

SMA Care, Outcomes, and Reported Experiences (CORE) Survey

Survey launch: February 2025

Intended audience: individuals (or primary caregivers of individuals) who are newly diagnosed with 5q SMA that reside in the U.S.

If you have any questions, please feel free to contact data-analytics@curesma.org

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Survey Eligibility

Data previously provided to Cure SMA may be pre-populated in this survey form. Please review and update as needed.

What is your relationship to the individual with SMA?*

- ☐ Self
- ☐ Parent
- ☐ Spouse
- ☐ Grandparent
- ☐ Relative
- ☐ Friend
- ☐ Other

Show if question “What is your relationship to the individual with SMA?*” is not (“Self”)

Are you the primary caregiver of an individual with SMA?*

- ☐ Yes
- ☐ No

If YES, display the following text:

If you care for more than 1 individual with SMA, please complete the survey for the individual specified in the email invitation. Thank you!

Do you / Do you and the individual with SMA currently reside in the United States?*

- ☐ Yes
- ☐ No

What type of SMA **do you/does the individual with SMA** have?*

- ☐ Type 0
- ☐ Type 1
- ☐ Type 2
- ☐ Type 3
- ☐ Type 4

- Distal
- Kennedy's
- SMARD
- SMA-LED
- Unknown because diagnosed before symptom onset
- Not sure
- Other, please specify

Show if question "What type of SMA do you/does the individual with SMA have?" is ("Other, please specify:")

Please specify the type of SMA: _____

Your / The individual's birth date: _____ [Format validation required](#)

Please provide in mm/dd/yyyy or m/d/yyyy format (for example: 1/1/2000)

Show disqualification text if question:

- “Are you the primary caregiver of an individual with SMA?” is (“No”) OR
- “Do you / Does the individual with SMA reside in the United States?” is (“No”) OR
- “What type of SMA do you/does the individual with SMA have?” is (“Distal”, “Kennedy’s”, “SMARD”, or “SMA-LED”) OR
- “What is your relationship to the individual with SMA?” is (“Self”) AND age < 18

Mark survey as complete

Thank you for your interest in this survey, but this survey is intended for individuals with 5q SMA that are at least 18 years old or primary caregivers of individuals with 5q SMA that currently reside in the United States.

We invite you to explore other relevant research opportunities at <https://www.curesma.org/research-study-opportunities/>

If you have any questions, please contact us at data-analytics@curesma.org

Your Contact Information

Your first name:*

Your last name:*

Your email address: [Format validation required](#)

Your street address:

Your city:

Your state of residence: [Dropdown - show list of abbreviated states](#)

Your zip code:

Your phone number:

Your date of birth:* [Format validation required](#)

Please provide in mm/dd/yyyy or m/d/yyyy format (for example: 1/1/2000)

How did you first hear of Cure SMA?

- ☐ Doctor or other healthcare provider
- ☐ Family/Friend
- ☐ Online/Website Search
- ☐ Other - Write In:

May we share **your / your family's** information with the Chapter in your area? We share your contact information (name, email and phone number), child's name (if applicable) and SMA type.

- ☐ Yes
- ☐ No

Information About the Individual with SMA

Show page if question "What is your relationship to the individual with SMA?*" is not ("Self")

The individual's first name:*

The individual's last name:*

The individual's street address:

The individual's city:

The individual's state: [dropdown]

[Dropdown - show list of abbreviated states](#)

The individual's zip code:

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Your Information (Continued) [show for self] / Information About the Individual with SMA (Continued) [show for primary caregivers]

What sex was assigned at birth?*

- ☐ Male
- ☐ Female
- ☐ Prefer not to answer

Show question if the individual with SMA is at least 2 years of age

What is your/the individual's current gender identity?

- ☐ Male
- ☐ Female
- ☐ Transgender man/transgender male
- ☐ Transgender woman/transgender female
- ☐ Prefer to be identified as (non-binary, gender-fluid, agender, please specify)
- ☐ Prefer not to answer

Show if question ("What is your/the individual's current gender identity?") is ("Prefer to be identified as (non-binary, gender-fluid, agender, please specify):")

Please specify, gender identity: _____

What is your/the individual's racial identity? If you identify/the individual with SMA identifies with more than one, please select 'Two or more races'

- ☐ American Indian or Alaska Native
- ☐ Asian
- ☐ Black or African American
- ☐ Native Hawaiian or Other Pacific Islander
- ☐ White
- ☐ Middle Eastern or North African
- ☐ Two or more races
- ☐ Race not listed

- Prefer not to answer

Show if question "What is your racial identity? If you identify with more than one, please select 'Two or more races'" is one of the following answers ("Two or more races")

If 'Two or more races' was selected in the question above, please select all races that apply:

- ☐ American Indian or Alaska Native
- ☐ Asian
- ☐ Black or African American
- ☐ Native Hawaiian or Other Pacific Islander
- ☐ White
- ☐ Middle Eastern or North African
- ☐ Race not listed

Do you/Does the individual with SMA identify as Hispanic or Latinx?

- Yes
- No
- Unknown
- Prefer not to answer

Is English the primary language spoken at home?

- Yes
- No

Show if question "Is English the primary language spoken at home?" is one of the following answers ("No")

What language is primarily spoken at home?

Free text

How many copies of the backup gene, *SMN2* do you/does the individual with SMA have?*
The SMN2 gene is also called a back-up gene.

- 1
- 2

- ☐ 3
- ☐ 4 or more
- ☐ I/The individual with SMA did not have a genetic test done
- ☐ Not sure

Show if question (“How many copies of the backup gene, SMN2 do you/does the individual with SMA have?*) is (“4 or more”)

Some labs report combined results when there are 4 or more copies of SMN2 present (for example: “5 or more copies”). Please choose the option that was shown on your lab report, if known

- ☐ 4 copies of SMN2
- ☐ “4 or more” copies of SMN2
- ☐ 5 copies of SMN2
- ☐ “5 or more: copies of SMN2
- ☐ 6 copies of SMN2
- ☐ Not applicable
- ☐ Unknown

Show if question "What is your relationship to the individual with SMA?" is not one of the following answers ("Self")

Did you / the individual with SMA have a positive prenatal screen (for example: amniocentesis or chorionic villus sampling [CVS])?*

- ☐ Yes
- ☐ No
- ☐ Unknown

If yes to prenatal screening:

What was the reason for screening for SMA prenatally?

- ☐ I have another child with SMA
- ☐ I am and/or my partner are carriers of the SMA gene mutation
- ☐ Fetal ultrasounds showed abnormal findings such as reduced fetal movement or contractures

- ☐ Prenatal screening was recommended by our OB/GYN and/or fertility specialist
- ☐ Other (please specify)
- ☐ Unknown

Show if other (please specify) is selected

Please specify, other reason: _____

Show if question "What is your relationship to the individual with SMA?" is not one of the following answers ("Self") AND year of birth \geq 2016

Did you / the individual with SMA have a positive newborn screen?*

- ☐ Yes
- ☐ No
- ☐ Unknown

Show if question "What is your relationship to the individual with SMA?" is not one of the following answers ("Self")

If you are completing this survey on behalf of an individual with SMA that has passed away, please provide the deceased date. Otherwise, please skip this question.

If date is input, show the following text:

Cure SMA offers support and resources for families that are grieving the loss of a loved one. Please contact community support at CommunitySupport@curesma.org for more information.

Show if a deceased date was provided

Would you like to end the survey at this point?

- ☐ Yes, I'd like to end the survey now
- ☐ No, I'd like to continue

If yes is selected, skip to "Wrap Up" page (2nd to last page)

SMA Diagnosis

Was there a known biological family history of SMA (e.g., parent, sibling, and/or other relative with SMA)?

- ☐ Yes
- ☐ No
- ☐ Unknown

Was there a positive carrier screen of either biological parent?

- ☐ Yes
- ☐ No
- ☐ Unknown

Show if question “Did you / the individual with SMA have a positive newborn screen?” is (“No”)

Was genetic testing used to confirm the diagnosis of SMA (also known as an SMA blood test, SMA Diagnostic Test, or SMN Diagnostic Test)?

- ☐ Yes
- ☐ No genetic testing was done
- ☐ Unknown

Show if question “Did you / the individual with SMA have a positive newborn screen?” is (“Yes”) OR if question “Was genetic testing completed (also known as an SMA blood test, SMA Diagnostic Test, or SMN Diagnostic Test)?” is (“Yes”)

On what date did you receive the results from genetic testing?*

Instructions for individuals identified via screening:

- *For individuals identified by newborn screening, please report the date of that a diagnosis of SMA was confirmed by genetic testing. This usually occurs after the newborn screening test.*
- *For individuals diagnosed prenatally, please report the age (in days) that diagnosis was confirmed after birth. If unknown/unavailable then please report the date of prenatal testing.*

Format validation required

Is this an estimated date?

- ☐ Yes

- No

Show if question “Did you / the individual with SMA have a positive prenatal screen (for example: amniocentesis or chorionic villus sampling [CVS])?” is (“No”) AND if question “Did you / the individual with SMA have a positive newborn screen?” is (“No”)

Were other methods used to diagnose SMA? If genetic testing was completed, please indicate which methods used to diagnosed SMA prior to genetic testing.

	Yes	No	Don't Know
Electromyography (EMG)			
Muscle biopsy			
SMA diagnosed by symptoms			
Other, please specify			

If question (“Other, please specify”) is (“Yes”)

Please specify, other diagnostic method: _____

Show if (“Yes”) was selected for any options in question “What other methods were used to diagnose SMA prior to genetic testing results?”

If you were/the individual was diagnosed with SMA by one of the above methods prior to receiving the results of genetic testing: When did you/the individual first receive an SMA diagnosis from a clinician? If you are unsure of the exact date, please enter an approximate date.

Mm/dd/yyyy

Show question if answer was provided for question “If you were diagnosed with SMA by one of the above methods prior to receiving the results of genetic testing: When did you first receive an SMA diagnosis from a clinician?”

Is this an estimated date?

- Yes
- No

Show if question “Did you / the individual with SMA have a positive prenatal screen (for example: amniocentesis or chorionic villus sampling [CVS])?” is (“No”) AND if question “Did you / the individual with SMA have a positive newborn screen?” is (“No”)

Were the symptoms of SMA previously incorrectly diagnosed as a different disorder?

- ☐ Yes
- ☐ No
- ☐ Unknown

Show if question “Did you / the individual with SMA have a positive prenatal screen (for example: amniocentesis or chorionic villus sampling [CVS])?” is (“No”) AND if question “Did you / the individual with SMA have a positive newborn screen?” is (“No”) AND year of birth \geq 2016

Was the SMA diagnosis missed by newborn screening? Some babies that are screened for SMA have a false negative newborn screening result due to a genetic mutation that was not picked up by newborn screening. If this describes your experience, please select yes.

- ☐ Yes
- ☐ No
- ☐ Unknown

Show if question “Was the SMA diagnosis missed by newborn screening?” is (“Yes”)

Please select how many SMN1 copies are present, if known. Spinal muscular atrophy (SMA) is caused by a mutation in the survival motor neuron gene 1 (*SMN1*). SMA occurs when both of an individual’s *SMN1* copies have missing or mutated segments. Except in very rare cases, this happens when that individual has inherited two faulty copies of the gene—one from each parent.

- ☐ 0
- ☐ 1
- ☐ 2
- ☐ Unknown

Prenatal / Newborn Screening Questions

Show page if individual was identified by newborn or prenatal screening

What was the gestational age of the baby at birth (in weeks)?

- ☐ Less than 28 weeks
- ☐ 28 to 31 weeks
- ☐ 32 to 36 weeks
- ☐ 37 weeks or greater

State of birth: [Dropdown - show list of abbreviated states](#)

Who first contacted you regarding a possible SMA diagnosis?

- ☐ Pediatrician or primary care office
- ☐ Neuromuscular specialty clinic
- ☐ Newborn screening state laboratory
- ☐ Unknown
- ☐ Other (please specify)

When was the individual first seen by an SMA specialist? An SMA specialist is a provider who typically confirms the SMA diagnosis and can provide treatment options.

- ☐ 0 to 3 days after birth
- ☐ 4 to 7 days after birth
- ☐ 8 to 14 days after birth
- ☐ 15 to 28 days after birth
- ☐ Greater than 28 days after birth
- ☐ Not seen by an SMA specialist
- ☐ Unknown

Show if “When was the individual first seen by an SMA specialist?” is NOT (“Not seen by an SMA specialist”)

How far did you travel (one way) to see the first SMA specialist?

- ☐ Less than 10 miles
- ☐ 10-29 miles

- 30-59 miles
- 60-99 miles
- 100-199 miles
- 200-299 miles
- 300 or more miles
- Internationally
- Unknown
- Not applicable

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SMA Treatment

Has SMA treatment been received?*

- ☐ Yes
- ☐ Not yet, but seeking to start treatment
- ☐ No, treatment was declined

Show if question “Has SMA treatment been received?” is one of (“No, treatment was declined”)

If you are willing to share, please provide the primary reason that treatment was declined?

- ☐ Recommended by provider
- ☐ Healthcare provider does not administer SMA treatment
- ☐ Insurance restrictions
- ☐ Out of pocket cost
- ☐ Elevated AAV9 antibody titers
- ☐ Abnormal lab values (e.g. liver enzymes, troponin levels)
- ☐ Parent/caregiver choice
- ☐ Personal or cultural reasons
- ☐ Concerns with safety or side effects
- ☐ Other (please specify)
- ☐ Prefer not to answer

Show if question (“If you are willing to share, please provide the primary reason that treatment was declined?”) is (“Other (please specify)”):

Please specify, other reason for treatment decline:

First SMA Treatment

Show page if “Has SMA treatment been received?” is (“Yes”)

Questions on this page are specific to the first SMA treatment received. If multiple SMA treatments have been utilized, we will ask about those on another page.

What was the first SMA treatment received?*

- ☐ Spinraza (nusinersen)
- ☐ Zolgensma (onasemnogene-abeparvovec-xioi)
- ☐ Evrysdi (risdiplam)
- ☐ Apitegromab (SRK-015)
- ☐ Taldefgrobep
- ☐ RO7204239 (also known as GYM329, treatment used in the Manatee clinical trial)
- ☐ NMD670 (treatment used in the NMD Pharma clinical trials)
- ☐ Unknown

What was the date when the first SMA treatment was initiated? If you are unsure of the exact date, please enter an approximate date.

Show if date is input:

Is this an estimated date?

- ☐ Yes
- ☐ No
- ☐ Unknown

Was the first SMA treatment intended to be used temporarily while waiting to start another SMA treatment? This is sometimes referred to as “bridge” treatment, and it can allow for an individual to start another SMA treatment earlier while waiting on possible delays caused by insurance or clinical reasons.

- ☐ Yes
- ☐ No
- ☐ Unknown

How was the SMA treatment first accessed?

- ☐ Through a clinical trial
- ☐ Through an early access program prior to FDA approval

- Treatment was prescribed by a physician after FDA approval
- Other (please specify)
- Unknown

Show if question “How was the SMA treatment first accessed?” is (“Through a clinical trial”) *[remove logic once FDA approval]* and “What was the first SMA treatment received?” is (“Spinraza (nusinersen)”)

What dosage of Spinraza (nusinersen) was received?

- 12/12 mg regimen dose (approved by FDA in 2016)
- 50/28 mg regimen dose (higher dose evaluated as part of the DEVOTE clinical trial)
- Unknown

Show if question “How was the SMA treatment first accessed?” is (“Through a clinical trial”) *[remove logic once FDA approval]* and “What was the first SMA treatment received?” is (“Zolgensma (onasemnogene-abeparvovec-xioi)”)

How was Zolgensma (onasemnogene-abeparvovec-xioi) administered?

- Intravenous (approved by the FDA in 2019)
- Intrathecal via spinal lumbar puncture (evaluated as part of the STRENGTH and STEER clinical trial)
- Unknown

[Question disabled until FDA approval]

Show if question “What was the first SMA treatment received?” is (“Evrysdi (risdiplam)”)

What formulation of Evrysdi (risdiplam) was received?

- Liquid (approved by the FDA in 2020)
- Tablet
- Unknown

Show if question “How was the SMA treatment first accessed?” is (“Treatment was prescribed by a physician”)

Please select your insurance provider(s) at the time of first SMA treatment initiation.

- ☐ Aetna
- ☐ Anthem
- ☐ BCBS Florida
- ☐ BCBS Massachusetts
- ☐ BCBS Michigan
- ☐ BCBS New Jersey
- ☐ BCBS North Carolina
- ☐ BCBS Tennessee
- ☐ Care First BCBS
- ☐ Centene
- ☐ Cigna
- ☐ Emblem Health
- ☐ Health Care Service Corporation (HCSC)
- ☐ Highmark Health
- ☐ Humana
- ☐ Independence BC
- ☐ Kaiser Permanente
- ☐ UnitedHealthcare
- ☐ State Medicaid or Medical Assistance
- ☐ Medicare
- ☐ Other (please specify)
- ☐ Unknown

Show if question ("Please select your insurance provider(s) at the time of first SMA treatment initiation") is ("Other (please specify)")

Please specify, other insurance provider:

Delay of First SMA Treatment

Show page if "What was the first SMA treatment received?" is one of the following answers ("Spinraza", "Evrysdi", "Zolgensma")) and "How was SMA treatment first accessed" is not ("Through a clinical trial")

How much time passed between when the physician prescribed the first SMA treatment and when you / the individual with SMA received treatment?

- ☐ Less than 1 week
- ☐ 1-2 weeks
- ☐ 3-4 weeks
- ☐ 1-3 months
- ☐ 4-6 months
- ☐ 7-12 months
- ☐ Greater than 1 year
- ☐ Unknown

Did any of the following reasons increase the time it took to receive the first SMA treatment? Please select all that apply.

- ☐ Insurance initially denied coverage
- ☐ Insurance review process
- ☐ Had to apply for patient assistance program / manufacturer free drug program
- ☐ Financial burden
- ☐ Elevated AAV9 antibody titers
- ☐ Abnormal lab values
- ☐ Scheduling challenges
- ☐ Distance from a treatment center
- ☐ Changed doctors and/or treatment center
- ☐ Treatment availability (challenges with care center obtaining treatment)
- ☐ Shipping delays
- ☐ Other (please specify)

Show question if “Did any of the following reasons increase the time it took to receive the first SMA treatment? Please select all that apply.” is (“Other (please specify)”)

Please specify, other reason that increased time to receive first treatment: _____

Show question if “Did any of the following reasons increase the time it took to receive the first SMA treatment? Please select all that apply.” is (“Insurance initially denied coverage”)

Which insurer(s) denied coverage?

- ☐ Pipe in insurers selected
- ☐ Unknown

Show question if > 1 answer was selected for question: “Did any of the following reasons increase the time it took to receive the first SMA treatment? Please select all that apply.”

Which factor resulted in the longest delay?

- ☐ [pipe in all answers selected]

Additional comments, if applicable: [\[open text\]](#)

Additional SMA Treatments

Show page if question (“Has SMA treatment been received”) is (“Yes”)

Were additional SMA treatments received after [\[insert first treatment name\]](#)?*

- ☐ Yes
- ☐ No
- ☐ Unknown

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Additional SMA Treatments

Show page if question (“Were additional SMA treatments received?”) is (“Yes”)

Information provided on the first SMA treatment has already been filled in. Please add information on additional SMA treatments and how they were accessed below.

Please select the additional SMA treatments that have ever been received. The first SMA treatment previously reported has been automatically filled in? *

[select “yes” for treatment reported as the first SMA treatment]

	Yes	No	Unknown
Spinraza (nusinersen)			
Zolgensma (onasemnogene abeparvovec-xioi)			
Evrysdi (risdiplam)			
Apitegromab (also known as SRK-015)			
Taldefgrobep			
RO7204239 (also known as GYM329, treatment used in the Manatee clinical trial)			
NMD670 (treatment used in the NMD Pharma clinical trials)			

How was [insert SMA treatment] accessed?

If [insert SMA treatment] was the first SMA treatment received, any responses from the "First SMA Treatment" page have been filled in. If [insert SMA treatment] was accessed in multiple ways, please indicate below.

[pre-fill answer selected for first treatment; repeat question for each selected SMA treatment]

	Yes	No	Unknown

Through a clinical trial			
Through an early access program prior to FDA approval [Spinraza, Zolgensma, Evrysdi, Apitegromab only]			
Treatment was prescribed by a physician after FDA approval [Spinraza, Zolgensma, Evrysdi only]			
Other (please specify)			

Show if: Question "Spinraza (nusinersen)" is ("Yes") and "How was Spinraza (nusinersen) accessed?" is ("Through a clinical trial") *[remove logic once FDA approval]*

What dosage of Spinraza was received?

[pre-fill answer selected for first treatment]

	Yes	No	Unknown
12/12 mg regimen dose (approved by FDA in 2016)			
50/28 mg regimen dose (higher dose evaluated as part of the DEVOTE clinical trial)			

Show if question "Zolgensma (onasemnogene abeparvovec-xioi)" is one of the following answers ("Yes") and "How was Zolgensma (onasemnogene abeparvovec-xioi) accessed?" is ("Through a clinical trial") *[remove logic once FDA approval]*

How was Zolgensma (onasemnogene abeparvovec-xioi) administered?

[pre-fill answer selected for first treatment]

- Intravenous (approved by the Food and Drug Administration for individuals in 2019)

- Intrathecal via spinal lumbar puncture (evaluated as part of the STRENGTH and STEER clinical trial)
- Unknown

[Question disabled until FDA approval]

Show if question “What was the first SMA treatment received?” is (“Evrysdi (risdiplam)”)

What formulation of Evrysdi (risdiplam) was received?

[pre-fill answer selected for first treatment]

	Yes	No	Don't know
Liquid (approved by the Food and Drug Administration in 2020)			
Tablet			

Treatment Discontinuation

Show page if question (“Has SMA treatment been received”) is (“Yes”)

Show if any treatment options were selected in question “Which SMA treatment(s) have been utilized? *”

Were any of the following treatments discontinued? Please mark yes if the treatment was stopped and restarted.*

	Yes	No	Don't know
Pipe in treatments selected in question “Which SMA treatment(s) have been utilized? *”			

Show if any treatment options were selected (“Yes”) in question “Were any of the following treatments discontinued? Please mark yes if the treatment was stopped and restarted.*

Were any treatments re-started after discontinuation?

- ☐ Yes
- ☐ No
- ☐ Unknown

Treatment Start and Stop Dates

Show page if at least one treatment is selected

Show question if individual is on more than one treatment or if individual is on one treatment and has noted discontinuation of treatment.

Please list treatment start and stop dates, where applicable. Start date is the date of first dose and stop date is the date of last dose.

If treatment has not been discontinued, please leave the field blank. Additional start and stop date columns have been provided if it was indicated that a treatment has been discontinued and re-started.

					Only show if they reported a treatment has been restarted
	Start Date	Stop Date	Start Date (2)	Stop Date (2)	
Pipe in treatments selected in question "Which SMA treatment(s) have been utilized? *"					

Show if Spinraza has been marked as discontinued

What was the primary reason for Spinraza (nusinersen) discontinuation?

- ☐ SMA symptoms did not improve
- ☐ SMA symptoms worsened
- ☐ Not well tolerated
- ☐ Difficult route of administration
- ☐ Insurance no longer covered treatment
- ☐ Clinical trial ended
- ☐ Required to enter a clinical trial for another SMA treatment
- ☐ Patient/parent/caregiver request
- ☐ It was intended to be temporarily used while waiting to start another SMA treatment
- ☐ Other

- Unknown

Show if Evrysdi has been marked as discontinued

What was the primary reason for Evrysdi (risdiplam) discontinuation?

- SMA symptoms did not improve
- SMA symptoms worsened
- Not well tolerated
- Difficult route of administration
- Insurance no longer covered treatment
- Clinical trial ended
- Required to enter a clinical trial for another SMA treatment
- Patient/parent/caregiver request
- It was intended to be temporarily used while waiting to start another SMA treatment
- Other
- Unknown

SMA Treatments Not Received

Were any of the following SMA treatments prescribed, but not utilized?

	Yes	No	Unknown
pipe in FDA-approved SMA treatments that were not utilized			

What was the primary reason that __[insert treatment name = “yes”]__ was not utilized?

- ☐ Insurance denied treatment
- ☐ Waiting on insurance approval
- ☐ Elevated AAV9 antibody titers [show for Zolgensma only]
- ☐ Abnormal lab values
- ☐ Scheduling
- ☐ Distance from a treatment center
- ☐ Changed doctors and/or treatment center
- ☐ Patient / caregiver declined
- ☐ Other (please specify)

Show if question (“What was the primary reason for not receiving __[insert treatment name = “yes”]__?”) is (“Other (please specify)”)

Please specify, other reason why [insert treatment name] has not been utilized:

Barriers to Treatment Continuation

Show if individual selected Spinraza or Evrysdi was utilized

Have you / Has the individual with SMA experienced any unplanned gaps in treatment in the past 12 months?

- ☐ Yes
- ☐ No
- ☐ Unknown

Show if yes

Which treatment(s)?

[pipe in treatments selected – restrict to Spinraza and Evrysdi]

	Yes	No	Unknown
Spinraza (nusinersen)			
Evrysdi (risdiplam)			

Please select the reasons for the significant gap(s) in [pipe in each treatment] utilization in the past 12 months.

- ☐ Insurance denied the treatment renewal
- ☐ Lost / switched insurance providers
- ☐ Scheduling conflicts
- ☐ Distance from treatment center
- ☐ Finding transportation to treatment center
- ☐ Changed physicians / treatment centers
- ☐ Cost of treatment
- ☐ Personal choice
- ☐ Shipping delays
- ☐ Treatment availability (challenges with care center obtaining treatment)
- ☐ Other (please specify)

Show question if (“Please select the reasons for the significant gap(s) in [pipe in treatment] utilization in the past 12 months.”) is (“Other (please specify)”):

Please specify, other reason for significant gaps in [pipe in each treatment name] utilization: _____

For each reason selected:

How long was the longest gap due to [pipe in each gap reason(s) selected]?

- ☐ Less than 1 week
- ☐ 1-2 weeks
- ☐ 3-4 weeks
- ☐ 1-2 months
- ☐ 3-5 months
- ☐ 6-12 months
- ☐ More than 1 year

In the last year, how often have significant treatment gaps occurred?

- ☐ One time
- ☐ 2-3 times
- ☐ More than 4 times

Symptoms

Have you / Has the individual with SMA exhibited any SMA symptoms? Examples include stiff joints, feeding difficulties, breathing difficulties, muscle weakness, etc.

- ☐ Yes
- ☐ No
- ☐ Unknown

Show if question “Have you / Has the individual with SMA exhibited any SMA symptoms? Examples include stiff joints, feeding difficulties, breathing difficulties, muscle weakness, etc.” is (“Yes”) AND age ≥ 18

At what age did your symptoms start? Age can be provided in either months or years. If less than 1 month, please enter 0.

___ months
___ years

Show if question “Have you / Has the individual exhibited any SMA symptoms? Examples include stiff joints, feeding difficulties, breathing difficulties, muscle weakness, etc.” is (“Yes”) AND age < 18

Please indicate which symptoms the individual with SMA has experienced and the earliest timing of symptom onset.

	Not experienced	At birth	After birth, but before any SMA treatment	Hide column if treatment == No or not yet After initiation of first SMA treatment
Joint or muscle contractures (stiff joints)				
Feeding difficulties				
Respiratory difficulties (breathing difficulties)				
Muscle weakness or delayed milestone achievement				
Other, please specify				

Show question if (“Other, please specify) is (“Yes”)

Please specify, other symptom: _____

SMA Care

Are any healthcare specialists currently caring for you / the individual with SMA? An example of a healthcare specialist is a neurologist.

- ☐ Yes
- ☐ No
- ☐ Unknown

Show if question (“Are any healthcare specialists currently caring for you / the individual with SMA?”) is (“Yes”)

Which of the following types of healthcare provider specialty areas are currently caring for you / the individual with SMA?

	Yes	No	Unknown
Pediatrician			
Neurology			
Medical Genetics/Genetic Counselor			
Pulmonology (focus on breathing and lungs)			
Care coordination (helps navigate clinic appointments and treatments)			
Nutrition			
Social work			
Emotional health (mental health providers such as psychiatrist, psychologist, etc.)			
Physical therapy			
Occupational therapy			
Speech / swallow therapy			
Physical Medicine and Rehabilitation / Physiatry (focus on enhancing and restoring functional ability)			
Gastroenterology (focus on digestive system, including esophagus, stomach, intestines, liver, bile ducts, and gallbladder)			
Orthopedics			
Endocrinology (focus on glands and organs that make hormones)			
Urology (focus on urinary and reproductive organs)			
Cardiology (focus on heart function)			
Obstetrics and Gynecology (OB/GYN)			
Other (please specify)			

Show question if “Which of the following types of healthcare provider specialty areas are currently caring for you / the individual with SMA? Please select all that apply.” Is (“Other (please specify)”)

Please specify, other specialist: _____

Show question if no responses were selected in question “Which of the following types of healthcare provider specialty areas are currently caring for you / the individual with SMA? Please select all that apply.”

Have you / Has the individual with SMA been referred to a specialist?

- ☐ Yes
- ☐ No
- ☐ Unknown

Show if: question " Have you / Has the individual with SMA been referred to a specialist?" is one of the following answers ("Yes")

Which specialist(s)?

	Yes	No	Unknown
Pediatrician			
Neurology			
Medical Genetics/Genetic Counselor			
Pulmonology (focus on breathing and lungs)			
Care coordination (helps navigate clinic appointments and treatments)			
Nutrition			
Social work			
Emotional health (mental health providers such as psychiatrist, psychologist, etc.)			
Physical therapy			
Occupational therapy			
Speech / swallow therapy			
Physical Medicine and Rehabilitation / Physiatry (focus on enhancing and restoring functional ability)			
Gastroenterology (focus on digestive system, including esophagus, stomach, intestines, liver, bile ducts, and gallbladder)			
Orthopedics			
Endocrinology (focus on glands and organs that make hormones)			
Urology (focus on urinary and reproductive organs)			
Cardiology (focus on heart function)			
Obstetrics and Gynecology (OB/GYN)			
Other (please specify)			

Show question if “Which specialist(s)?” Is (“Other (please specify)”)

Please specify, other specialist: _____

On average, how far do you / does the individual with SMA travel (one way) for SMA-related healthcare services?

- ☐ Less than 10 miles
- ☐ 10-29 miles
- ☐ 30-59 miles
- ☐ 60-99 miles
- ☐ 100-199 miles
- ☐ 200-299 miles
- ☐ 300 or more miles
- ☐ Internationally
- ☐ Unknown
- ☐ Not applicable

Breathing and Nutrition

Are any of the following used for breathing?

- ☐ Cough assist machine
- ☐ BIPAP or ventilator, less than 8 hours per day
- ☐ BIPAP or ventilator, 8-16 hours per day
- ☐ BIPAP or ventilator, more than 16 hours per day
- ☐ Tracheostomy
- ☐ None of the above

Do you/ Does the individual with SMA currently have trouble swallowing?

- ☐ Always (all the time) (100%)
- ☐ Usually (80-99%)
- ☐ Often (60-79%)
- ☐ About half the time (40-59%)
- ☐ Sometimes (20-39%)
- ☐ Rarely (1-19%)
- ☐ Never (0%)

How often is **your / the individual's** voice soft, weak or hard to be heard?

- ☐ Always (all the time) (100%)
- ☐ Usually (80-99%)
- ☐ Often (60-79%)
- ☐ About half the time (40-59%)
- ☐ Sometimes (20-39%)
- ☐ Rarely (1-19%)
- ☐ Never (0%)

During the past month, which of the following describes **your / the individual's** feeding routine on a typical day?

- ☐ Nothing by mouth
- ☐ Tube dependent with minimal attempts of food or liquid.
- ☐ Tube dependent with consistent oral intake of food or liquid.

- Total oral diet of a single consistency.
- Total oral diet with multiple consistencies, but requiring special preparation or compensations.
- Total oral diet with multiple consistencies without special preparation, but with specific food limitations.
- Total oral diet with no restrictions.

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Motor Milestones

Can you/the individual with SMA currently perform the following motor functions?

	Yes	No	Unknown
Head control (able to hold head up on own)			
Voluntary grasping			
Voluntary kicking			
Roll over completely			
Sitting without support			
Hands and knees crawling			
Standing with assistance			
Walking with assistance			
Standing alone			
Walking alone			

Which of the following motor functions have you/ has the individual with SMA ever achieved?

[prepopulate current motor milestones marked "Yes"]

	Yes	No	Unknown
Head control (able to hold head up on own)			
Voluntary grasping			
Voluntary kicking			

Roll over completely			
Sitting without support			
Hands and knees crawling			
Standing with assistance			
Walking with assistance			
Standing alone			
Walking alone			

Show question if age < 5

At what age did the individual with SMA attain the ability to achieve the following milestone(s)? Please report the age in months.

Only show rows where ever achieved = yes

Holds up head	Textbox (force positive, whole number)
Voluntary or independent grasping	Textbox (force positive, whole number)
Voluntary or independent kicking	Textbox (force positive, whole number)
Rolling (back to tummy)	Textbox (force positive, whole number)
Rolling (tummy to back)	Textbox (force positive, whole number)
Sitting supported	Textbox (force positive, whole number)
Sitting independently	Textbox (force positive, whole number)
Hands and knees crawling	Textbox (force positive, whole number)
Standing with assistance	Textbox (force positive, whole number)
Walking with assistance	Textbox (force positive, whole number)
Standing alone	Textbox (force positive, whole number)
Walking alone	Textbox (force positive, whole number)

PROMIS

Show page if individual with SMA is 5 -17 years old

This page contains questions from a standardized measurement tool called the Patient-Reported Outcomes Measurement Information System (PROMIS) to evaluate physical function.

PROMIS Parent Proxy Upper Extremity – Short Form 8a

Please respond to each question or statement by marking one box per row.

In the past 7 days...

	With no trouble	With a little trouble	With some trouble	With a lot of trouble	Not able to do
My child could button his/her shirt or pants (trousers)	<input type="checkbox"/> 4	<input type="checkbox"/> 3	<input type="checkbox"/> 2	<input type="checkbox"/> 1	<input type="checkbox"/> 1
My child could open a jar by himself/herself	<input type="checkbox"/> 4	<input type="checkbox"/> 3	<input type="checkbox"/> 2	<input type="checkbox"/> 1	<input type="checkbox"/> 1
My child could open the rings in school binders	<input type="checkbox"/> 4	<input type="checkbox"/> 3	<input type="checkbox"/> 2	<input type="checkbox"/> 1	<input type="checkbox"/> 1
My child could pour a drink from a full pitcher (jug)	<input type="checkbox"/> 4	<input type="checkbox"/> 3	<input type="checkbox"/> 2	<input type="checkbox"/> 1	<input type="checkbox"/> 1
My child could pull a shirt on over his/her head without help.....	<input type="checkbox"/> 4	<input type="checkbox"/> 3	<input type="checkbox"/> 2	<input type="checkbox"/> 1	<input type="checkbox"/> 1
My child could pull open heavy doors	<input type="checkbox"/> 4	<input type="checkbox"/> 3	<input type="checkbox"/> 2	<input type="checkbox"/> 1	<input type="checkbox"/> 1
My child could put on his/her shoes without help	<input type="checkbox"/> 4	<input type="checkbox"/> 3	<input type="checkbox"/> 2	<input type="checkbox"/> 1	<input type="checkbox"/> 1
My child could use a key to unlock a door.....	<input type="checkbox"/> 4	<input type="checkbox"/> 3	<input type="checkbox"/> 2	<input type="checkbox"/> 1	<input type="checkbox"/> 1

Show for ambulatory individuals only:

PROMIS Parent Proxy Mobility – Short Form 7a

Please respond to each question or statement by marking one box per row.

In the past 7 days...

	With no trouble	With a little trouble	With some trouble	With a lot of trouble	Not able to do
My child could get up from the floor	<input type="checkbox"/> 4	<input type="checkbox"/> 3	<input type="checkbox"/> 2	<input type="checkbox"/> 1	<input type="checkbox"/> 1
My child could walk up stairs without holding on to anything.....	<input type="checkbox"/> 4	<input type="checkbox"/> 3	<input type="checkbox"/> 2	<input type="checkbox"/> 1	<input type="checkbox"/> 1
My child has been physically able to do the activities he/she enjoys most	<input type="checkbox"/> 4	<input type="checkbox"/> 3	<input type="checkbox"/> 2	<input type="checkbox"/> 1	<input type="checkbox"/> 1
My child could keep up when he/she played with other kids.....	<input type="checkbox"/> 4	<input type="checkbox"/> 3	<input type="checkbox"/> 2	<input type="checkbox"/> 1	<input type="checkbox"/> 1
My child could stand up on his/her tiptoes.....	<input type="checkbox"/> 4	<input type="checkbox"/> 3	<input type="checkbox"/> 2	<input type="checkbox"/> 1	<input type="checkbox"/> 1
My child could move his/her legs.....	<input type="checkbox"/> 4	<input type="checkbox"/> 3	<input type="checkbox"/> 2	<input type="checkbox"/> 1	<input type="checkbox"/> 1
My child could stand up without help	<input type="checkbox"/> 4	<input type="checkbox"/> 3	<input type="checkbox"/> 2	<input type="checkbox"/> 1	<input type="checkbox"/> 1

SMAIS (Self Report)

Show page if age ≥ 18 and relationship is “Self”

Display text: This page contains questions from a standardized measurement called the SMA independence scale (SMAIS) tool to evaluate functional independence.

SMAIS not shown in this version

SMAIS (Caregiver Report)

Show page if individual with SMA is ≥ 18 years and relationship is not “Self”

Display text: This page contains questions from a standardized measurement called the SMA independence scale (SMAIS) tool to evaluate functional independence.

SMAIS not shown in this version

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Hospitalizations

Show page if age < 5

How many times has the individual with SMA been hospitalized in the past 12 months for any reason related to SMA other than treatment administration?

- ☐ 0
- ☐ 1
- ☐ 2
- ☐ 3
- ☐ 4
- ☐ More than 4
- ☐ Unknown

Show question if “How many times has the individual with SMA been hospitalized in the past 12 months for any reason related to SMA other than treatment administration?” is (“More than 4”)

How many times has the individual with SMA been hospitalized in the past 12 months?

Show if question “Have you / Has the individual with SMA been hospitalized for any reason other than treatment administration?” is (“1”, “2”, “3”, “4”, “More than 4”)

**Please provide additional detail on the hospitalizations from the past 12 months.
If more than 4 hospitalizations have occurred in the past 12 months, please provide details on the first 4.**

The following is asked for each hospitalization:

Month: [Dropdown](#)

Year: _____

Reason:

- ☐ Respiratory illness (e.g., RSV, pneumonia)
- ☐ Respiratory failure
- ☐ Complication of treatment
- ☐ Elective / planned surgery or other procedure (e.g., spinal fusion)

- ☐ Non-planned surgery or other procedure (e.g., appendectomy)
- ☐ Dehydration or malnutrition
- ☐ Gastrointestinal / bowel issues
- ☐ Feeding tube problems
- ☐ Heart issues
- ☐ Seizures
- ☐ Headache
- ☐ Trauma, fracture, or external injury
- ☐ Other (please specify)

Show if question (“Reason”) is (“Other (please specify)”) **CONFIDENTIAL**

Other reason: _____

Length of stay:

- ☐ Less than 2 days
- ☐ 2-7 days
- ☐ 8-14 days
- ☐ 15-28 days
- ☐ More than 28 days
- ☐ Unknown

Wrap Up and Thank you

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