LOKMAN HEKIM HEALTH SCIENCES

DOI: 10.14744/lhhs.2025.48841
Lokman Hekim Health Sci 2026;6(1):27–37

ORIGINAL ARTICLE

lokmanhekimhs.com

Awareness and Attitudes Toward Spinal Muscular Atrophy Carrier and Newborn Screening Programs in Türkiye: A Cross-Sectional Study in Family Health Centers

Cansu Özbaş, Cansu Gingir, Asiye Uğraş Dikmen

Department of Public Health, Gazi University Faculty of Medicine, Ankara, Türkiye

Abstract

Introduction: Spinal Muscular Atrophy (SMA) is an autosomal recessive genetic disorder characterized by progressive loss of motor neurons. Both premarital carrier screening and newborn screening programs were recently implemented in Türkiye. This study aims to assess public knowledge, awareness, and attitudes toward SMA and screening programs, as well as to examine factors influencing these attitudes.

Methods: This cross-sectional study was conducted through a face-to-face survey in the individuals who visited various Family Health Centers in Ankara. The questionnaire included items on sociodemographic information, knowledge, attitudes, and behaviors regarding SMA and its screening. The data obtained were presented through descriptive analyses, and the factors influencing knowledge, attitudes, and behaviors regarding SMA and screening were examined. Univariate and multivariate logistic regression analyses were then conducted, using "hypothetical acceptance to undergo SMA screening" as the dependent variable.

Results: The study included 1643 individuals. The majority of participants reported having never heard of SMA screening. Knowledge and positive attitudes were among the factors that increased the likelihood of accepting SMA screening. Hearing of SMA, demonstrating higher levels of knowledge about it, and knowing that screening is provided free of charge by the government significantly influenced willingness to undergo screening.

Discussion and Conclusion: The effectiveness of SMA screening programs is directly related not only to service delivery but also to public knowledge and attitudes. This study demonstrates that developing strategies to raise public awareness can increase the success of screening programs. Such studies, conducted with a public health approach, contribute to the sustainability of screening programs.

Keywords: Attitudes; Genetic carrier screening; Health knowledge; Mass screening; Practice; Primary health care; Public health; Spinal muscular atrophies of childhood

Spinal Muscular Atrophy (SMA) is a genetic disorder characterized by the progressive degeneration of motor neurons, typically inherited as an autosomal recessive pattern.[1] Its prevalence is reported to be approximately 1 in 11,000 live births worldwide.[2] SMA poses a significant burden on public health as a leading genetic cause of infant and child mortality. The disease significantly impacts the quality and duration of life, creating widespread impacts on the individual, family, and societal levels.

Cite this article as: *Özbaş C, Gingir C, Uğraş Dikmen A. Awareness and Attitudes Toward Spinal Muscular Atrophy Carrier and Newborn Screening Programs in Türkiye: A Cross-Sectional Study in Family Health Centers. Lokman Hekim Health Sci 2026;6(1):27–37.*

Correspondence: Cansu Özbaş, M.D. Gazi Üniversitesi Tıp Fakültesi, Halk Sağlığı Anabilim Dalı, Ankara, Türkiye
E-mail: cansuultav@gazi.edu.tr

Submitted: 18.08.2025 **Revised:** 01.09.2025 **Accepted:** 13.10.2025 **Available Online:** 16.03.2026

To effectively manage SMA, awareness of the disease and screening programs must be increased. The best treatment response to the disease occurs before symptoms appear.[3] Therefore, various newborn SMA screening programs are implemented in countries such as Australia, Belgium, Canada, Germany, Italy, Japan, Taiwan, and the United States.[4]

In Türkiye, SMA carrier rates are high at 1/50.[5] As of May 2022, SMA was added to the list of diseases screened for by the National Newborn Screening Program currently being conducted in Türkiye. The Premarital Spinal Muscular Atrophy Carrier Screening Program in Türkiye was implemented in all 81 provinces by the Department of Child and Adolescent Health, General Directorate of Public Health (HSGM), Ministry of Health of the Republic of Türkiye, starting on December 27, 2021.[6] From this date until September 4, 2023, 1,834 million people in Türkiye were screened for SMA.[7]

Screening programs play an essential role in public health. However, the success of such programs depends on the level of knowledge of individuals in society about the disease, their risk perception, and their attitudes toward screening programs.[8,9] Therefore, developing strategies to increase SMA awareness by taking into account the demographic, socioeconomic, and cultural characteristics of the population is a critical necessity. The development of these strategies is possible through studies investigating the level of public knowledge and attitudes towards SMA screening programs.

This study aims to assess the knowledge and attitudes of individuals visiting Family Health Centers in Ankara regarding SMA and its screening programs, and to identify factors influencing these perceptions. It aims to contribute not only to health policies in Türkiye but also to screening programs aimed at preventing genetic diseases globally.

Materials and Methods

This cross-sectional study was conducted between January 22-24, 2024, with the participation of individuals aged 18 and over who applied for any reason to designated Family Health Centers in the districts of Sincan, Etimesgut, Yenimahalle, and Gölbaşı within the borders of Ankara province. The sample size was 1493, calculated with a 95% confidence interval, α= 0.05, d=3%, a design effect of 1.0, and a 50% unknown frequency using the Open Epi program. Considering the 10% risk of data loss, the target was to reach 1642 people, and 1643 people participated in the study.

The survey form "Assessment of Knowledge and Attitudes of Individuals Applying to Family Health Centers About SMA Disease and Screening" was used as the data source in the study. The survey consisted of four sections and included a total of 33 questions. The research team prepared the survey form after reviewing national and international literature. The questions designed to measure individuals' knowledge about SMA disease and SMA screening were based on the Spinal Muscular Atrophy Carrier Screening Program Field Guide published in 2023 by the General Directorate of Public Health, Department of Child and Adolescent Health of the Ministry of Health of the Republic of Türkiye.[10] A pilot survey was conducted with 20 individuals to test the feasibility of the study, assess the readability and clarity of the survey, and estimate the approximate time required to complete the survey. Following the pilot, the primary data collection phase was carried out by 42 trained medical students under the supervision of the investigators. Prior to the fieldwork, all interviewers received standardized training on the study protocol, questionnaire content, and interview techniques to ensure consistency and data quality. Each interviewer was responsible for approximately 13 participants per day, making it feasible to complete the data collection of 1,643 participants within the planned 3-day period. The reporting of this study follows the Strengthening the Reporting of Observational Studies in Epidemiology (STROBE) statement.[11]

The questionnaire used in this study consisted of four sections. The first section gathered information on participants' demographic characteristics and identified potential risk factors for Spinal Muscular Atrophy (SMA). The second section assessed participants' knowledge about SMA. It included two multiple-response items addressing the risk factors and clinical features of the disease, as well as three single-response multiple-choice items regarding the estimated carrier frequency of SMA in Türkiye, the possible onset period of the disease, and the optimal time to initiate treatment. The third section evaluated participants' knowledge of SMA screening programs. This section comprised two multiple-response items on the target groups and characteristics of screening, and one single-response multiple-choice item about the primary point of application for premarital screening. The fourth section examined participants' attitudes toward SMA screening. It included one multiple-response item on the perceived importance of screening for various population groups, two three-point Likert scale items on the relevance of screening for family planning and newborn health, and one three-point Likert scale item assessing the perceived importance of public awareness in increasing screening acceptance.

In this study, each correct answer in the knowledge sections was assigned a score of '1', while incorrect or incomplete answers were assigned a score of '0'. In the attitude section, the options 'Disagree' and 'No Opinion' were assigned a score of 0, and 'Agree' was assigned a score of 1. After scoring the second, third, and fourth sections of the survey, the median value was used as the cut-off value, and those scoring at or below the median were classified as 'low' or 'negative.' In contrast, those scoring above were classified as 'high' or 'positive'.[12]

Study data were analyzed using the SPSS 29.0 (SPSS Inc. Chicago, USA) statistical package program. Descriptive statistics were used to summarize the sociodemographic characteristics of the participants and their responses regarding SMA knowledge, screening knowledge, and attitudes. Categorical variables were presented as numbers and percentages.

The association between participants' sociodemographic characteristics and their SMA knowledge, screening knowledge, and attitudes was examined using Chi-square tests. To evaluate the factors influencing participants' hypothetical acceptance of SMA screening for themselves and their children, univariate logistic regression analyses were first performed. Variables with a significance level of $p<0.05$ in the univariate analysis were then included in the multivariate logistic regression model. Results were presented as odds ratios (OR) with 95% confidence intervals (CI). A two-sided p-value <0.05 was considered statistically significant.

The study was conducted under the Helsinki Declaration. The Gazi University Ethics Committee reviewed and approved it with decision number 2024-135.

Results

This study was completed with 1643 participants. Among the participants, 208 individuals (12.7%) reported having a family history of congenital anomalies, while 1029 individuals (87.3%) reported no such history. Regarding having heard of SMA disease, 1420 participants (86.4%) stated that they had heard of SMA, whereas 223 participants (13.6%) had not.

Table 1 summarizes the descriptive characteristics of the 1,643 participants. Most were female (53.3%), aged 18–39 years (46.5%), and had at least a high school education (73.3%). Nearly half (48%) were employed, and two-thirds (67.1%) were married. Among participants who were married or had ever been married, 12.7% reported consanguineous marriage. While 68.5% had children, only 7.3% had a child born after 2022. A family history of congenital anomalies was reported by 12.7%. More than half (54%) reported equal income and expenses, while 23.7% had higher income and 22.3% had lower income than expenses. Most participants (86.4%) had heard of SMA.

Higher SMA knowledge was associated with younger age ($p<0.001$), higher education ($p=0.026$), being unmarried ($p=0.017$), having no children ($p=0.006$), higher income ($p=0.046$), and non-consanguineous marriage ($p=0.002$). Knowledge about SMA screening was higher among participants without children ($p=0.008$) and those in non-consanguineous marriages ($p=0.006$). Positive attitudes toward SMA screening were more frequent among younger ($p=0.009$), more educated ($p=0.001$), childless ($p<0.001$), and non-consanguineous individuals ($p=0.015$) (Table 1).

Table 2 summarizes participants' knowledge and attitudes toward SMA and screening. The most frequently recognized risk factors were genetic predisposition (66.9%), consanguineous marriage (61.3%), and having a child with SMA (32.0%), while 21.7% reported no knowledge of the disease. Commonly identified symptoms included muscle weakness (64.0%) and difficulty walking (53.6%). Only 9.1% knew that SMA can onset at any time from conception to adulthood, and 61.4% correctly stated that treatment is most effective before symptoms appear.

Knowledge about screening was limited: 59.5% knew it was offered premaritally, 41.2% for newborns, and 33.2% for pregnant women; 50.4% were aware that screening is free of charge. Attitudes toward screening were generally positive: 70.7% supported screening for all couples before marriage, 56.0% before pregnancy, and 53.0% for all newborns. More than 90% agreed that awareness should be increased, screening is essential for future children, and it is necessary for informed family planning (Table 2).

The study evaluated the risk factors affecting participants' hypothetical willingness to have their children screened for SMA using both univariate and multivariate regression analyses (Table 3). Males were found to be less likely to have SMA screening compared to females (OR=0.571, 95% CI: 0.388-0.841, $p=0.004$). Employment status indicated that employed individuals were more likely to agree to have their children screened for SMA compared to unemployed individuals (OR=1.882, 95% CI: 1.212-2.922, $p=0.005$). Individuals who have heard of SMA disease are much more likely to accept SMA screening for their children compared to those who have not heard of it (OR=4.406, 95% CI: 2.911-6.671, $p<0.001$) (Table 3).

Table 1. Participants' characteristics and their SMA knowledge and screening attitudes

	Total n (%)*	Knowledge score of SMA, n (%)** High	Knowledge of SMA screening, n (%)** High	Attitudes toward SMA screening, n (%)** Positive
Gender				
Female	876 (53.3)	559 (63.8)	450 (51.4)	531 (60.6)
Male	767 (46.7)	460 (60.0)	395 (51.5)	460 (60.0)
		p=0.110	p=0.958	p=0.791
Age group				
18–39	764 (46.5)	513 (67.1)	415 (54.3)	490 (64.1)
40–64	680 (41.4)	393 (57.8)	334 (49.1)	393 (57.8)
≥65	199 (12.1)	113 (56.3)	96 (48.2)	108 (54.3)
		p<0.001	p=0.090	**p=0.009**
Educational status				
Illiterate	24 (1.5)	14 (58.3)	12 (50.0)	14 (58.3)
Literate, no formal education	43 (2.6)	23 (53.3)	24 (55.8)	26 (60.5)
Primary or secondary school graduate	372 (22.6)	209 (56.2)	184 (49.5)	191 (51.3)
High school and above	1204 (73.3)	773 (64.2)	625 (51.9)	760 (63.1)
		p=0.026	p=0.792	**p=0.001**
Employment status				
Employed	789 (48.0)	515 (65.3)	415 (52.6)	491 (62.2)
Housewife	404 (24.6)	237 (58.7)	204 (50.5)	235 (58.2)
Retired	237 (14.4)	127 (53.6)	113 (47.7)	134 (56.5)
Unemployed	213 (13.0)	140 (65.7)	113 (53.1)	131 (61.5)
		p=0.003	p=0.546	p=0.318
Marital status				
Single	540 (32.9)	357 (66.1)	292 (54.1)	342 (63.3)
Married	1103 (67.1)	662 (60.0)	553 (50.1)	649 (58.8)
		p=0.017	p=0.134	p=0.080
Having children				
Yes	1126 (68.5)	673 (59.8)	554 (49.2)	647 (57.5)
No	517 (31.5)	346 (66.9)	291 (56.3)	344 (66.5)
		p=0.006	**p=0.008**	**p<0.001**
Income level				
Income < Expenses	366 (22.3)	221 (60.4)	196 (53.6)	219 (59.8)
Income = Expenses	888 (54.0)	536 (60.4)	439 (49.4)	520 (58.6)
Income > Expenses	389 (23.7)	262 (67.4)	210 (54.0)	252 (64.8)
		p=0.046	p=0.214	p=0.110
Consanguinity with spouse (n=1231)				
Yes	156 (12.7)	76 (48.7)	62 (39.7)	77 (49.4)
No	1075 (87.3)	661 (61.5)	555 (51.6)	641 (59.6)
		p=0.002	**p=0.006**	**p=0.015**

*: Column percentage; **: Row percentage; SMA: Spinal muscular atrophy.

Table 2. Distribution of participants' knowledge and attitudes regarding SMA and Its screening

Question and option	n	%
Knowledge of SMA		
1. Risk factors for SMA (multiple choice)		
Genetic	1099	66.9
Consanguineous marriage	1007	61.3
Having a child with SMA	526	32.0
Don't know	357	21.7
2. Clinical features of SMA (multiple choice)		
Muscle weakness	1051	64.0
Difficulty walking	881	53.6
Hypotonia	614	37.4
Motor delay in infants	484	29.5
Don't know	462	28.1
3. Prevalence of SMA carriers in Türkiye		
Normal	345	21.0
High	517	31.5
Low	243	14.8
Don't know	538	32.7
4. Onset time of SMA		
Prenatal	533	32.5
Early infancy	371	22.6
Any time from prenatal to adulthood	150	9.1
Childhood	83	5.0
Don't know	506	30.8
5. When is the optimal time to start SMA treatment?		
Before symptom onset	1008	61.4
After symptoms	224	13.6
Don't know	411	25.0
Knowledge of SMA screening		
1. Who should be screened for SMA? (multiple choice)		
Premarital couples	978	59.5
Couples planning pregnancy	713	43.4
Pregnant women	545	33.2
Newborns	677	41.2
Don't know	413	25.1
2. First point of contact for premarital screening		
Family physician	739	45.0
Medical geneticist	335	20.4
Pediatrician	117	7.1
Don't know	452	27.5
3. Which of the following are true regarding SMA screening? (multiple choice)		
It is free of charge	828	50.4
Helps detect the disease before symptoms	755	46.0
Newborn screening is via heel-prick test	530	32.3
The first blood sample is taken from the male partner	325	19.8
Don't know	582	35.4
Attitudes toward SMA screening		
1. Who should undergo screening? (multiple choice)		
All premarital couples	1162	70.7
Preconception screening for those not previously screened	920	56.0
Pregnant women not previously screened	685	41.7
All newborns	871	53.0
Don't know-None	265	16.1
2. Understanding premarital screening is important for informed family planning		
Agree	1501	91.4
Disagree	13	0.8
Don't know	129	7.8
3. Undergoing screening is important for the health of future children		
Agree	1530	93.1
Disagree	11	0.7
Don't know	102	6.2
4. Raising public awareness is important for the acceptance of screening		
Agree	1506	91.7
Disagree	11	0.7
Don't know	126	7.6

SMA: Spinal muscular atrophy.

Individuals who know someone outside the family with an SMA diagnosis are significantly more likely to agree to have their children screened for SMA (OR=5.898, 95% CI: 1.421-24.478, p=0.015). Individuals who had heard of the SMA screening program had a higher probability of accepting SMA screening for their children compared to those who had not heard of it (OR=2.212, 95% CI: 1.427-3.428, p<0.001). Individuals with a positive attitude towards SMA screening were significantly more likely to accept SMA screening for their children compared to individuals

Table 3. Regression analysis of factors hypothetically affecting the status of accepting SMA screening for their child

	Univariate analysis (OR 95%CI)	**p**	**Multivariate analysis (OR 95%CI) (Included model)**	**p**
Age	0.957 (0.948-0.966)	**<0.001**	1.002 (0.986-1.019)	0.776
Gender		**0.023**		**0.004**
Female	1		1	
Male	0.705 (0.522-0.952)		0.571 (0.388-0.841)	
Marital status		0.050		0.625
Married	1		1	
Single	1.398 (0.999-1.955)		1.128 (0.696-1.830)	
Having children		**<0.001**		0.450
No	1		1	
Yes	0.359 (0.240-0.537)		0.766 (0.384-1.529)	
Educational status		**<0.001**		0.078
Below high school	1		1	
High school and above	4.512 (3.313-6.146)		1.493 (0.956-2.331)	
Employment status		**<0.001**		**0.005**
Unemployed	1		1	
Employed	2.851 (2.042-3.980)		1.882 (1.212-2.922)	
Income level		**<0.001**		0.321
Income ≤ Expenses	1		1	
Income > Expenses	2.530 (1.611-3.971)		1.292 (0.779-2.144)	
Having heard of SMA disease		**<0.001**		**<0.001**
No	1		1	
Yes	11.678 (8.341-16.351)		4.406 (2.911-6.671)	
Having a child diagnosed with SMA in the family		0.530		
No	1		–	
Yes	0.670 (0.192-2.335)			
Knowing someone diagnosed with SMA outside the family		**<0.001**		**0.015**
No	1		1	
Yes	14.223 (3.502-57.765)		5.898 (1.421-24.478)	
Having heard of SMA screening		**<0.001**		**<0.001**
No	1		1	
Yes	6.401 (4.423-9.264)		2.212 (1.427-3.428)	
Knowledge score of SMA disease		**0.003**		0.885
Low	1		1	
High	1.575 (1.166-2.127)		0.970 (0.642-1.466)	
Knowledge score of SMA screening		0.086		0.092
Low	1		1	
High	1.301 (0.964-1.756)		0.683 (0.438-1.065)	
Attitude score of SMA screening		**<0.001**		**<0.001**
Negative	1		1	
Positive	3.457 (2.522-4.739)		3.350 (2.223-5.048)	

OR: Odds ratio; CI: Confidence intervals; SMA: Spinal muscular atrophy.

with a negative attitude (OR=3.350, 95% CI: 2.223-5.048, p<0.001). Although factors such as age, marital status, having children, education level, income level, and knowledge about SMA were significant in single analyses, they lost significance in multiple analyses. The presence of a child diagnosed with SMA in the family, knowledge about SMA, and knowledge about SMA screening did not show significant differences in both univariate and multivariate analyses (Table 3).

In the study, risk factors affecting participants' acceptance of SMA screening were evaluated using both univariate and multivariate regression analyses (Table 4). Regarding gender, male individuals were found to be less likely to undergo SMA screening than female individuals (OR=0.456, 95% CI: 0.303-0.685, p<0.001). Regarding educational status, a higher level of education increased the likelihood of accepting screening (OR=1.636, 95% CI: 1.022-2.619, p=0.040). When employment status was examined, employed individuals were more likely to undergo SMA screening compared to unemployed individuals (OR=1.990, 95% CI: 1.251-3.168, p=0.004). Individuals who have heard of SMA are significantly more likely to accept screening than those who have not (OR=5.610, 95% CI: 3.634-8.661, p<0.001). Individuals who have heard of the SMA screening program are more likely to accept SMA screening than those who have not (OR=2.008, 95% CI: 1.258-3.206, p=0.003) (Table 4).

Individuals with a positive attitude toward SMA screening are significantly more likely to accept screening than those with a negative attitude (OR=4.032, 95% CI: 2.607-6.235, p<0.001). Factors such as age, having children, income, knowing someone outside the family diagnosed with SMA, knowledge about SMA, and knowledge about SMA screening were significant in single analyses, but they lost significance in multiple analyses. Marital status and the presence of a child diagnosed with SMA in the family did not show a significant difference in both univariate and multivariate analyses (Table 4).

Discussion

This study explored the factors influencing participants' decisions regarding SMA screening for their children. Female participants were more likely to express willingness to undergo screening for themselves and their children. Prior research has reported similar patterns, with women often participating more actively in health screenings, including colorectal cancer programs.[13–16] A study on colon cancer screening in Singapore found that while men were more likely to be screened, women were more likely to be screened if they were unemployed and given information about Screening.[17] These findings may reflect women's greater sense of responsibility and engagement in health matters, especially when children's well-being is involved. Overall, this highlights the role of gender in shaping attitudes toward genetic screening.

Employed individuals were found to be more likely to have their children and themselves screened for SMA than unemployed individuals. Low socioeconomic status negatively impacts both individuals' health literacy and their access to healthcare. Furthermore, low health literacy also creates an additional barrier to accessing healthcare.[18] The generally better economic situation of employed individuals compared to unemployed individuals may have facilitated their access to healthcare and health information, leading to their adoption of health services such as genetic screening. Furthermore, greater exposure to health-related information in their workplace and higher levels of health literacy may also account for this difference.

Individuals who have previously heard of SMA were significantly more likely to agree to have SMA screenings for themselves and their children, and those who knew someone with a similar diagnosis of SMA were substantially more likely to agree to have their children screened for SMA. A study on SMA awareness, knowledge, and attitudes found that the public and healthcare providers were generally unfamiliar with SMA.[19] A study examining the views of families affected by SMA regarding community screening found that families affected by SMA were largely supportive of screening. The belief that screening would raise public awareness of SMA was a prominent reason for supporting pre-pregnancy screening.[20] Another study conducted to find factors affecting the potential use of genetic testing for cancers included the perceived severity of the cancers in question, the perceived effectiveness of screening, and family experience with cancer.[21] In a study conducted in Romania to find out the factors associated with the popularity, perceptions and acceptance or rejection of prenatal screening for Down syndrome (DS), misperception of the risk of DS was found to be one of the main factors associated with the refusal of the test.[22] In a study conducted in Indonesia to determine the level of acceptance of cervical cancer screening among women, demographic characteristics such as having experience with the disease and exposure to information were found to have a significant relationship with knowledge, perception and acceptance of HPV vaccination and cervical cancer screening.[23] Accordingly, individuals with direct or

Table 4. Regression analysis of factors hypothetically affecting the status of accepting SMA screening

	Univariate analysis (OR 95%CI)	**p**	**Multivariate analysis (OR 95%CI) (included model)**	**p**
Age	0.954 (0.944-0.963)	**<0.001**	1.000 (0.982-1.017)	0.959
Gender		**<0.001**		**<0.001**
Female	1		1	
Male	0.601 (0.440-0.820)		0.456 (0.303-0.685)	
Marital status		0.128		0.690
Married	1		1	
Single	1.303 (0.927-1.833)		1.108 (0.670-1.831)	
Having children		**<0.001**		0.799
No	1		1	
Yes	0.375 (0.249-0.566)		0.910 (0.440-1.881)	
Educational status		**<0.001**		**0.040**
Below high school	1		1	
High school and above	4.912 (3.570-6.758)		1.636 (1.022-2.619)	
Employment status		**<0.001**		**0.004**
Unemployed	1		1	
Employed	2.899 (2.053-4.093)		1.990 (1.251-3.168)	
Income Level		**<0.001**		0.362
Income ≤ Expenses	1		1	
Income > Expenses	2.600 (1.625-4.158)		1.284 (0.751-2.195)	
Having heard of SMA disease		**<0.001**		**<0.001**
No	1		1	
Yes	14.377 (10.165-20.335)		5.610 (3.634-8.661)	
Having a child diagnosed with SMA in the family		0.458		
No	1		–	
Yes	0.623 (0.179-2.172)			
Knowing someone diagnosed with SMA outside the family		**<0.001**		0.104
No	1		1	
Yes	6.453 (2.367-17.591)		2.369 (0.837-6.706)	
Having heard of SMA screening		**<0.001**		**0.003**
No	1		1	
Yes	6.726 (4.567-9.904)		2.008 (1.258-3.206)	
Knowledge score of SMA disease		**0.005**		0.429
Low	1		1	
High	1.558 (1.144-2.122)		0.839 (0.543-1.296)	
Knowledge score of SMA screening		**0.027**		0.269
Low	1		1	
High	1.417 (1.040-1.931)		0.768 (0.480-1.227)	
Attitude score of SMA screening		**<0.001**		**<0.001**
Negative	1		1	
Positive	4.039 (2.898-5.630)		4.032 (2.607-6.235)	

OR: Odds ratio; CI: Confidence intervals; SMA: Spinal muscular atrophy.

indirect experience of severe and relatively rare diseases like SMA appear to be more likely to seek preventive healthcare services as their disease awareness increases. This demonstrates the impact of personal experiences on health behavior decision-making processes.

Individuals who had heard about the SMA screening program earlier were found to be more likely to agree to have their children screened for SMA. A systematic review examining the factors influencing decisions to accept cystic fibrosis carrier screening found that the most frequently identified acceptance factors included greater perceived benefits of cystic fibrosis carrier screening, lack of knowledge about the test, and a weaker perception of the benefits of screening, all of which were associated with refusal of the test.[24] In a study conducted in the Netherlands investigating the acceptance of prenatal screening for congenital anomalies, the positive characteristics of the screening test were determined to be the second most crucial reason affecting acceptance.[25]

Individuals with positive attitudes toward SMA screening were found to be more likely to agree to have their children and themselves screened for SMA. A study conducted to identify factors associated with the acceptability of HPV screening for cervical cancer showed that factors related to test acceptability included attitudes toward the test, perceived high testing benefits, and negative attitudes toward increased screening intervals.[26] A study investigating the factors affecting the acceptance of colorectal cancer screening in Thailand found that reluctance to undergo screening was among the reasons for refusing Screening.[27] This may be because individuals make health decisions based not only on knowledge but also on their attitudes and beliefs. According to the "Health Belief Model", widely accepted in the health behavior literature, an individual's likelihood of adopting a healthcare service is directly related to their perceived benefit, perceived risk, and personal attitudes.[28] In this context, the importance of awareness-raising campaigns to develop positive attitudes towards SMA screening for screening acceptance becomes evident.

Individuals with higher education levels have been found to be more likely to be screened for SMA. A study conducted in Türkiye to determine the level of knowledge and awareness about SMA in premarital health screenings found a significant correlation between variables such as education level and the likelihood of screenings being appropriate and the willingness to undergo testing.[29] A study conducted to determine the factors influencing women's acceptance of extended alpha-fetoprotein testing during pregnancy found that skepticism about the usefulness of test results, low education level, and misunderstanding of the purpose of the test were among the factors significantly associated with test refusal.[30] This may be due to the fact that as individuals' education level increases, their ability to access and understand health-related information improves, leading to an increased tendency to utilize health services. This demonstrates the importance of education level in affecting health awareness and informed decision-making.

This study has some limitations. First, its cross-sectional design does not allow for causal inferences. Second, the use of self-reported data may have introduced social desirability bias. Third, the study population was limited to individuals attending Family Health Centers in Ankara, which may limit the generalizability of the findings. Moreover, the knowledge and attitude questions, although developed based on literature, were not derived from a previously validated instrument. Also, incorrect and "no opinion" responses were grouped together and scored as 0. This approach was preferred in order to emphasize accurate knowledge, which is the main determinant in transforming awareness into attitudes and behaviors in health services. However, this strategy may have limited our ability to distinguish between participants who had incorrect knowledge and those who had no knowledge at all. Future studies may adopt alternative scoring approaches to explore these differences further. Finally, participants' intention to undergo SMA screening was evaluated based on hypothetical scenarios, which may not fully reflect real-life behavior.

Conclusion

In conclusion, this study found that women, employed individuals, individuals who know someone diagnosed with SMA outside the family, individuals who have heard of the SMA screening program, and individuals with positive attitudes toward SMA screening are more likely to agree to have their children screened for SMA. Furthermore, women, individuals with higher education levels, employed individuals, individuals who have heard of SMA, individuals who have heard of the SMA screening program, and individuals with positive attitudes toward SMA screening are significantly more likely to agree to SMA screening themselves.

By revealing the factors influencing individuals' decisions to undergo SMA screening, this study provides essential information on how health policies and awareness campaigns should be directed. Increasing awareness campaigns about SMA and screening, particularly among men and low-educated groups, is critical for increasing public acceptance of screening programs. Strengthening communication between primary health care providers and the public and promoting positive health attitudes toward screening are crucial for increasing the frequency of SMA screening. In addition, further studies are needed to examine the acceptance of screening and other factors that may affect it for some special groups that may be prone to SMA and pose a risk.

Ethics Committee Approval: The Gazi Ethics Committee granted approval for this study (date: 13.08.2025, number: 2024-135).

Informed Consent: Written informed consent was obtained from participants.

Conflict of Interest: None declared.

Financial Disclosure: The authors declared that this study has received no financial support.

Use of AI for Writing Assistance: Artificial intelligence-supported technologies were not used in this study.

Authorship Contributions: Concept: CÖ, CG, AUD; Design: AUD, CG, CÖ; Supervision: CG, CÖ, AUD; Data Collection or Processing: CG, CÖ; Analysis or Interpretation: CG; Literature Search: CG; Writing: CG, CÖ; Critical Reviews: AUD, CÖ.

Acknowledgments: We want to thank all the individuals who participated in this study for their valuable time and contributions. We also express our gratitude to the healthcare staff working at the Family Health Centers in Ankara for their support in the data collection process.

Peer-review: Double blind peer-reviewed.

References

1. National Institute of Neurological Disorders and Stroke. Spinal Muscular Atrophy. Available at: https://www.ninds.nih.gov/health-information/disorders/spinal-muscular-atrophy. Accessed September 5, 2025.
2. 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;20(1):27-32. [CrossRef]
3. De Vivo DC, Topaloglu H, Swoboda KJ, Bertini E, Hwu WL, Crawford TO, et al. Nusinersen in infants who initiate treatment in a presymptomatic stage of spinal muscular atrophy (SMA): interim efficacy and safety results from the phase 2 Nurture study (S25.001). Neurology 2019;92(15 Suppl):S25.001. [CrossRef]
4. Dangouloff T, Vrščaj E, Servais L, Osredkar D; SMA NBS World Study Group. Newborn screening programs for spinal muscular atrophy worldwide: where we stand and where to go. Neuromuscul Disord 2021;31(6):574-82. [CrossRef]
5. Türkiye SMA Foundation. Carrier screening. Available at: https://www.sma.org.tr/tasiyicilik-taramasi. Accessed September 5, 2025. [In Turkish]
6. Republic of Türkiye Ministry of Health, General Directorate of Public Health. Field Guide for the Premarital Spinal Muscular Atrophy Carrier Screening Program (2023). Available at: https://hsgm.saglik.gov.tr/depo/birimler/cocuk-ergen-sagligi-db/Programlar/EO_SMA_TASIYICI_TARAMA_PROGRAMI_SAHA_REHBERI.pdf. Accessed September 5, 2025. [In Turkish]
7. Republic of Türkiye Ministry of Health. Premarital SMA screening reached over 1 million people in two years. Available at: https://www.saglik.gov.tr/TR,99556/evlilik-oncesi-sma-taramasi-2-yilda-1-milyonu-askin-kisiye-ulasti.html. Accessed September 5, 2025. [In Turkish]
8. Almadi MA, Mosli MH, Bohlega MS, Al Essa MA, AlDohan MS, Alabdallatif TA, et al. Effect of public knowledge, attitudes, and behavior on willingness to undergo colorectal cancer screening using the health belief model. Saudi J Gastroenterol 2015;21(2):71-7. [CrossRef]
9. Rijal BM, Dawadi P. Knowledge, attitudes and associated factors regarding cervical cancer and its screening practice among women of central Nepal. Heliyon 2024;10(14):e34886. [CrossRef]
10. Republic of Türkiye Ministry of Health, General Directorate of Public Health, Department of Child and Adolescent Health. Premarital SMA Carrier Screening Program-information page. Available at: https://hsgm.saglik.gov.tr/tr/tarama-programlari/evlilik-oncesi-sma-tasiyici-tarama-programi.html. Accessed September 5, 2025. [In Turkish]
11. von Elm E, Altman DG, Egger M, Pocock SJ, Gøtzsche PC, Vandenbroucke JP; STROBE Initiative. The Strengthening the Reporting of Observational Studies in Epidemiology (STROBE) statement: guidelines for reporting observational studies. Lancet 2007;370(9596):1453-57. [CrossRef]
12. DeCoster J, Gallucci M, Iselin AMR. Best practices for using median splits, artificial categorization, and their continuous alternatives. J Exp Psychopathol 2011;2(2):197-209. [CrossRef]
13. Clark GRC, Steele RJC, Fraser CG. Strategies to minimise the current disadvantages experienced by women in faecal immunochemical test-based colorectal cancer screening. Clin Chem Lab Med 2022;60(10):1496-505. [CrossRef]
14. Guiriguet C, Pera G, Castells A, Toran P, Grau J, Rivero I, et al. Impact of comorbid conditions on participation in an organised colorectal cancer screening programme: a cross-sectional study. BMC Cancer 2017;17(1):524. [CrossRef]
15. Rollet Q, Tron L, De Mil R, Launoy G, Guillaume É. Contextual factors associated with cancer screening uptake: a systematic review of observational studies. Prev Med 2021;150:106692. [CrossRef]
16. Koskenvuo L, Malila N, Pitkaniemi J, Miettinen J, Heikkinen S, Sallinen V. Sex differences in faecal occult blood test screening for colorectal cancer. Br J Surg 2019;106(4):436-47. [CrossRef]
17. Tan K, Wong G, Lau J, et al. Gender differences associated with colorectal cancer screening uptake in Singapore. Gut 2024;73(Suppl 2):A285. [CrossRef]

18. Simmons RA, Cosgrove SC, Romney MC, Plumb JD, Brawer RO, Gonzalez ET, et al. Health literacy: cancer prevention strategies for early adults. Am J Prev Med 2017;53(3 Suppl 1):S73-S77. [CrossRef]
19. Moultrie RR, Kish-Doto J, Peay H, Lewis MA, Treadwell-Deering D, Brewer CA, et al. A review on spinal muscular atrophy: awareness, knowledge, and attitudes. J Genet Couns 2016;25(5):892-900. [CrossRef]
20. Boardman FK, Young PJ, Griffiths FE. Population screening for spinal muscular atrophy: a mixed methods study of the views of affected families. Am J Med Genet A 2017;173(2):421-34. [CrossRef]
21. Patenaude AF, Schneider KA, Kieffer SA, Calzone KA, Stopfer JE, Basili LA, et al. Acceptance of invitations for p53 and BRCA1 predisposition testing: factors influencing potential utilization of cancer genetic testing. Psychooncology 1996;5(3):241-50. [CrossRef]
22. Pop-Tudose ME, Armean P, Pop IV. Prenatal screening for Down syndrome: popularity, perceptions and factors associated with acceptance or refusal of testing. J Matern Fetal Neonatal Med 2022;35(25):6505-09. [CrossRef]
23. Endarti D, Satibi S, Kristina SA, Farida MA, Rahmawanti Y, Andriani T. Knowledge, perception, and acceptance of HPV vaccination and screening for cervical cancer among women in Yogyakarta Province, Indonesia. Asian Pac J Cancer Prev 2018;19(4):1105-11.
24. Chen LS, Goodson P. Factors affecting decisions to accept or decline cystic fibrosis carrier testing/screening: a theory-guided systematic review. Genet Med 2007;9(7):442-50. [CrossRef]
25. van den Berg M, Timmermans DR, Kleinveld JH, Garcia E, van Vugt JM, van der Wal G. Accepting or declining the offer of prenatal screening for congenital defects: test uptake and women's reasons. Prenat Diagn 2005;25(1):84-90. [CrossRef]
26. Tatar O, Thompson E, Naz A, Perez S, Shapiro GK, Wade K, et al. Factors associated with human papillomavirus (HPV) test acceptability in primary screening for cervical cancer: a mixed methods research synthesis. Prev Med 2018;116:40-50. [CrossRef]
27. Saengow U, Chongsuwiwatvong V, Geater A, Birch S. Preferences and acceptance of colorectal cancer screening in Thailand. Asian Pac J Cancer Prev 2015;16(6):2269-76. [CrossRef]
28. Alyafei A, Easton-Carr R. The Health Belief Model of Behavior Change. In: StatPearls [Internet]. Treasure Island (FL): StatPearls Publishing; 2025-. Available at: https://www.ncbi.nlm.nih.gov/books/NBK606120/. Accessed September 5, 2025.
29. Yanmaz G, Gürsoy E. Knowledge and awareness of spinal muscular atrophy in pre-marital health screenings: a cross-sectional study. J Basic Clin Health Sci 2024;8(2):441-47. [CrossRef]
30. Li DK, Karlberg K, Wi S, Norem C. Factors influencing women's acceptance of prenatal screening tests. Prenat Diagn.2008;28(12):1136-43. [CrossRef]