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“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

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Abstract

Background: With the approval of three treatments for spinal muscular atrophy (SMA) and several promising therapies on the horizon, the SMA adolescent and young adult populations are expected to evolve in the coming years. It is imperative to understand this cohort as it exists today to provide optimal care and resources, as well as to assess possible treatment effects over time. In 2018, Cure SMA launched two initiatives geared towards understanding adolescents and young adults with SMA, ages 12–25. First, Cure SMA launched a Quality of Life (QoL) survey to capture quantitative and qualitative information on this specific age demographic. Concurrently, Cure SMA invited SMA-affected individuals, ages 12–25, to create a three-minute video on their everyday experiences living with SMA. An inductive thematic analysis of the free-text survey questions along with the video contest findings are reported here.

Results: Eighty-five individuals—6 type Is, 58 type IIs, and 21 type IIIs—completed the Quality of Life free-response, while six individuals participated in the SMA awareness video contest. In both settings, individuals detailed a variety of challenges, including but not limited to forming or maintaining close relationships, experiencing feelings of isolation, challenges with accessibility, independence, and dealing with the stigma of being perceived as mentally disabled. Individuals also discussed their successes, including but not limited to higher education enrollment and attendance, development of quality friendships, and perseverance through obstacles. Additionally, notably in the survey, 39% of respondents requested the creation of an SMA peer support group in efforts to connect with each other as well as collectively navigate the aforementioned challenges they face.

Conclusion: Together, these findings provide a rare glimpse into the unique mindsets, challenges and motivations of SMA adolescents and young adults, via patient-reported measures instead of caregiver proxy. The adolescent and young adult age demographics assessed represent a critical transition period in life and in SMA care. No one understands the needs of an adolescent or young adult with SMA better than the individuals themselves, and it is critical to encapsulate their insights to affect change.

Keywords: Spinal muscular atrophy, Quality of life in SMA adolescents and young adults, Qualitative research, Emotional health, Disability, Peer-support group, Accessibility, Fatigue, Dependence

Background

Spinal muscular atrophy (SMA) is an autosomal recessive neurodegenerative disease characterized by progressive muscle weakness often resulting in loss of movement, significant disability, and premature death

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Journal : **BMCTwo 13023**
Article No : **1701**
MS Code :

Dispatch : **16-2-2021**
 LE
 CP

Pages : **15**
 TYPESET
 DISK

[1–3]. SMA is estimated to affect between 8500 and 10,300 children and adults in the US [4–6]. SMA is traditionally classified into three main subtypes based on age of symptom onset and maximal achieved motor abilities [7]. In SMA type I, individuals are diagnosed within the first 6 months of life and never achieve independent sitting. Traditionally, without treatment, these individuals require intensive supportive care [2, 8–11]. In SMA type II, (Kugelberg–Welander disease) patients experience symptom onset within the first 18 months of life and have achieved the ability to sit independently but will not achieve the ability to walk unaided. Over time, these patients may develop difficulties involving chewing and swallowing, respiration, as well as progressive scoliosis [12, 13]. In SMA type III, individuals achieve the ability to stand and walk independently but may lose these abilities over time [12, 13]. In SMA type IV, otherwise known as adult-onset SMA, symptom onset typically occurs at age 30 or later, and as such, this type was not identified within our target age population [12]. Regardless of subtype, the disease burden of SMA is substantial, and complexly impacts affected individuals and their loved ones across multiple dimensions [6, 14–17].

With the approval of three treatments as of August 2020, the phenotypic presentation and prevalence of SMA in adolescent and young adult populations is expected to change. Due to this shift, disease classification is moving away from subtype and towards demographics based on motor function. All data presented below will be displayed in accordance with motor function while information related to SMA type is used to validate.

In previous years, a variety of initiatives have qualitatively captured clinical meaningfulness and the experiences of the adult SMA population [16, 18–20]. However, to our knowledge, this study is the first to directly capture information specific to quality-of-life directly from affected adolescents (12–18) and young adults (19–25), as the adolescent perspective tends to be captured via caregiver proxy, while the young adult perspective is captured along with that of adults across various ages [5, 14, 16–20]. It is necessary to further understand the factors this population deems most important in the overall improvement of quality-of-life. A clearer understanding may provide an opportunity for researchers to develop or optimize outcome measures and resources to further support adolescents and young adults affected by SMA.

Methods

Quality of Life (QoL) Survey

The Quality of Life Survey consists of four open-ended questions. The objective of this survey was to provide an opportunity for adolescents and young adults affected by SMA to share, in their own words, individual experiences about living with SMA as well as highlight topics important and specific to their stage of life. The four free text questions inquired on biggest obstacles, the impact of SMA on schooling and socialization, and potential resources to improve quality-of-life. No guidelines were given towards these responses in order to help identify a variety of themes. The Quality of Life Survey was distributed as part of a larger survey which was comprised of three sections: (1) demographics information, (2) SMA Health Index instrument (SMA-HI, a patient reported outcome measure developed by Dr. Chad Heatwole at the University of Rochester) (Heatwole [15, 18, 21] abstract), and (3) free text response. The SMA-HI information collected will be presented in a future publication.

The survey was generated through Survey Monkey and approved by the Western Institutional Review Board (Approval Number 1-11240001-1). Written informed consent was obtained online from all adult respondents prior to the conduct of any research related activities. For minor adolescent respondents (ages 12–18), written consent was obtained online from a parent/guardian, and assent was obtained from the survey respondent thereafter.

The survey was advertised through targeted email blasts sent to over 400 families using Cure SMA's membership database as well as Cure SMA's social media platforms. Cure SMA is a non-profit patient advocacy organization for SMA, which currently maintains a robust database that features data from over 8000 individuals affected with SMA [6]. As of June 1, 2018, 412 individuals between the ages of 12–25 were contacted within the database. Eighty-three percent of this cohort is composed of self-reported type II and III affected individuals [6]. Confidence intervals were calculated to provide power for each comparison. Sample size calculations, however, were not done prior to the study because open-ended responses were evaluated here and therefore, effect size could not be assessed using prior studies. Based on an expected response rate of about 20%, we expected about 80 responses that would elicit a 95% confidence level of and a 9.8% margin of error. Email blasts and social media reminders were disseminated biweekly. Materials directed at consenting adults contained a direct link to the survey, while materials designed for families of minors contained information regarding the parent/caregiver consenting and the participant assenting process prior to the survey. All consents and assents



138 were collected electronically. The survey remained open
 139 from November 2018 through January 2019 for a total
 140 of 8 weeks—an initial 6-week submission period with a
 141 2-week extension.

142 An inductive thematic approach was adopted for quali-
 143 tative analysis of the free text collected in the survey. All
 144 questions were evaluated for themes independently of
 145 one another. For each question, all responses were manu-
 146 ally sorted by the primary author into themes through the
 147 identification of key words and sentiments. Key words
 148 were identified retroactively of survey completion, for
 149 purposes of data analyses. Although the prompts were
 150 open-ended, there was a high concordance in responses
 151 across participants, particularly regarding the final ques-
 152 tion. Once all responses were categorized, they were
 153 sorted by age, SMA type, and respondent’s maximum
 154 motor function at time of diagnosis and time of survey
 155 completion, with all demographic information being self-
 156 identified. Maximum motor function status was defined
 157 as; non-independent sitter (head control, maintain seated
 158 position supported), sitter/non-independent walker
 159 (maintain seated position unsupported, crawl, cruise),
 160 and independent walker (walk independently). Statistical
 161 significance based on response age and maximum motor
 162 function at time of survey completion was assessed using
 163 Fischer’s exact test. For detailed information regarding
 164 key words and phrases included in each theme as well as
 165 statistical significance results, please see Additional file 1:
 166 Tables S1–S4.

167 **SMA awareness video contest**

168 The objective of the SMA awareness video contest was
 169 to raise awareness about the impact/burden of SMA on
 170 adolescents and young adults, and learn about the strate-
 171 gies they use/have developed to help navigate their day to
 172 day life and the challenges that come with managing this
 173 multifaceted and complex disease. Contest guidelines
 174 included general topic suggestions that participants could
 175 discuss within a 3-min time limit. Topics suggestions
 176 included but were not limited to schooling, everyday rou-
 177 tine, relationships with family and friends, and activities/
 178 interests. Post-completion, this project was submitted
 179 for and received IRB exemption for the purposes of ret-
 180 rospective analysis (Approval Number 1-1305771-1). For
 181 more information on contest guidelines, please review
 182 Appendix 1.

183 The contest was announced on Cure SMA’s website
 184 and social media platforms in October 2018 and ran
 185 through January 2019. Targeted e-mail blasts featuring
 186 contest guidelines were disseminated to over 400 fami-
 187 lies of SMA on a biweekly basis, using the Cure SMA
 188 membership database; recruitment reminders were also
 189 posted on Cure SMA’s web and social media pages. The

190 contest submission period was 6 weeks in duration, after
 191 which point all videos were reviewed and approved by
 192 Cure SMA staff for video length and use of appropriate
 193 language, and subsequently uploaded to Cure SMA’s You-
 194 Tube channel. All submitters received an Amazon gift
 195 card, as a thank-you for their participation. The three vid-
 196 eos with the most "likes" received an additional incentive.
 197 Video submissions are still accessible; a full link along
 198 with contest results can be found in Appendix 2. Video
 199 submissions were analyzed for key content themes as
 200 well as percentage of time spent on each theme.

201 In the results detailed below, individuals involved with
 202 the survey will be referred to as ‘respondents’, while indi-
 203 viduals involved with the video contest will be referred
 204 to as ‘participants.’ All individuals quoted will be identi-
 205 fied by their age, gender, and motor function level at the
 206 time of completion, and all quotes cited are directly their
 207 words.

208 **Results**

209 **Quality of life survey**

210 **Participation rates and sample characteristics**

211 The QoL survey elicited eighty-five (85) responses, repre-
 212 senting a 21.3% response rate, with a 95% confidence level
 213 and a 9.4% margin of error. Of the respondents, forty-
 214 four (44) were adolescent minors (ages 12–18, mean age
 215 15), while forty-one (41) were young adults (ages 19–25,
 216 mean age 21). Fifty-three (53) individuals identified as
 217 female, thirty-one (31) as male, and one as non-binary.
 218 The most prevalent maximum motor function level at
 219 both time of diagnosis and at time of survey completion
 220 was sitter/non-independent walker. As respondents were
 221 recruited from the survey through the Cure SMA data-
 222 base, demographic information could be verified. Sample
 223 demographics and clinical characteristics can be found
 224 on Table 1.

225 *What is the most difficult aspect of balancing SMA symp-*
 226 *toms with everyday life?* The first free text question
 227 of the QoL survey addressed the most difficult aspect
 228 of balancing SMA symptoms with the everyday lives of
 229 adolescents and young adults (Table 2, Additional file 1:
 230 Table S1). The highest cited factor overall was dependence
 231 on others and the lack of independence. One respondent
 232 wrote, “At this point I don’t know any different, but it’s the
 233 fact that you always have to rely on someone that starts
 234 to become overwhelming” (Respondent #63; sitter, age 22,
 235 female). The respondents who indicated dependency were
 236 most likely to be sitter/non-walker (p = 0.008).

237 Other highly cited factors included accessibility,
 238 fatigue, and social concerns. Regarding accessibil-
 239 ity, one respondent wrote, “There is an awful lot of
 240 planning that has to go into getting around a huge

Table 1 Quality of life survey demographics

	Total age cohort	Adolescents (12–18)	Young adults (19–24)
Total	85	44 (52%)	41 (48%)
SMA type			
Type I	6 (7%)	4 (5%)	2 (2%)
Type II	56 (66%)	28 (33%)	28 (33%)
Type III	23 (27%)	12 (14%)	11 (13%)
Gender			
Female	53 (62%)	28 (33%)	25 (29%)
Male	31 (37%)	15 (18%)	16 (19%)
Non-binary	1 (1%)	1 (1%)	0
Motor function at time of diagnosis ^a			
Non-independent sitter	25 (29%)	13 (15%)	12 (14%)
Sitter/non-independent walker	37 (44%)	20 (24%)	17 (20%)
Walker	23 (27%)	11 (13%)	12 (14%)
Motor function at time of survey completion*			
Non-independent sitter	31 (37%)	17 (20%)	14 (17%)
Sitter/non-independent walker	40 (47%)	19 (22%)	21 (25%)
Walker	14 (16%)	8 (9%)	6 (7%)

85 individuals responded to the QoL survey. Non-binary was included as a gender option, and describes individuals who identify with no one gender

^a Motor function abilities were divided into three categories: non-independent sitter (head control, maintain seated position supported), sitter/non-independent walker (maintain seated position unsupported, crawl, cruise), and walkers (walk independently). All demographic information was self-identified. All percentages are based on the total surveyed population of 85 respondents

Table 2 Responses to QoL free text question 1

	Age			Motor function			p value
	Adolescents n = 44% (95% CI)	Young Adults n = 41% (95% CI)	p value	Non independent sitter n = 31% (95% CI)	Sitter/non-independent walker n = 40% (95% CI)	Independent walker n = 14% (95% CI)	
Accessibility	15.91 (6.64–30.07)	14.63 (5.57–29.17)	0.87	0	20 (9.05–35.65)	35.71 (12.76–64.86)	0.002*
Advocating	2.27 (0.06–12.02)	2.44 (0.06–12.9)	1	3.23 (0.08–16.70)	0	7.14 (0.18–33.9)	0.147
Balance	6.82 (1.43–18.66)	7.32 (1.54–19.92)	1	3.23 (0.08–16.70)	12.5 (4.2–26.8)	0	0.256
Dependence	22.73 (11.47–37.84)	24.39 (12.36–40.30)	0.857	16.13 (5.45–33.73)	37.5 (22.73–54.20)	0	0.008^
Everyday	9.09 (2.53–21.67)	9.76 (2.72–23.13)	1	9.68 (2.04–25.75)	5 (0.61–16.92)	21.43 (4.66–50.80)	0.151
Fatigue	6.82 (1.43–18.66)	17.07 (7.15–32.06)	0.186	19.35 (7.45–37.47)	7.5 (1.57–20.39)	7.14 (0.18–33.9)	0.258
Finding care	0	9.76 (2.72–23.13)	0.05*	9.68 (2.04–25.75)	2.5 (0.06–13.16)	0	0.374
Mental health	6.82 (1.43–18.66)	0	0.242	6.45 (0.79–21.42)	0	7.14 (0.18–33.9)	0.181
other	6.82 (1.43–18.66)	0	0.242	3.23 (0.08–16.7)	2.5 (0.06–13.16)	7.14 (0.18–33.9)	0.567
Pain	9.09 (2.53–21.67)	7.32 (1.54–19.92)	1	12.9 (3.63–29.83)	7.5 (1.57–20.39)	0	0.451
Social	13.64 (5.17–27.35)	7.32 (1.54–19.92)	0.486	16.13 (5.45–33.73)	5 (0.61–16.92)	14.29 (1.78–42.81)	0.25

Dependence and the lack of independence was the highest-cited difficult aspect, incurring 20 total respondents. Percentages are out of total number of respondents (n = 85). The table values represent statistically significant differences in responses between either the age groups (2 bins) or between the motor function groups (3 bins) using a chi square or Fisher’s exact test. Highlighted findings were found to have statistical significance; asterisk(*) indicates significance by Fisher’s exact test and carrots(^) indicates significance by Chi2 test

241 campus like where I go to school. I worry all the time building and I will slip on the ice, what happens if I 244
 242 about stupid things like whether or not my chair lift fall in the shower, will my smart drive be charged to 245
 243 in my car will work, whether it will snow outside by get me through the day” (Respondent #86, walker, age 246

247 20, female). There was a statistically significant dif-
 248 ference in discussing accessibility by motor function
 249 ($p=0.005$), as the majority of individuals reporting
 250 accessibility had a maximum motor function of inde-
 251 pendent walker. Fatigue was most frequently men-
 252 tioned by individuals with a maximum motor function
 253 status of sitter/non-independent walker than an inde-
 254 pendent walker. One respondent remarked, “Every-
 255 thing is difficult because depending on the day, my
 256 muscles get tired which makes me tired” (Respondent
 257 30, non-independent sitter, age 12, female). Regard-
 258 ing socialization, one respondent wrote, “It’s hard to
 259 always keep up with my friends” (Respondent 20, sit-
 260 ter, age 14, non-binary).

261 Other factors mentioned with less frequency
 262 included the burden of pain and physical symptoms,
 263 finding proper care and finding a healthy life balance.
 264 Multiple non-independent sitting individuals cited
 265 pain as their most difficult aspect of balancing life,
 266 one writing, “Just dealing with the pain.” (Respondent
 267 01, non-independent sitter, age 15, male). There was a
 268 statistically significant difference between adolescents
 269 and young adults citing finding one-on-one care to be
 270 the most difficult aspect of balancing life, with only
 271 young adults citing this factor ($p < 0.05$). Responses are
 272 categorized by age and motor function at the time of
 273 survey completion in Table 2 and the themes by type
 274 in Additional file 1: Table S1.

275 **How does SMA affect schooling?**

276 The second question of the QoL free text inquired about
 277 the impact of SMA and SMA symptoms on schooling
 278 (Table 3, Additional file 1: Table S2). The largest response
 279 category was no effect with 13 responses. Some respond-
 280 ents simply indicated “it doesn’t”, while others elaborated
 281 on their situations. “SMA does not affect my schooling.
 282 I am currently enrolled in college and taking 14 units
 283 on campus” (Respondent 36, sitter, age 18, male). This
 284 response was primarily indicated by individuals with a
 285 maximum motor function of non-independent sitter or
 286 sitter/non-independent walker.

287 Conversely, the other most frequent responses indi-
 288 cated much higher burden. These answers include acces-
 289 sibility, aid and assistance, homeschooling, and fatigue.
 290 Accessibility received 9 responses, mainly comprised of
 291 individuals with various levels of motor function, but
 292 who identified as Type III. One respondent wrote, “I use
 293 an elevator and a rolling backpack, and the other stu-
 294 dents do not” (Respondent 72, walker, age 12, male). Aid
 295 and assistance also received 9 responses. One respondent
 296 wrote, “It forces me to require a 1 on 1 aide” (Respond-
 297 ent 05, non-independent sitter, age 18, male). 8 individu-
 298 als discussed homeschooling, for reasons including germ
 299 exposure and placement struggles. This response was pri-
 300 marily indicated by adolescents and is statistically signifi-
 301 cant by age groups ($p=0.006$). A respondent detailed, “I
 302 stopped going to public school, now I do it at home. They

Table 3 QoL free text responses to question 2

How does SMA affect your schooling?

	Age			Motor function			
	Adolescents n = 44% (95% CI)	Young Adults n = 41% (95% CI)	p value	Non independent sitter n = 31% (95% CI)	Sitter/non- independent walker n = 40% (95% CI)	Independent walker n = 14% (95% CI)	p value
Accessibility	11.36 (3.79–24.56)	9.76 (2.72–23.13)	0.55	6.45 (0.79–21.42)	7.5 (1.57–20.39)	28.57 (8.39–58.10)	0.077
Activity limitation	13.64 (5.17–27.35)	2.44 (0.06–12.9)	0.11	3.23 (0.08–16.70)	15 (5.71–29.84)	0	0.163
Aid	9.09 (2.53–21.67)	12.2 (4.08–26.20)	0.733	12.9 (3.63–29.83)	12.5 (4.2–26.8)	0	0.457
Everyday	11.36 (3.79–24.56)	4.88 (0.60–16.53)	0.435	9.68 (2.04–25.75)	2.5 (0.06–13.16)	21.43 (4.66–50.80)	0.073
Fatigue	2.27 (0.06–12.02)	17.07 (7.15–32.06)	0.026*	6.45 (0.79–21.42)	12.5 (4.2–26.8)	7.14 (0.18–33.9)	0.79
Homeschool	18.18 (8.19–32.71)	0	0.006*	6.45 (0.79–21.42)	12.5 (4.2–26.8)	7.14 (0.18–33.9)	0.79
Keep up	6.82 (1.43–18.66)	9.76 (2.72–23.13)	0.707	9.68 (2.04–25.75)	7.5 (1.57–20.39)	7.14 (0.18–33.9)	1
More effort	4.55 (0.56–15.47)	14.63 (5.57–29.17)	0.147	16.13 (5.45–33.73)	7.5 (1.57–20.39)	0	0.255
No effect	15.91 (6.64–30.07)	14.63 (5.57–29.17)	0.87	16.13 (5.45–33.73)	17.5 (7.34–32.78)	7.14 (0.18–33.9)	0.642
Other	4.55 (0.56–15.47)	7.32 (1.54–19.9)	0.669	9.68 (2.04–25.75)	2.5 (0.06–13.16)	7.14 (0.18–33.9)	0.381
Writing	2.27 (0.06–12.02)	7.32 (1.54–19.9)	0.349	3.23 (0.08–16.70)	2.5 (0.06–13.16)	14.29 (1.78–42.81)	0.217

Although the most common response was ‘no effect’ with 13 total participants, responses were distributed evenly across a variety of factors including accessibility, aid, and fatigue. When categorized by motor function at time of survey completion, sitters were the main respondents of ‘no effect’, while non-sitters more commonly detailed accessibility, aid, and time/effort challenges. Percentages are out of total number of respondents (n = 85). The table values represent statistically significant differences in responses between either the age groups (2 bins) or between the motor function groups (3 bins) using a chi square or Fisher’s exact test. Highlighted findings were found to have statistical significance; asterisk (*) indicates significance by Fisher’s exact test and carrots (^) indicates significance by Chi2 test



303 put me with all of the other special needs kids because
 304 I was in a wheelchair” (Respondent 64, sitter, age 14,
 305 female). Fatigue was also discussed by 8 individuals, this
 306 time primarily indicated by young adults. This finding
 307 is also statistically significant ($p=0.026$) by age groups.
 308 One respondent stated, “Because of my lack of energy,
 309 I could not handle being a full-time student” (Respond-
 310 ent 74, SMA-III, age 23, female). Although these indi-
 311 viduals have faced academic challenges, many of them
 312 report being highly successful. Of the young adult survey
 313 respondents, 16 are currently enrolled in college, with an
 314 additional 12 individuals having graduated college and 3
 315 individuals holding a graduate degree. Responses are cat-
 316 egorized by age and motor function at the time of survey
 317 completion in Table 3 and the themes by type in Addi-
 318 tional file 1: Table S2.

319 **How does SMA affect socialization?**

320 The third free-text question asked how SMA affects
 321 socialization (Table 4, Additional file 1: Table S3). The
 322 highest responses were accessibility and no effect, each
 323 detailed by 16 respondents. Accessibility respondents
 324 once again primarily consisted of individuals with the
 325 maximum motor function of independent walker. One
 326 respondent remarked, “Since I use a wheelchair now, I
 327 have to think about accessibility. Are my friends’ homes
 328 accessible for me? Are the locations of social activi-
 329 ties accessible? I definitely won’t have the same amount

of independence as a sixteen-year-old who doesn’t have
 SMA” (Respondent 72, sitter, age 16, male).

A high rate of respondents indicated no effect, incur-
 ring responses from all levels of motor function. One
 respondent stated, “It does not really affect my social life
 because my friends are cool” (Respondent 06, non-sitter,
 age 14, female). Conversely, some survey respondents
 felt the opposite, with little to no socialization. “What
 social life?” one respondent remarked (Respondent 03,
 non-sitter, age 15, male). Another respondent wrote,
 “SMA affects my social life very much. I never want to
 go out because everyone stares at me everywhere I go”
 (Respondent 19, non-sitter, age 12, female). This answer
 was discussed by 6 respondents of all motor function
 levels.

Other factors impacting socialization include activi-
 ty limitations, communication difficulties, and feeling
 judged or misunderstood. While describing activity limi-
 tations, one respondent wrote, “I am not able to do many
 activities. It affects my social life and my love life (by that
 I mean I don’t have one)” (Respondent 85, walker, age
 22, female). Additionally, one respondent noted com-
 munication difficulties, stating, “People have a hard time
 understanding me” (Respondent 11, non-sitter, age 20,
 male). Regarding feelings of judgement, one participant
 wrote, “Many adults and teens speak to me as though I’m
 much younger or will talk to the person I’m with acting
 as though I’m not really there” (Respondent 47, sitter,
 age 18, female). Dependence as a response in this section

Table 4 QoL free text responses to question 3

	Age			Motor function			
	Adolescents n = 44% (95% CI)	Young Adults n = 41% (95% CI)	p value	Non independent sitter n = 31% (95% CI)	Sitter/non- independent walker n = 40% (95% CI)	Independent walker n = 14% (95% CI)	p value
Accessibility	11.36 (3.79–24.56)	26.83 (14.22–42.94)	0.068	16.13 (5.45–33.73)	22.5 (10.84–38.45)	14.29 (1.78–42.81)	0.708
Activity limitation	18.18 (8.19–32.71)	14.63 (5.57–29.17)	0.659	12.9 (3.63–29.83)	15 (5.71–29.84)	28.57 (8.39–58.10)	0.423
Communication difficulties	4.55 (0.56–15.47)	9.76 (2.72–23.13)	0.349	12.9 (3.63–29.83)	5 (0.61–16.92)	0	0.312
Dating	2.27 (0.06–12.02)	2.44 (0.06–12.9)	1	0	5 (0.61–16.92)	0	0.653
Dependence	6.82 (1.43–18.66)	7.32 (1.54–19.9)	1	0	15 (5.71–29.84)	0	0.04*
Feeling judged	11.36 (3.79–24.56)	0	0.056	3.23 (0.08–16.70)	7.5 (1.57–20.39)	7.14 (0.18–33.9)	0.705
Misunderstood	6.82 (1.43–18.66)	12.2 (4.08–26.20)	0.474	19.35 (7.45–37.47)	2.5 (0.06–13.16)	7.14 (0.18–33.9)	0.056
No effect	27.27 (14.96–42.79)	9.76 (2.72–23.13)	0.053	19.35 (7.45–37.47)	15 (5.71–29.84)	28.57 (8.39–58.10)	0.533
No social life	9.09 (2.53–21.67)	4.88 (0.60–16.53)	0.677	12.9 (3.63–29.83)	2.5 (0.06–13.16)	7.14 (0.18–33.9)	0.209
Other	2.27 (0.06–12.02)	12.2 (4.08–26.20)	0.102	3.23 (0.08–16.70)	10 (2.79–23.66)	7.14 (0.18–33.9)	0.542

‘No effect’ on socialization was most commonly cited by non-independent sitter, though it was followed closely by major effects including accessibility concerns and an absence of socialization. Percentages are out of total number of respondents (n = 85). The table values represent statistically significant differences in responses between either the age groups (2 bins) or between the motor function groups (3 bins) using a chi square or Fisher’s exact test. Highlighted findings were found to have statistical significance; asterisk (*) indicates significance by Fisher’s exact test and carrots (^) indicates significance by Chi2 test

359 once again obtained statistical significance by motor
 360 function categories, with all respondents being sitter/
 361 non-independent walker ($p=0.04$) rather than independ-
 362 ent walker. Responses are categorized by age and motor
 363 function at the time of survey completion in Table 4
 364 and the themes by type in Additional file 1: Table S3,
 365 respectively.

366 **What resources should exist for teens and young adults**
 367 **with SMA?**

368 The final question of the QoL free text asked respond-
 369 ents what resources they would like to see created for
 370 this specific SMA population (Table 5, Additional file 1:
 371 Table S4). Most often mentioned, 33 responses, across
 372 all ages and levels of motor function, suggested the crea-
 373 tion of peer support groups. One respondent elaborated,
 374 “A group where we can just talk to each other about the
 375 issues we have, how we deal with them, joke with each
 376 other, and just connect over it” (Respondent 44, sitter,
 377 age 18, male). Another respondent remarked, “Support
 378 groups to help cope with the emotional and mental toll
 379 of SMA” (Respondent 53, sitter, age 22, female). Separate,
 380 but closely related, was the idea of a big/little buddy sup-
 381 port program. One respondent wrote, “It would be use-
 382 ful to pair young adults with older individuals with SMA,
 383 like a buddy to learn from” (Respondent 13, non-sitter,
 384 age 23, female).

385 Another frequent suggestion was the creation of col-
 386 lege and independent living transition materials. This
 387 response incurred 11 responses of all motor function lev-
 388 els. One respondent stated, “More info on going to col-
 389 lege independently; as an adult I have met others with
 390 SMA who were surprised I went to college and lived
 391 independently” (Respondent 52, sitter, age 25, female).
 392 Other ideas included improved access to equipment and
 393 technology, accessible transportation maps and activities,
 394 and accessible video games. Regarding improved access
 395 to equipment and technology, one respondent wrote,
 396 “Help with better access. Help to get a van for using
 397 power chair away from home and school. Grants for
 398 families” (Respondent 69, sitter, age 18, female). Regard-
 399 ing accessible activities, respondents suggested the crea-
 400 tion of a wheelchair accessible waterpark, video game
 401 console and sport specifically modified to fit the needs of
 402 all SMA severities. Lastly, a few respondents wrote that a
 403 cure would be an excellent resource. Responses are cat-
 404 egorized by age and motor function at the time of survey
 405 completion in Table 5 and the themes by type in Addi-
 406 tional file 1: Table S4.

407 **Awareness video contest**

408 The video contest received six submissions: four from
 409 adolescent minors and two from young adults. Five of
 410 the six individuals identified as type II; the remaining
 411 participant was a young adult with type III. Ambulatory

Table 5 QoL free text responses to question 4

What resources do you think should exist for teens and young adults with SMA?

	Age			Motor function			p value
	Adolescents n = 44% (95% CI)	Young Adults n = 41% (95% CI)	p value	Non independent sitter n = 31% (95% CI)	Sitter/non- independent walker n = 40% (95% CI)	Independent walker n = 14% (95% CI)	
A Cure	9.09 (2.53–21.67)	0	0.117	0	5 (0.61–16.92)	14.29 (1.78–42.81)	0.116
Accessible activities	6.82 (1.43–18.66)	0	0.242	6.45 (0.79–21.42)	0	7.14 (0.18–33.9)	0.181
Accessible transit	0	7.32 (1.54–19.92)	0.108	6.45 (0.79–21.42)	2.5 (0.06–13.16)	0	0.755
Big/little support	6.82 (1.43–18.66)	2.44 (0.06–12.9)	0.617	3.23 (0.08–16.70)	7.5 (1.57–20.39)	0	0.654
Caregivers	0	7.32 (1.54–19.92)	0.108	6.45 (0.79–21.42)	2.5 (0.06–13.16)	0	0.755
College/independ- ence materials	0	100.00%	0*	12.9 (3.63–29.83)	12.5 (4.2–26.8)	14.29 (1.78–42.81)	1
Equipment/technolog- y	18.18 (8.19–32.71)	4.88 (0.60–16.53)	0.057	6.45 (0.79–21.42)	15 (5.71–29.84)	14.29 (1.78–42.81)	0.591
Other	2.27 (0.06–12.02)	4.88 (0.60–16.53)	0.607	3.23 (0.08–16.70)	5 (0.61–16.92)	0	1
Support groups	40.91 (26.34–56.75)	36.59 (22.12–53.06)	0.683	38.71 (21.85–57.81)	42.5 (27.04–59.11)	28.57 (8.39–58.10)	0.655
Unknown	6.82 (1.43–18.66)	9.76 (2.72–23.13)	0.707	9.68 (2.04–25.75)	5 (0.61–16.92)	14.29 (1.78–42.81)	0.516
Video games	9.09 (2.53–21.67)	0	0.117	6.45 (0.79–21.42)	2.5 (0.06–13.16)	7.14 (0.18–33.9)	0.502

The overwhelming majority response to Question 4 was peer-support groups. Question 4 exhibited the largest group consensus. Percentages are out of total number of respondents (n = 85). The table values represent statistically significant differences in responses between either the age groups (2 bins) or between the motor function groups (3 bins) using a chi square or Fisher's exact test. Highlighted findings were found to have statistical significance; asterisk (*) indicates significance by Fisher's exact test and carrots (^) indicate significance by Chi2 test

412 status was assessed from the video submissions. Please
 413 see Table 6 for full demographic information.

414 Although the video contents had a much smaller sam-
 415 ple size, the findings align with those of the QoL survey.
 416 Video content was divided into 6 subthemes discussed by
 417 all participants: social engagement, everyday life, physical
 418 health, emotional health, other obstacles and overcom-
 419 ing obstacles (Table 6). Of note, all video submissions
 420 discussed social engagement in detail, with participants
 421 spending a minimum of 20% of their submission time
 422 on the topic. Participants highlighted the importance of
 423 friendships, as well as the importance of family members,
 424 pets, paras and other key relationships. Regarding the
 425 impact of SMA on socialization, once again participants
 426 spoke to both ends of the spectrum and the high level of
 427 impact it had on their emotional well-being. Once partic-
 428 ipant said, “I have a few really close friends, and they’re
 429 absolutely my best friends. I think that we’re closer than
 430 someone who wasn’t in a wheelchair because they walk
 431 through SMA with me” (Participant 01, SMA-II, age 15,
 432 female). Conversely, another discussed, “I realized that
 433 not being included was much harder than it seemed to
 434 be. It’s not that I was bullied, but that no one wanted me
 435 to be their friend.” (Participant 06, SMA-II, age 17, male).

436 Submissions highlighted fatigue and feeling unable to
 437 participate in everyday life as difficult aspects of man-
 438 aging SMA. One participant detailed the difficulty of
 439 transitioning from a fully-independent walker to need-
 440 ing assistive support. “It’s really hard to get up and down
 441 stairs, to bring my walker places, to need friends to wait
 442 or slow down. I never had to think about any of it before.
 443 It’s a challenge I overcome, but I did not think it would
 444 be so soon” (Participant 04, SMA-III, age 22, female).
 445 Additionally, participants spoke to both ends of the spec-
 446 trum while describing the impact of SMA on school-
 447 ing. One participant noted, “I needed an aid as well as
 448 many other accommodations, which resulted in a fair
 449 amount of arguments with people who just don’t under-
 450 stand” (Participant 03, SMA-II, age 18, female) while

451 another detailed, “I don’t want to be known as the girl
 452 in the wheelchair, I want to be known as this cool teen-
 453 age girl. I’m really into student government, yearbook,
 454 French club, Humane society...” (Participant 02, SMA-II
 455 age 15, female). While the video submissions did not dis-
 456 cuss specific resource recommendations, an inspiration
 457 for quite a few participants to create videos was to be a
 458 role model for struggling members of the Cure SMA and
 459 disabled adolescent communities. One participant said,
 460 “I hope to be an inspiration to all the kids out there that
 461 grew up like me, who are in wheelchairs and have self-
 462 doubt.” (Participant 05, SMA-II, age 20, female).

463 One element of the video contest that was not covered
 464 in the QoL survey is overcoming adversity. Each video
 465 participant highlighted challenges they experienced
 466 while also discussing their motivations and strategies of
 467 resilience. One participant said, “I keep a blog and make
 468 YouTube videos to show you can have a disability and still
 469 have a life that’s full and fulfilling. I hope to be an inspira-
 470 tion to all the kids out there that grew up like me, who
 471 are in wheelchairs and have self-doubt.” (Participant 05,
 472 age 20, female). Another participant detailed, “My family
 473 and I, we love to get out and travel. We don’t let inacces-
 474 sible routes or people tell us ‘You can’t go that way.’ Usual-
 475 ly, we figure out a way to do it. There are challenges, but
 476 there are ways to help, that’s what SMA is all about” (Par-
 477 ticipant 01, SMA-II, age 15, female). For more informa-
 478 tion on the topics discussed in the video contest, please
 479 refer to Table 7.

Impact on mental health

480 Across the platforms, mental health was discussed by
 481 twelve different survey respondents. When detailing dif-
 482 ficult obstacles, one survey respondent elaborated, “I was
 483 just diagnosed with depression and anxiety mostly from
 484 the constant worrying about my life and having SMA,
 485 I think that in some cases having SMA makes me want
 486 to work harder and prove others wrong, but at the same
 487 time there are a lot of days where I just want to give up
 488 and say what’s the point” (Respondent 86, walker, age
 489 20, female). Other respondents were more succinct but
 490 equally impactful. One respondent wrote in the difficult
 491 obstacles question, “Depression. It makes everything
 492 worse and less bearable” (Respondent 09, SMA-II, age 16,
 493 female). Survey respondents also discussed the emotional
 494 impact of being considered disabled simply by having a
 495 wheelchair. In the socialization question, one respondent
 496 explained, “I feel like people judge me just by seeing the
 497 wheelchair. People treat me like I’m a baby or mentally
 498 disabled or too innocent and I’m none of those things!”
 499 (Respondent 61, sitter, age 19, female). Another respond-
 500 ent wrote, “It makes me sad when people stare at me. I
 501

Table 6 Video contest demographics

Awareness video contest demographics				
	Age	Gender	Type	Ambulatory status
Respondent 1	15	Female	II	Non-ambulant
Respondent 2	15	Female	II	Non-ambulant
Respondent 3	17	Female	II	Non-ambulant
Respondent 4	22	Female	III	Ambulant
Respondent 5	20	Female	II	Non-ambulant
Respondent 6	17	Male	II	Non-ambulant

6 individuals participated in the video contest

Table 7 Video contest topic breakdown

Awareness video contest topic breakdown (n=6)			
Theme	% of participants addressing theme	Subtheme	Sample quote
Social engagement	100	Friendship	"My friends treat me the same way they treat anyone else, that's why we're best friends"
	100	Key relationship (family, pet, helper)	"My brother is a runner and he runs for me"
	50	Lack of social engagement	"No one invited me to be their friend"
Everyday life	100	Schooling	"With school, I have to have a full-time para"
	100	Activities	"One of my passions is the arts, I like to paint"
	83.3	Everyday routine	"I still do chores, I still help out around the house"
Physical health	100%	Mobility	"A wheelchair can't go that way"
	100%	Overall disease burden	"I have all the physical demands of being disabled"
Emotional health	50	Frustration	"It's hard to go upstairs, and I get mad at myself sometimes when I can't do it"
	33.3	Depression	"Realizing how my peers treated me, depression overcame me"
Other obstacles	100	Dependence	"I have to have someone do everything for me"
	83.3	Being labeled/treated "disabled"	"Stop with these misconceptions that everyone who is disabled is mentally slow or just sitting around"
Overcoming obstacles	100	Defying other's expectations	"Just because you can't physically do something doesn't mean you can't follow your dreams"
	100	Accepting + adapting	"I've learned to adapt and teach myself to write with both hands"

Contest participants discussed various topics regarding life as adolescents and young adults living with SMA. A sample of direct quotes from each topic is shown here

502 know they're probably just 'curious,' but still, it makes me
 503 upset" (Respondent 18, non-sitter, age 18, female).

504 In the awareness video contest, mental health issues
 505 were discussed in-depth by two participants. One partici-
 506 pant elaborated, "After losing my aide of three years and
 507 realizing how I was being treated by my peers, depression
 508 overcame me. I could no longer stay well" (Participant 03,
 509 SMA-II, age 18, female). Another said, "As soon as middle
 510 school started, I was thrown into a deep depression, and
 511 I still have not fully recovered...It is the physical demands
 512 of being disabled that would eventually shape my person-
 513 ality to being an introvert." (Participant 06, SMA-II, age
 514 17, male).

515 **Successes and accomplishments**

516 Although not directly addressed in either incitive, it
 517 is evident that many adolescents and young adults
 518 affected by SMA are leading productive lives despite
 519 challenges. In the schooling section of the QoL survey,
 520 a few individuals highlighted their academic accom-
 521 plishments. One survey respondent wrote, "Gradu-
 522 ated from the Wharton School at the University of

Pennsylvania" (Respondent 29, sitter, age 23, male) 523
 while another wrote, "I get As and Bs" (Respondent 06, 524
 non-sitter, age 14, female). Many survey respondents 525
 also emphasized their interests outside of their disease, 526
 such as video games, swimming and wheelchair sports. 527
 In response to the question regarding socialization, one 528
 respondent wrote, "I still go dancing!!!!!" (Respondent 529
 68, sitter, age 23, female). 530

531 Similar successes were also emphasized in the aware-
 532 ness video contest. One video contest participant high-
 533 lighted their future plans. "After I finish my degree, I plan
 534 on working with children who are similar to me, hope-
 535 fully inspiring them to do what they want to do with
 536 their lives. Just because you can't physically do something
 537 doesn't mean that you can't follow your dreams" (Partici-
 538 pant 05, SMA-II, age 20, female). All contest participants
 539 detailed their interests, ranging from clubs to YouTube,
 540 attending concerts, and traveling. Additionally, many
 541 participants highlighted unique experiences. One par-
 542 ticipant reflected, "If you want to live twenty hours away
 543 from your parents, go for it. If you want to go hunting
 544 for [sic] your dad, do it. If you want to go skydiving, why

545 not? My only advice is, don't be afraid of a challenge. Be
546 excited for it" (Participant 06, age 17, male).

547 **Discussion**

548 In comparing our population to the SMA adult popula-
549 tion captured in previous publications, our top responses
550 coincide with their key findings. Both accessibility and
551 fatigue were reported as significant in multiple questions.
552 Accessibility was a high response in our study in rela-
553 tion to schooling and socialization. This correlates with
554 the identification of accessibility and limited mobility as
555 symptoms with the greatest impact on SMA adults in the
556 SMA Patient-Reported Impact of Symptoms (PRISM-
557 SMA) study (n = 359) [18]. Fatigue is also often identified
558 as clinically meaningful, and it is highlighted both in the
559 above study as well as in our findings in relation to great-
560 est challenge and schooling. Furthermore, dependence
561 and the lack of independence was found to be a difficult
562 challenge in our study in correlation to motor function,
563 and the existing literature. Achieving greater independ-
564 ence with treatment was considered to be extremely sig-
565 nificant, and therefore, clinically meaningful to parents/
566 caregivers as well as affected adults [8, 14]. Similarly,
567 results from a global polling with SMA parents/car-
568 egivers and affected adults at an externally led Patient-
569 Focused Drug Development Meeting corroborated the
570 finding that any treatment that increases independence is
571 acutely meaningful [17].

572 **Impact of age and motor function**

573 When breaking our population into cohorts of adoles-
574 cents (ages 12–18) and young adults (ages 19–25), it is
575 evident that certain themes are most relevant to certain
576 age groups versus others. Only young adults requested
577 the creation of college and independent transition mate-
578 rials and young adults also more frequently reported
579 fatigue issues in school and issues finding care. Con-
580 versely, adolescents were more likely to report being
581 homeschooled. With these differences noted between
582 age groups, majority of the key factors impacting the day
583 to day lives of adolescents and young adults were largely
584 shared by both groups.

585 There was a statistically significant difference between
586 maximum motor function and identifying accessibility
587 and dependence as the most difficult aspect of balancing
588 SMA; accessibility concerns were mainly voiced by par-
589 ticipants who previously walked independently or were
590 able to walk independently at the time of survey com-
591 pletion. While accessibility was a main issue for these
592 individuals, the sentiment was not shared by those with
593 a maximum motor function of non-independent sitters,
594 who more frequently detailed pain or communication
595 difficulties. Non-walkers were the most likely to voice

596 challenges regarding dependence or reliance on others,
597 and these results were significant with respect to sociali-
598 zation and considered a difficult aspect of disease man-
599 agement. While motor function status was correlated
600 with some responses, many responses had no correlation.
601 At least one individual of every motor function group
602 discussed difficulties regarding mental health, feeling left
603 out, or needing more time and effort in school. Addition-
604 ally, the no effect responses detailed in schooling and
605 socialization all incurred varied responses from all levels
606 of motor function.

607 **No effect vs severe effect of SMA on schooling
608 and socialization**

609 In both the schooling and socialization questions of the
610 free text, a parabola effect was observed in high fre-
611 quency answers. A similar number of respondents felt
612 that either, SMA greatly affected, or had no effect on
613 their schooling or socialization, two significant aspects
614 of life in this age period. The majority of those who felt
615 that SMA did not impact schooling did not overlap
616 with individuals who felt that SMA did not impact their
617 socialization. It is important to note that neither of these
618 questions received a large group consensus unlike the
619 categories of everyday life or resources, with similar fre-
620 quencies reported amongst many responses.

621 **Impact on mental health**

622 Although mental health was not the most frequently
623 cited factor in any category, the impact of SMA on men-
624 tal health can be severe [16, 17, 19, 20]. Similar concerns
625 regarding mental health have been observed in both the
626 adult SMA population as well as the larger adolescent
627 population. It is estimated that approximately one in five
628 adolescents struggles with mental health or a diagnos-
629 able mental health disorder [22]. Additionally, in a poll
630 conducted among caretakers and individuals with SMA,
631 during the SMA Patient Focused Drug Development
632 Meeting with the FDA, 105 adults with SMA reported
633 having experienced anxiety (79%), depression (61%), and
634 social isolation (63.8%) as a result of coping with SMA
635 and SMA symptoms [17]. Furthermore, in a series of 25
636 interviews conducted with SMA-affected young adults
637 and adults in Australia, mental health was highlighted as
638 a major unmet need, particularly in stressful times related
639 to SMA symptoms including loss of function, social iso-
640 lation, stigma and questions of self-worth [20]. While this
641 impact was not found to affect all individuals, those who
642 were affected described being severely impacted, noting
643 effects ranging from withdrawal from mainstream school
644 to ongoing battles with depression. Within our survey,
645 amongst those respondents that indicated mental health

646 concerns, there were no similarities in responses to other
647 questions.

648 The specific psychosocial and developmental chal-
649 lenges observed in our research are similar to those
650 found in the larger adolescent and young adult popula-
651 tions. These population archetypes struggle with feeling
652 left out at school, navigating adulthood, and finding inde-
653 pendence [22]. Our results indicate that SMA affected
654 adolescents and young adults experience typical age-
655 demographic challenges compounded by the burden of
656 coping with and managing a significant disability.

657 **Overcoming adversity**

658 As discussed in the corresponding results section, most
659 of the SMA adolescent and young population surveyed
660 are leading successful and productive lives. The resilient
661 spirit exhibited in our findings complements existing
662 publications in which adversity, resilience and grit were
663 found to be key components of adolescence and young
664 adulthood with SMA affected individuals. Study partici-
665 pants described strategies including focusing on abilities
666 rather than disabilities, and prioritizing self-worth and
667 value [19].

668 **Potential impact of peer support groups**

669 A peer support group was the most requested and most
670 desired resource to support this population. Such a
671 resource is not uncommon for disease populations. In a
672 July 2013 publication from Patient Education and Coun-
673 seling, researchers found that while there were benefits
674 to both mentor/mentee programs and age-matched sup-
675 port groups, the latter had the greatest potential for sym-
676 metrical and mutually beneficial relationships leading to
677 greater emotional support [23]. This is consistent with
678 our findings, although a mentoring buddy program was
679 requested, the requests for a peer support program were
680 much greater.

681 Furthermore, peer support groups are common
682 resources for this age demographic within and outside
683 of the rare disease community. In a 2015 study regarding
684 cystinosis support groups for emerging adults (n=49),
685 participants reported valuing the connection and sup-
686 port they received from those who understand their lived
687 experience [24]. Additionally, in a 2018 study involving
688 a literature review regarding 15 studies on the impact
689 of peer support groups on adolescents struggling with
690 mental health, support groups were shown to be positive
691 influences in the management of short-term and long-
692 term problems [22]. These articles are consistent with our
693 population's interest and demonstrate the strong poten-
694 tial for benefit were this resource to be created. Undoubt-
695 edly, teens and young adults with SMA view peer support

696 groups as preferred mechanism to cope with the reported
697 isolation they experience as a result of their disease.

698 **Study limitations**

699 There are several important limitations to note with
700 our research. Although the size of our population is a
701 strength, there is a low participation of type I individu-
702 als. Traditionally, most of those affected with SMA type I
703 do not survive past the age of two and those who do sur-
704 vive present with a very severe phenotype (requiring res-
705 piratory and feeding support and little to no movement)
706 that would make direct participation in these studies very
707 challenging. Furthermore, there is a greater number of
708 females than males in our study. This limitation makes it
709 difficult to summarize our findings to the greater SMA
710 population, as it does not represent the entire popula-
711 tion. Additionally, because this was a qualitative research
712 study using open ended responses, rather than a validated
713 patient reported outcome measures with prior usage in
714 the SMA population, predications about possible differ-
715 ences in responses between sub-populations were not
716 feasible, making a priori power calculations impossi-
717 ble. Instead, confidence intervals were calculated when
718 comparing sub-populations to provide indication of sta-
719 tistical precision. Furthermore, as the survey is directly
720 patient-reported, there may have been mis-reported data
721 on SMA type and other demographic information. Some
722 information that could impact results, such as mental
723 health status, economic status, and motor function gain/
724 loss was not collected. Lastly, the awareness video con-
725 test response rate is hypothesized to be much lower than
726 the survey response rate due to the high demands of the
727 contest design, including creating and editing a video
728 submission, publicly posting to YouTube, and the subse-
729 quent contest campaigning. Although the small cohort
730 is not intended to represent the entire SMA population,
731 their anecdotes and insights are intended to supplement
732 a fuller picture of this age cohort and largely align with
733 the findings of the larger QoL free response findings.

734 **Implications and future directions**

735 The findings of this research corroborate previous
736 findings on disease burden and quality of life in adults
737 and caregivers with SMA [14–16, 19]. These find-
738 ings also point to gaps and tangible opportunities to
739 guide patient organizations and other key SMA stake-
740 holders to generate important community and school
741 resources that may help to improve the experience of
742 teens and young adults living with SMA. For instance,
743 the consensus and enthusiasm for a peer support group
744 was unmatched and demonstrates a clear need in
745 this community. Equally salient is the creation of col-
746 lege and independent living transition materials and

747 information about accessibility in schools and college
 748 campuses. Additionally, these studies provide a per-
 749 spective captured for the first time specifically on teens
 750 and young adults with SMA, along key dimensions that
 751 impact the quality-of-life in this population, at a unique
 752 point in time with the recent approval by the FDA of
 753 therapies for SMA. There is much more to be explored,
 754 both regarding the current generation as well as future
 755 adolescents receiving transitional care in the land-
 756 scape of SMA’s changing phenotypes. Future awareness
 757 efforts are needed to combat the stigma that comes
 758 with a disability, and the barriers regarding accessibil-
 759 ity, to ensure this population fully experiences the best
 760 life they possibly can. Lastly, developing programs that
 761 support optimal functioning and mental health among
 762 teenagers and young adults living with a complex dis-
 763 ease is of utmost importance.

764 **Conclusion**

765 As SMA treatments and care expand, more and more
 766 SMA-affected children will live to become teenagers
 767 and young adults with bright futures ahead. It is impera-
 768 tive to understand the current life experience in order
 769 to improve the quality-of-life for this and future gener-
 770 ations. It is evident that this population has a big voice
 771 with valuable insights, and no one understands the
 772 needs of the SMA community better than those directly
 773 affected. Despite all of the challenges and complexi-
 774 ties that come with living with SMA as adolescents and
 775 young adults, these teens are living full and successful
 776 lives. Indeed, via this work, we have learned that these
 777 individuals have SMA but SMA does not have them.

778 **Supplementary Information**

779 The online version contains supplementary material available at <https://doi.org/10.1186/s13023-021-01701-y>.

781 **Additional file 1.** contains Supplementary Tables I-IV, denoting the qual-
 782 ity of life survey questions’ key words and response breakdown by SMA
 783 type.

784 **Acknowledgements**

785 The authors would like to thank Angela Paradis for her advice and expertise
 786 on manuscript feedback. The authors and Cure SMA also express sincere grati-
 787 tude to every survey respondent and video contest participant for their time,
 788 efforts, and candid honesty. Their insightful perspectives are instrumental in
 789 understanding the lived experiences and quality-of-life of adolescents and
 790 young adults living with SMA in the United States.
 791

792 **Authors’ contributions**

793 AM, MC and RC developed and conducted the Clinical Meaningfulness
 794 Survey and Awareness Video Contest. AM, MC, and LB analyzed survey and
 795 video contest results and findings. JJ provided guidance and oversight for all

activities. AM, MC, LB, RC, and JJ contributed to authorship of the manuscript. All authors have read and approved the final manuscript. 796 797

Funding

This study was financially supported by the SMA Industry Collaboration with funding to Cure SMA. During the project period, members of the SMA Industry Collaboration included Novartis Gene Therapies, Biogen, Genentech/Roche Pharmaceuticals, Novartis Pharmaceuticals, Astellas, Cytokinetics, and Scholar Rock. 798 799 800 801 802 803

Availability of data and materials

Additional data is available from the authors upon request. 804 805

Ethics approval and consent to participate

The clinical meaningfulness survey was approved by a US Institutional Review Board (Approval Number 1-11240001-1). Written informed consent was obtained from all adult respondents prior to the conduct of any research related activities. For minor adolescent respondents (ages 12–18), written consent from a parent/guardian was obtained, as well as the survey respondent’s assent thereafter. Regarding the awareness video contest, the project was submitted for and received IRB exemption for the purposes of retrospective analysis (Approval Number 1-1305771-1). All contest participants consented to publication and posting their videos on the Cure SMA YouTube page. 806 807 808 809 810 811 812 813 814 815

Consent for publication

All survey respondents and video contest participants. All participants provided written consent for participation in the study and the publication of the findings prior to research participation. 816 817 818 819

Competing interests

AM, MC, LB, RC, and JJ are employees of Cure SMA and report grants for the Industry Collaboration, which provided funding for this project. The support received to conduct this study was noted and it does not result in a conflict of interest for any of the authors. The authors have indicated that they have no other conflicts of interest regarding the content of this article. Study sponsor did not participate in study design, collection, data analysis or interpretation of data or writing of the manuscript, it was the decision of Cure SMA to submit this manuscript for publication. 820 821 822 823 824 825 826 827 828

Appendix 1: Cure SMA teen video contest guidelines

Cure SMA is hosting a video contest to raise SMA awareness and provide the opportunity for teens and young adults (aged 12–21) to share their experiences. We want you to participate!! 829 830 831 832 833

The purpose of this contest is to raise awareness about the impact that SMA has on teens and young adults, and the strategies you use to help navigate your day to day life. Your voice is crucial. Your video will help to increase awareness of the disease. We hope that your peers can learn from you! 834 835 836 837 838 839

Who can enter? 840

- You must have a diagnosis of Spinal Muscular Atrophy (SMA) 841 842 843
- You must be 12–21 years old 844
- You must live in the United States 845
- You may work individually or as a group (up to 3 individuals), but one person must apply and represent on behalf of the group. This individual must have a prior diagnosis of SMA 846 847 848 849

850	• Parents and adults can be advisors, but we respectfully request the teen/young adult be the primary driver	893
851		894
852		895
853	• Parental permission is required for contestants under 18 years old	896
854		897
855	• Immediate family and staff of Cure SMA are ineligible to submit	898
856		899
857	How to enter?	900
858	• We want to know more about all of the creative techniques you utilize to remain an active teenager or young adult. Create a short (3 min or less) video that highlights the following:	901
859		902
860		903
861		904
862	1. How does SMA impact your goals as a teenager/young adult as you deal with the issues that others your age face including (but not limited to):	905
863		906
864		907
865	a. School/work	908
866	b. Friendships	
867	c. Independence	
868	d. Dating	
869	In addition to item #1 above, all videos must also include one or more of the following topics.	
870		
871	2. What care/treatment options are the most effective for you as you manage your symptoms? (physical therapy, hydrotherapy, massage therapy, breathing assistance, nutrition etc.) Please refrain from mentioning any drug name or clinical trial; videos including this information will be disqualified from the contest.	
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878	3. Even with the most well thought out plan, life gets in the way! What challenges do you face along the way and how do you manage?	
879		
880		
881	4. In addition to treatment, do you have additional strategies that contribute to your daily joy/improved quality of life? (dog companion, helper, devices, treatments, etc.)	
882		
883		
884		
885	• Upon completion, please submit your video, via email, to CREATE EMAIL ADDRESS. All videos will be reviewed prior to Cure SMA staff posting the content to the Cure SMA's YouTube® account. Participants under the age of 18 years of age must have parental consent. A signed consent form along with a copy of identification is required with each submission; those over the age of 18 must also consent and	
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	provide a copy of identification. Please contact Mary Curry at mary.curry@curesma.org or 847.264.1189. Notification of acceptance will be provided via email along with a direct link to the uploaded content.	910
	• Individuals or groups may submit more than one video as long as each video covers different topics. Please see above for the list of subject areas.	911
	• Winners will be chosen based on total "Likes" obtained by each video; every "Like" equals one vote. All participants are encouraged to share their video with friends and family members to increase the total number of votes received. Only videos submitted to Cure SMA and uploaded to Cure SMA's YouTube® account will be considered.	912
	• Make sure you read and approve the terms and conditions. Easy!!	913
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		922
	Exclusion criteria.	923
		924
	• All entrants 18 years of age and older are required to submit a signed consent and photo ID via email to Mary Curry at mary.curry@curesma.org; entrants under the age of 18 year of age are required to obtain permission from all parents and/or legal guardians as well as a copy of a photo ID. Those that do not comply are excluded from participation.	925
	• All videos will be reviewed for content prior to Cure SMA's acceptance of video entry. All videos that do not feature the required subjects will be disqualified. Any videos that mention any drug name or clinical trial will be disqualified. The participant will be notified via email if disqualified.	926
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	Timeline is as follows:	
	Submission Period: October 15, 2018 to November 20, 2018.	
	Cure SMA Video Upload Date: November 28, 2018.	
	Time Period to Market Your Film: November 28, 2018 to December 9, 2018.	
	Announcement of Finalists and Grand Prize Winner: December 21, 2018.	
	Prizes:	
	Winners will be chosen based on the total "Likes" obtained on their video between November 28, 2018 at 12:00 am CST to December 9, 2018 11:59 pm CST.	
	Prizes will be awarded as follows:	
	1st Place: Amazon Gift Card for \$150.	
	2nd Place: Amazon Gift Card for \$125.	
	3rd Place: Amazon Gift Card for \$100.	
	Winners will be notified by e-mail.	
	Terms and Conditions.	
	For optimal video upload guidelines visit the YouTube® website.	

943 Participants of Cure SMA's Video Contest agree to
 944 YouTube's® Community Guidelines (<https://www.YouTube.com/yt/about/policies/#community-guidelines>)
 945 and Terms of Service (<https://www.YouTube.com/static?gl=US&template=terms>). YouTube® is not a sponsor
 946 of your contest and all participants are required to release
 947 YouTube® from any and all liability related to this contest.
 948

949 Entrants may only submit their own original works.

950 All submitted videos are subject to approval and may
 951 not be accepted if they are found to be offensive or in vio-
 952 lation of copyright. This includes the use of copyrighted
 953 music, digital, movie, or printed works. See "Acceptable
 954 Site Use" in ACS Terms of Use.
 955

956 Cure SMA reserves the right to use submitted vid-
 957 eos on the Cure SMA website and in other promotions,
 958 including but not limited to the Cure SMA channel on
 959 YouTube®, Cure SMA's SMARt Moves microsite, and the
 960 Cure SMA publications. Cure SMA and the organiza-
 961 tion's industry partners are release from any and all liabil-
 962 ity related to this contest.

963 Any personal information supplied by you to Cure
 964 SMA will be subject to Cure SMA's privacy policy posted
 965 at <http://www.curesma.org/privacy-policy.html>. By
 966 entering the Contest, the Entrant/Parent/Legal Guard-
 967 ian grants Cure SMA permission to share your E-mail
 968 address and any other personally identifiable information
 969 with You Tube or with any co-sponsor solely for the pur-
 970 pose of administration and prize fulfillment. Cure SMA
 971 will not sell, rent, transfer or otherwise disclose your
 972 personal data to any third party other than as described
 973 above herein or in the privacy policy.
 974

975 Please contact Mary Curry at mary.curry@curesma.org
 or 847.264.1189 with any questions.

976 **Appendix 2: Cure SMA teen video link and results**



977 Cure SMA recently hosted a video contest to raise SMA
 978 awareness and provide the opportunity for teens and
 979 young adults (aged 12–21) to share their experiences. The
 980 purpose of this contest is to raise awareness about the

981 impact that SMA has on teens and young adults, and the
 982 strategies used to help navigate day to day life.

983 Special thanks to everyone who participated in our
 984 contest and helped to make it a success! Thank you for
 985 sharing your journey. We continue to learn from each of
 986 your stories.

987 Although the contest has ended, please continue to
 988 view and share all of the contest entries on Cure SMA's
 989 YouTube Channel at https://www.youtube.com/watch?v=u9baqO4lno8&list=PLQVcp9RApBmz3QN_CBFXGUAWeoTlmKMn&index=2
 990
 991

992 Congratulations to all the participants and thank you to
 993 all that voted!
 994

995 Received: 27 July 2020 Accepted: 20 January 2021

996 Published online: 22 February 2021

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