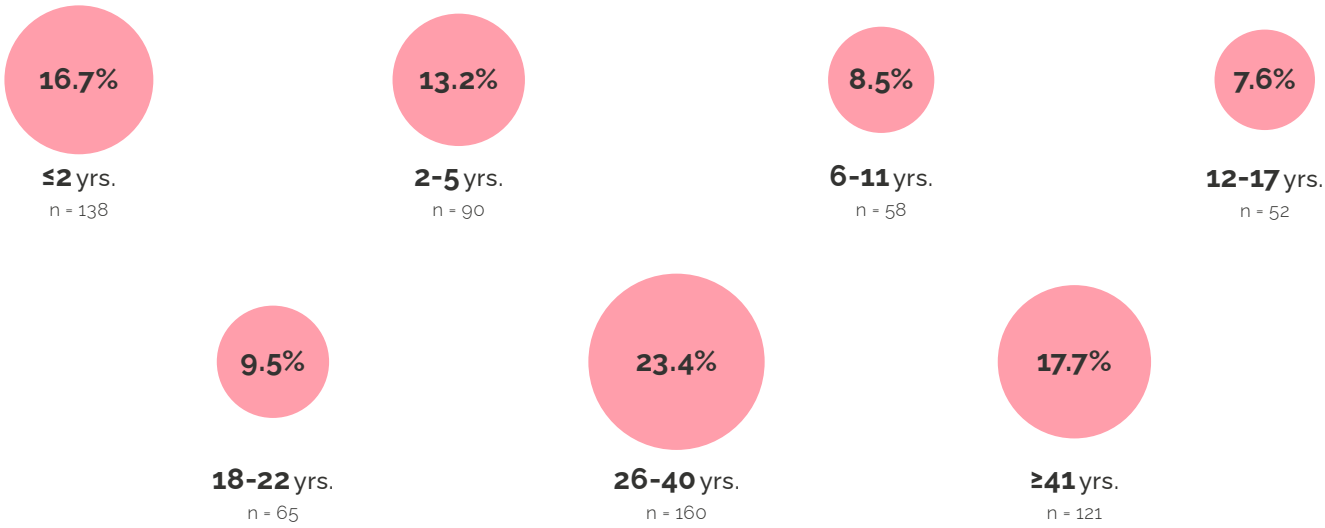
STAGE 3

Starting the first treatment

Once a treatment decision has been made, patients and families enter a new stage of the SMA treatment journey: the actual beginning of therapy. This moment is often characterised by a mix of relief, hope, and uncertainty, as individuals prepare for a significant transition in their care. The survey data show how respondents experienced the initiation of treatment, what they expected at the outset, and how they perceived the risks and opportunities associated with starting a DMT.

Age at first treatment

Respondents began their first SMA medicine at a wide range of ages, reflecting different treatment eras, diagnostic timelines, and national access pathways (n=826).



This variability highlights that while many children today have the opportunity to start treatment early, many adults living with SMA began treatment much later, often after years of progression without therapeutic options.

Starting treatment: risk or opportunity?

Families living with SMA often begin treatment with both hope and concern. Respondents reported that starting treatment felt like (n=640).

Indeed, uncertainty about long-term outcomes, eligibility rules, and treatment burden often co-exist with optimism about stabilisation or improvement.

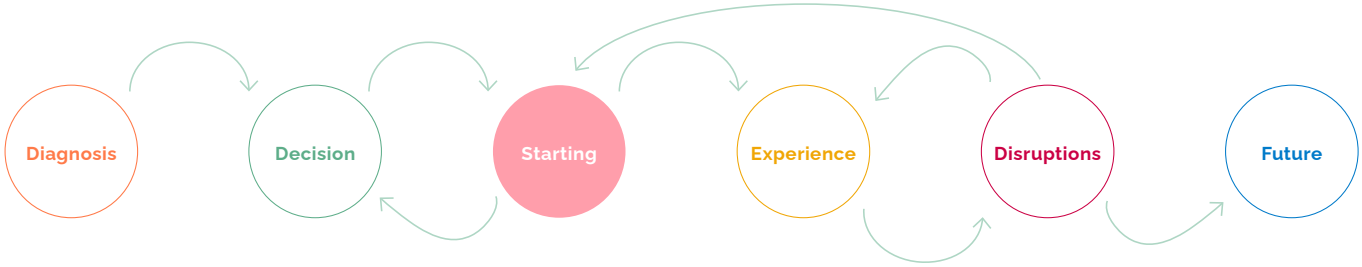
Expectations before starting a treatment

Respondents' expectations about the likely impact of treatment played a major role in their decision-making (n = 688):

55.5 % Expected **improvement** n= 382

34.2 % Expected **stabilisation** n= 236

10.3 % Expected their condition to **worsen** n= 71



What this stage of the journey shows

Age at first treatment varies, showing that children today can start treatment early, but adults began treatment much later. This means that care needs are very diverse.

Starting a treatment is largely seen as a valuable opportunity.

Most people expect that treatment will improve or stabilise disease progression.

These are welcome outcomes at any stage, independent of age.

STAGE 4

Living with treatment: experiences over time

Once treatment begins, individuals and families enter a new phase of the SMA treatment journey, one defined by gradual changes, stability, variability, and daily-life impact. While experiences differ based on age, SMA type, functional ability, and treatment type, common themes emerge across the survey: stabilisation is meaningful, improvements occur across multiple domains, and some challenges persist.

Overall perception of treatment impact

Respondents described a wide spectrum of experiences after starting treatment (n = 687):

39.6 % Expectations were met **'a lot' or 'a great deal**
n = 272

51.1 % Expectations were met **'a little' or 'a moderate amount**
n = 351

9.3 % Expectations were entirely **unmet**
n = 64

The overwhelming majority perceived some degree of benefit, ranging from stabilisation to significant improvement.

Stabilisation as meaningful progress

Stabilisation, defined as a halt in decline, is strongly valued (n = 792):

89.6 % Consider stabilisation to be meaningful progress
n = 710

This finding underscores a major shift in what SMA treatment means:

Not getting worse is already getting better.

Stabilisation as progress

SMA is a progressive condition: untreated people living with SMA gradually lose function and abilities.

For many people living with SMA, living with a progressive condition means experiencing uncertainty about the future, about what functions will be lost next, and how this will impact independent living and overall health.

Clinical trials for SMA medicines were designed to focus on measuring improvement, interpreted as a measurable gain in function and abilities. However, while regaining function is certainly desirable, many people living with SMA, based on their lived experience, claim that **stabilising disease progression constitutes a meaningful improvement.**

Stopping the disease from progressing further or slowing down the progression rate reduces uncertainty and opens opportunities. Knowing what current abilities they have, and that they will not be lost, allows people to imagine a future for themselves and make more ambitious plans for the long-term.

People living with SMA and their families should have a say in defining what meaningful progress is to them.

Patient organisations are uniquely positioned to represent the range of perspectives that the community holds in defining meaningful progress and have a key role in ensuring that this shift in perspective is reflected in clinical trial design. Reflecting these patient perspectives into trial design is key, because clinical trial results can influence regulatory and access decisions, as well as access continuation over time.

Changes across functional domains

Respondents reported how treatment had changed their experience in 15 physical and functional areas: improvement, stabilisation, or worsening.

Improvements and stabilisation were common; some domains showed mixed change. Below is a consolidated overview, grouped by major functional categories.

Motor abilities and strength

Gross motor function
n = 681

↗	→	↘
47.0 %	44.0 %	9.0 %

Fine motor function
n = 677

↗	→	↘
57.3 %	37.4 %	5.3 %

Muscle strength
n = 683

↗	→	↘
60.9 %	29.3 %	9.8 %

Endurance and fatigue

Endurance
n = 684

↗	→	↘
67.3 %	25.0 %	7.7 %

Fatigue
n = 683

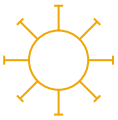
↗	→	↘
51.4 %	38.7 %	10.0 %

Improved Stabilised Worsened

Respiratory function

Breathing
n = 679

↗	→	↘
43.0 %	53.0 %	4.0 %



Respiratory infections
n = 677

↗	→	↘
30.0 %	66.3 %	3.7 %

Bulbar function

Swallowing
n = 678

↗	→	↘
27.7 %	69.6 %	2.7 %

Eating
n = 678

↗	→	↘
27.9 %	69.0 %	3.1 %

Speaking
n = 676

↗	→	↘
27.7 %	71.4 %	0.9 %

Musculoskeletal health

Scoliosis
n = 676

↗	→	↘
6.7 %	70.4 %	22.9 %

Contractures
n = 669

↗	→	↘
10.5 %	72.5 %	17.0 %

Fractures
n = 661

↗	→	↘
6.2 %	91.7 %	2.1 %

Weight and nutritional status

Weight gain
n = 675

↗	→	↘
23.0 %	62.3 %	14.7 %

Weight loss
n = 661

↗	→	↘
8.2 %	87.4 %	4.4 %

Improved Stabilised Worsened

Pain

Pain
n = 669

↗	→	↘
14.3 %	76.4 %	9.3 %

Heart symptoms

Heart symptoms
n = 159*

↗	→	↘
22.6 %	69.2 %	8.2 %

* 'Other' domain subset

Patterns across domains

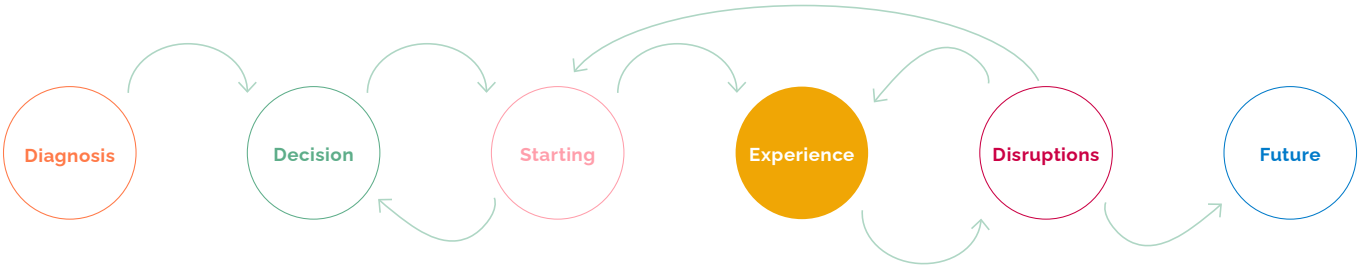
Across functional areas, the data reveal several consistent patterns:

Most common improvements: ↗ **Endurance, fine motor function, muscle strength, fatigue, some respiratory measures**

Most common stabilisation: → **Breathing, eating and swallowing, pain, bulbar function overall**

Domains with mixed or limited change: ↗ → **Scoliosis, contractures, weight-related outcomes**

Meaning of stabilisation:
high value is placed on stabilisation, especially in domains where deterioration is normally expected.



What this stage of the journey shows

For many respondents, treatment has led to:

Improvement across several physical and daily-life domains.

Stabilisation where decline was previously the norm.

Subtle but meaningful functional gains that impact independence.

Persistent challenges, especially in musculoskeletal health and endurance and fatigue.

Variability depending on age, SMA type, baseline function, and treatment timing.

Living with treatment is therefore characterised by a combination of progress, stability, and ongoing needs, all of which shape needs and expectations for future therapies.

STAGE 5

Treatment switching and add-on use

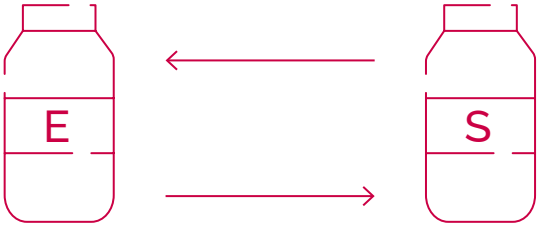
This chapter describes how respondents reported changes in their SMA treatment over time. These changes included switching between ongoing DMTs (Spinraza and Evrysdi), receiving a DMT before or adding a DMT after Zolgensma. Because Zolgensma remains active after administration, any therapy given after gene therapy is considered add-on use rather than switching.

How treatments for SMA may be used over time: key definitions

The SMA treatment landscapes is complex, and treatment trajectories and regimens vary. Here are some key terms that have been used to describe some of the more frequent uses of SMA medicines.

- **Monotherapy**: only one treatment is taken, either in one administration (Zolgensma), or at regular intervals over the life course (Spinraza and Evrysdi).
- **Switching**: A DMT is stopped to initiate another one (A —> B). Applies to Spinraza to Evrysdi, Evrysdi to Spinraza, or a *SMN2* splicing modifier to Zolgensma.
- **Bridging**: When a DMT is planned for temporary use prior to another therapy (e.g. planned use of a *SMN2* splicing modifier prior to Zolgensma).
- **Add-On Therapy**: Adding a second treatment without stopping the first one (A + B). In SMA this may occur also when a therapy is started after receiving, for instance Zolgensma, while its gene-addition effect remains. It can also describe DMT + non-DMT therapy when used together.

Switching between SMA medicines



29.3% n = 629

Have the option to switch between treatments (Evrysdi/Spinraza) or to add a second medicine after Zolgensma
n = 184

17.5% n = 644

Switched between splicing-modifier medicines, i.e. Evrysdi to Spinraza, or Spinraza to Evrysdi
n = 104

Why do people switch SMA medicines?

Among respondents who decided to switch medicines, 87.1% (n=88) reported that they were seeing improvements with their first treatment regimen. However, they still decided to switch for a variety of overlapping reasons, including clinical, practical, and emotional factors.

78.2%

Administrative or procedural burden

Switched due to how the medicine was administered (e.g., difficulty tolerating intrathecal administration, logistics of repeated dosing)
n = 79

9.9%

Lifestyle considerations

Switched because treatment interfered with daily life, work, school, or independence
n = 10

15.8%

Side effects or tolerability

Switched due to side effects
n = 16

13.9%

Hope for better outcomes

Switched with the hope of achieving better stabilisation or functional gains
n = 14

14.9%

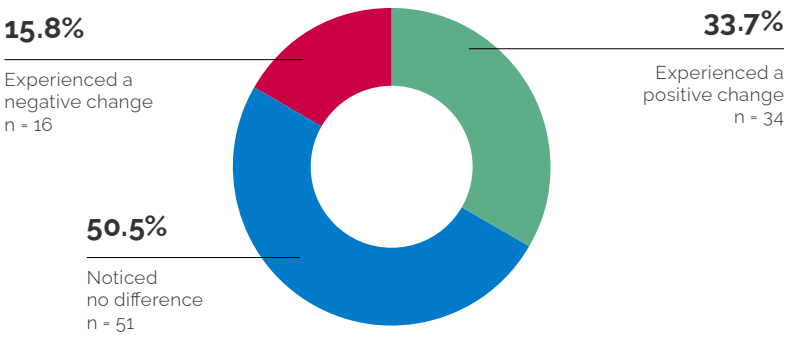
Opportunity-driven switching

Switched because a new option became available to them
n = 10

No one reported switching because they wanted to get pregnant or have children. These reasons show that switching is not only medically driven, but also shaped by burden, access, expectations, curiosity, and hope for more.

How switching was experienced

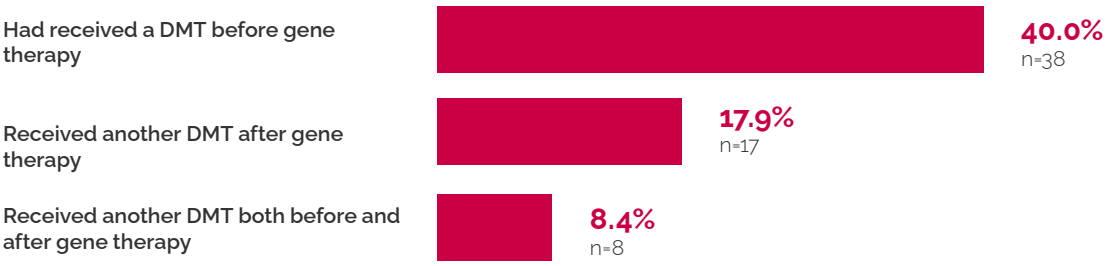
Among those who switched (n = 101):



Switching therefore seems to bring benefits for some, but outcomes remain variable.

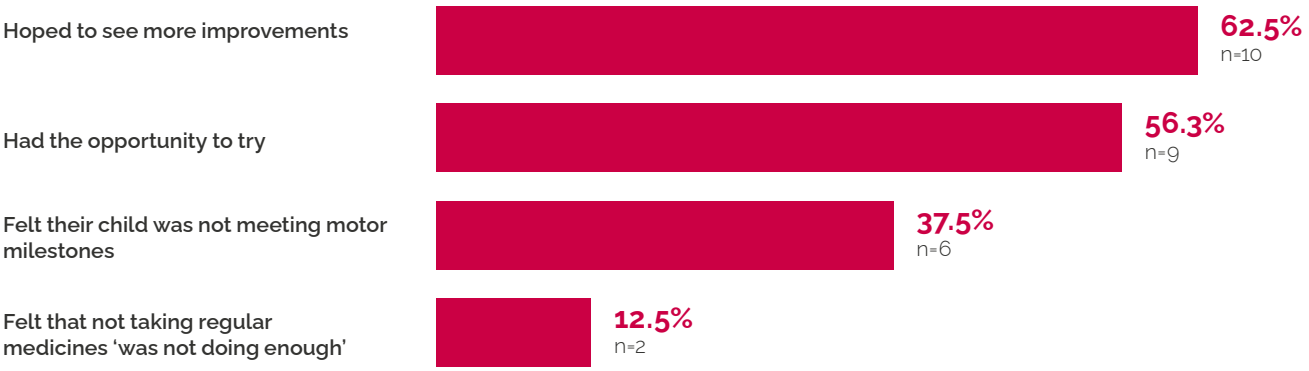
Receiving another medicine before or after gene therapy

Among respondents who had received Zolgensma (n = 95).



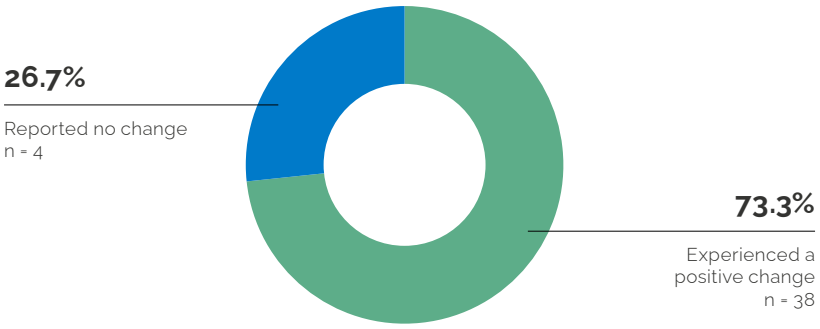
Why add a medicine after gene therapy?

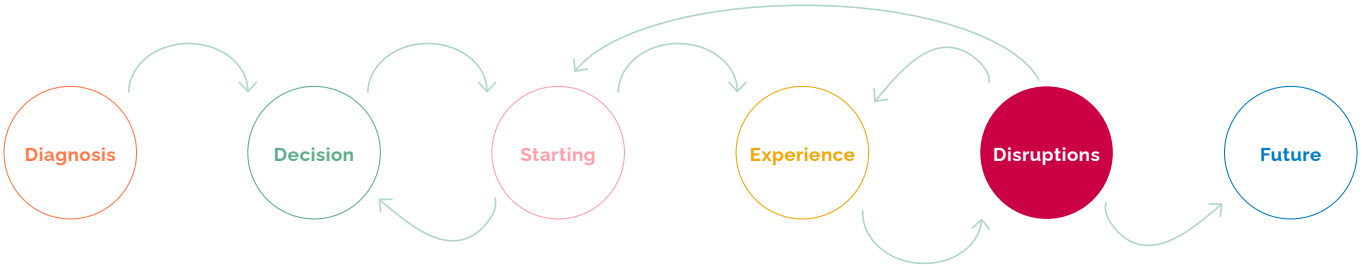
While most respondents experienced improvement after receiving gene therapy (93.8 %, n=16), they decided to add another SMA medicine because they:



How adding-on was experienced

Among those who added another DMT to Zolgensma, with no negative changes reported (n = 15).





What this stage of the journey shows

Switching and add-on use remains relatively uncommon and an option that is available to a small percentage of people living with SMA.

People who switched or added on after gene therapy were usually experiencing benefits from being on treatment.

Still, they switched or added on for a variety of reasons, both practical and related to treatment effects.

For those who switched, switching led to a positive change or no change.

For those who added on, the majority experienced a positive change.

Evidence about the benefits of switching and adding-on is still scarce, but of vital importance to inform treatment decision-making as well as access pathways.

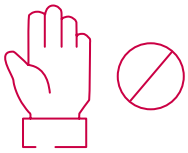
STAGE 5

Disruptions in treatment

Stopping or interrupting SMA treatment is a significant moment in the treatment journey. For some respondents, discontinuation reflects personal choice or concerns about effectiveness, for others, it is imposed by clinical criteria, reimbursement rules, or logistical challenges. Regardless of the reason, interrupting treatment carries emotional, physical, and practical consequences.

Stopping or interrupting treatment

Among respondents who had experience with Spinraza or Evrysdi.



n = 637

19.5% Stopped or interrupted treatment at least once
n = 122

Duration of treatment disruption

Among those who stopped or interrupted treatment (n=118), reported disruption length was:

16.1% Less than **1 month**
n = 19

17.8% Less than **3 months**
n = 21

66.1% More than **3 months**
n = 78

Two-thirds experienced prolonged interruption, often due to system delays or unresolved criteria.

Information about the consequences of treatment disruption

Whether or not the decision was voluntary, not enough is known about the consequences of stopping or interrupting treatment.

n = 118

46.6% Reported that they **did not have enough information** about the potential consequences of stopping or interrupting treatment
n = 54

This highlights a major communication gap around the risks associated with treatment disruptions.

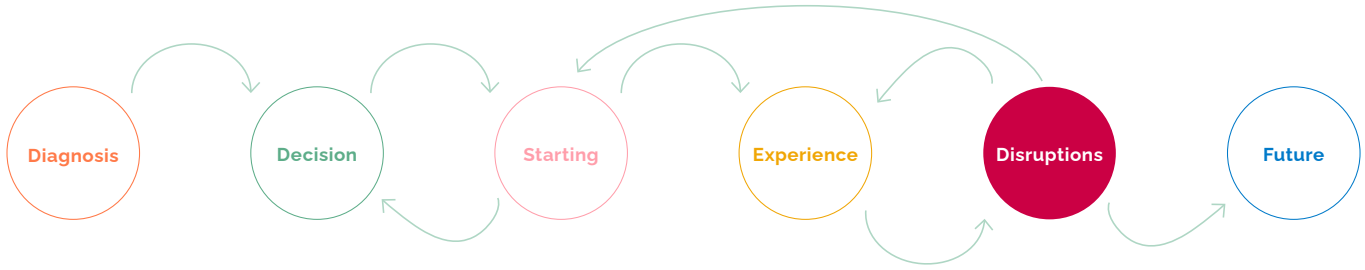
Concerns about future treatment loss

Concerns about the possibility of losing treatment access in the future are high among people living with SMA:

n = 778

50.4% Reported experiencing **fear** about being forced to stop treatment in the future
n = 392

Fears were grounded in concerns that governments or insurances would stop reimbursing treatment, or that disease progression may lead to meeting stop criteria.



What this stage of the journey shows

While splicing modifying treatments are meant to be administered continuously, some patients are experiencing disruptions in treatment:

Little is known about the consequences of stopping or interrupting treatment.

Access to medicines is important to initiate treatment, as well as in the long term, to ensure that treatment can be continued in the long term.

Continuity of treatment matters.

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STAGE 6

What people want next: priorities for future SMA medicines

While today's SMA treatments have transformed the landscape of care, respondents made it clear that their journey is not finished. Many still face daily challenges, live with uncertainty about the future, and see clear gaps that current medicines do not yet address.

This chapter explores what people living with SMA and their families want from future medicines and add-on therapies, and how these expectations connect to their lived experience.

Feeling anxious about the future

Most respondents reported feeling anxious about the possibility that their or their child's SMA symptoms may get worse in the future (n= 801).

2.1 %
Not anxious at all (score 1)
n = 17

5.2 %
Not anxious
n = 42

14.2 %
Neutral
n = 114

20.1 %
Anxious
n = 161

58.3%
Very anxious (score 5)
n = 467

Taken together, 78.4% of respondents indicated high levels of concern about future worsening. This emotional backdrop helps explain why there is such a strong call for ongoing innovation and next-generation medicines.

A clear need for future medicines

Despite the benefits of existing SMA treatments, most respondents see a continuing need for new options.

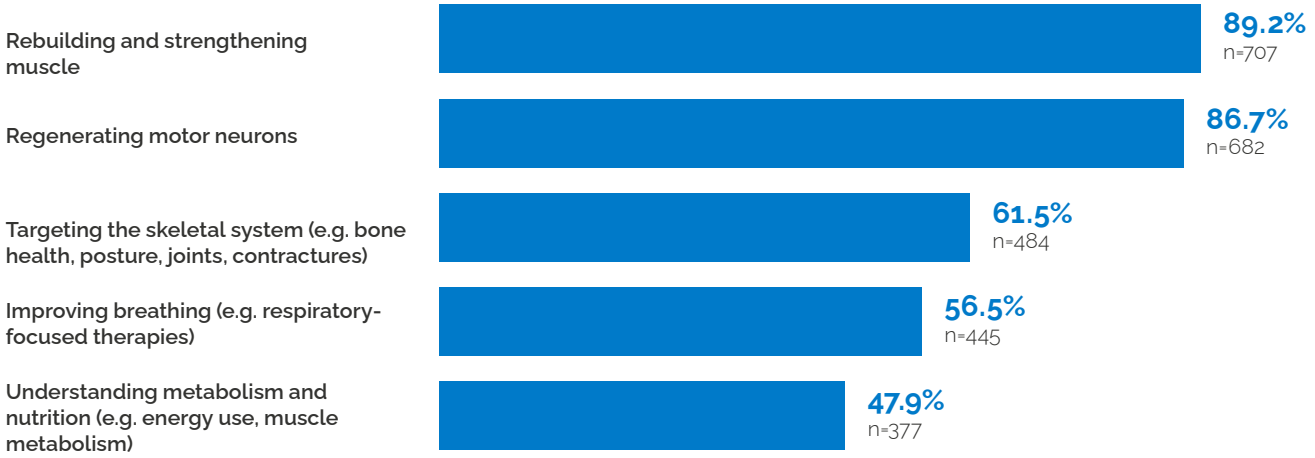
n = 799

83.3%
Agree that there is a need for future SMA medicines.
n = 641

This does not reflect dissatisfaction. Rather, it reflects a realistic and forward-looking view: treatments have changed the trajectory of SMA, but they have not resolved all aspects of the condition.

Targets for new medicines

When asked about potential targets for future medicines, respondents showed strong support for approaches that go beyond SMN alone and address multiple aspects of SMA (n = 787).



These findings underline that the community wants medicines that address the various domains that are affected by SMA.

Preferences for treatment choices

As the treatment landscape matures, many respondents do not think in terms of a single "one-off" treatment, but rather in sequences and combinations over time.

Respondents agreed that people living with SMA, under medical advice, should be able to (n = 780).

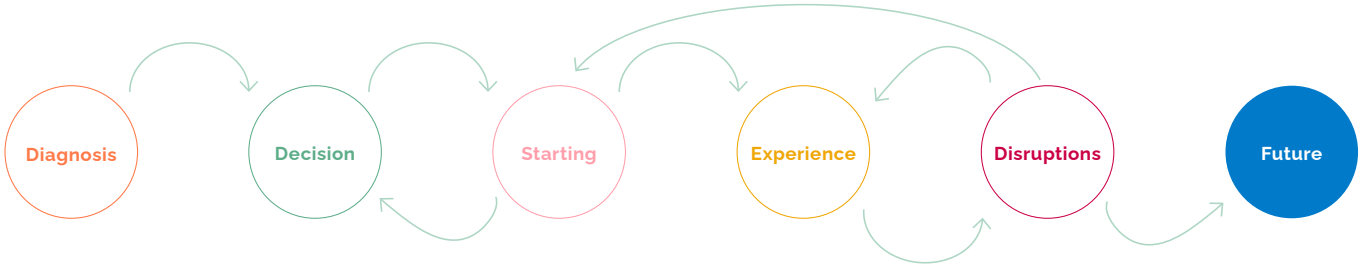
Supporting research and care on SMA

The availability of current SMA treatments is the result of extensive fundamental research, which has advanced our understanding of the underlying biological and pathophysiological mechanisms of the condition. For 20 years, SMA Europe has supported fundamental research and clinical advances on SMA to improve care and treatments for SMA. We are proud to continue to do this through our:

- **Call for Research:** a recurring funding initiative that supports high-quality scientific projects aimed at improving understanding of SMA and advancing solutions that matter to people living with SMA. The Call primarily funds fundamental and translational research, including studies that explore disease mechanisms, therapeutic targets, biomarkers and biologically grounded aspects of care, with the objective of building knowledge that can inform future discoveries and benefit the community.

+ Info abour Call for Research

- **International Scientific Congress on SMA:** brings together scientists, young researchers, clinicians, other healthcare professionals, patient advocates, and industry representatives to share the latest scientific and clinical developments in the field of SMA.
- **International Clinical Care Symposium on SMA:** brings together SMA key opinion leaders, healthcare professionals, researchers, industry stakeholders, and patient advocates to discuss the medical and supportive care needs of people with SMA and their caregivers, which frequently extend beyond clinical treatments and require a multidisciplinary approach.



What this stage of the journey shows

Current DMTs have proved effective in stabilising disease course, but…

Most people living with SMA are still anxious about future disease progression.

Treatments have not resolved all aspects of living with SMA.

The community expresses a need for new medicines that address SMA holistically, as a multisystem condition.

People have different needs, priorities, and preferences in regard to medicines.

This suggests that individualised therapeutic approaches, that take into account personal needs and decision making, are necessary.

Conclusions

The EUPESMA 2025 Treatment Experience Survey provides a comprehensive and community-driven overview of how people across Europe experience the modern SMA treatment landscape. By following the SMA treatment journey step by step, from diagnosis to future expectations, this report highlights both the progress achieved and the challenges that remain.

Across all chapters, a consistent narrative emerges: treatment has changed the trajectory of SMA, but not the complexity of living with it.

STAGE 1
Diagnosis: A journey shaped by unequal starting points

Different starting conditions lead to unequal paths. Many respondents reported that no SMA medicines or clinical trials were available at the time of diagnosis, and for some, the only way to access treatment was to leave their home country. These early inequalities create divergent pathways, determining who can start treatment early, who must wait, and who must navigate additional burdens just to gain access. In a progressive condition, time is of the essence and early intervention facilitates the preservation of abilities and function.

STAGE 2
Decision-making: Hope, uncertainty, and constrained choice

The emotional and cognitive intensity of deciding whether and how to start treatment is enormous. While most respondents felt informed and supported, their decisions were shaped by:

- limited treatment choice in many countries
- uncertainties due to incomplete evidence
- the emotional weight of hope, anxiety, and responsibility

Despite these challenges, most respondents would make the same decision again, showing trust in their decision process and their care teams.

STAGE 3
Starting treatment: Expectations meet reality

Respondents described starting treatment as a moment defined simultaneously by opportunity and risk. Expectations varied, with many hoping for an improvement in function, but strongly valuing stabilisation. Treatment initiation, however, is not guaranteed: access barriers, reimbursement restrictions, and clinical criteria continue to prevent some from starting treatment at all, even when medicines are technically available.

STAGE 4
Living with treatment: Improvements, stabilisation, and persistent unmet needs

Stabilisation, considered as meaningful progress by the vast majority of respondents, stood out as a central outcome. The findings shows that treatment generally brings meaningful benefits, most frequently in:

- endurance and fatigue
- muscle strength
- fine and gross motor function

STAGE 5
Switching or adding treatments: Navigating burden, hopes, and practical realities

A minority of respondents reported switching between DMTs, typically citing reasons such as administration challenges, side effects, interference with daily routines, or limited perceived improvement. Respondents who added a treatment after gene therapy mentioned hopes for additional gains, emerging opportunities to try another option, or concerns that progress was not meeting expectations.

Treatment disruption: A vulnerable moment

This survey unveiled a stunning finding: a meaningful number of participants experienced a discontinuation of treatments that are meant to be taken continuously. Little information is available about the consequences of interrupting treatment, and nearly half of the respondents did not feel adequately informed about the potential consequences of treatment interruption, underscoring the need for clearer communication and more stable access pathways.

STAGE 6
Future medicines: A multisystem vision for SMA

While current therapies improve or stabilise many aspects of SMA, they do not yet address the full range of needs that people living with SMA experience. Respondents overwhelmingly called for next-generation and add-on therapies targeting:

- regeneration of the neuromuscular system
- muscle function
- breathing
- endurance and fatigue
- skeletal health

The community envisions a future in which multimodal and combination therapies complement SMN-targeting treatments.

Progress is real but the journey is not yet complete

A coherent story emerges:

- People value stabilisation and seek continued improvement.
- Treatment benefits are clear, and fragile when access is interrupted.
- Inequalities persist at every stage, from diagnosis to clinical trial participation.
- Future innovation must address the multisystem nature of SMA.
- Access, continuity, and personalisation matter as much as the medicines themselves.
- The community is engaged and willing to contribute, but they need better systems and clearer, more equitable pathways.

EUPESMA 2025 offers more than evidence: it offers direction. By listening to lived experience, we see where progress has been made and where it must continue. The SMA community has articulated its needs clearly. Now, it is up to all stakeholders, clinicians, researchers, regulators, policymakers, and industry, to translate these insights into action.

SMA Europe will continue to systematically monitor the community's needs and to support research to advance therapies and care for people living with SMA.

The journey is evolving. With continued collaboration and commitment, the future of SMA treatment can become more inclusive, comprehensive, and aligned with what matters most to the people living with SMA.

Calls to action

Ensure timely and equitable access to SMA treatments across Europe

The treatment journey begins unequally. Many respondents lacked access or had to move countries for treatment. Countries should harmonise access pathways, eligibility criteria, and reimbursement processes so that treatment is based on need, not geography.

Support shared, informed, and personalised treatment decisions

Respondents want collaborative decision-making but still face evidence gaps and limited choice. Clinicians, regulators, and patient organisations should provide clear, up-to-date information, ensure shared decision-making tools are available, and personalise recommendations to each person's goals and situation.

Safeguard treatment continuity and prevent unnecessary interruptions

Treatment disruption is not uncommon, yet little is known of its consequences. Health systems must minimise interruptions caused by delays, reimbursement renewals, or eligibility changes, ensuring continuity of care.

Drive innovation toward multisystem and add-on therapies

Current treatments improve or stabilise SMA, but unmet needs remain in many domains. Research and industry should prioritise therapies that target these needs including regeneration of neuromuscular system, metabolism or respiration, complementing SMN-based approaches.

List of acronyms and abbreviations

Add-On Therapy: Adding a second treatment without stopping the first one (A + B). In SMA this may occur also when a therapy is started after receiving, for instance, Zolgensma while its gene-addition effect remains. It can also describe DMT + non-DMT therapy when used together.

Adult Committee of SMA Europe: SMA Europe volunteer group contributing to the projects and initiatives of the organisation, composed of adults living with SMA.

Bridging: When a DMT is planned for temporary use prior to another therapy (e.g. planned use of a *SMN2* splicing modifier prior to Zolgensma).

DNA: Deoxyribonucleic Acid (the hereditary material that carries genes, which contains the instructions essential for growth, development, functioning, and reproduction).

DMT (disease-modifying therapy): A treatment that targets the underlying causes of a disease to slow or alter its natural course and improve patient outcomes. In SMA, currently approved DMTs are Spinraza, Evrysdi, Zolgensma.

EMA: European Medicines Agency.

Ethical Approval: Permission from an ethics committee to conduct a study.

ERB: Ethics Review Board (for EUPESMA: Tilburg University Ethics Review Board).

EUPESMA: European Patient Experience Survey on SMA.

Evrysdi® (risdiplam): Oral SMN protein enhancer targeting *SMN2* splicing; approved in 2021.

FDA: Food and Drug Administration (United States).

Gene Therapy: Treatment that introduces or modifies genetic material to produce a therapeutic effect. In SMA, this typically refers to onasemnogene abeparvovec.

Monotherapy: only one treatment is taken, either in one administration (Zolgensma), or at regular intervals over the life course (Spinraza and Evrysdi).

n: A statistical abbreviation representing the 'number' or count of respondents for a particular answer.

OdySMA: an SMA Europe platform that collects information on access to treatment and care for SMA across Europe.

Pan-European: Covering multiple European countries.

Patient journey: describes the series of experiences, decisions, challenges, and changes a person goes through from the moment of first symptoms or diagnosis to their daily life with SMA and their outlook on their future.

PED (patient experience data): information that reflects the experiences, needs and priorities of people living with SMA and their families, including their daily lives, challenges, expectations, and treatment experiences.

% (Percentage): Indicates proportion or share in survey results.

® (Registered Trademark): indicates that a name or logo is officially registered as a trademark with the relevant authority.

SMA: spinal muscular atrophy.

SMA Europe: a non-profit umbrella organisation of SMA patient organisations from across Europe.

SMA Europe Delegates: Representatives from SMA Europe's member organisations.

SMN: Survival Motor Neuron (protein critical for motor neuron health).

***SMN1*:** survival motor neuron 1 gene (defective in SMA).

***SMN2*:** survival motor neuron 2 gene ('backup' gene partially compensating SMN1 deficiency).

Spinraza® (nusinersen): intrathecal SMN protein enhancer targeting *SMN2* splicing; approved in 2016.

Splicing modifier: treatment that modulates the splicing process, resulting in a different form of a protein. In SMA, this refers to Spinraza and Evrysdi, which act on *SMN2* splicing to increase the production of functional SMN protein.

Switching: A DMT is stopped to initiate another one (A —> B). Applies to Spinraza to Evrysdi, Evrysdi to Spinraza, or a *SMN2* splicing modifier to Zolgensma.

Treatment Committee (TC): SMA Europe group composed by expert patient advocates focusing on therapy-related matters.

Zolgensma® (onasemnogene abeparvovec): Gene therapy delivering a functional *SMN1* gene; approved in 2019.

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Additional information

For any questions, or more detailed data, you can send a request to SMA Europe

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