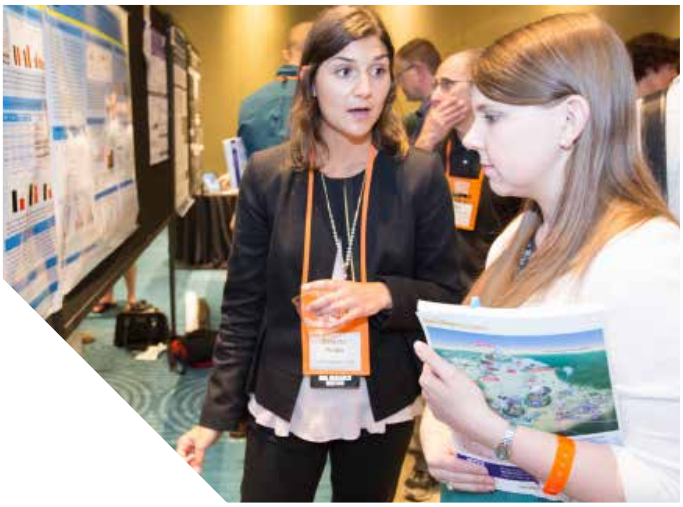


CURE SMA STATE OF SMA

2024 REPORT

PUBLICATION DATE: MARCH 31, 2025

DEAR CURE SMA COMMUNITY,

2024 marked another transformative year in research, advocacy, treatment, and care in the SMA community. Cure SMA has been busy preparing the patient, caregiver, payer, researcher and healthcare provider communities for the next wave of SMA treatments expected to become available in 2025.

Cure SMA has connected with individuals worldwide through three separate databases: a patient-reported outcomes database with data from over 11,000 affected individuals worldwide that also incorporates longitudinal data from our annual community update survey; an electronic medical record (EMR) sourced registry that compiles clinical data from U.S.-based SMA Care Center Network sites; and a newborn screening registry with data from parents of babies with SMA identified through statewide SMA newborn screening. The data from these databases has been presented in 11 poster presentations and 2 oral presentations at 7 scientific conferences in 2024. These achievements were made possible because of the SMA community and your willingness to share your stories, experiences, hopes, and needs with Cure SMA.

The data presented in this fourth annual State of SMA focuses on the current landscape of the SMA community, provides support and evidence for the previous milestones we have reached, and highlights ongoing unmet needs. These key unmet needs are emphasized in **blue bubbles** throughout the report.

While the data presented here summarizes the entire population of those who have previously connected with us and are diagnosed with SMA, we understand that not every graph and figure in this report is representative of every unique individual and their own personal story. Cure SMA is hard at work to engage with new members of the SMA community and develop new surveys to collect new outcomes from those recently diagnosed with SMA and collect viewpoints from those caring for those with SMA.

Every participant counts and lends insight into further understanding the changing landscape of SMA. **This work celebrates you.**

Thank you all for your commitment to Cure SMA.

Sincerely,

Lisa Belter, MPH
Vice President,
Data Analytics

Sarah Whitmire, MS
Director,
Data Analytics

Erin Welsh, MPH
Specialist,
Data Analytics

Mary Schroth, MD, FAAP, FCCP
Chief Medical Officer

TABLE OF CONTENTS

ACKNOWLEDGMENTS

Cure SMA is thankful to all individuals with spinal muscular atrophy (SMA) and their families who have generously shared their data. Their willingness to provide details about how SMA impacts their families and daily lives allows us to advance the understanding of this disease and lays the foundation for continued progress on behalf of our community.

Cure SMA is grateful for the support and funding provided by the Cure SMA Real World Evidence Collaboration (RWEC) and the Cure SMA Industry Collaboration (SMA-IC) for research initiatives.

Cure SMA is also grateful to the SMA Care Center Network (CCN) for their commitment to improving care for people with SMA and contributing consented patient data. Cure SMA acknowledges the Care Center Network Registry Committee for their review and editing of this work.

THE CURE SMA REAL WORLD EVIDENCE COLLABORATION

The Cure SMA Real World Evidence Collaboration (RWEC) was established in 2021 to leverage the experience, expertise and resources of pharmaceutical and biotechnology companies and nonprofit organizations involved in the development of SMA therapeutics to guide the future direction of real world evidence collection and use in SMA. Funding for the development of the State of SMA was provided by the Cure SMA RWEC. Current members of the RWEC include Cure SMA, Biogen, Genentech/Roche, and Novartis.

THE CURE SMA INDUSTRY COLLABORATION

The Cure SMA Industry Collaboration (SMA-IC) was established in 2016 to leverage the experience, expertise, and resources of pharmaceutical and biotechnology companies, as well as other nonprofit organizations involved in the development of spinal muscular atrophy (SMA) therapeutics to more effectively address a range of scientific, clinical, and regulatory challenges. Funding for the research included in the State of SMA report was provided by the 2024 SMA-IC. Members of the 2024 SMA-IC include Cure SMA, Scholar Rock, Biogen, Biohaven Pharmaceuticals, Genentech/Roche, Novartis, Alcyone Therapeutics, NMD Pharma, and SMA Europe.

ACKNOWLEDGMENTS

CURE SMA CARE CENTER NETWORK

Since 2018, Cure SMA has partnered with hospitals across the U.S. with the goal to improve healthcare for people with SMA. Every Care Center Network site submits consented patient information and data to the Cure SMA Clinical Data Registry. This data is then analyzed to drive healthcare improvements.

The SMA Care Center Network includes the following sites:

ADULT & PEDIATRIC CENTERS

Boston Children's Hospital, Boston, MA

Columbia University, New York, NY

Connecticut Children's Medical Center, Hartford, CT

Duke University Medical Center, Durham, NC

Gillette Children's Specialty Healthcare, St. Paul, MN

The Children's Hospital of Philadelphia, Philadelphia, PA

The University of Michigan, Ann Arbor, MI

University of California, Los Angeles (UCLA), Los Angeles, CA

University of Miami, Miami, FL

University of New Mexico, Albuquerque, NM

University of Rochester Medical Center, Rochester, NY

University of Utah, Program for Inherited Neuro Disorders, SLC, UT

Washington University/ St. Louis Children's Hospital, St. Louis, MO

PEDIATRIC CENTERS

Advocate Children's Hospital, Park Ridge, IL

Arkansas Children's Hospital, Little Rock, AR

Children's Healthcare of Atlanta, Atlanta, GA

Children's Hospital Colorado, Aurora, CO

Children's National Medical Center Washington, DC

Children's of Alabama, Birmingham, AL

Phoenix Children's Hospital, Phoenix AZ

Seattle Children's Hospital, Seattle, WA

Stanford Children's Health, Palo Alto, CA

University of Texas Southwestern /Children's Health, Dallas, TX

Vanderbilt University Medical Center, Nashville, TN

Yale Pediatric Neuromuscular Clinic, New Haven, CT

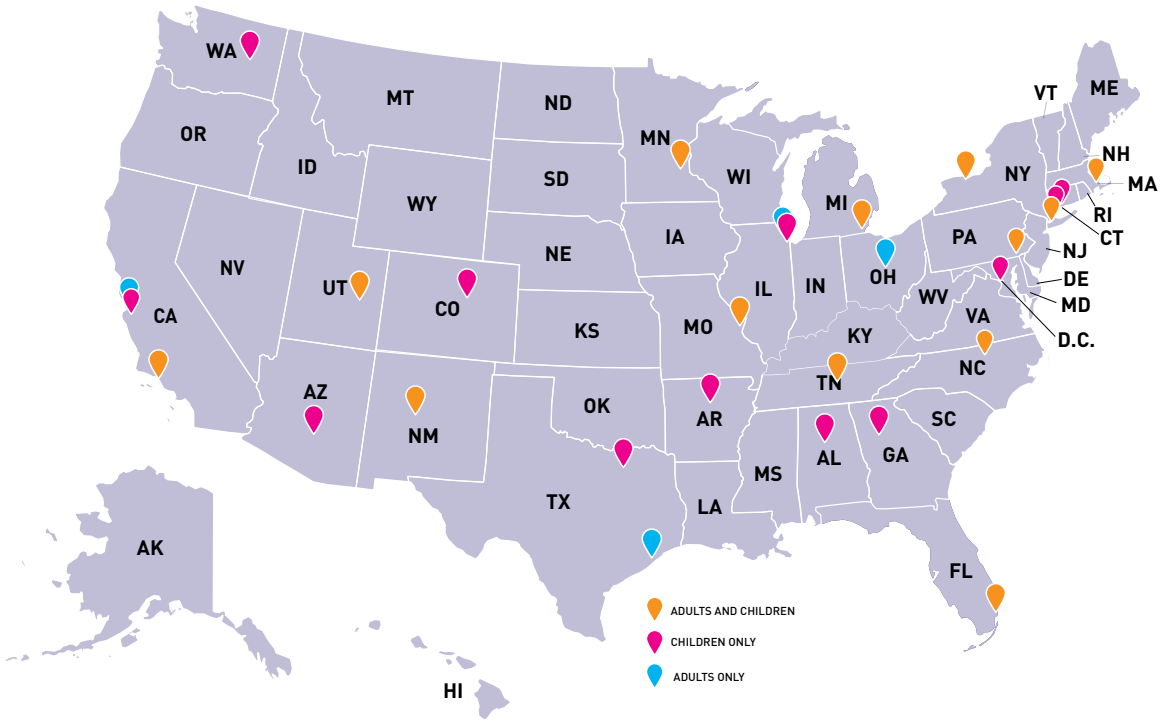
ADULT CENTERS

Baylor College of Medicine, Houston, TX

Northwestern University, Chicago, IL

Stanford Health, Palo Alto, CA

The Ohio State University, Wexner Medical Center, Columbus, OH



Additional Acknowledgments

Funding for the Cure SMA Care Center Network has been provided in part by the Erin Trainor Memorial Fund, and the Tyler William Orr Memorial Fund.

ABOUT THIS REPORT

This report is based on internal data from Cure SMA data sources and output from the SMA model, which Cure SMA created to estimate demographic and clinical characteristics of individuals with SMA in the United States.

Both the patient-reported data and the clinician-reported data are presented in this report. In some of our analyses, we have combined the data sources. Even though data comes from multiple sources and perspectives, previous analyses have shown high reliability between the patient-reported and clinician reported data.[1]

The data in this report includes individuals who are:

- Diagnosed (both self-reported and/or clinically confirmed) with 5q SMA
- Included in one or more of our databases as of December 31, 2024
- Residents of the United States

The data in the report describes individuals as:

- Children (ages 0-12 years)
- Teens (ages 13-17 years)
- Pediatric (ages 0-17 years) and
- Adults (ages 18 years and older)

The Cure SMA databases are dynamic and rely on the community's participation to share up to date information on the lived experiences of SMA. This report reflects the most current information housed in Cure SMA's databases, but there may be delays in collecting new information, and differences in numbers presented in this report and in previous and future State of SMA reports may be observed.

Additionally, all the analyses within this report are descriptively showing what we see in the data, but they do not adjust for factors that may bias the results. Caution should be taken when interpreting the results.

References:

1. *Belter L, Whitmire S, Curry M, Schroth M. Comparing Spinal Muscular Atrophy (SMA) outcomes between a patient-reported membership database and a clinical data registry. Poster presented at the American Academy of Neurology in Boston, MA in April 2023.*

Here are a few terms that you will see as you read this report:

CLINICIAN REPORTED DATA:
Data that is gathered from clinician reported medical records about a patient seeking care, commonly through medical records, case report forms, or surveys.

DURABLE MEDICAL EQUIPMENT:
Medical devices or supplies that are prescribed for repeated long-term use, such as a wheelchair or BPAP machine.

EFFICACY:
How well a treatment works in a controlled setting, such as a clinical trial.[1]

ELECTRONIC CASE REPORT FORM (eCRF):
A digital questionnaire used to collect data.

ELECTRONIC MEDICAL RECORD (EMR):
Digital version of a patient's healthcare chart.[2]

INSTITUTIONAL REVIEW BOARD (IRB):
A group that has been formally designated to review and monitor biomedical research involving human subjects. The IRB has the authority to approve, modify, or disapprove research.[3]

MEAN:
The average value in a dataset.

MEDIAN:
The middle value in a set of numbers.

MORTALITY RATE PER YEAR:
The frequency of the occurrence of death within a subgroup of individuals within a one-year period.

PATIENT-REPORTED DATA:
Data that is gathered directly from a patient, commonly through online surveys and questionnaires.

QUALITATIVE DATA:
Non-numerical or descriptive data, such as information gathered from interviews, focus groups, etc.[4]

SMA BIRTH PREVALENCE:
The proportion of individuals born in a specific time period that have SMA.

SMA INCIDENCE:
The number of individuals who are diagnosed with SMA per year.

SMA PREVALENCE:
The number of individuals that are currently living with SMA.

SOCIAL DETERMINANTS OF HEALTH (SDOH):
The conditions in which people are born, grow, live, work, and play that affect their health and well-being.

STANDARD DEVIATION (SD):
A measure of how far away the data points are from the average value in a dataset.[5]

References:

1. *https://pubmed.ncbi.nlm.nih.gov/14586394/*
2. *https://www.forbes.com/advisor/business/software/what-is-an-emr/ -*
3. *https://www.fda.gov/about-fda/center-drug-evaluation-and-research-cder/institutional-review-boards-irbs-and-protection-human-subjects-clinical-trials*
4. *https://www.nnlm.gov/guides/data-glossary/qualitative-data*
5. *https://www.sciencedirect.com/topics/mathematics/standard-deviation -*

2024 KEY FINDINGS IN CURE SMA DATABASES

51% of the SMA community were adults (page 13)

The rates of osteoporosis and chronic pain were higher in the SMA community than the U.S. general population (page 43)

Since 2016, the proportions of individuals living with SMA Type 1 and Unknown SMA Type continued to rise (page 18)

Among individuals diagnosed with hip dysplasia, we observed an increase in the proportion of those with Type 1 SMA after 2017 (page 37)

43% of individuals working part-time reported doing so to stay below an income threshold (page 20)

72% of individuals diagnosed in 2024 were identified by newborn or prenatal screening (page 23)

The average time from diagnosis to first treatment dropped to 18 days for individuals diagnosed in 2024 (page 30)

The average age at first treatment for all individuals diagnosed in 2024 was 23 days (page 25)

The mortality rate of SMA has dropped nearly 80% in the last 10 years (page 44)

Delays in time from diagnosis to treatment for individuals that were diagnosed symptomatically and diagnosed at older ages still remain (page 22)

Pediatric individuals with SMA reported traveling greater distances to their SMA care than adults (page 36)

Approximately half of caregivers reported travel expenses affected them financially

Approximately 75% of the population have been treated with at least one FDA-approved treatment (page 24)

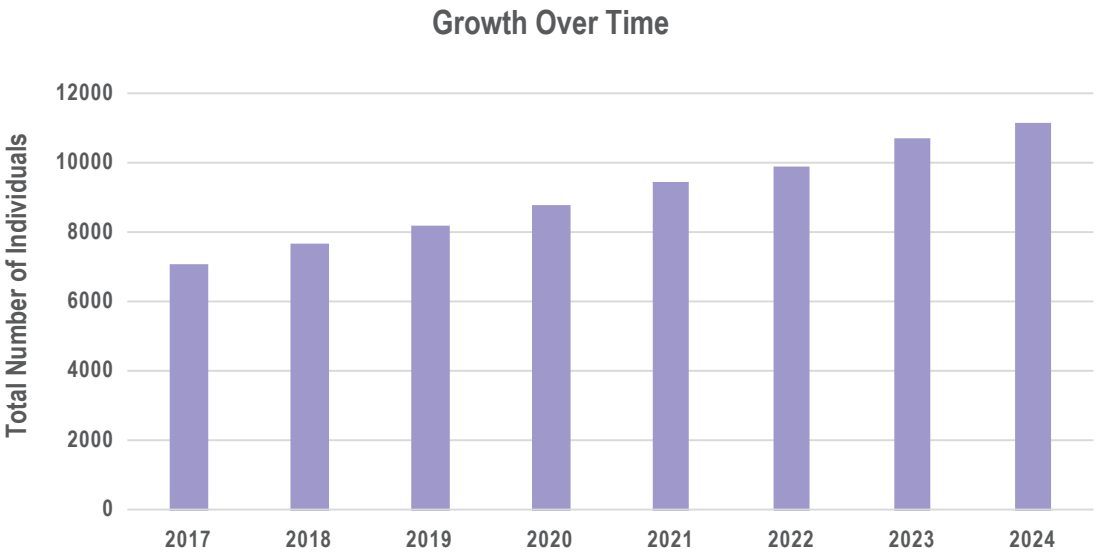
52% of treated individuals have reported receiving an insurance denial for their SMA treatment(s) (page 31)

45% of adults who needed mental health services reported not knowing where to access the needed services (page 39)

Note: connected bubbles on this page are used to highlight some of the unmet needs within the SMA community.

CURE SMA MEMBERSHIP DATABASE

The Cure SMA membership database constitutes one of the largest patient-reported data repositories for people living with SMA worldwide. It was launched in 1996. Since then, an average of 40 newly diagnosed individuals have contacted Cure SMA each month to share information. Patient-reported data captures real world patient experiences and can represent a broad spectrum of patients. Research projects use de-identified patient data from the membership database and receive Institutional Review Board (IRB) approval prior to project start.



DATA TYPE:
Patient- and caregiver-provided data

YEARS:
1996-Present

SIZE:
>11,100 individuals diagnosed with SMA

GEOGRAPHIC SPAN:
International

CORE DATA FIELDS:
Demographics, diagnosis date, SMA type, number of *SMN2* copies, and deceased date (if applicable)

COMMUNITY UPDATE SURVEY (CUS)

Since 2017, Cure SMA has conducted an annual online Community Update Survey to capture longitudinal data from the patient's perspective and develop additional data that can support assessment of SMA disease impact. Survey participants include both new and existing Cure SMA members.

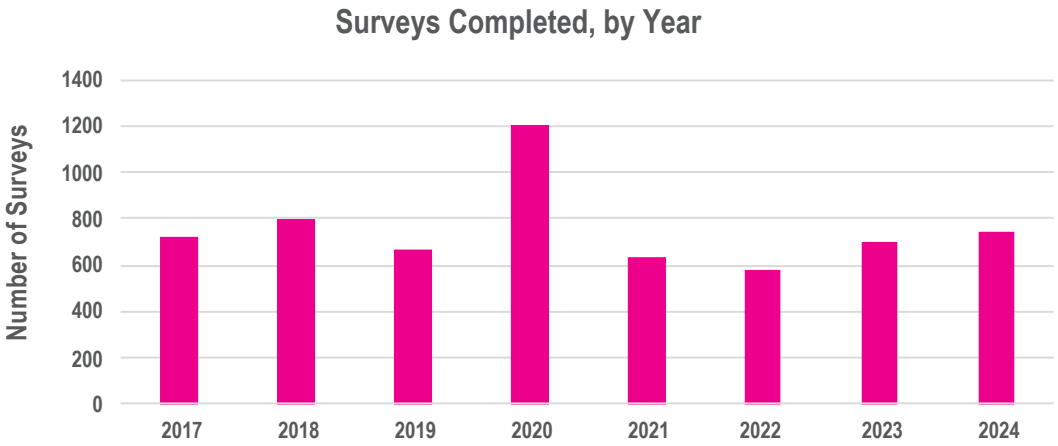
DATA TYPE:
Survey

YEARS:
2017-2024

SIZE: 2,864

GEOGRAPHIC SPAN:
International

CORE DATA FIELDS:
Demographics, surgeries, hospitalizations, motor function, treatment, and quality of life

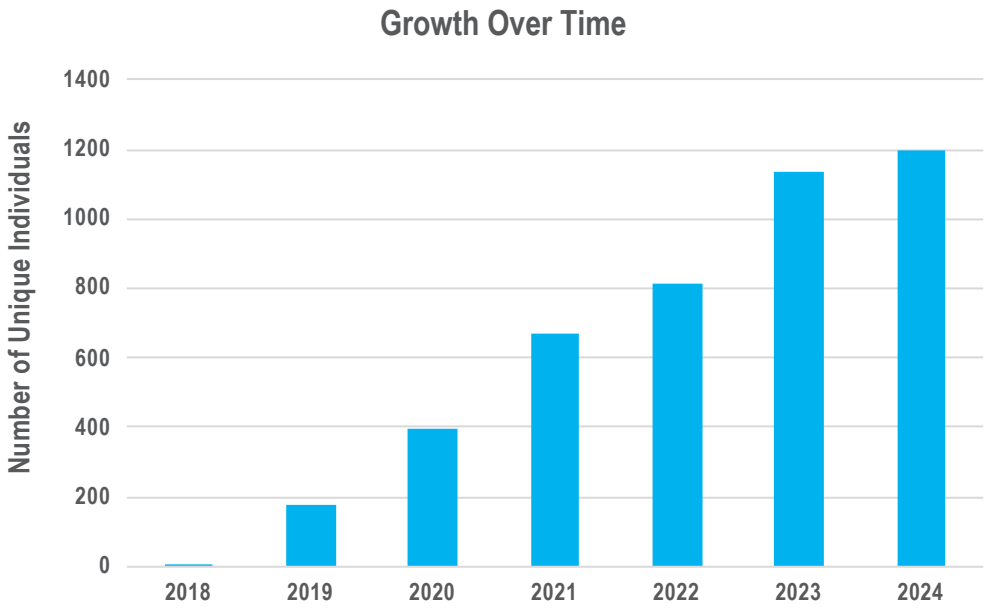


**The Community Update Survey had the largest response rate in 2020 at the start of the COVID-19 pandemic*

CURE SMA DATABASES

CLINICAL DATA REGISTRY (CDR)

The CDR is an Institutional Review Board (IRB) governed database for individuals with SMA comprised of electronic medical records (EMR) sourced data from Care Center Network sites and clinician-entered electronic case report forms to gather additional information that is not easily found in the EMR. The registry was launched in October 2018.



DATA TYPE: Electronic medical records and electronic case report form

YEARS: 2018-2024

SIZE: ~1200 individuals

GEOGRAPHIC SPAN: United States*

CORE DATA FIELDS: Demographics, genetic information, motor function, diagnoses, medications, procedures, laboratory studies, vitals signs, etc.

**Data from a few international patients who receive care in the U.S are included in this registry.*

NEWBORN SCREENING REGISTRY DATABASE (NBSR)

The NBSR is a caregiver-reported data repository comprised of individuals with SMA identified via newborn screening. This database was launched in 2019 and allows for the collection of real world data that can be used to track outcomes in this population.

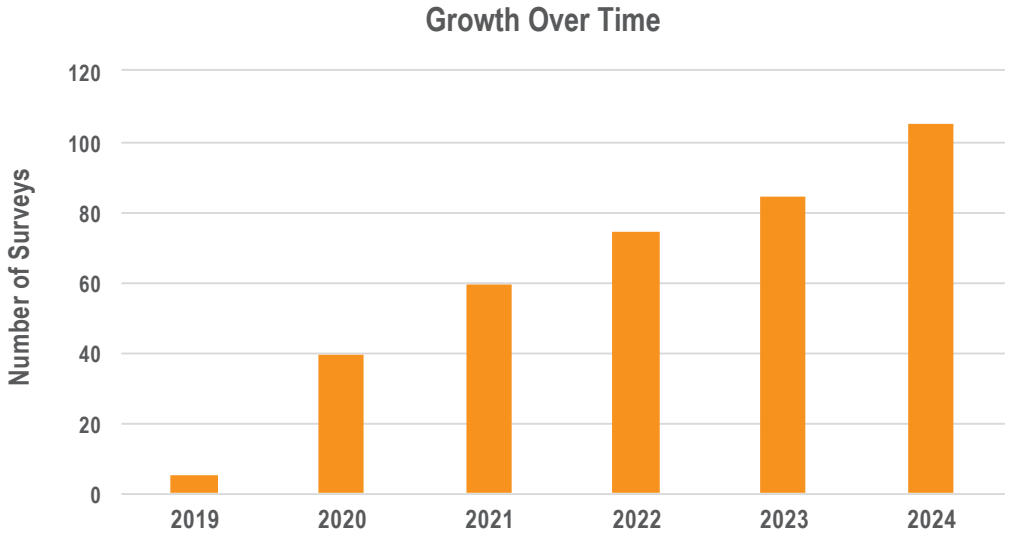
DATA TYPE: Survey

YEARS: 2019-2024

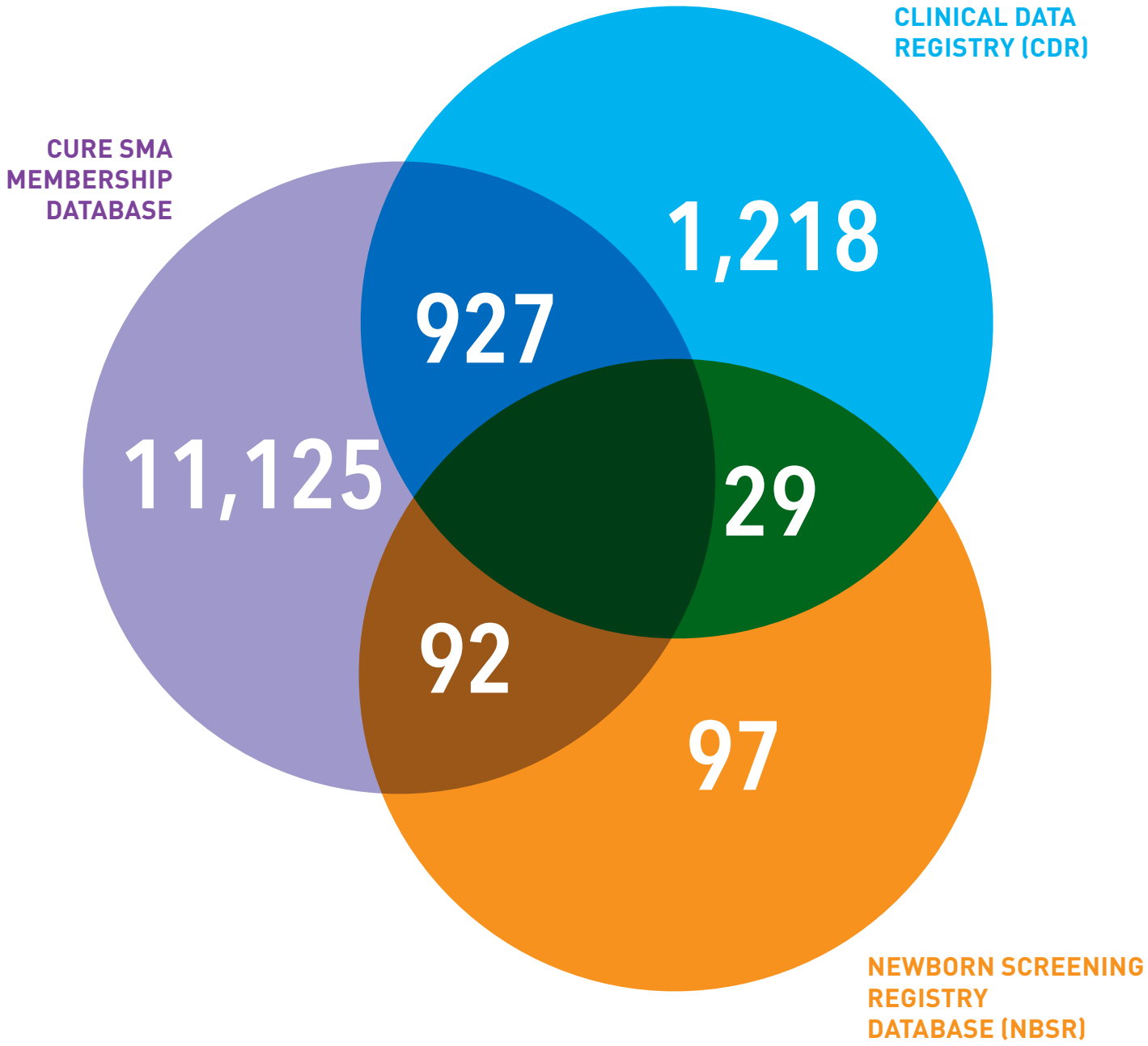
SIZE: >95 individuals

GEOGRAPHIC SPAN: United States

CORE DATA FIELDS: Demographics, *SMN2* copy number, diagnosis date, treatment, treatment date, motor function milestones



UNIQUE INDIVIDUALS ACROSS CURE SMA DATA SOURCES

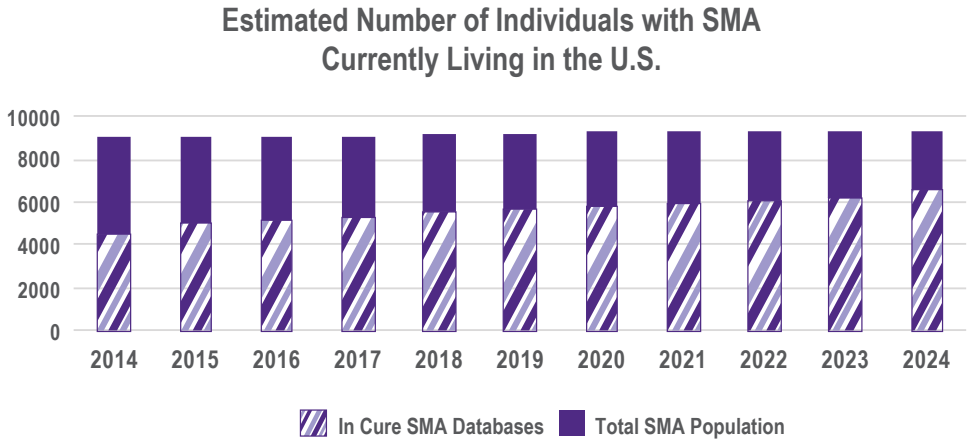


Within the Cure SMA databases, there are currently 11,125 unique individuals within the Membership database, 1,218 unique individuals in the CDR, and 97 unique individuals within the NBSR. Databases are not mutually exclusive, and some individuals may participate in more than one database. There are currently 92 individuals in both the NBSR and Membership database, 29 individuals in both the NBSR and CDR, and 927 individuals in both the CDR and Membership database. Across all data sources, there are 988 unique individuals who are in two or more databases.

SMA MODEL

The "SMA Model" is not a database, but a model that was created by Cure SMA to estimate the prevalence, survival and characteristics of the population with SMA in the United States to gauge the coverage of the Cure SMA databases.

Cure SMA estimates there are currently 9,000-9,500 individuals with SMA currently living in the U.S.



Results from the model are based on the following inputs:

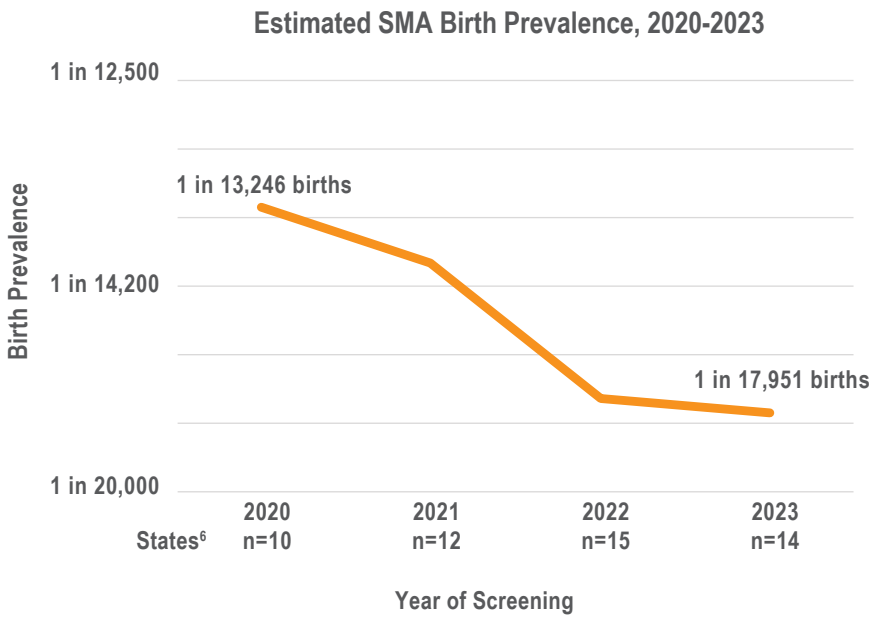
- Annual births from the National Vital Statistics System (NVSS)[1]
- Historical ("pre-SMA NBS") incidence rate of 1 in 11,000 decreasing to a current incidence rate of 1 in 15,000 from U.S. Public Health State labs[2]
- A decreasing mortality rate as reported by the CDC WONDER database[3] and the Cure SMA data, see page 44.
- SMA subtype incidence of 50% for Type 1, 35% for Type 2, and 15% for Type 3/4[4]
- Race and ethnicity SMA carrier rates described by Sugarman et al.[5]

Cure SMA's databases cover about 70% of the current U.S. SMA population

DATA FROM U.S. PUBLIC HEALTH STATE LABS

Cure SMA collects data from U.S. public health state labs for data on the number of individuals screened and the number of individuals that screened positive for SMA through statewide newborn screening (NBS). This data helps to understand the number of individuals diagnosed with SMA in the United States each year (birth prevalence). As of December 2023, more than 5 million infants have been screened for SMA with an estimated 362 infants screened positive and SMA diagnosed confirmed.[6]

The overall estimated birth prevalence of SMA is approximately 1 in 16,200 births.[6]

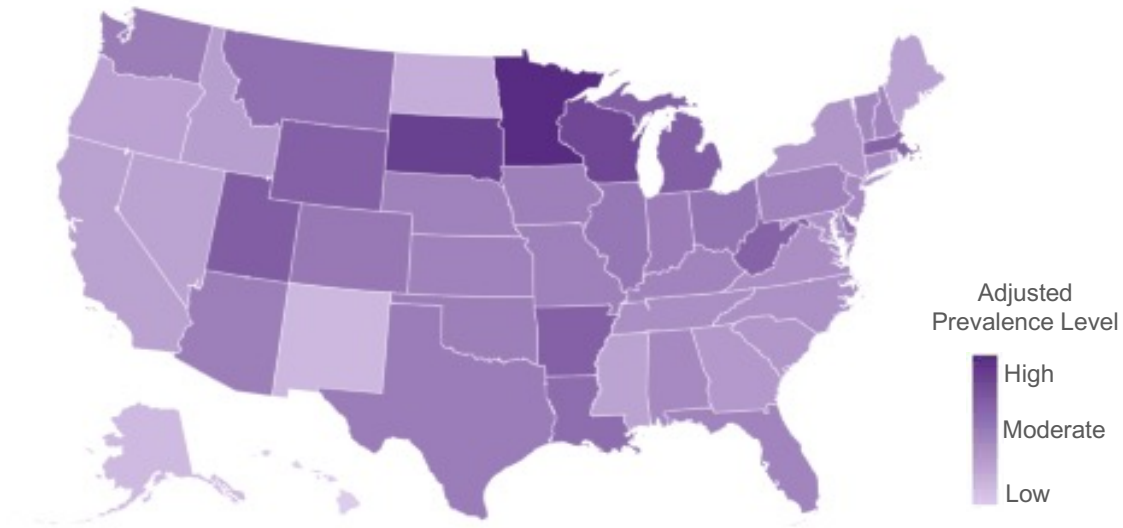


1. https://www.cdc.gov/nchs/nvss/births.htm
2. Belter L, Taylor JL, Jorgensen E, et al. Newborn Screening and Birth Prevalence for Spinal Muscular Atrophy in the US. JAMA Pediatr. 2024;178(9):946–949.
3. https://wonder.cdc.gov/
4. Verhaart IEC, Robertson A, Wilson IJ, Aartsma-Rus A, Cameron S, Jones CC, Cook SF, Lochmüller H. Prevalence, incidence and carrier frequency of 5q-linked spinal muscular atrophy - a literature review. Orphanet J Rare Dis. 2017 Jul 4;12(1):124.
5. Sugarman EA, Nagan N, Zhu H, Akmaev VR, Zhou Z, Rohlfs EM, et al. Pan-ethnic carrier screening and prenatal diagnosis for spinal muscular atrophy: clinical laboratory analysis of >72,400 specimens. Eur J Hum Genet. 2012 Jan;20(1):27-32. .
6. Data is from 17 states who provided data on newborn screening for SMA across years 2018-2023. Data submission varied by year.

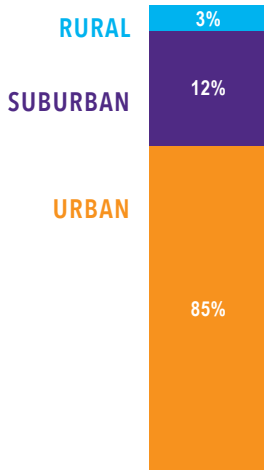
U.S. DISTRIBUTION

Cure SMA currently has 37 chapters throughout the United States and Puerto Rico, providing community and support for families and individuals living with SMA.

U.S. Distribution of Individuals Living with SMA, Adjusted for State Population (n=6,294)



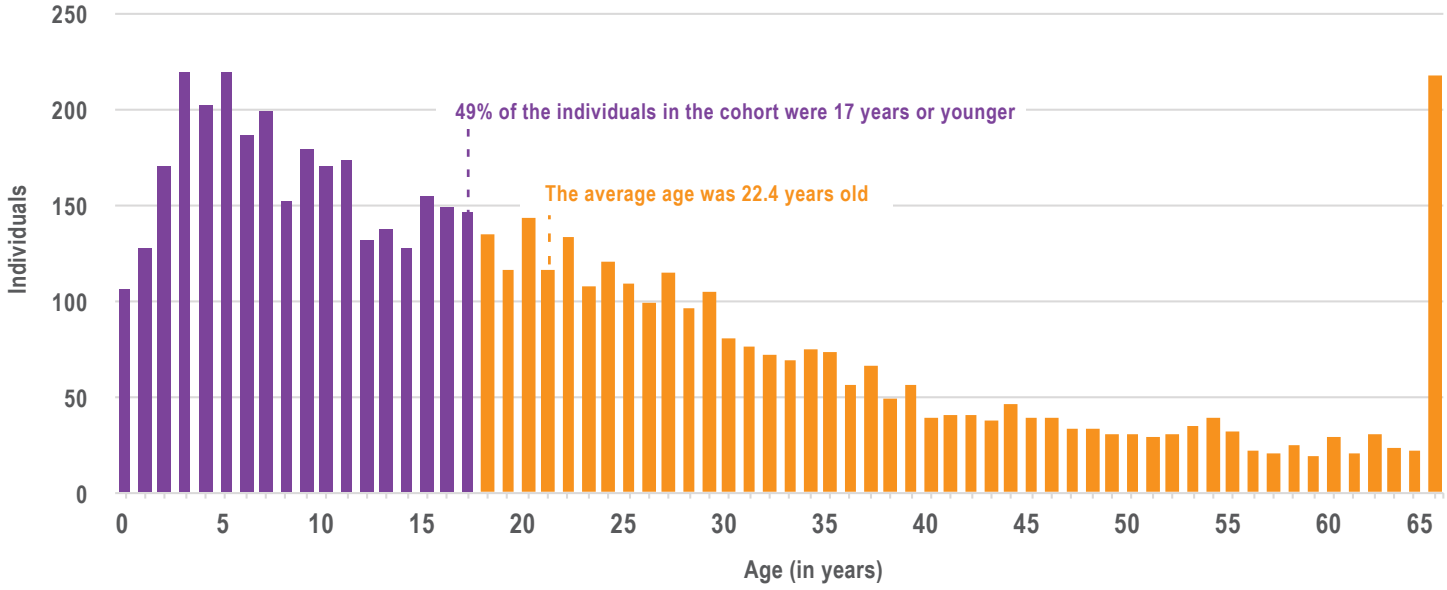
Urban/Rural Residence[1,2] (n=6,255)



Minnesota, South Dakota, Utah, Wisconsin, and Michigan had the highest proportion of individuals living with SMA when adjusted for the state population. Additionally, majority of the population resides in an urban area (85%), similar to the general U.S. population (80.7%)

AGE

Age Distribution of Individuals Living with SMA in Cure SMA Data in 2024 (n=6,000)



Average Age: 22.4 years | Standard Deviation: 18.2 years | Median Age: 18 years

Cure SMA Data Sources: Membership data, CUS, CDR, and NBSR data combined. Individuals participating in multiple sources were de-duplicated.

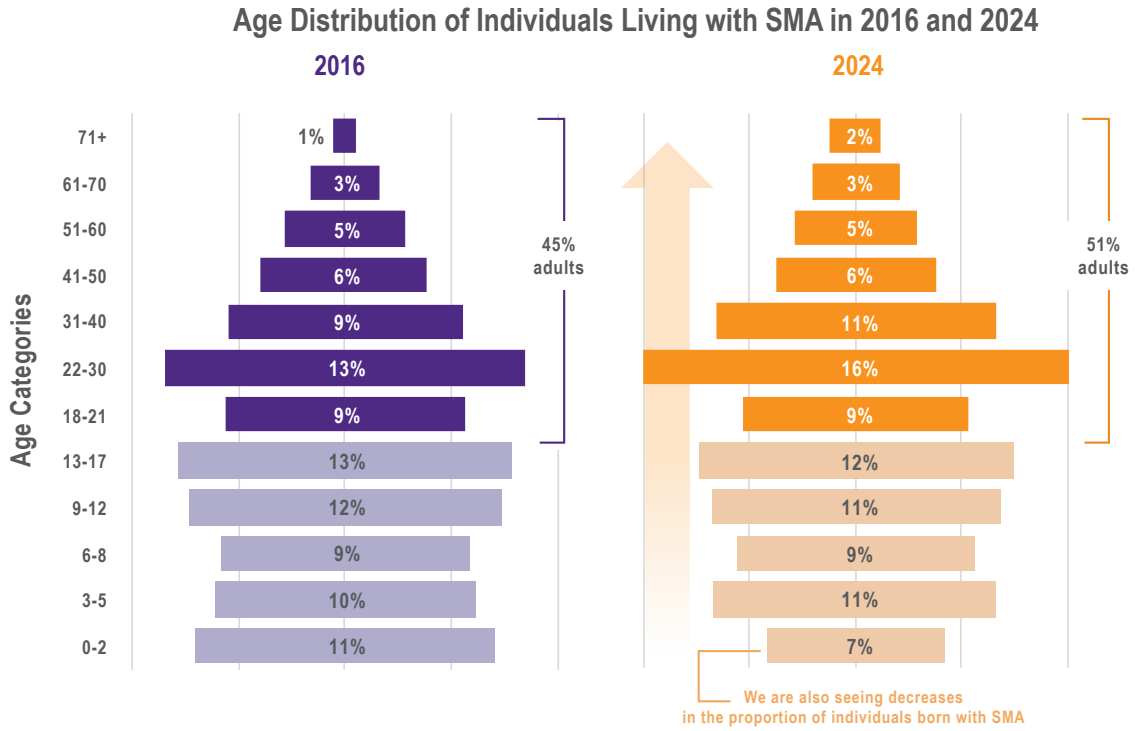
1. Urban/rural residence was determined using zip-code information (if available) and classifications determined by the U.S. Department of Agriculture (USDA)
2. *Rural-urban commuting area codes.* USDA ERS - Rural-Urban Commuting Area Codes. (2023, September 23). https://www.ers.usda.gov/data-products/rural-urban-commuting-area-codes/

Analysis Notes:

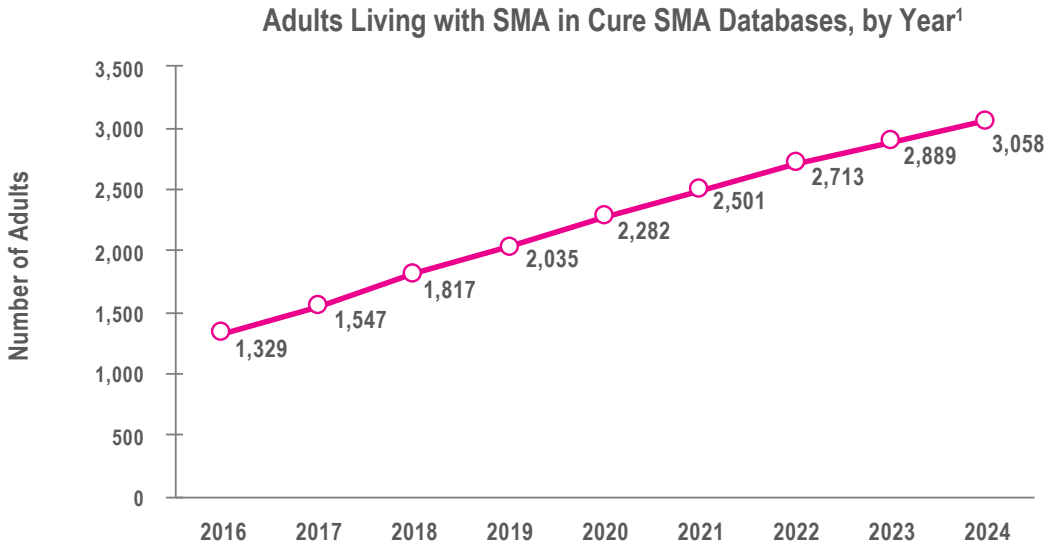
- If state of residence was reported in multiple databases, CDR listing was prioritized
- All graphics include individuals that were alive as of December 31, 2024

AGE TRENDS

The SMA landscape is changing, and we are seeing an increase in the individuals with SMA who are living into adulthood in our databases.



The number of adults living with SMA in the Cure SMA databases have also steadily increased each year.



This increase of adults living with SMA in our databases is important because we strive to ensure our databases are reflective of the SMA community.

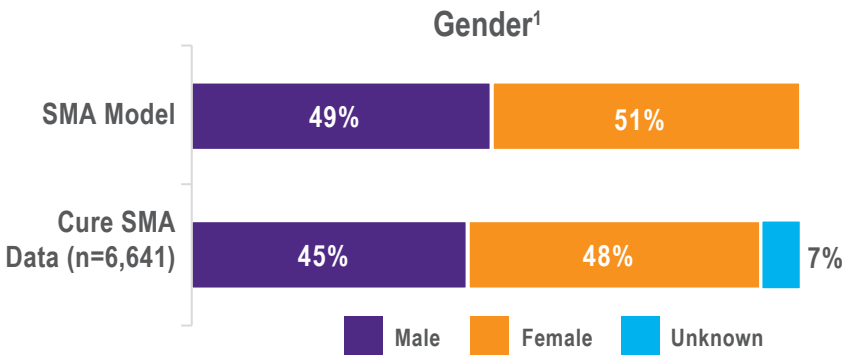
Cure SMA Data Sources: Membership data, CUS, CDR, and NBSR data combined. Individuals participating in multiple sources were de-duplicated.

1. Individuals without a reported birthdate, death date (if applicable) were not included. If an individual was missing date of first contact to Cure SMA, the date that a contact record was created was used, which may not always reflect the date of first contact.

Analysis Notes:

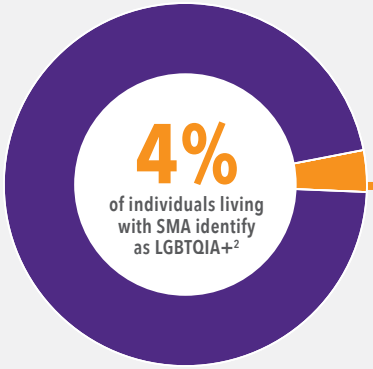
- Graphics include individuals that were alive as of 12/31 of the year specified.

GENDER / GENDER IDENTITY



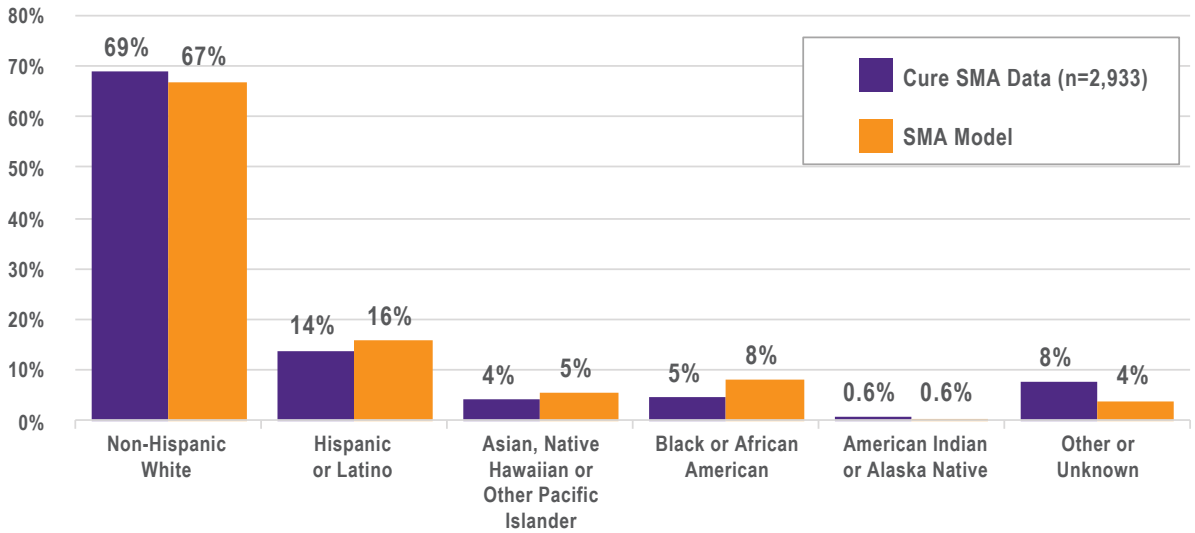
1.2% of individuals living with SMA identify with a gender that is different than their birth sex

PATIENT/CAREGIVER REPORTED



- Transgender Female
- Non-binary
- Asexual Biromantic
- Pansexual
- Demisexual
- Genderqueer
- Gay
- Queer
- Lesbian
- Transgender Male
- Fictoromantic
- Asexual
- Homosexual

RACE/ETHNICITY CATEGORIES



Cure SMA Data Sources: Membership data, CUS, CDR, and NBSR data combined. Individuals participating in multiple sources were de-duplicated.

1. The CDR collects gender used for administrative purposes (ie. Insurance) that may or may not align with a person's gender in all context.
2. LGBTQIA+ population was determined by those that had a gender listed as "Non-binary or genderqueer", a gender identity that was different from their sex assigned at birth, or a sexual orientation outside of straight or heterosexual

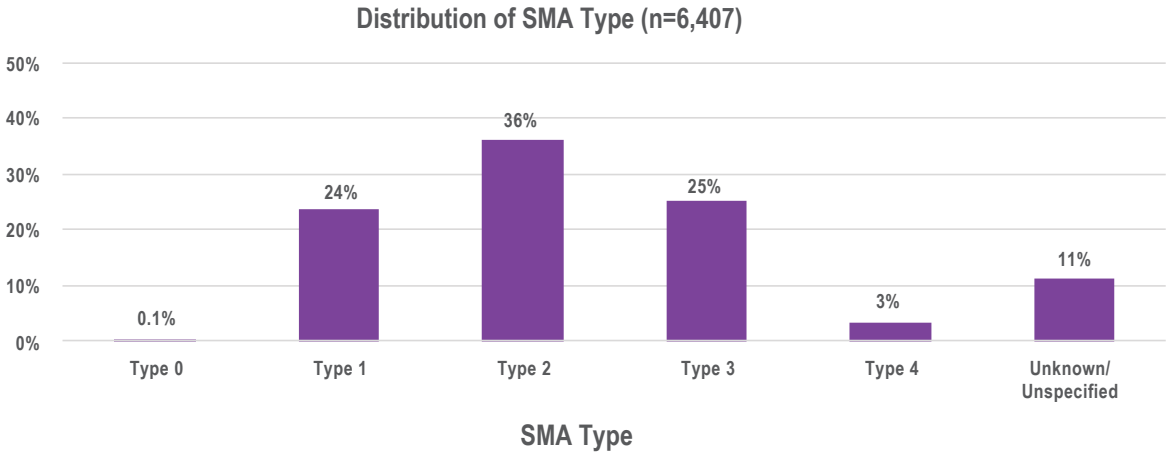
Analysis Notes:

- All graphics include individuals that were alive as of December 31, 2024
- If gender or race/ethnicity was provided for the same individual in multiple databases, the self-reported data was prioritized.

PREVALENCE OF SMA TYPE AND *SMN2* COPY NUMBER:

SMA TYPE:

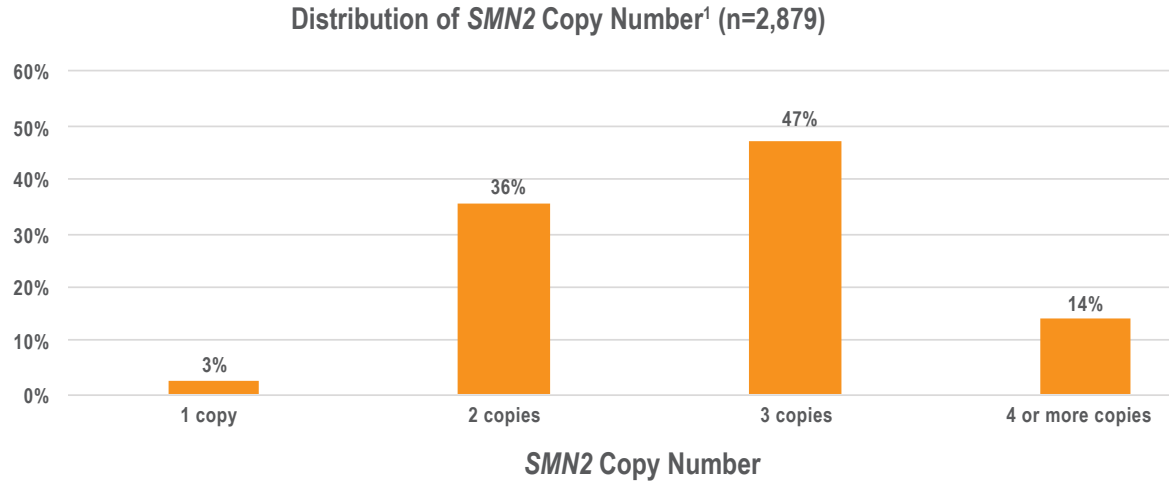
Historically, SMA was characterized by a classification system for describing age of symptom onset and maximum motor function achieved. This classification divides SMA into five types: Types 0, 1, 2, 3, and 4.



The largest proportion of individuals living with SMA in Cure SMA data sources have Type 2 SMA.

SMN2 COPY NUMBER:

***SMN2* is an inefficient variant of the *SMN1* gene. This means that *SMN2* cannot fully make up for the mutated *SMN1* gene. The number of *SMN2* genes can vary from person to person, and individuals with more *SMN2* copies usually have a less severe form of SMA than those with fewer copies. However, there are exceptions.**



The largest proportion of individuals living with SMA in Cure SMA data sources have 3 copies of *SMN2*.

Cure SMA Data Sources: Membership data, CUS, CDR, and NBSR data combined. Individuals participating in multiple sources were de-duplicated.

1. Individuals with unknown *SMN2* copy number were not included in this graphic

Analysis Notes:

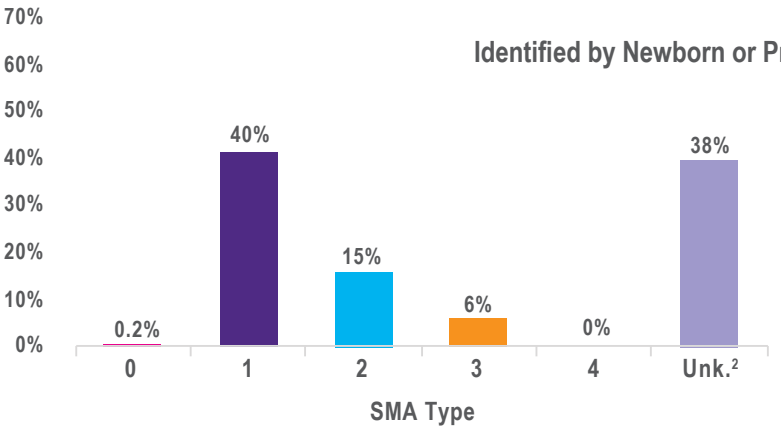
- All graphics include individuals that were alive as of December 31, 2024

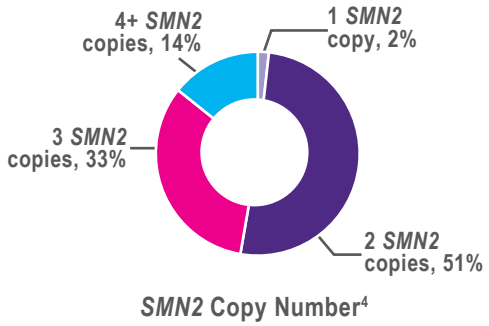
PREVALENCE OF SMA TYPE AND *SMN2* COPY NUMBER:

The prevalence of SMA Type and *SMN2* copy number vary by current age and method of diagnosis

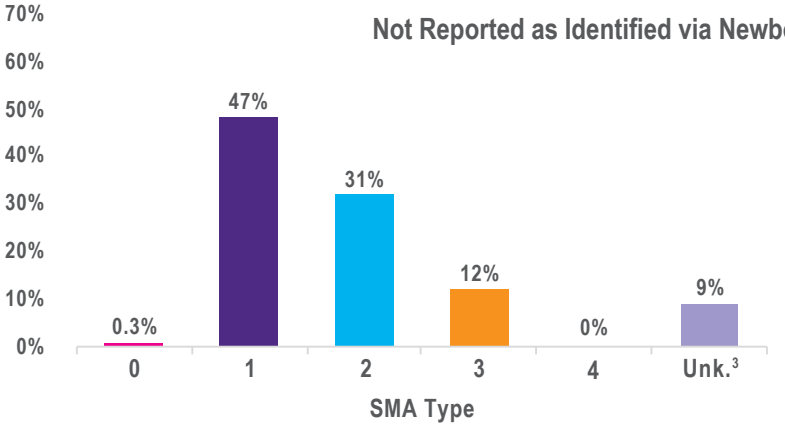
Children Living with SMA (Currently 0-12 Years Old)

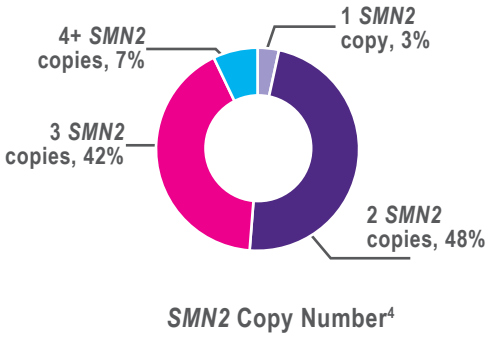
Identified by Newborn or Prenatal Screening[1], n=628



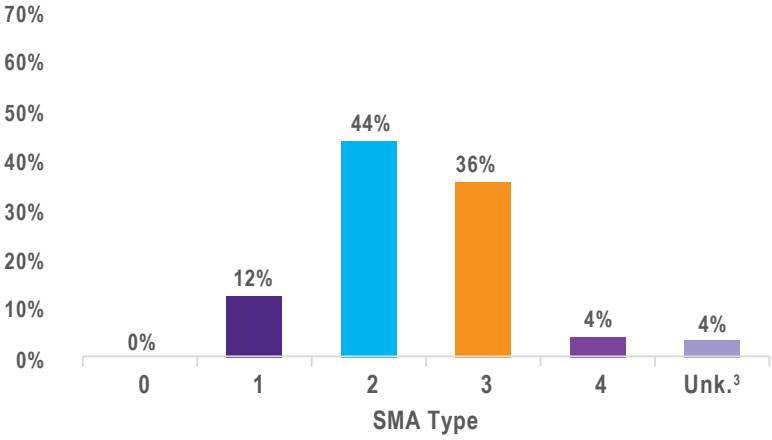


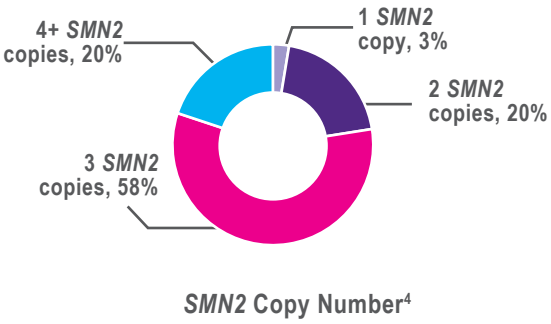
Not Reported as Identified via Newborn or Prenatal Screening, n=1,589





Teens and Adults Living with SMA (Currently 13+ Years Old), n=3,770





Cure SMA Data Sources: Membership data, CUS, CDR, and NBSR data combined. Individuals participating in multiple sources were de-duplicated.

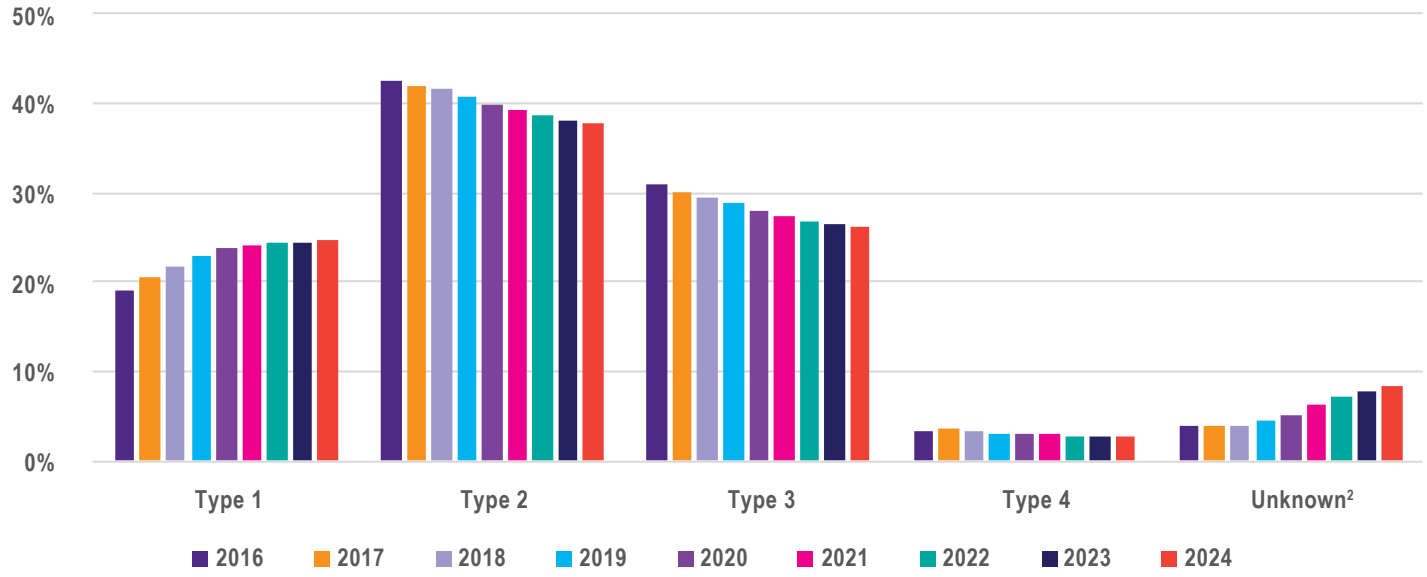
1. Newborn screening status is patient-reported. Graph restricted to individuals diagnosed before 2 years of age
2. Unknown SMA type includes the following: Unknown, Unspecified, Pre-symptomatic, and Unknown because diagnosed < 6 months of age before symptom onset. If does not include missing values.
3. Unknown SMA type includes the following: Unknown, Unspecified. If does not include missing values.
4. Individuals with unknown SMN2 copy number were not included (children via screening, n=61; children not via screening, n=621; teens/adults, n=2448)

Analysis Notes:

- All graphics include individuals that were alive as of December 31, 2024

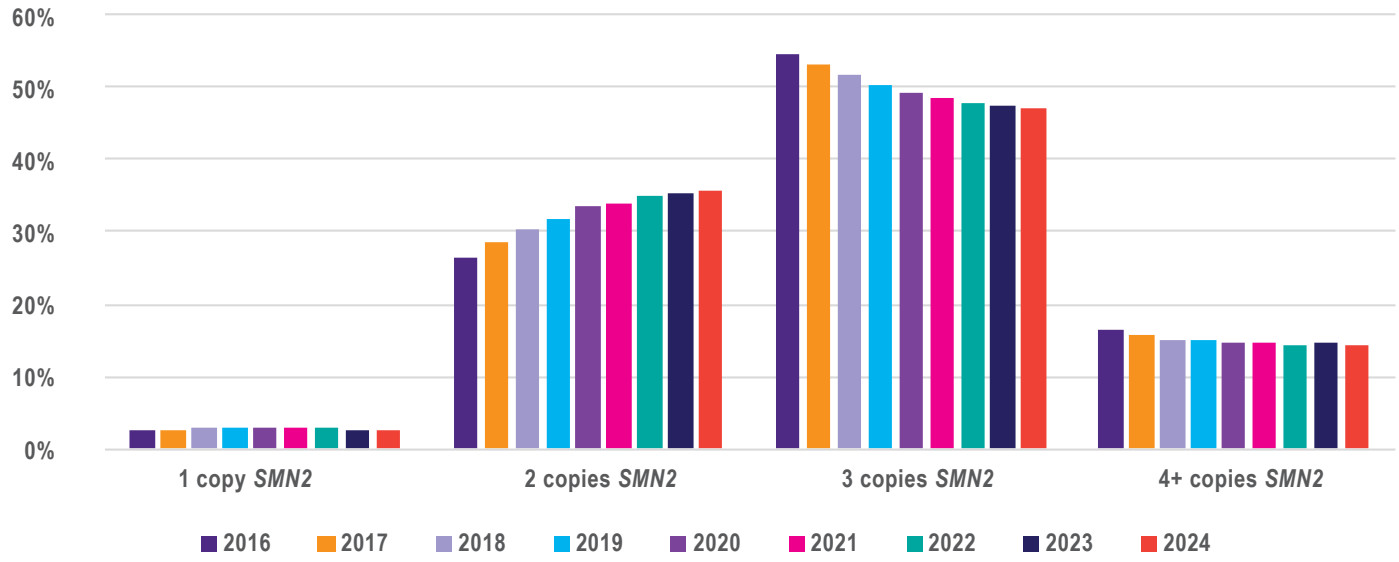
TRENDS IN OUR DATA: SMA TYPE & *SMN2* COPY NUMBER

Prevalence of SMA Type Over Time[1]



We are continuing to observe rising proportions of individuals with SMA Type 1 and Unknown SMA Type and individuals with 2 *SMN2* copies. While we didn't analyze what factors might be driving this change, an increase in survival of individuals due to the availability of treatments and diagnosis before symptom onset may be possible factors.

Prevalence of *SMN2* Copy Number Over Time[3]



Cure SMA Data Sources: Membership data, CUS, CDR, and NBSR data combined. Individuals participating in multiple sources were de-duplicated.

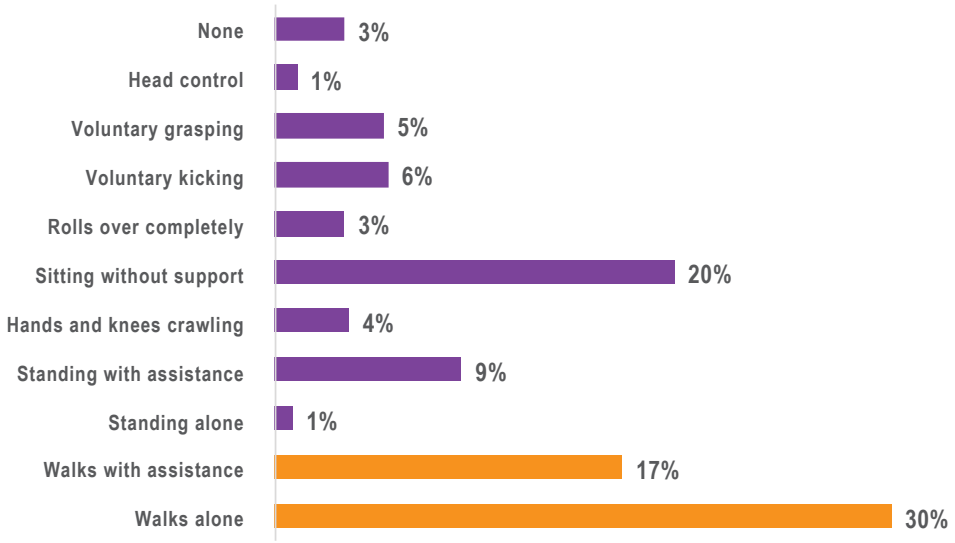
1. SMA Type 0 was not included in the graphic due to small sample size.
2. Unknown SMA type includes the following: Unknown, Unspecified, Pre-symptomatic, and Unknown because diagnosed < 6 months of age before symptom onset. If does not include missing values.
3. Individuals with unknown SMN2 copy number were not included in this graphic

Analysis Notes:

- All graphics include individuals that were alive as of December 31st of each specified year

MOTOR FUNCTION STATUS

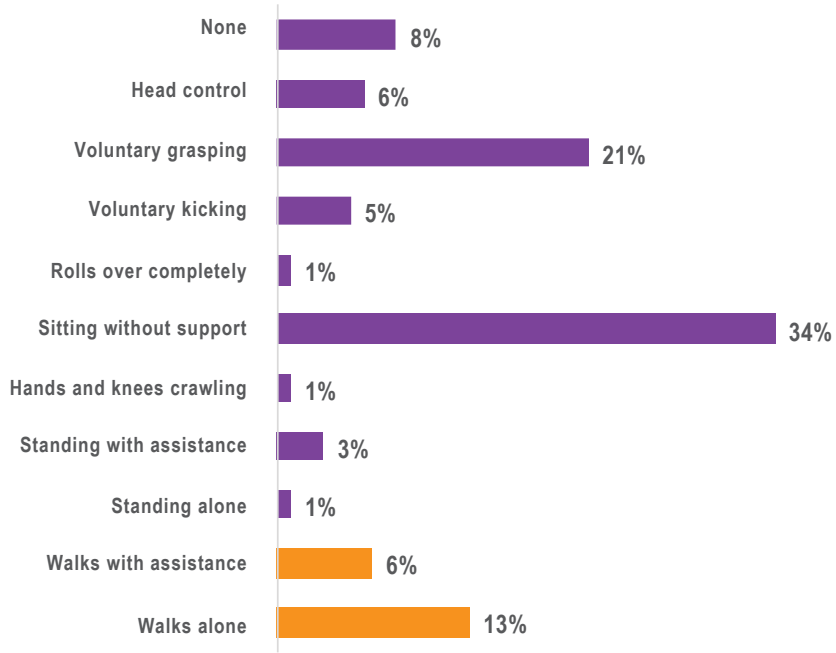
MAXIMUM CURRENT MOTOR FUNCTION OF CHILDREN[1]



Approximately 47% of children living with SMA are currently ambulatory while 19% of adults living with SMA are currently ambulatory.[3]

89% of adults report achieving new motor function is their greatest unmet need[4]

MAXIMUM CURRENT MOTOR FUNCTION OF ADULTS[2]



Cure SMA Data Sources: Membership data

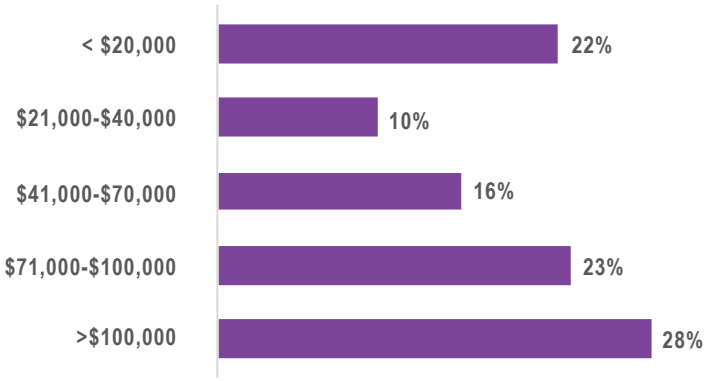
Analysis limited to those 2 years of age and older and excludes those with SMA Type 0 and Type 4 due to small sample size.

1. n=466
2. n=540
3. Ambulatory defined as those who can walk with assistance or walk alone
4. Data from the 2024 Community Update Survey, n=311

SOCIAL DETERMINANTS OF HEALTH AMONG ADULTS

PATIENT/CAREGIVER REPORTED

Household Income (n=307)

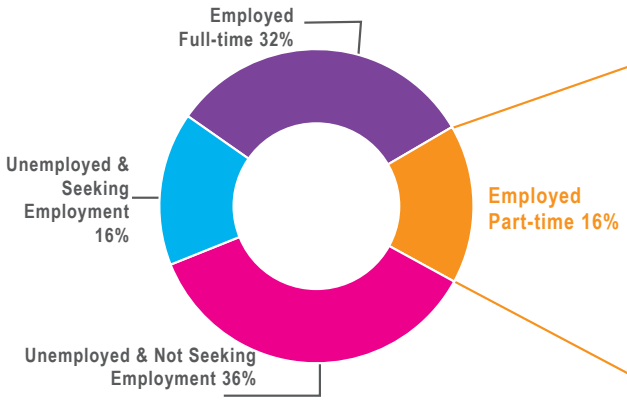


8% of adults with SMA reported that in the last 12 months they had to skip buying medications or going to doctor's appointments to save money[1]

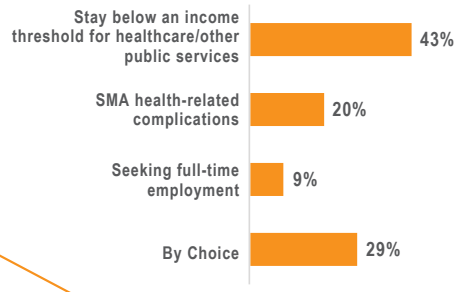
11% of adults with SMA reported in the last 12-months they worried that their food could run out before they got money to buy more[1]

When asked why an individual is working part-time, the proportion of those reporting the reason is to stay below an income threshold for healthcare/ other public services increased from 31% in 2023, to 43% in 2024, while those reporting the reason is due to SMA related health complications decreased from 37% in 2023 to 20% in 2024.

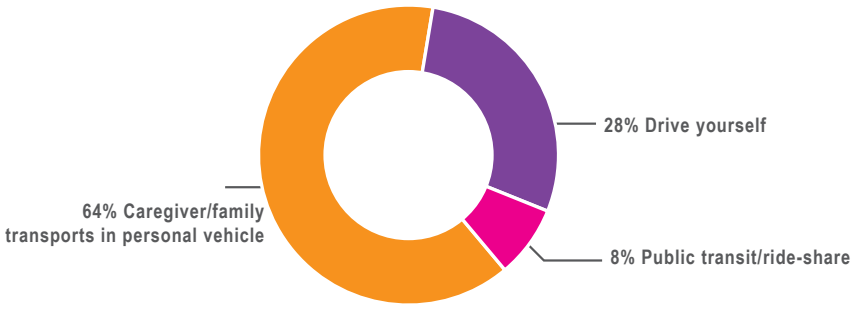
Employment Status (n=342)



Reasons for Working Part-Time (n=56)



Transportation: How the Cure SMA Community is Accessing Their Healthcare Services (n=295)



14% of adults with SMA reported that in the last 12 months lack of transportation kept them from medical appointments or getting their medications[2]

Transportation method was not shown to affect an individual's ability to access SMA treatment

Cure SMA Data Sources: 2024 CUS

1. Data is from adults who self-completed questions from the Montefiore SDOH Assessment
2. US Department of Health and Human Services. (n.d.). Reduce the proportion of people living in poverty - SDOH-01. Reduce the proportion of people living in poverty - SDOH-01 - Healthy People 2030. https://health.gov/healthypeople/objectives-and-data/browse-objectives/economic-stability/reduce-proportion-people-living-poverty-sdoh-01

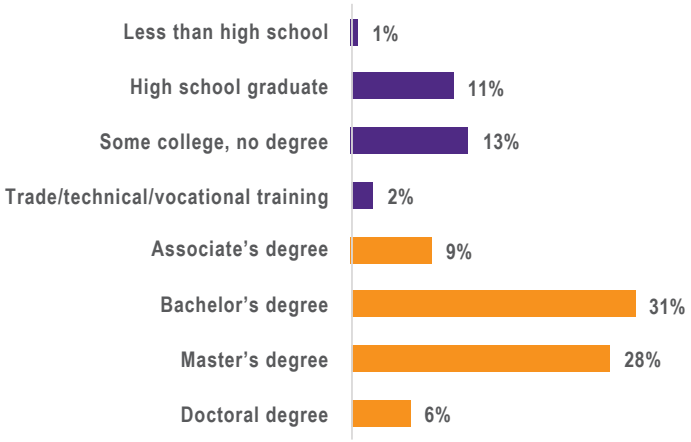
Analysis Notes:

- All graphics include individuals that were 18 years of age and older and living when completing the 2024 CUS

PATIENT/CAREGIVER REPORTED

SOCIAL DETERMINANTS OF HEALTH AMONG ADULTS

Highest Level of Education Completed Among Adults 25 Years and Older (n=301)



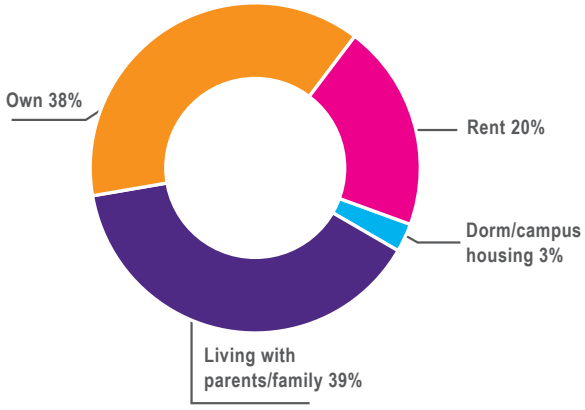
73% of adults with SMA aged 25 years and older have gone on to complete some form of higher education. This is higher than the U.S. Census estimate of 44.9%.[1]

5% of adults with SMA reported that in the past 12-months their electric, gas, oil, or water company has threatened to shut off services to their home.[2]

3% of adults with SMA reported that they are worried they may not have a safe or stable place to live in the next 2 months (i.e. eviction, being kicked out, homelessness).[2]

3% of adults with SMA reported that they are worried the place they are currently living is making them sick (i.e. mold, bugs/rodents, water leaks, not enough heat).[2]

Housing: Where the Cure SMA Community is Living (n=321)



Safety and Well-Being[2]

Adults with SMA who completed the 2024 CUS themselves were asked a series of questions on their health and well-being using the Montefiore SDOH assessment. The following are self-reported statistics on the safety and well-being among adults with SMA.

5% of adults with SMA reported needing legal assistance

14% of adults with SMA reported that it is hard to get along with a partner, spouse, or family members

3% of adults with SMA reported that someone in their life hurts, threatens, or frightens them, or makes them feel unsafe

If you are feeling unsafe at home, 24/7 confidential help can be reached by calling 1-800-799-SAFE (7233) or 911. You are not alone.

Cure SMA Data Sources: 2024 CUS

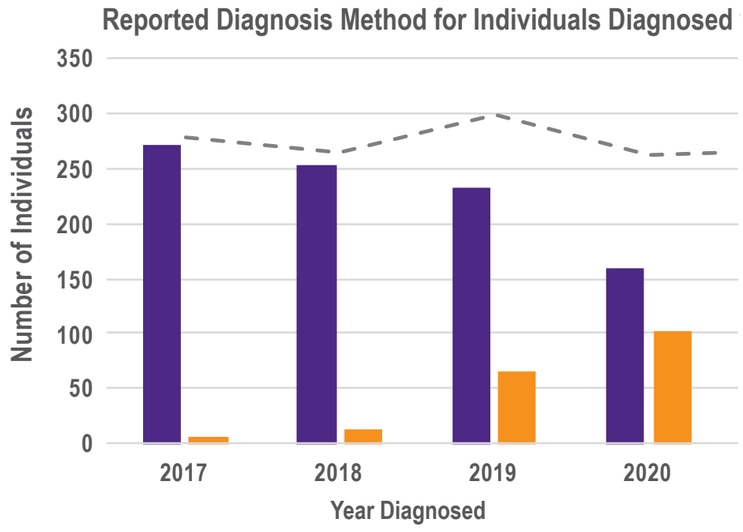
1. https://data.census.gov/table/ACSST1Y2023.S1501?q=Educational%20Attainment. Accessed 2/18/2025
2. Data is from adults who self-completed questions from the Montefiore SDOH Assessment

Analysis Notes:

- All graphics include individuals that were 18 years of age or older and living when completing the 2024 CUS.
- Higher education was considered completion of as Associate's, Bachelor's, Master's, or Doctorate degree

DIAGNOSIS OF SMA

In the Cure SMA databases, we observed a decline in the number of individuals diagnosed with SMA in the last few years.

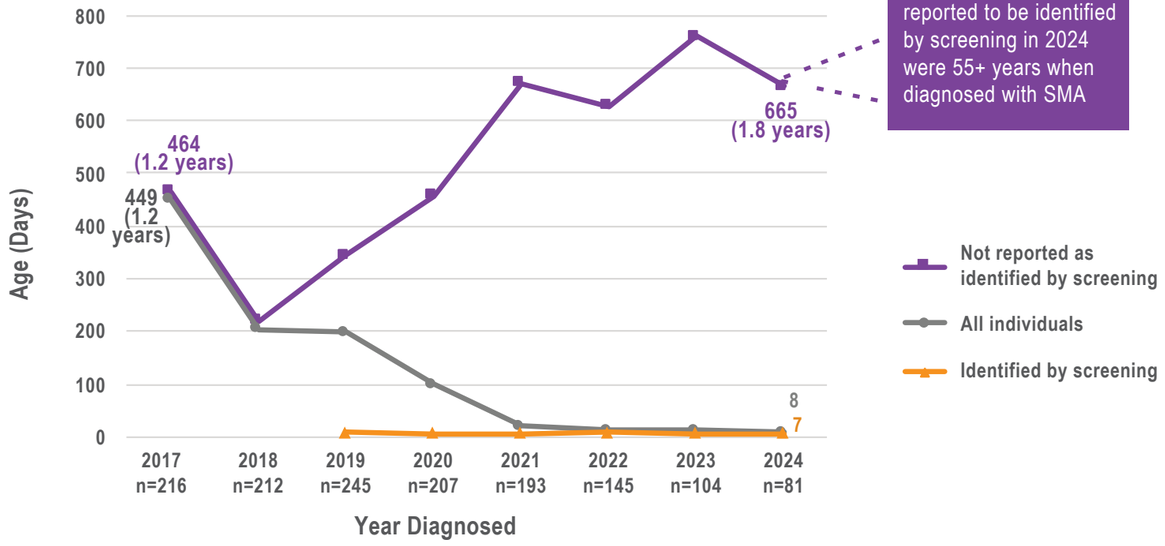


We are also seeing that the majority of diagnoses are identified via newborn or prenatal screening.

Overall, the average age at SMA diagnosis has decreased, which is driven by early diagnosis through screening. However, we continue to see different trends for individuals not identified by screening, and we anticipate the average age at SMA diagnosis may continue to increase as more individuals diagnosed symptomatically may be diagnosed at an older age due to later symptom onset.

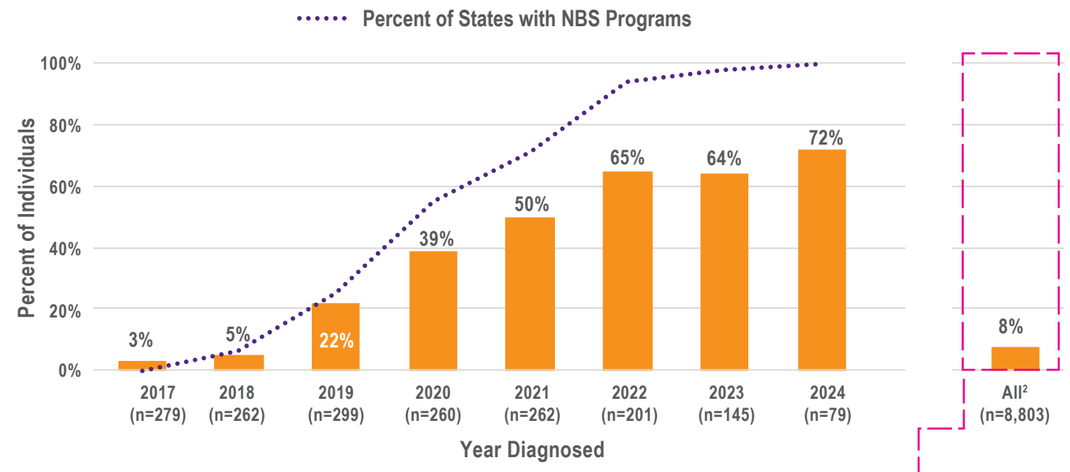
The number of U.S. states implementing newborn screening programs increased to 100% in 2024, and we have seen increases in the percentage of individuals identified via screening in our databases. We have also observed increases in prenatal screening. In the Community Update Survey, 21.6% of diagnoses reported between 2020-2024 were identified by prenatal screening. To provide best practice recommendations for SMA diagnosis, Cure SMA published in Neurology Clinical Practice, "Spinal muscular atrophy (SMA) Update in Best Practices: Recommendations for Diagnosis Considerations."

Median Age at SMA Diagnosis[1]



3 individuals (15%) not reported to be identified by screening in 2024 were 55+ years when diagnosed with SMA

Percent of Individuals Identified by SMA Newborn or Prenatal Screening, by Year of Diagnosis[1]



Although we are seeing an increase in diagnoses via screening in recent years, individuals identified by screening make up a very small part of the individuals in our databases

Cure SMA Data Sources: Membership data, CUS, CDR, and NBSR data combined. Individuals participating in multiple sources were de-duplicated.

1. Included individuals where the date of diagnosis was not reported to be estimated

Analysis Notes:

- Newborn and prenatal screening status are patient reported. If status was missing, it was assumed the individual was not identified by screening.
- Diagnosis date could not be more than 1 year prior to birthdate

Cure SMA Data Sources: Membership data, CUS, CDR, and NBSR data combined. Individuals participating in multiple sources were de-duplicated.

1. Sample includes anyone reported to be identified via newborn or prenatal screening
2. All individuals in Cure SMA databases

Analysis Notes:

- Newborn and prenatal screening status are patient reported. If status was missing, it was assumed the individual was not identified by screening.

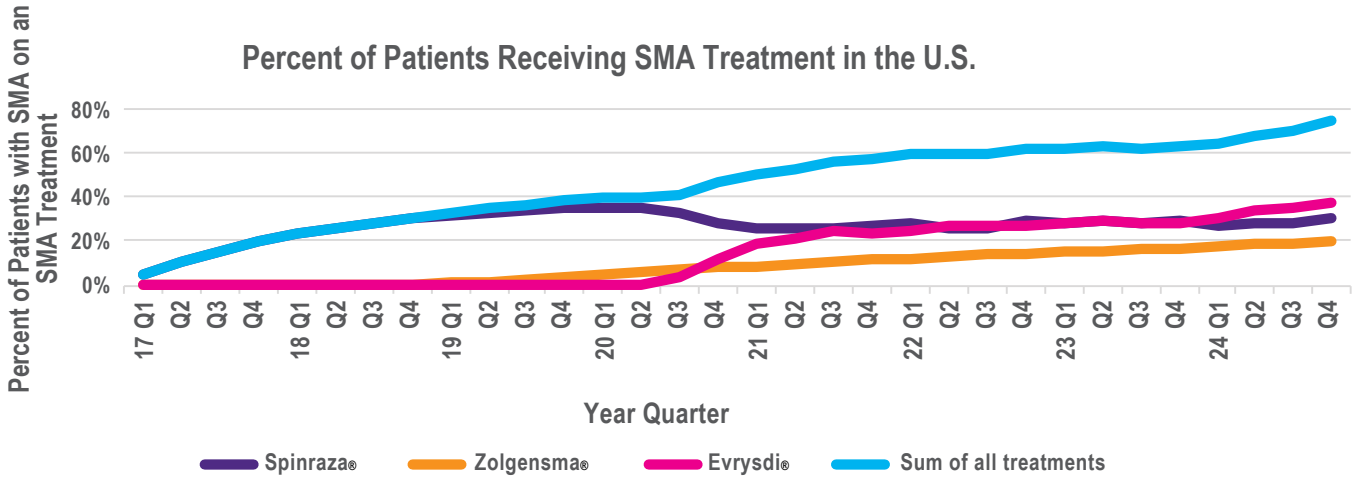
SMA TREATMENTS IN THE U.S.

SMA Treatment Pipeline[1]

Organization/Drug Name	Mechanism of Action	Age Coverage	SMA Type Coverage	Stage of Development			
				Phase 3	FDA Submission	FDA Approval Date	To Patients
Biogen - SPINRAZA®	Modulation of *SMN2*	All ages	All SMA types			12/23/2016	
Novartis - ZOLGENSMA® (IV)	Modulation of *SMN2*	Individuals < 2 years old	All SMA types			5/24/2019	
Roche-Genentech - EVRYSDI® (oral solution)	Modulation of *SMN2*	All ages	All SMA types			8/7/2020	
Roche-Genentech - EVRYSDI® (tablet formulation)	Modulation of *SMN2*					2/12/2025	
Biogen – Higher Dose 50/28mg nusinersen	Modulation of *SMN2*						
Scholar Rock – apitegromab	Muscle-directed therapy						
Novartis - OAV101 (IT)	Gene Therapy						
Roche-Genentech - GYM329	Muscle-directed therapy						

There are multiple clinical trials that continue to evaluate new therapies.
To learn more, please visit: https://www.curesma.org/cure-sma-clinical-trials/

Approximately 75% of individuals with SMA in the U.S. had received an FDA approved treatment as of Q4 2024[2-4]



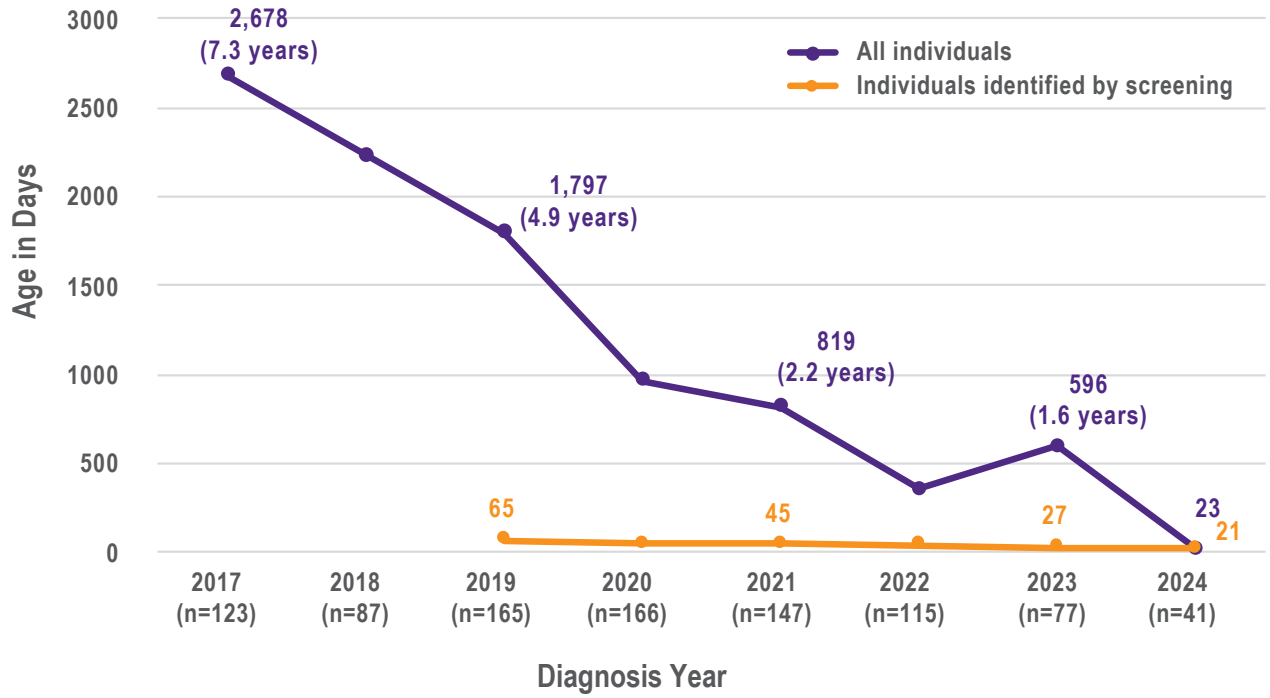
Data Sources: https://www.curesma.org/cure-sma-clinical-trials/ and published industry earnings reports from Biogen, Roche, and Novartis. These are estimates and may over or underrepresent treatment utilization

1. SMA Pipeline as of 3/1/2025
2. The sum of all treatments data is accounting for an estimated 15% of concurrent treatment use
3. Anyone treated with Zolgensma was categorized as "currently on treatment" for all quarters following treatment
4. Data here is presented by standard calendar quarters: January, February, and March (Q1) April, May, and June (Q2) July, August, and September (Q3) October, November, and December (Q4)

TREATMENT: OBSERVATIONS IN OUR DATA

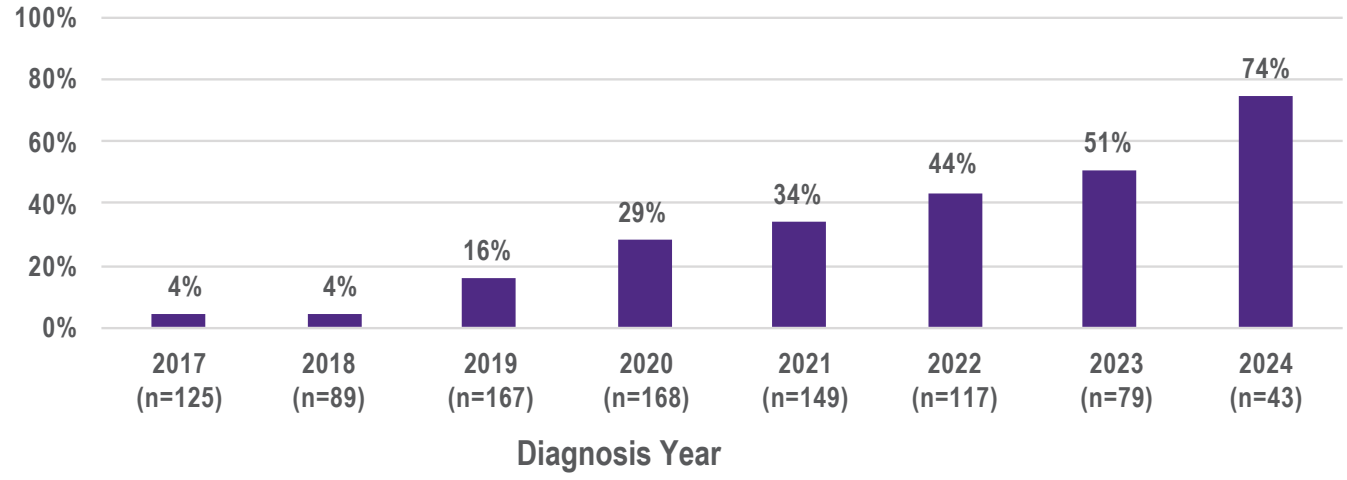
In Cure SMA databases, the average age at first treatment has decreased since 2017. For individuals diagnosed with SMA in 2024, the average age at first treatment was under 30 days.

Average Age at First SMA Treatment, by Year Diagnosed[1-2]



We are observing increased proportions of individuals with SMA treated by 30 days old, likely due to the increase of diagnoses via newborn or prenatal screening.

Percent of Individuals with SMA Receiving Treatment by 30 Days Old, by Year Diagnosed



Cure SMA Data Sources: Membership data, CUS, CDR, and NBSR data combined. Individuals participating in multiple sources were de-duplicated.

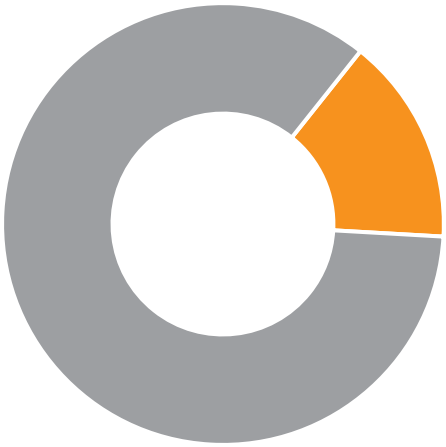
1. Newborn and prenatal screening status are patient reported. If status was missing, it was assumed the individual was not identified by screening.
2. The minimum and maximum values were removed for each reporting year.

Analysis Notes:

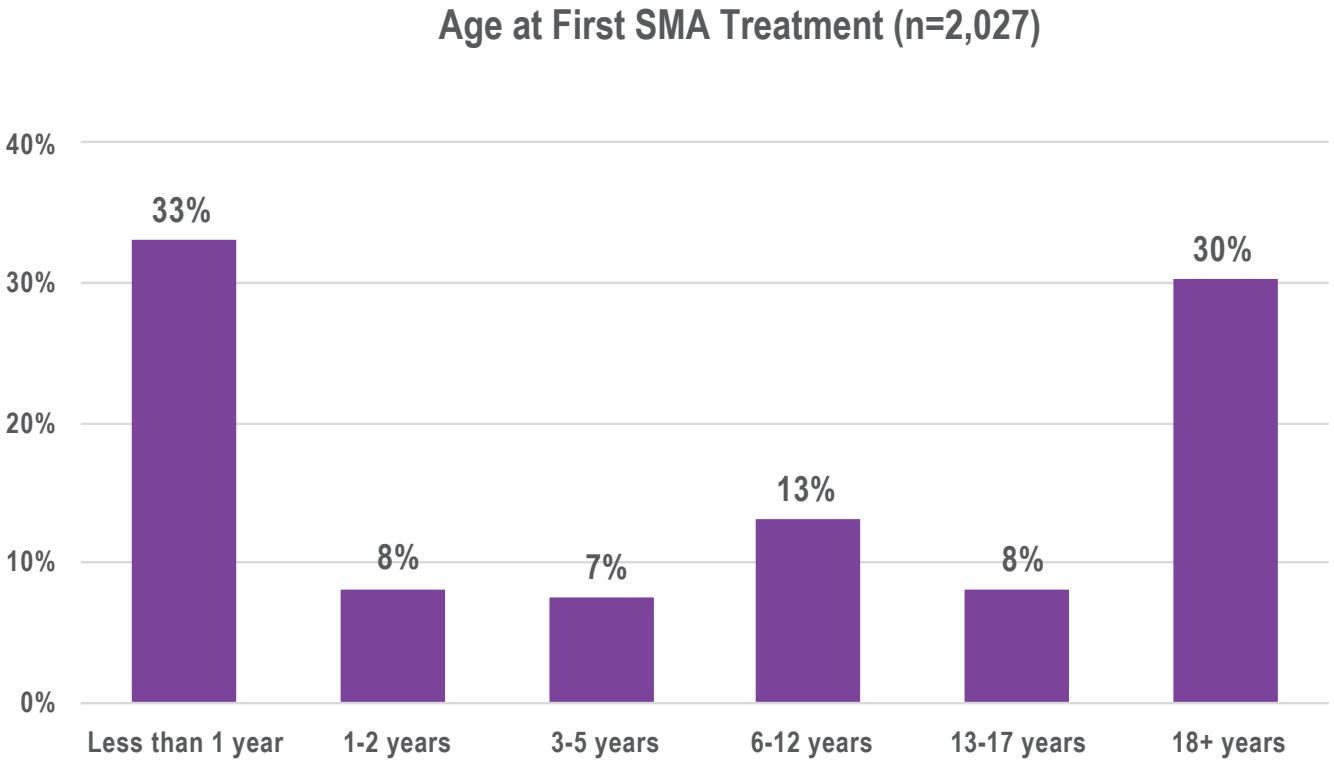
- Includes SMA treatments that were FDA approved as of 12/31/2024
- Graphs include individuals who have not participated in clinical trials and where all treatment dates are known

TREATMENT: OBSERVATIONS IN OUR DATA

Even though the average age at first treatment has decreased to less than 30 days in 2024 and we are seeing increases in the proportion of individuals receiving treatment before 30 days old, this does not reflect the treatment journey for most individuals with SMA in our databases.



15% of individuals with SMA in Cure SMA databases that did not participate in a clinical trial received treatment by 30 days old (n=1709)[1]



Cure SMA Data Sources: Membership data, CUS, CDR, and NBSR data combined. Individuals participating in multiple sources were de-duplicated.

1. Restricted to individuals where all treatment start dates were known.

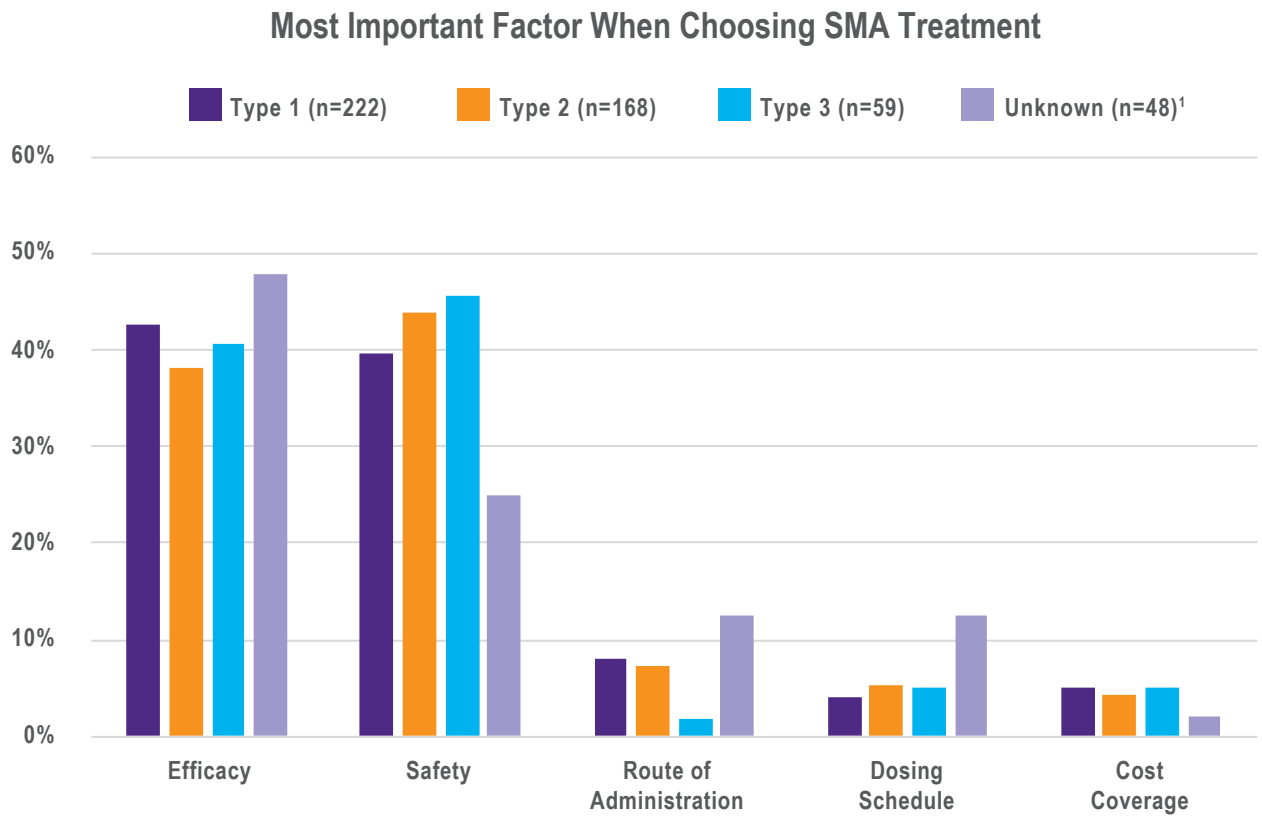
Analysis Notes:

- Includes SMA treatments that were FDA approved as of 12/31/2024
- Graphs include individuals who have not participated in clinical trials

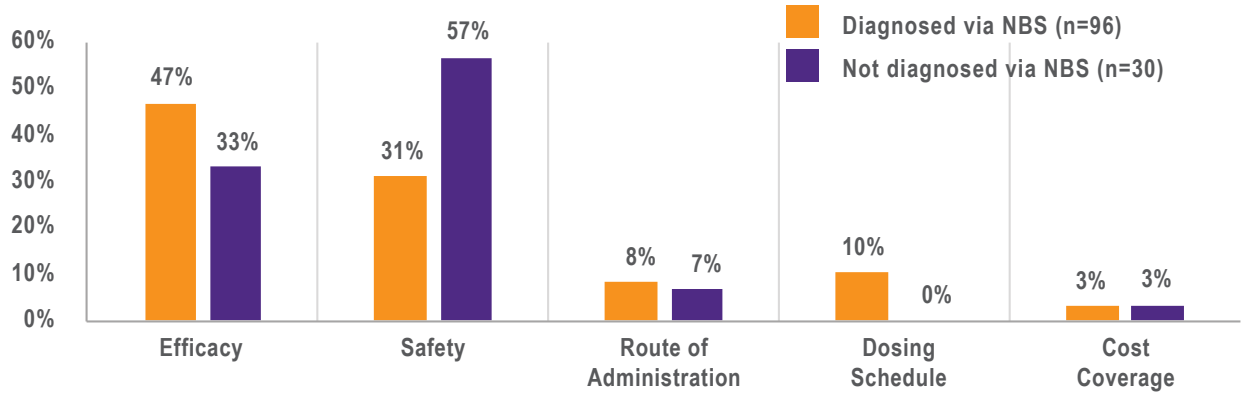
TREATMENT: OBSERVATIONS IN OUR DATA

PEDIATRIC (0-17 YEARS OLD)

Across SMA Types, caregivers of children with SMA ranked efficacy and safety as the most important factors when choosing an SMA treatment.



In individuals aged 0-2 years, a higher percentage of caregivers of children diagnosed with SMA via NBS ranked efficacy as the most important factor. A higher percentage of caregivers of children not identified via SMA NBS ranked safety as the most important factor.



Data Sources: 2022-2024 CUS Data. The most recent survey was used for any individuals with multiple surveys.

1. Unknown SMA type includes the following: Unknown, Unspecified, Pre-symptomatic, and Unknown because diagnosed < 6 months of age before symptom onset. If does not include missing values.

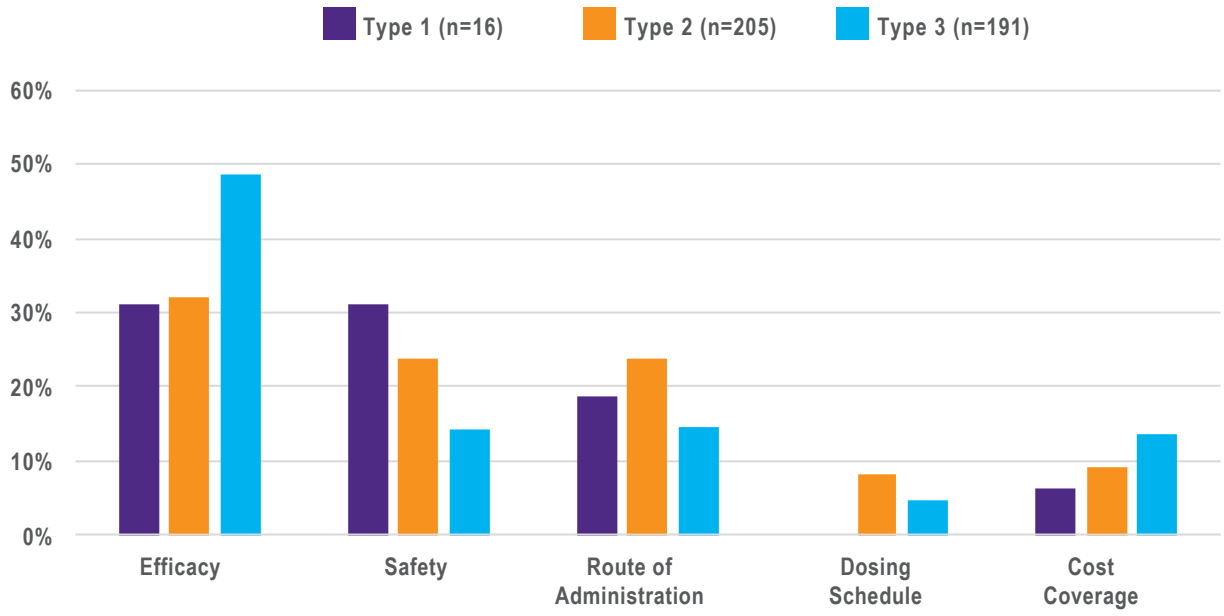
TREATMENT: OBSERVATIONS IN OUR DATA

PATIENT/CAREGIVER REPORTED

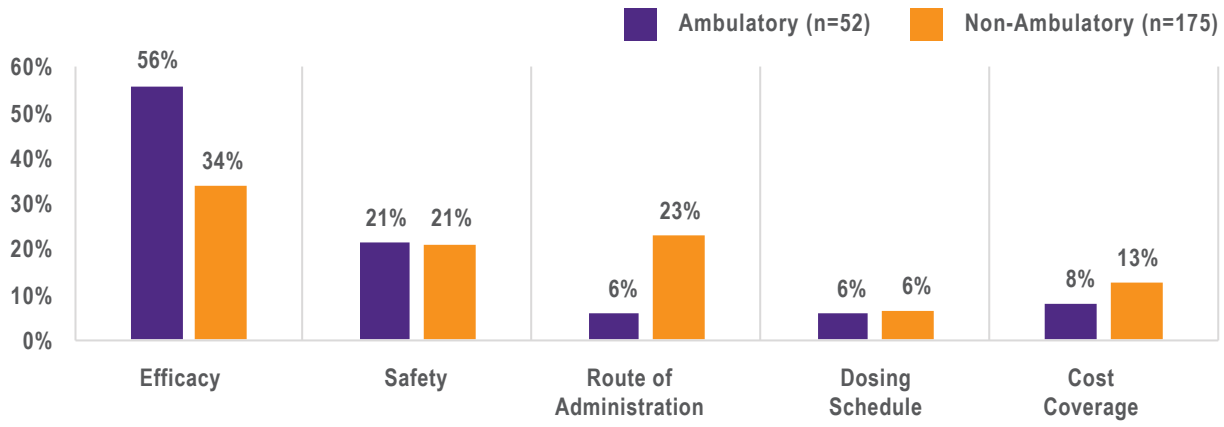
ADULTS

In general, we observed that most adults living with SMA report efficacy as the most important factor when choosing SMA treatment. Adults with Type 1 and 2 SMA had a broader distribution across efficacy, safety, and route of administration.

Most Important Factor When Choosing SMA Treatment[1]



In part- and full-time employed adults, efficacy was ranked as the most important factor in both ambulatory and non-ambulatory adults living with SMA. However, 23% of non-ambulatory individuals ranked route of administration as the most important factor compared to 6% of ambulatory individuals.



Cure SMA Data Sources: 2022-2024 CUS Data. The most recent survey was used for any individuals with multiple surveys.

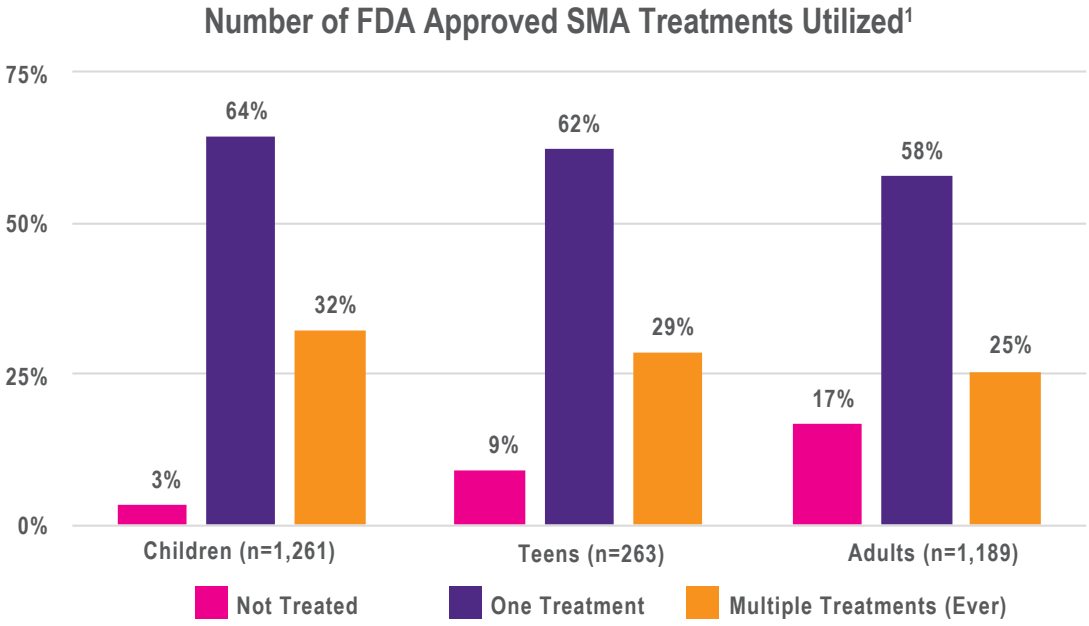
1. Individuals with SMA Type 4 were not included in the graphic due to low sample size

Analysis Notes:

- Analyses include individuals who self-completed the CUS

TREATMENT: USE OF MULTIPLE SMA TREATMENTS

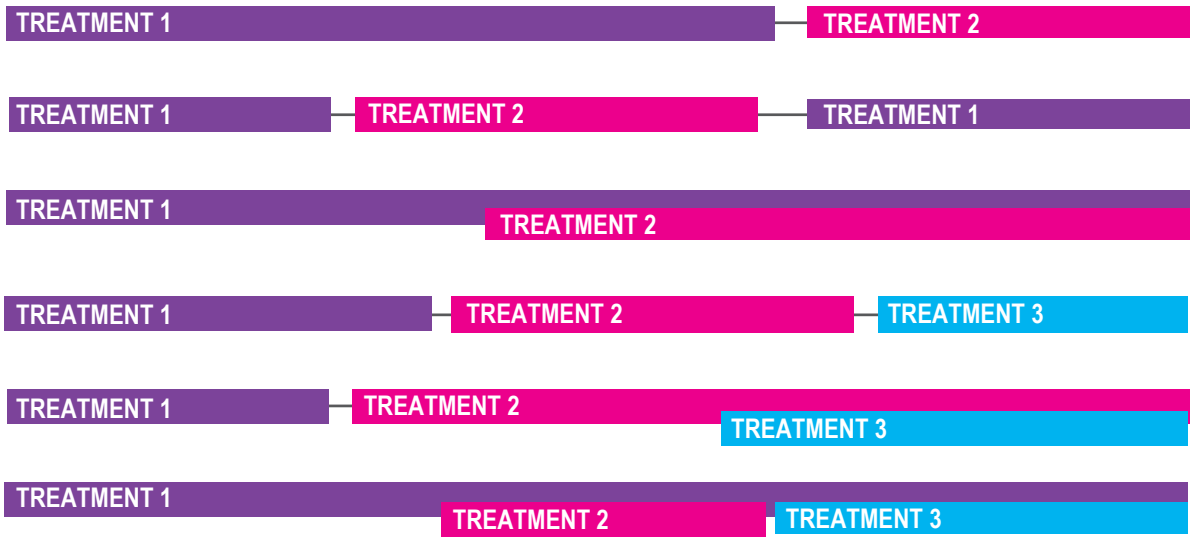
In our data, approximately 29% of individuals with SMA that have not participated in a clinical trial have received 2 or more FDA approved SMA treatments.



48% of those on one FDA-approved treatment have reported interest in receiving an additional genetic modifying therapy[2]

POTENTIAL PATHWAYS OF MULTIPLE TREATMENT USE

With 3 treatments available, there are many possible treatment pathways and durations an individual could possibly take. Not all pathways are appropriate for everyone, as each clinical journey is different. In 2024, Cure SMA published in Neurology Clinical Practice "Spinal Muscular Atrophy (SMA) Update in Best Practices: Recommendations for Treatment Considerations" to help support healthcare professionals in treatment decision making. With muscle-enhancing treatments on the horizon, we expect the number of possible treatment pathways and decision making to increase.



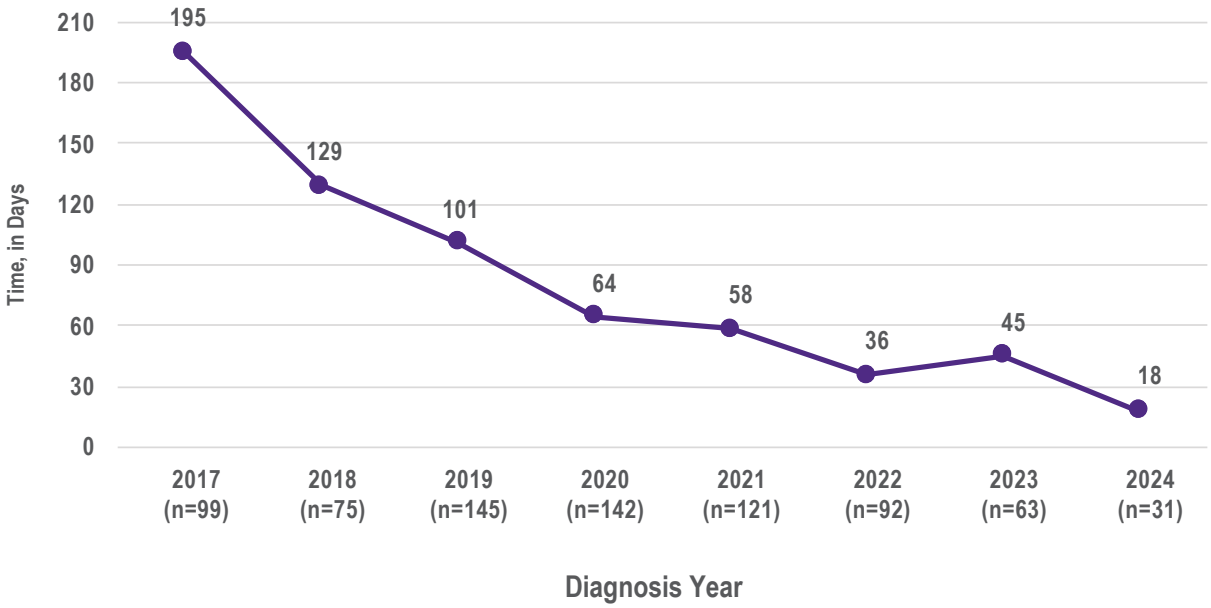
Cure SMA Data Sources: Membership data, CUS, CDR, and NBSR data combined. Individuals participating in multiple sources were duplicated.

1. Based on data from individuals with treatment status information on FDA approved treatments only. Individuals who have participated in a clinical trial have been removed from this analysis.
2. n=167

ACCESS

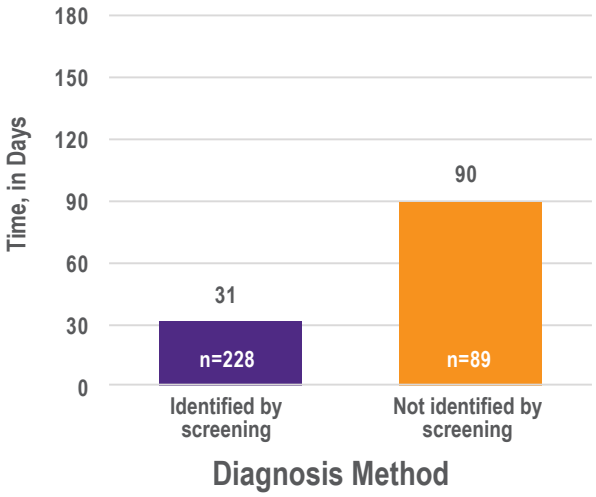
In our data, we observed a decrease in time between diagnosis and first treatment since 2017. For individuals diagnosed with SMA in 2024, the average time between diagnosis and first treatment was 18 days.

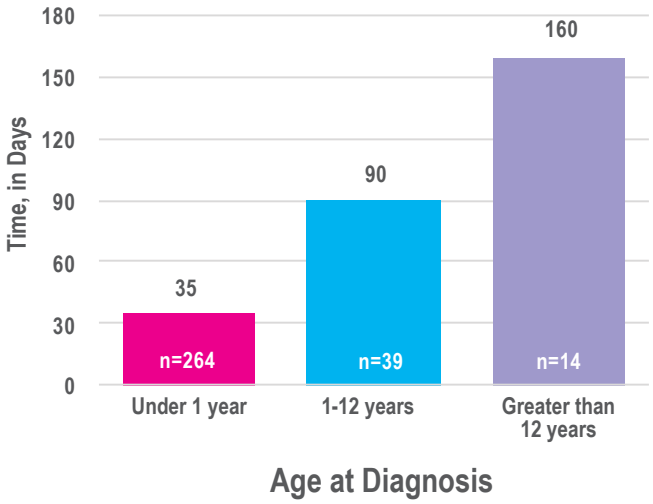
Average Time between Diagnosis and First SMA Treatment[1]



We are still seeing delays in time from diagnosis to treatment for individuals with SMA that were diagnosed symptomatically and diagnosed at older ages.

Average Time between Diagnosis and First SMA Treatment, by Subgroup[2] (Included individuals diagnosed with SMA between 2021-2024)





Cure SMA Data Sources: Membership data, CUS, CDR, and NBSR data combined. Individuals participating in multiple sources were de-duplicated.

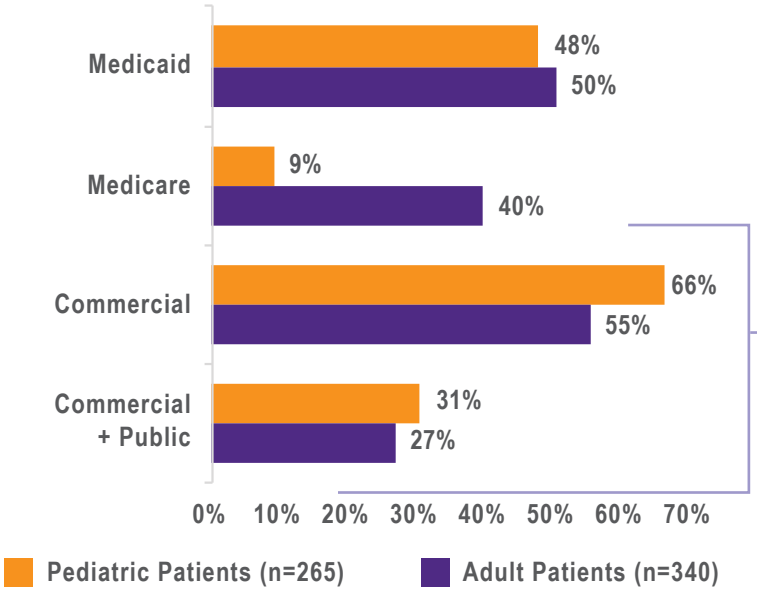
1. The minimum and maximum values were removed from each year
2. Newborn and prenatal screening status are patient reported. If status was missing, it was assumed the individual was not identified by screening.

Analysis Notes:

- Includes SMA treatments that were FDA approved as of 12/31/2024
- Graphs include individuals who have not participated in clinical trials and where both diagnosis and treatment dates are known
- Analyses include individuals where the date of diagnosis occurred on or before the date of first treatment

SMA care is expensive and includes many out-of-pocket costs for outpatient medical care, hospitalizations, and medications. Most children with SMA are enrolled in government-funded insurance programs.

Types of Insurance Among Pediatrics and Adults with SMA[1,2,3]



99% of individuals with SMA report having one or more forms of health insurance in 2024.

42% of individuals were on more than one insurance plan in 2024.

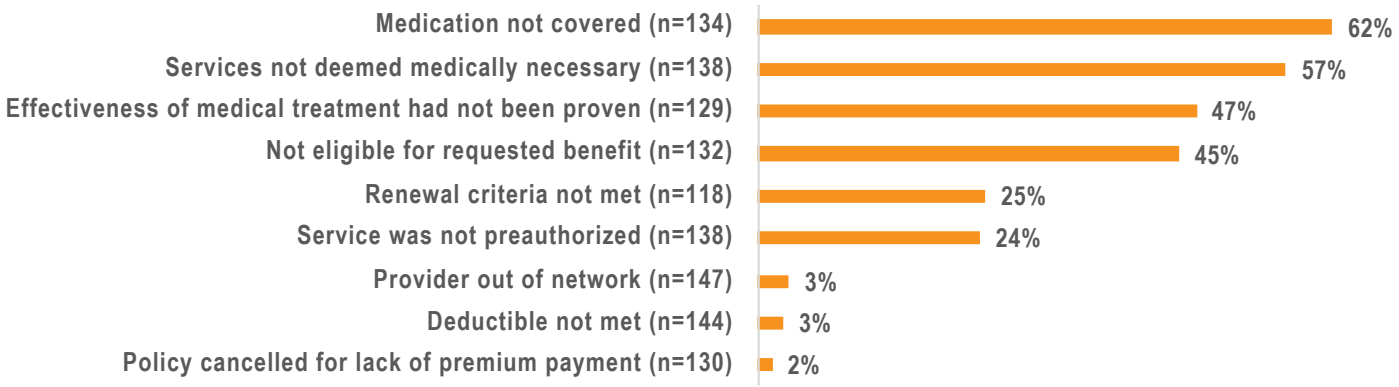
Among individuals on commercial insurance plans, 50% were on a PPO, 17% on an HMO, 4% on a HDHP, and 2% on a POS plan[4]

40% of adults report issues with insurance coverage or reimbursement for SMA treatments is their greatest barrier related to the treatments of SMA[7]

TREATMENT DENIALS

In the 2024 Access Survey (n=328), 52% of individuals with SMA reported ever receiving an insurance denial for their SMA treatment. Among those who experienced a denial (n=171), 44% had a denial at treatment initiation, 33% at treatment renewal, and 17% at both initiation and renewal[5]. The most frequently reported reasons for treatment denials were the medication not being covered (62%) and the services were not deemed medically necessary (57%).

Reasons Given for Initial Denial(s) of Treatment[6]



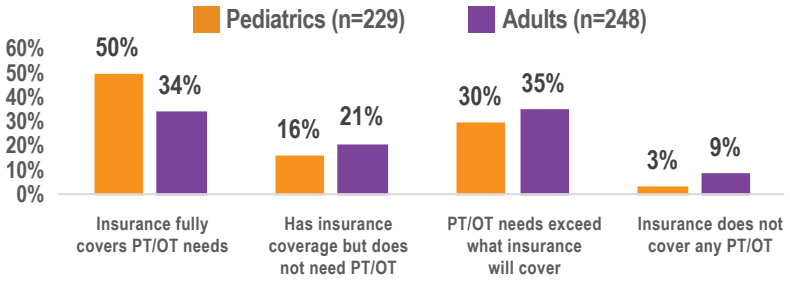
Cure SMA Data Sources: 2024 CUS data and 2024 Access Survey data

1. Insurance categories are not mutually exclusive
2. Categories in insurance coverage of "Don't know" and "Not applicable" were excluded from the graph, but were used in calculating proportions
3. For this analysis, the adult group was 18-64 years of age
4. PPO stands for preferred provider organization; HMO stands for health maintenance organization; HDHP stands for high deductible health plan; POS stands for point of service
5. Categories of treatment denials are not mutually exclusive and an individual may have received denials at different periods for more than one treatment.
6. Reasons for treatment denial may have occurred for a single or multiple treatments
7. n=241

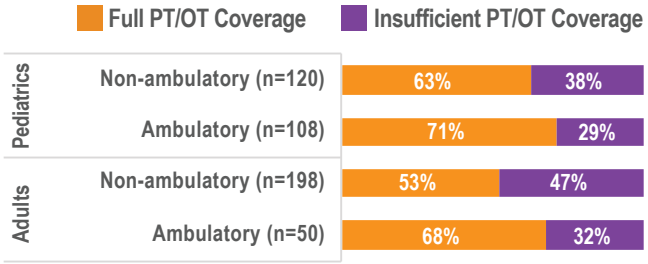
PHYSICAL THERAPY (PT) / OCCUPATIONAL THERAPY (OT) COVERAGE

We are observing a greater proportion of pediatric individuals with reported full insurance coverage for PT/OT services when compared to adults (50% vs. 34%).

Insurance coverage for PT/OT[1]



Insurance Coverage for PT/OT by Ambulation Status



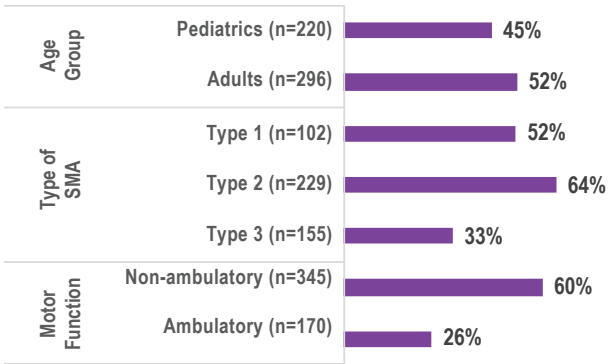
When PT/OT coverage was assessed by ambulation status, a greater proportion of both pediatric (38%) and adult (47%) individuals who are non-ambulatory reported insufficient PT/OT coverage compared to those who are ambulatory.

DURABLE MEDICAL EQUIPMENT (DME)

82% of individuals with SMA report using one or more forms of DME. The most used equipment was a power wheelchair (73%), bath/shower chair (72%), toilet chair (40%), and manual wheelchair (38%). Among those who use DME, 51% reported receiving an initial denial of their DME.

47% of adults report challenges in obtaining necessary assistive devices as a significant barrier for managing SMA (n=241)

DME Denials by Age Group, Type of SMA, and Motor Function



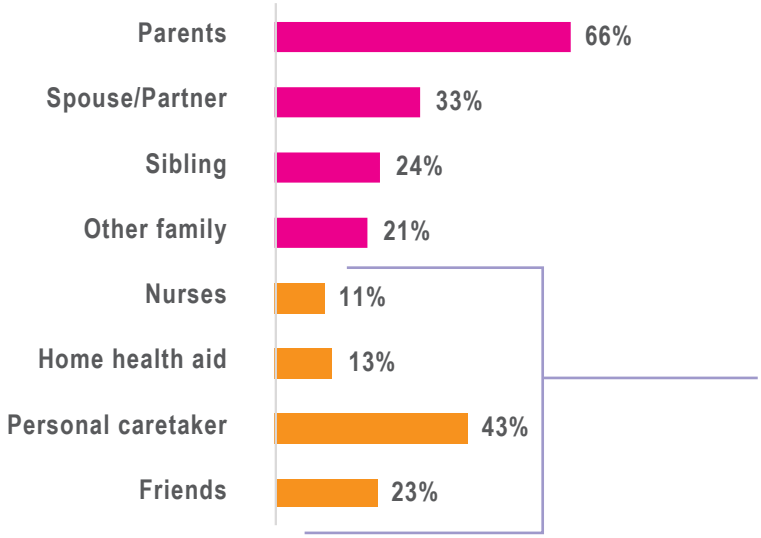
Denials for DME occurred most frequently among those with Type 2 SMA (64%), and those that are non-ambulatory (60%).

When the initial DME denial was appealed, 69% of individuals were able to have coverage approved, 26% still had coverage denied, and 5% are pending.

Cure SMA Data Sources: 2024 CUS data

1. If someone reported their insurance covers PT/OT regardless of if they need therapy, they were grouped into the category of "Insurance full covered PT/OT needs"

Types of Caregivers Utilized by Adults (n=234)



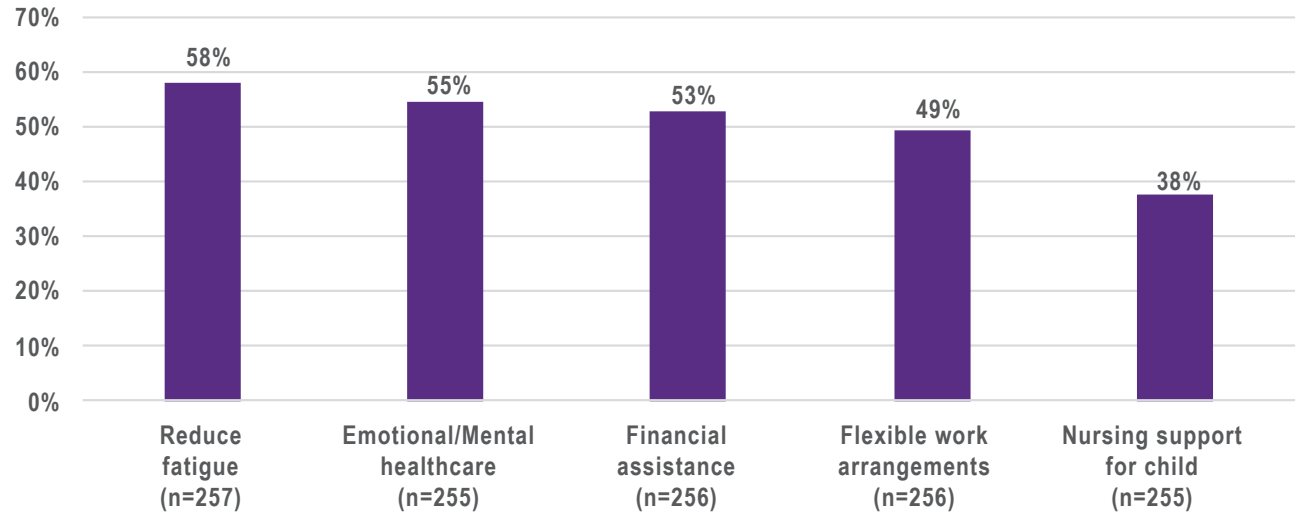
Average Number of Caregivers: 3
Minimum: 1 | Maximum: 20

85% of adults with SMA report having one or more caregivers

Caregivers outside of the immediate family are utilized in different capacities, with 25% being used < 20 hours weekly, 19% at 21-40 hours weekly, 24% at 41-80 hours weekly, and 32% at > 80 hours weekly.

UNMET NEEDS OF THE CAREGIVER

Caregivers are faced with many challenges and unmet needs of their own. When asked what the top 5 most significant unmet needs that caregivers face while caring for an individual with SMA, the following were the most commonly reported unmet needs.



Cure SMA Data Sources: 2024 CUS data

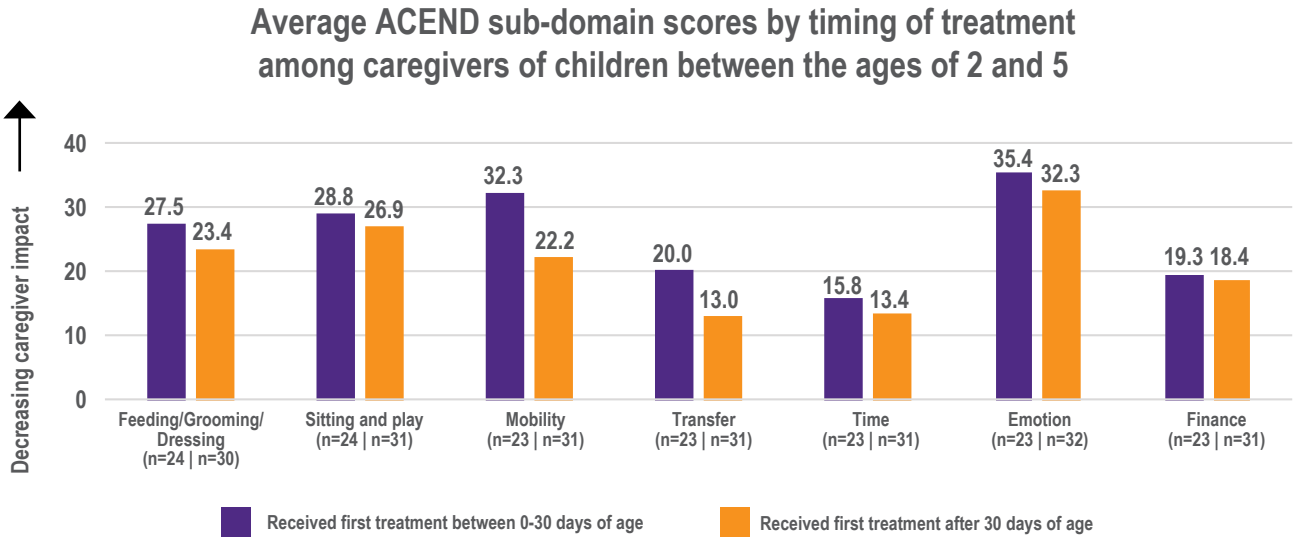
CAREGIVING

PATIENT/CAREGIVER REPORTED

The Assessment of Caregiver Experience with Neuromuscular Disease (ACEND) is a caregiver impact-based reported outcome measure to quantify a caregiver's perception of function and quality of life pertaining to caring for a child with a neuromuscular disease and pertaining to time, finance and emotion.[1]

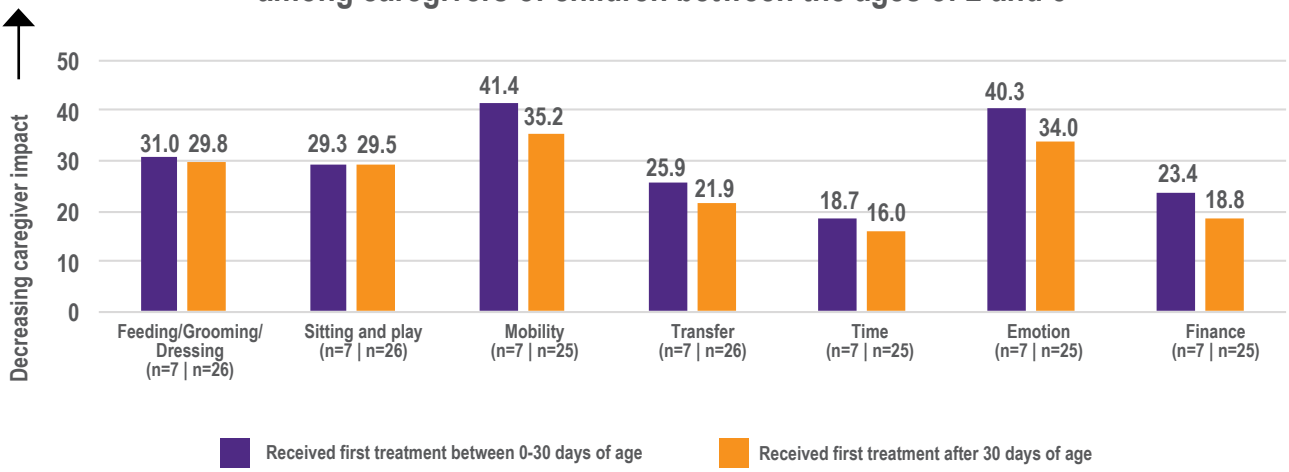
Higher scores indicate caregivers experience less intense caregiving impact and higher quality of life.

1-2 *SMN2* COPIES



The average ACEND scores are higher (less caregiving impact) among those that received treatment under 30 days of age compared to those that received treatment greater than 30 days of age.

3 *SMN2* COPIES

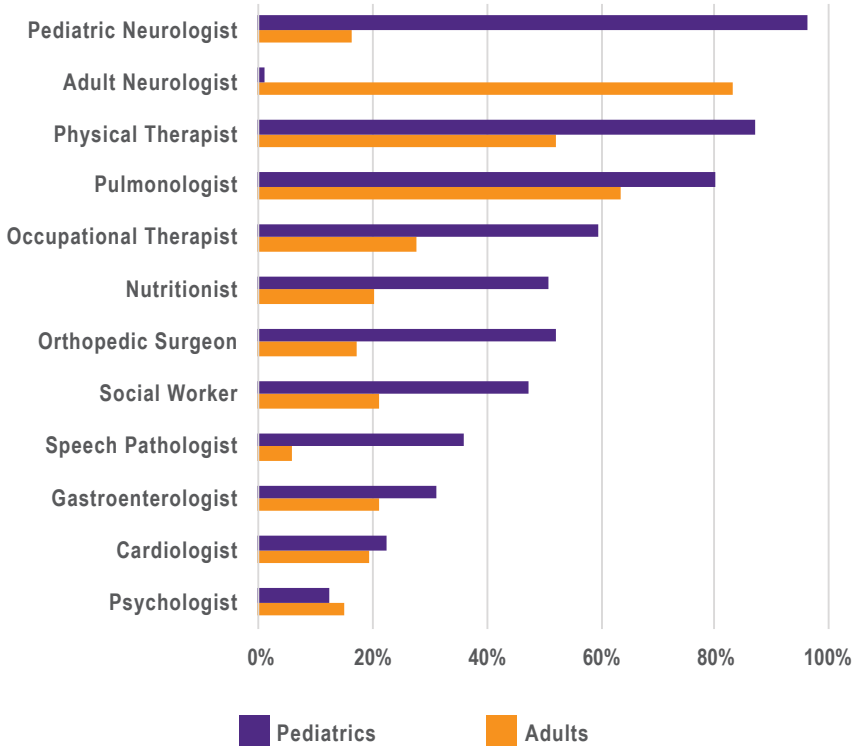


Cure SMA Data Sources: 2024 CUS

1. Matsumoto H, Clayton-Krasinski DA, Klinge SA, Gomez JA, Booker WA, Hyman JE, Roye DP Jr, Vitale MG. Development and initial validation of the assessment of caregiver experience with neuromuscular disease. J Pediatr Orthop. 2011 Apr-May;31(3):284-92.

In 2024, 97% of pediatrics with SMA and 83% of adults with SMA reported having an in-person appointment with a physician or specialist for SMA related care.

Types of Specialists Seen Among Pediatrics and Adults Living With SMA



The average number of specialists within a pediatric care team was 5, while the average number of specialists in an adult care team was 3.

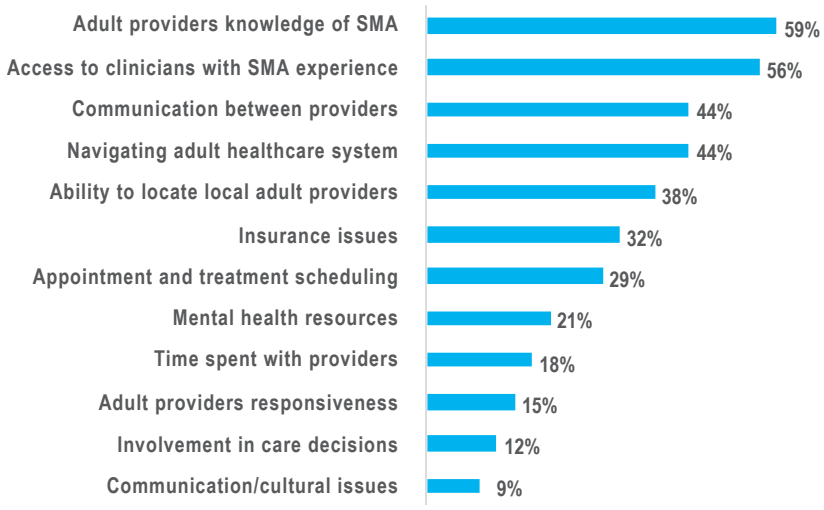
16% of adults over 18 years of age reported seeing a pediatric neurologist. The average age among these adults was 25.3 years (s.d. 7.8), and consisted primarily of those with Type 2 SMA (60%), followed by Type 3 (30%).

74% of adults report support from a dedicated team of healthcare professionals specializing in SMA is the most helpful when managing SMA[1]

TRANSITION OF CARE

In the 2024 Access Survey, 64% of individuals 18 to 29 years of age (n=53) reported having fully transitioned from pediatric to adult care, 25% partially transitioned, and 11% continue to see pediatric providers. Among those who fully transitioned (n=34), over half (59%) transitioned between 18-20 years of age, and among those not transitioned, most plan to transfer between 18-20 years old (48%) or 21-24 years old (39%). Most common issues experienced when transitioning to adult care were adult providers knowledge on SMA (59%), and access to clinicians with SMA experience (56%).

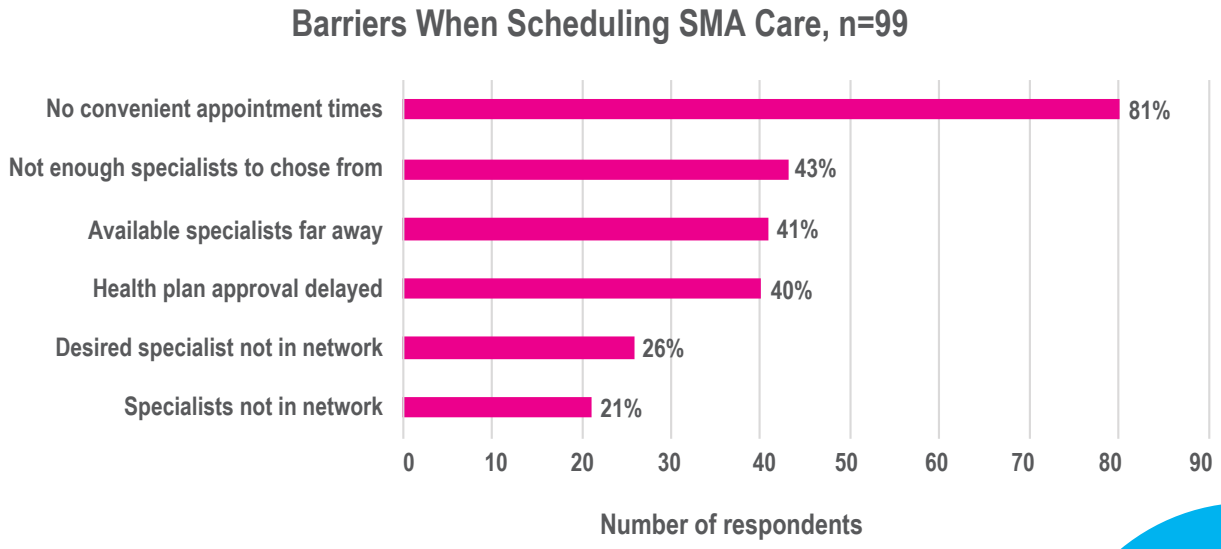
Transition of Care Reported Issues (n=34)



Cure SMA Data Sources: 2024 CUS and 2024 Access Survey data

1. n=242

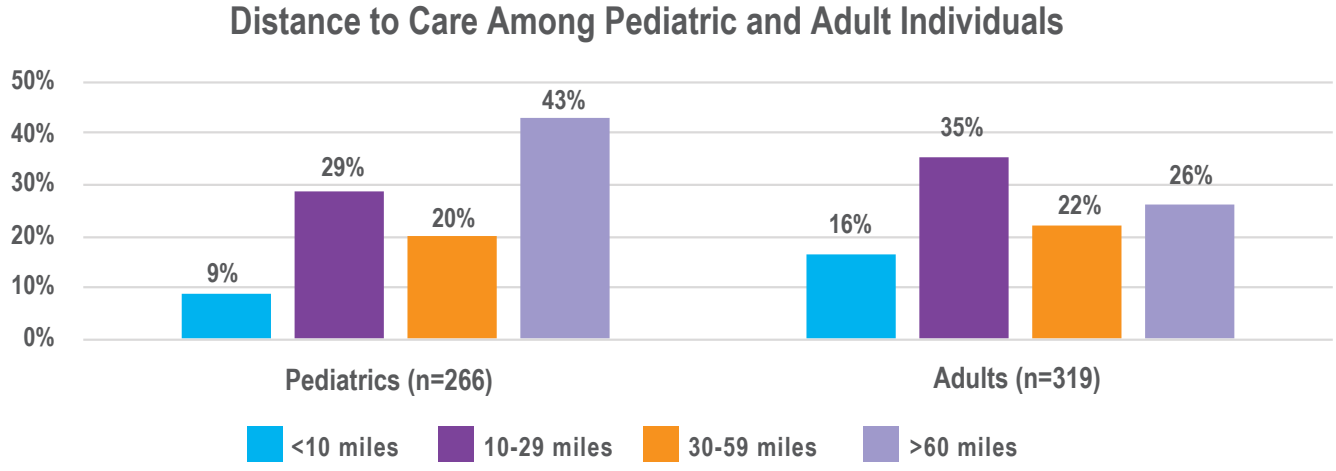
29% of affected individuals or their caregivers reported barriers when attempting to schedule an appointment with a healthcare specialist. The most commonly reported barriers were lack of convenient appointment times and not enough specialists to choose from within their health plan/network.



46% of caregivers rated that travel expenses to and from health providers has had moderate to extreme impact financially[1]

DISTANCE TO CARE

Affected individuals and their caregivers were asked to report on average how far they travel for their SMA related care (n=588). Overall, 13% reported traveling less than 10 miles, 32% travel 10 to 29 miles, 21% travel 30 to 59 miles, and 34% travel more than 60 miles. We also observed a greater proportion of pediatrics traveling greater distances for their care compared to adults.



Cure SMA Data Sources: 2024 Access Survey and 2024 CUS data

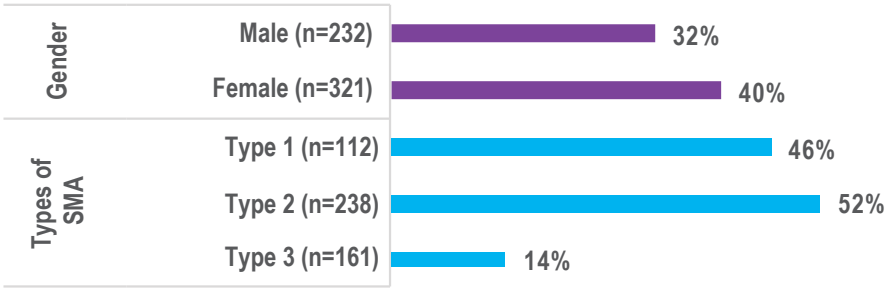
1. n=288

PATIENT/CAREGIVER REPORTED

HIP DYSPLASIA

A condition where the hip socket does not fully cover the ball portion of the upper thighbone, leading the hip joint to become unstable and partially or completely dislocated.[1]

Proportion of Individuals Diagnosed with Hip Dysplasia by Gender and Type of SMA

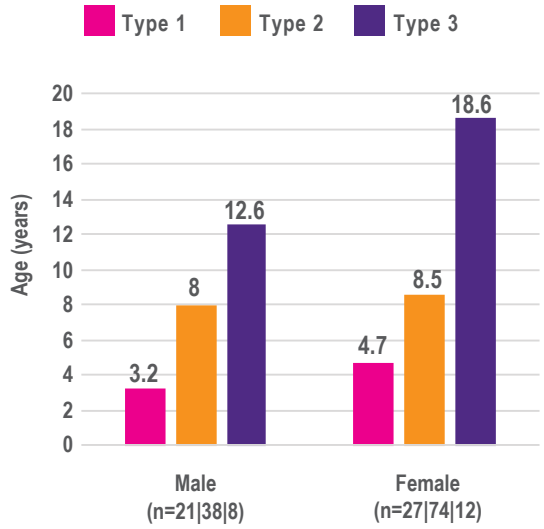


36% of affected individuals (n=554) reported being diagnosed with hip dysplasia, with diagnoses being reported more frequently among females and individuals with Type 1 and Type 2 SMA.

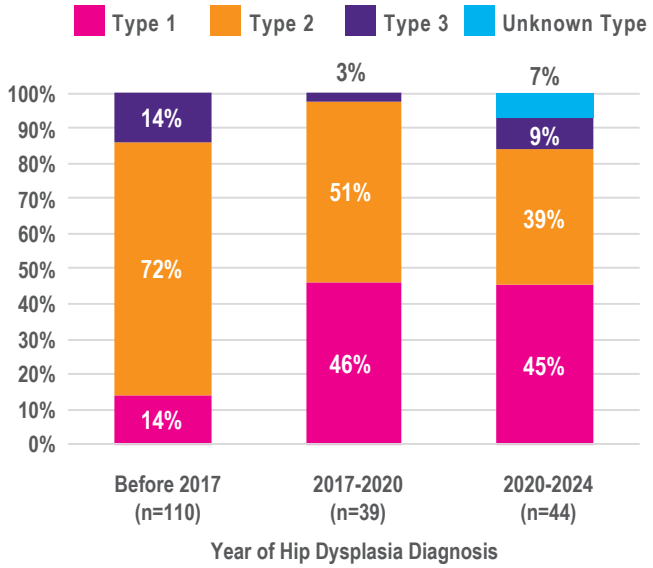
HIP DYSPLASIA DIAGNOSIS

The majority (93%) of individuals were diagnosed with SMA prior to hip dysplasia. Additionally, over-half (58%) reported being diagnosed before starting treatment for SMA (n=173). The average age of hip dysplasia diagnosis increases from Type 1 to Type 3 among both males and females. Among individuals diagnosed with hip dysplasia, we are observing an increase in the proportion of those with Type 1 SMA after 2017.

Average Age of Hip Dysplasia Diagnosis Among Males and Females by SMA Type



For Individuals with a Reported Hip Dysplasia Diagnosis: Distribution of SMA Type, by Year of Hip Dysplasia Diagnosis



Cure SMA Data Sources: 2024 CUS Data

1. https://www.mayoclinic.org/diseases-conditions/hip-dysplasia/symptoms-causes/syc-20350209

Analysis notes:

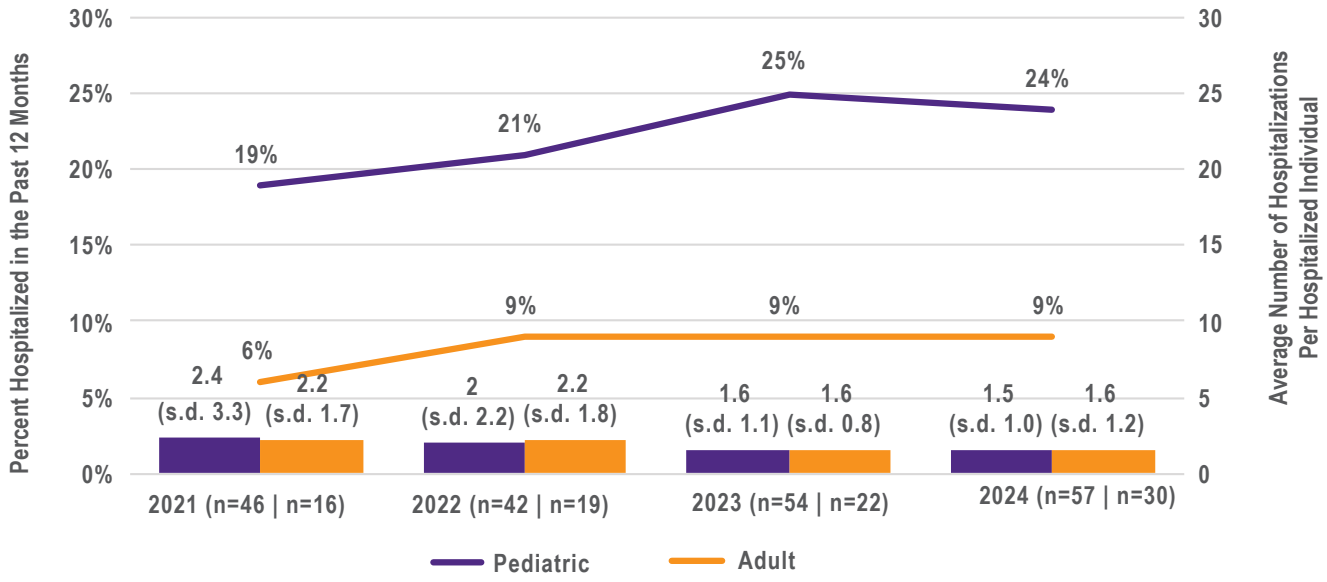
- Individual with Type 4 SMA have been excluded from the graphics due to small sample size

HOSPITALIZATIONS

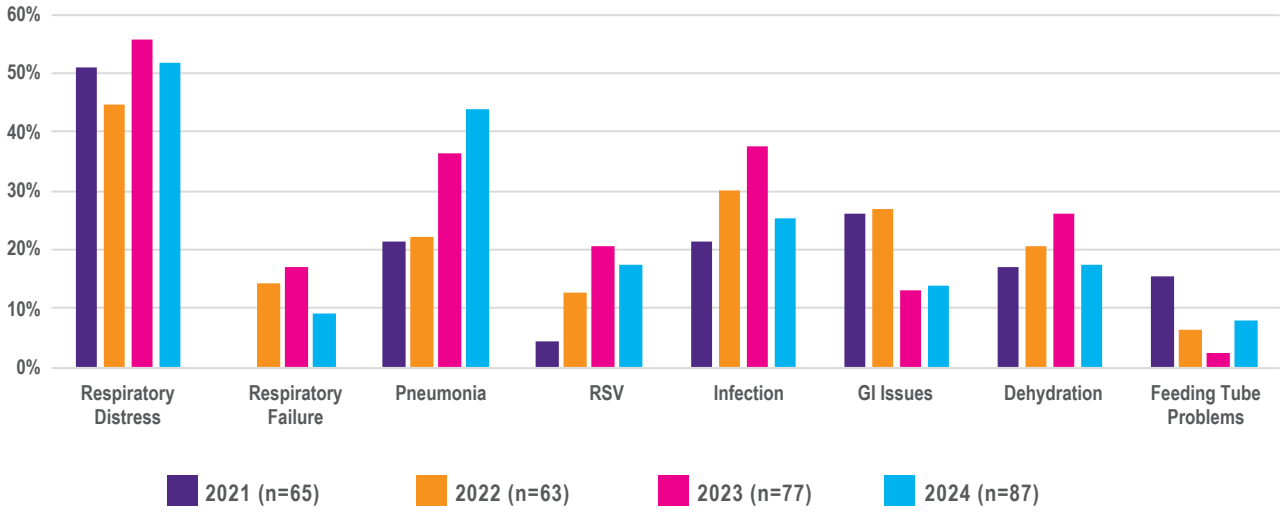
PATIENT/CAREGIVER REPORTED

In 2024, 14% of all individuals with SMA reported being hospitalized within the past 12-months. Among the pediatric population, 24% reported a hospitalization, and among adults 9% reported a hospitalization.

Hospitalization Rate Among Pediatrics and Adults and Average Number of Hospitalizations per Year: 2021-2024[1]



Trends in Reason for Hospitalization: 2021-2024



Among individuals who reported being hospitalized within the previous 12-months between years 2021-2024, we are observing an increase in the proportion of individuals hospitalized due to pneumonia (22% vs 44%) and RSV (5% vs. 17%), and a decrease in the proportion of individuals hospitalized due to GI issues (26% vs. 14%).

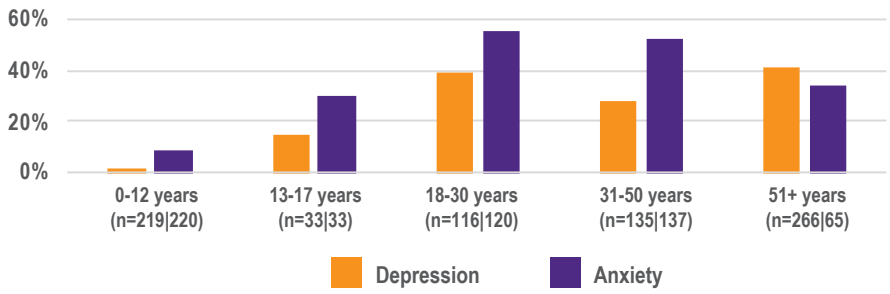
Cure SMA Data Sources: CUS data (2021-2024)

1. This is not truly longitudinal as the same individuals are not included in every time period

MENTAL HEALTH

A qualitative study by Cure SMA found that individuals with SMA reported their mental health to be severely impacted by SMA[1]

Prevalence of Depression and Anxiety Diagnoses by Age Group

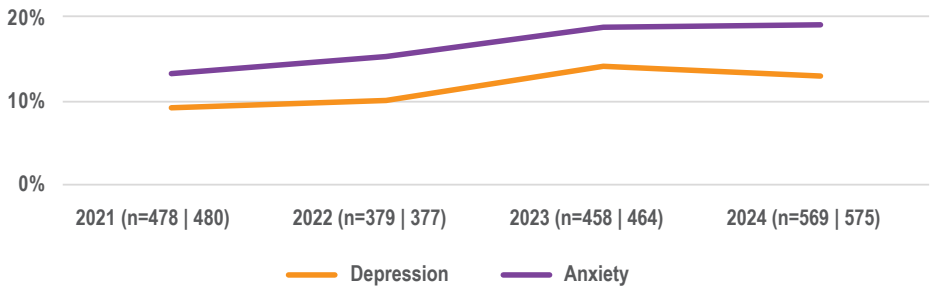


26% of the full population reported ever being diagnosed with depression, and 48% ever diagnosed with anxiety. Both depression and anxiety were most prevalent among those 18-30 years old, followed by those 31-50 years old.

The average age at diagnosis of depression was 22.9 years old (s.d. 11.6 years), and average age at diagnosis of anxiety was 22.9 years old (s.d. 13.3 years).

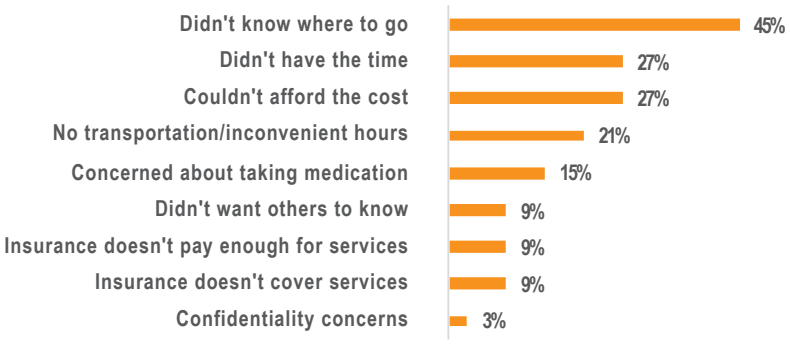
Proportion of Individuals Receiving Treatment for Depression or Anxiety by Year

Since 2021, we have observed a slight increase in the proportion of the population actively being treated for depression (9% vs.13%) and/or anxiety (13% vs. 19%) within a given year.



UNMET MENTAL HEALTH NEEDS

Reasons for Not Accessing Mental Health Services, (n=33)



45% of adults self-reported that they felt they needed mental health services to help with anxiety, depression or other issues in the past 12-months. Among these individuals, 31% reported not being able to get the mental health services they needed. The most commonly reported reason for not getting services was not knowing where to go, followed by lack of time and associated costs.

If you are experiencing anxiety or depression, please contact your healthcare provider. You are not alone.

Cure SMA Data Sources: 2024 CUS data

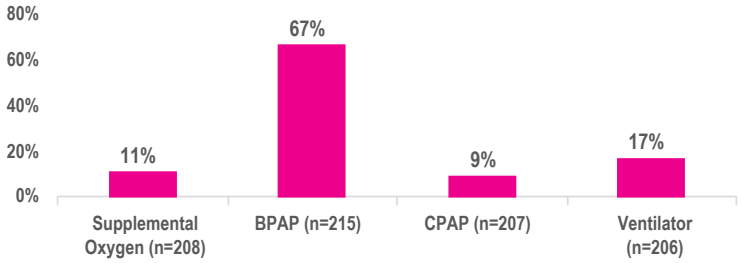
1. Mazzella A, Curry M, Belter L, Cruz R, Jarecki J. "I have SMA, SMA doesn't have me": a qualitative snapshot into the challenges, successes, and quality of life of adolescents and young adults with SMA. Orphanet J Rare Dis. 2021;16(1):96.

RESPIRATORY FUNCTION

PATIENT/CAREGIVER REPORTED

Individuals with SMA often have weakened intercostal muscles between their ribs, making breathing more difficult and requiring additional breathing support.

Forms of Breathing Support Among Individuals Currently Requiring Breathing Support[1]

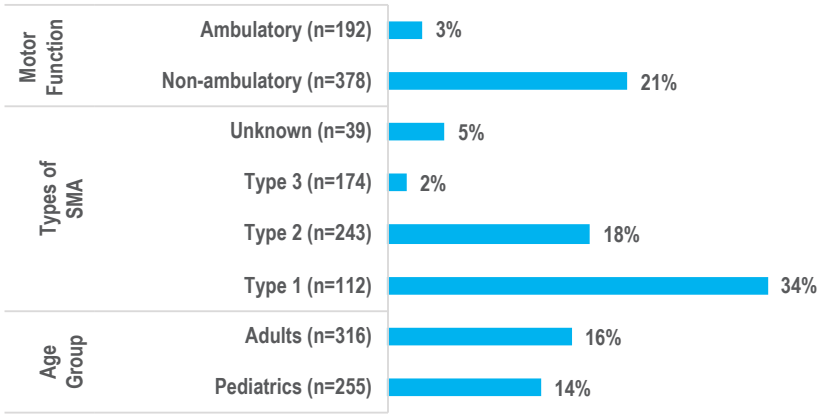


36% of individuals with SMA report currently using some form of breathing support. Among those on breathing support, BPAP use is the most prevalent form.

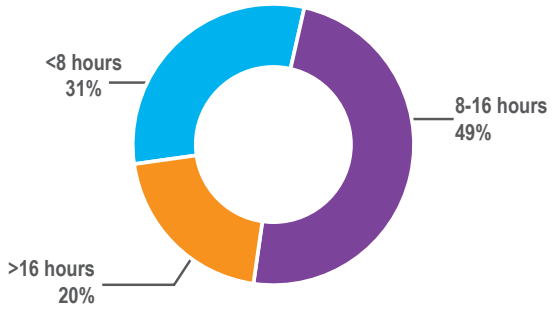
CHRONIC RESPIRATORY FAILURE

15% of individuals with SMA report a diagnosis of chronic respiratory failure. Chronic respiratory failure was most common among those with Type 1 SMA (34%), and those non-ambulatory (21%). The average age at diagnosis was 8.1 years (s.d. 9.6 years).

Proportions of those with Chronic Respiratory Failure



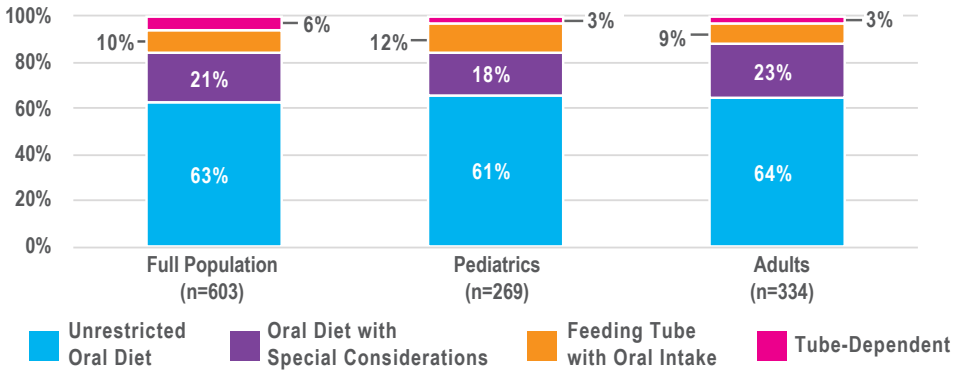
Hours per Day of Breathing Support Among Individuals with Chronic Respiratory Failure, n= 78



Among those with chronic respiratory failure, 92% report using some form of breathing support. About half (49%) of these individuals require breathing support 8-16 hours per day.

Cure SMA Data Sources: 2024 CUS Data

Daily Feeding Routine[1]



63%

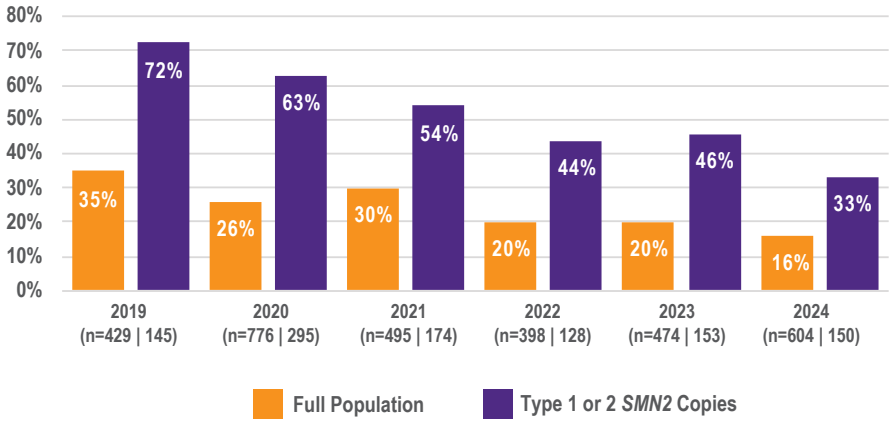
of individuals report being on an unrestricted oral diet. When stratified by age, a higher proportion of pediatric individuals are on tube-feeding compared to adults.

FEEDING TUBE UTILIZATION

Feeding tubes may be necessary for some individuals with SMA to provide partial or total nutritional needs. Types of feeding tubes include a gastrostomy tube or nasogastric tube into the stomach, or a jejunostomy tube into the small intestine.

Individuals with Type 1 SMA or 2 copies of *SMN2* report much higher feeding tube utilization than the overall population. Our data shows the proportion of individuals using a feeding tube to be decreasing overall.

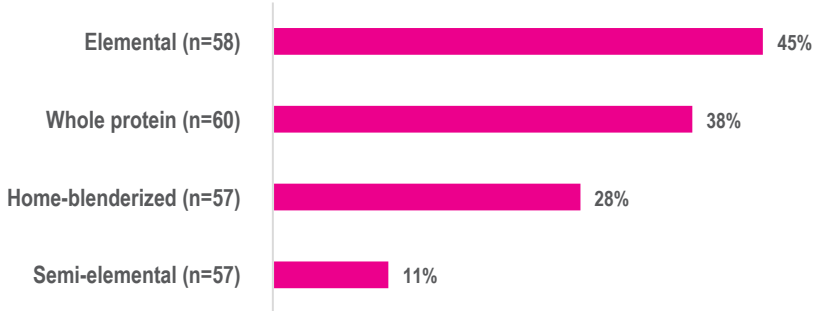
Proportion of Individuals Using a Feeding Tube: 2019-2024[2]



DIET

Whole protein and elemental diets were most frequently reported among affected individuals utilizing a feeding tube.

Diet Types Among Feeding Tube Users



Cure SMA Data Sources: CUS data (2019-2024)

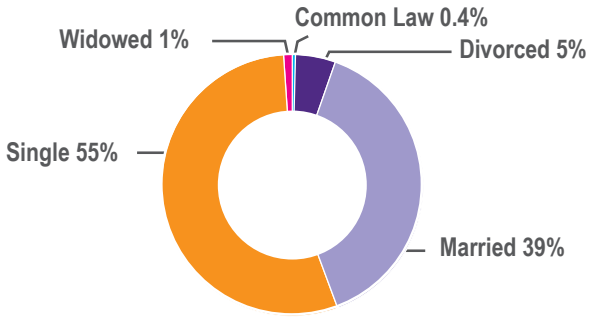
1. Data on daily feeding routine and diet type is of current data from 2024 CUS only
2. This is not truly longitudinal as the same individuals are not included in every time period

FAMILY PLANNING / FERTILITY

PATIENT/CAREGIVER REPORTED

39% of adults living with SMA are currently married or in a common law relationship.

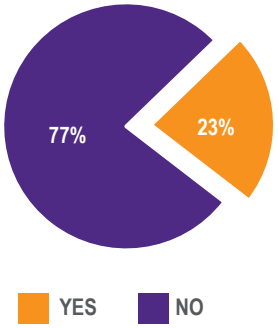
Marital Status[1] (n=1,028)



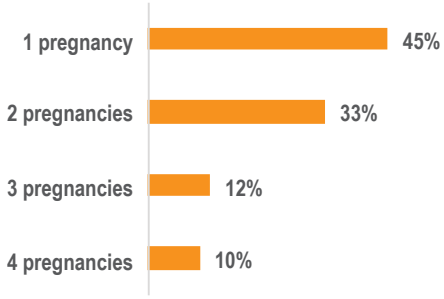
PREGNANCY

Cure SMA collects data on history of pregnancy in hopes to understand pregnancy rates and provide data for those considering a pregnancy in the future.

23% of women living with SMA report a history of ever being pregnant[2]

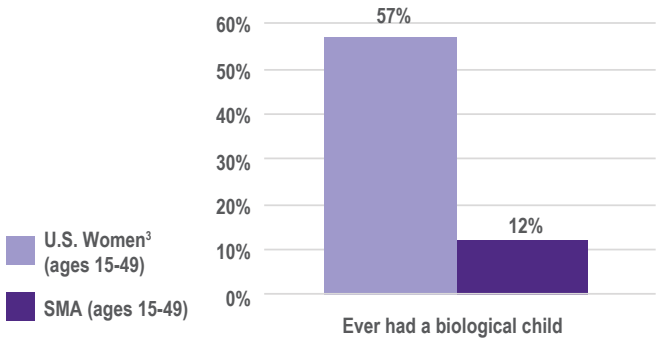


Number of pregnancies among women living with SMA with a history of pregnancy

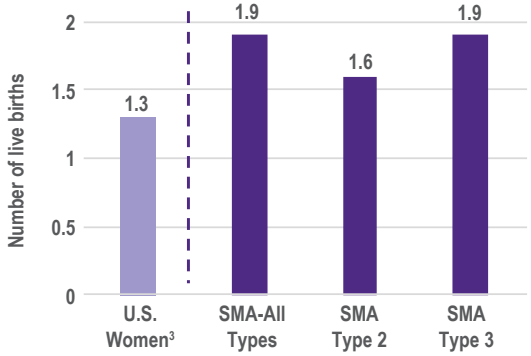


HISTORY OF LIVE BIRTHS

History of having a biological child among women



Average number of children born alive to women ages 15-49[4]



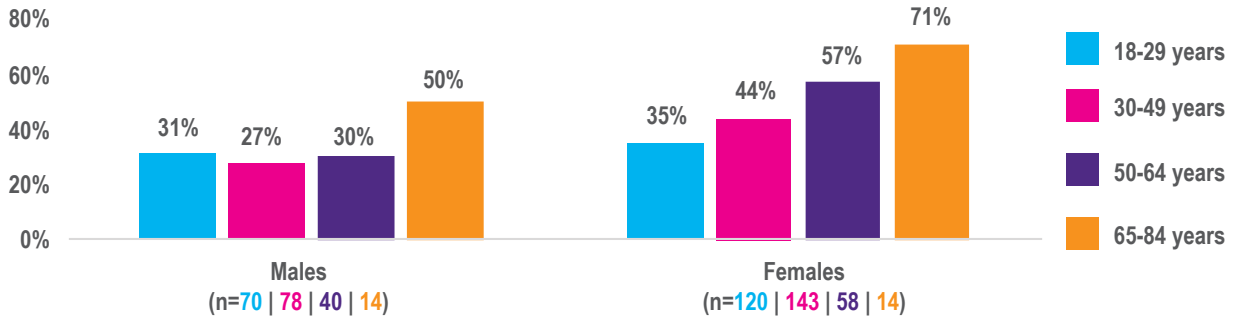
Cure SMA Data Sources: Membership Database & 2024 CUS data

1. Marital status is reported for those 18 years and older
2. n=51
3. https://www.cdc.gov/nchs/data/nhsr/nhsr179.pdf
4. Average number of births among those with SMA type 1 and a reported "unknown" SMA type are not shown due to small sample size

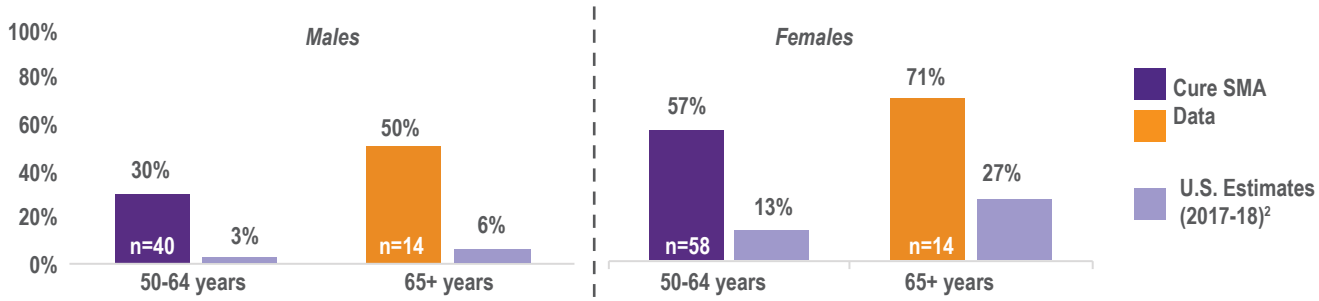
OSTEOPOROSIS: a disease that causes weakening of the bones, leading to increased risk of fractures[1]

We observed increases in osteoporosis as individual with SMA aged. Rates of osteoporosis were higher in Cure SMA data than the U.S. population.

Rates of Osteoporosis in Adults with SMA in Cure SMA Databases, by Age Categories

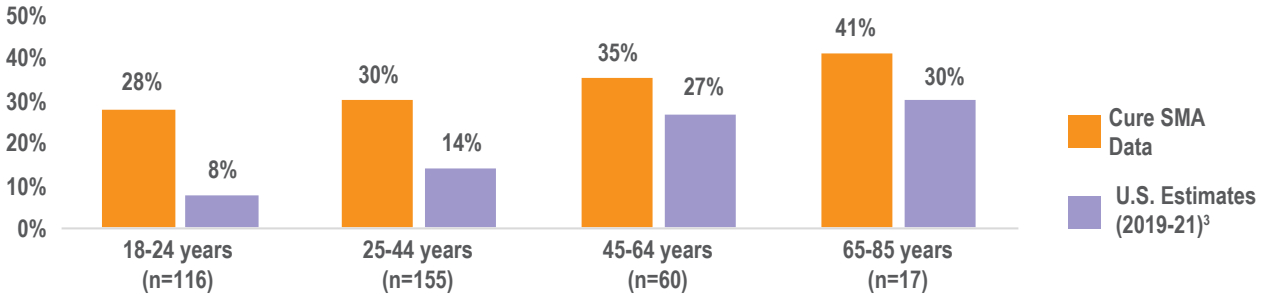


Rates of Osteoporosis in Cure SMA Databases and the U.S. Population



CHRONIC PAIN

Percent of Adults with SMA Reporting Chronic Pain in Cure SMA Databases and the U.S. Population



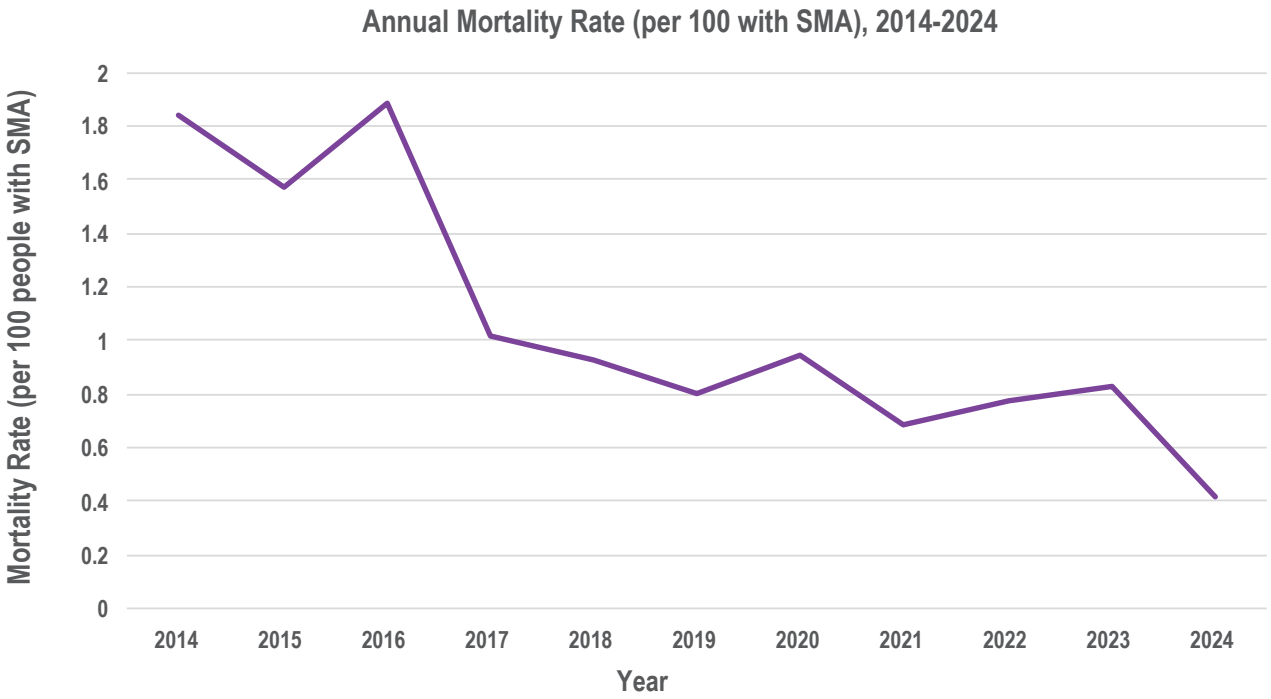
Rates of chronic pain were higher in Cure SMA databases than the U.S. population, especially at younger ages.

Cure SMA Data Sources: Osteoporosis: 2022-2024 CUS data | Chronis pain: 2024 CUS data; Denominator included individuals who answered a question in the comorbidity module; excluded individuals that reported "Don't know" for osteoporosis or chronic pain

1. https://www.niams.nih.gov/health-topics/osteoporosis
2. Sarafrazi N, Wambogo EA, Shepherd JA. Osteoporosis or low bone mass in older adults: United States, 2017–2018. NCHS Data Brief, no 405. Hyattsville, MD: National Center for Health Statistics. 2021. DOI: https://dx.doi.org/10.15620/ cdc:103477
3. Rikard SM, Strahan AE, Schmit KM, Guy GP Jr.. Chronic Pain Among Adults — United States, 2019–2021. MMWR Morb Mortal Wkly Rep 2023;72:379–385. DOI: http://dx.doi.org/10.15585/mmwr.mm7215a1.

MORTALITY

The mortality rate of SMA in 2024 has dropped nearly 80% since 2014, having decreased from 1.84 per 100 individuals to 0.42 per 100 individuals with SMA.



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.

There has been significant progress made in the SMA community but we will not stop until we have a cure.

Cure SMA Data Sources: Membership Data & CDR

Analysis Notes:

- Mortality data is captured from the CDR or family reported in the membership database.