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RESEARCH ARTICLE

KNOWLEDGE AND AWARENESS OF SPINAL MUSCULAR ATROPHY IN PRE-MARITAL HEALTH SCREENINGS: A CROSS-SECTIONAL STUDY

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ABSTRACT

Purpose: Early diagnosis of Spinal Muscular Atrophy (SMA) is crucial. This study assesses the awareness and attitudes toward SMA screening among individuals preparing for marriage.
Material and Methods: This cross-sectional study was conducted in three family health centers in the central district of Erzincan from June to September 2023. A researcher-designed 15-item instrument was utilized to collect demographic data and assess knowledge and attitudes regarding SMA among participants.
Results: The study was participated in by 197 individuals, with an average age determined to be 29.45±7.48. The proportion of those supporting the implementation of SMA screening was 84.8% (n=167), while the intention to undergo the test was identified at 72.6% (n=143).
A significant relationship was found between the willingness to be tested and variables such as the level of education, the absence of disabled close relatives, not considering SMA to be genetically inherited, finding screenings appropriate, the perceived impact of test results on marriage, and the belief that screening could not be conducted after marriage.
Conclusion: While most recognized SMA's severity, there's a notable deficit in understanding its preventability, suggesting the need for targeted educational interventions to enhance genetic screening uptake.

Keywords: Spinal Muscular Atrophy, Genetic Screening, Diagnostic Screening Programs, Secondary Prevention

INTRODUCTION

Spinal Muscular Atrophy (SMA) is a genetic and progressive neuromuscular disorder characterized by the loss of motor neuron function due to mutations in the SMN1 (survival motor neuron 1) gene (1). This condition results in a progressive weakening and atrophy of the muscles. The clinical spectrum of SMA exhibits significant variability in terms of the onset and severity of symptoms, which presents considerable challenges in the diagnosis and treatment of the disease(2).

The disease has four main types: Type I (Werdnig-Hoffmann disease) is the most severe form, beginning in infancy with limited life expectancy due to respiratory failure. Type II (Intermediate SMA) manifests in infancy or early childhood, where children can sit but are unable to stand or walk unassisted. Type III (Kugelberg-Welander disease)

starts after early childhood; individuals can walk but may lose this ability over time, yet they have a normal life expectancy. Type IV begins in adulthood and is the mildest form, progressing slowly with a normal life expectancy. Each type of SMA is associated with mutations in the SMN1 gene, which leads to a deficiency in the SMN protein critical for motor neuron survival, and the copy number of the SMN2 gene can influence the severity of the disease (3). SMA is a genetic disorder that exhibits varying incidence rates across different populations. These rates can fluctuate depending on factors such as genetic diversity, diagnostic methods, and characteristics of the population (4). Nevertheless, it can be stated that SMA occurs at an approximate rate of 7.8-10 per 100,000 live births worldwide (5-7).

SMA is a genetic condition where early diagnosis is particularly crucial. In the early stages of SMA, non-neuromuscular symptoms can be detected prior to the clinical signs of neuromuscular degeneration, potentially offering a sequence for the progression of the disease (8). Early diagnosis enables the timely initiation of interventions to slow disease progression and improve quality of life. As knowledge of the pathophysiology of SMA increases, promising strides in its treatment are also emerging (9). Therefore, early diagnosis of SMA is of critical importance in the management of the disease and the development of treatment strategies.

In Turkey, with the directive issued by the Ministry of Health on December 27, 2021, SMA screening has become mandatory nationwide during the pre-marital and newborn periods. Through this program, couples who are carriers for SMA can be identified, allowing for the provision of genetic counseling and treatment options before or after pregnancy. SMA screening is conducted free of charge at Family Health Centers. Nevertheless, comprehensive studies on the societal acceptance of screening programs and the challenges encountered in this process have not been conducted.

The purpose of our study is to assess the knowledge levels and attitudes towards SMA screening among individuals applying for a marriage certificate at family health centers in the central district of Erzincan, thereby contributing to the more effective shaping of relevant health policies and educational programs.

MATERIAL AND METHODS

Study design and execution

This descriptive and cross-sectional study was conducted at three different family health centers affiliated with the central district of Erzincan between June and September 2023.

Study population

The research population consisted of adults aged 18-65 years residing in the central district of Erzincan who applied for a health report for marriage purposes. Individuals without a diagnosis of mental retardation who voluntarily agreed to participate were included in the study.

Data collection instrument

Table 1 and Figure 1 present items from a 15-item questionnaire developed by researchers following a literature review, which was employed to measure participants' demographic characteristics and their knowledge and attitudes about SMA. The survey was designed using a five-point Likert scale for attitude scales and was converted to a three-point Likert scale for analysis. The scale does not have a total score, and the surveys were administered via face-to-face interviews.

Sample size and sampling method

According to TUIK 2022 population data, the number of marriages in Erzincan was recorded as 1309. It was projected that approximately 350 marriages would occur in the central district during the study period. The sample size was calculated to be a minimum of 184 with a 5% sampling error and p=0.5, q=0.5 values, and interviews were conducted with 197 individuals within the scope of the study.

Ethical approval

The research received ethical approval from the Erzincan Binali Yıldırım University Clinical Research Ethics Committee (Decision Date: 13.04.2023, Number: 2023-08/4). Both verbal and written consent were obtained from participants, and the principles of the revised Declaration of Helsinki were adhered to throughout the study.

Table 1. Demographic Data of Participants

		n	%
Gender	Female	103	52.3
	Male	94	47.7
Education	Illiterate - primary school	9	4.6
	Middle school	21	10.7
	High school	65	33
	University	102	51.8
Profession	Employee	64	32.5
	Officer	53	26.9
	Retired	5	2.5
	Housewife	22	11.2
	Self-employment	26	13.2
	Not working	27	13.7
Is there a consanguineous marriage (2nd and 3rd degree)	Yes	26	13.2
	No	171	86.8
Do you have any chronic diseases	Yes	4	2
	No	193	98
Do you have a disabled person in your 1st and 2nd degree relatives?	Yes	12	6.1
	No	185	93.9
Have you ever heard of SMA disease?	Yes	178	90.4
	No	19	9.6
Resource	Television-social media	142	72.1
	Physician and other healthcare personnel	12	6.1
	Friend-other	24	12.2

Data analysis

Data analysis was performed using IBM SPSS Statistics 23 (SPSS Inc., Chicago, IL, USA). The normal distribution of data was tested with the Kolmogorov-Smirnov test, numerical data were reported as mean ± standard deviation or median (min-max), and categorical variables were reported as frequency and percentage.

Statistical methods

Descriptive statistical analyses were conducted. The relationship between two categorical variables was examined with Chi-Square and Fisher's Exact tests, and the means of two independent samples were compared using the Independent Samples t-test. The level of statistical significance was set at $p<0.05$.

RESULTS

A total of 197 individuals were included in the study. The average age of the participants was 29.45±7.48 years, and 178 individuals who had previously heard of SMA had been aware of the disease for an average of 4.37 years (max=10, min=1). The demographic data of the participants are presented in Table 1.

A total of 84.8% (n=167) of the participants agreed with the implementation of SMA screening, while 45.2% (n=89) responded with "I don't know" to the statement "SMA is a preventable disease." On the other hand, the response rate for "Will you get tested for SMA?" with "Yes" was 72.6% (n=143). The responses to the attitude and knowledge questions by the participants are illustrated in Figure 1.

No significant relationship was found between the duration of awareness about SMA in years and the desire to undergo SMA testing (p=0.376). Similarly, no significant relationship was detected between age and the desire to be tested (p=0.148).

The desire to be tested and its association with various variables were evaluated using Chi-Square analysis, as presented in Table 2. Accordingly, a significant relationship was found between the desire to be tested and factors such as increased education level, absence of disabled individuals among close relatives, not considering SMA to be a genetically inherited disease, approving of SMA screenings, believing that a negative screening test would be a barrier to marriage, and the belief that screening could not be performed after marriage.

DISCUSSION

In this study, the knowledge levels and attitudes towards SMA screening among individuals applying for a marriage certificate at family health centers in the central district of Erzincan were investigated. The

Table 2. Comparison of variables with desire to have SMA screening

		Will you have an SMA screening test? Yes		No		
		n	%	n	%	p
Gender	Male	72	76.6	22	23.4	0.228
	Female	71	68.9	32	31.1	
Education	Illiterate-primary school	6	67.7	3	33.3	**0.017**
	Middle school	13	61.9	8	38.1	
	High school	40	61.5	25	38.5	
	University	84	82.4	18	17.6	
Do you have a consanguineous marriage?	Yes	21	80.8	5	19.2	0.316
	No	122	71.3	49	28.7	
Do you have a disabled person in your 1st and 2nd degree relatives?	Yes	5	41.7	7	58.3	**0.020***
	No	138	74.6	47	25.4	
Have you ever heard of SMA** disease?	Yes	132	74.2	46	25.8	0.131
	No	11	57.9	8	42.1	
SMA is a genetically inherited disease	I totally agree - I agree	63	78.8	17	21.3	**0.039**
	No idea	65	65	35	35	
	Disagree - completely disagree	15	88.2	2	11.8	
SMA is a preventable disease	I totally agree - I agree	72	77.4	21	22.6	0.133
	No idea	63	70.8	26	29.2	
	Disagree - completely disagree	8	53.3	7	46.7	
I find SMA campaigns correct	I totally agree - I agree	108	78.3	30	21.7	**0.024**
	No idea	24	58.5	17	41.5	
	Disagree - completely disagree	11	61.1	7	38.9	
I think it is correct to perform an SMA screening	I totally agree - I agree	130	77.8	37	22.2	**<0.001**
	No idea	10	43.5	13	56.5	
	Disagree - completely disagree	3	42.9	4	51.7	
If the SMA test is positive, it is an obstacle to marriage	I totally agree - I agree	27	87.1	4	12.9	**0.001**
	No idea	50	59.5	34	40.5	
	Disagree - completely disagree	66	80.5	16	19.5	
SMA test can also be done after marriage	I totally agree - I agree	76	77.6	22	22.4	**0.005**
	No idea	38	58.5	27	41.5	
	Disagree - completely disagree	29	85.3	5	14.7	

findings indicate that a significant portion of the participants lacks adequate awareness about the importance of SMA screening, perceiving it as a mandatory or routine procedure in the pre-marital process. This can be considered a significant deficiency in terms of preventing genetic diseases and promoting the birth of healthy generations.

Without asking for detailed knowledge, participants were only queried whether they had heard of SMA before and for how long. Out of 197 individuals, 178 (90.4%) acknowledged prior awareness of SMA, averaging 4.37 years of knowledge about the disease (min=1, max=10).

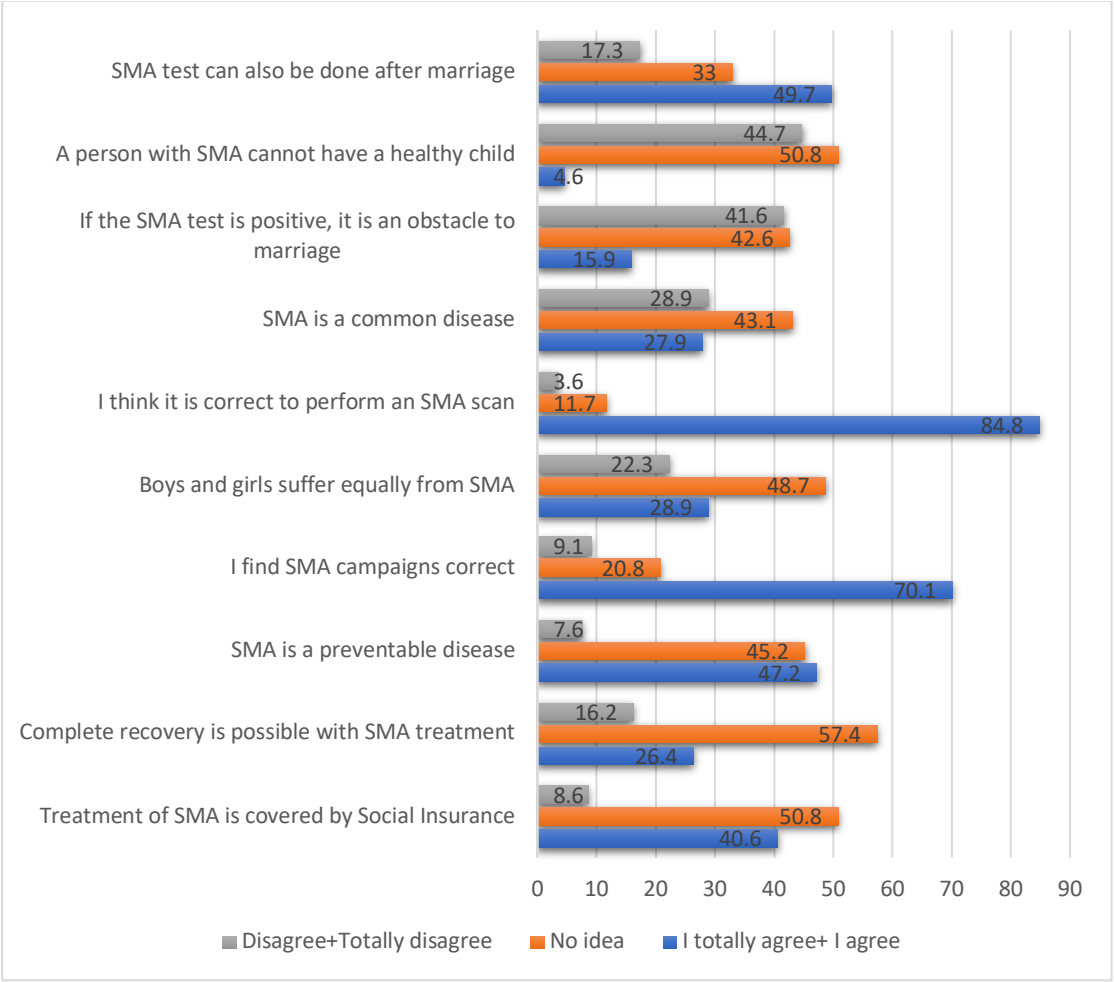

Figure 1. Participants' answers to attitude and knowledge questions

Conversely, 19 individuals (9.6%)reported never having heard of SMA before. It is also noteworthy that TV and social media were the primary sources of information. These findings suggest that while there is a general awareness of SMA in the population, this awareness is still not at a sufficient level. Previous studies have highlighted the critical importance of the public's knowledge level in the early diagnosis and treatment of genetic diseases(10). Additionally, research on the effectiveness of genetic counseling and screening programs has revealed that the level of knowledge within the community is a decisive factor in the success of these programs(11). On the other hand, no significant relationship was found between the duration of awareness of the disease and the desire to undergo SMA screening. This may suggest that the knowledge held is superficial. In this context, the results of our study underscore the importance of increasing public awareness regarding the prevention and management of genetic diseases, highlighting the significant responsibility that falls on health professionals.

While 84.8% (n=167) of participants supported SMA screening, 45.2% (n=89) were unsure about the preventability of SMA. However, 72.6% (n=143) indicated they would consider getting tested. This high level of support could be interpreted as a result of mandatory pre-marital screening procedures, despite participants not having sufficient knowledge about the disease. Factors influencing participation in health screenings include perceived risk, physician recommendations, personal vulnerability, concerns about receiving adverse results, and belief in the benefits of early detection(12, 13). However, it is understood from this study that the participation of the participants in the screening programs stems from the mandatory nature of these programs as part of the marriage process. While this situation has the potential to increase the effectiveness of screening programs and encourage participation, it also

indicates the need for greater efforts to enhance the community's level of knowledge and awareness about the disease.

Public health campaigns and informative programs in the field of SMA screenings are crucial for enhancing societal awareness. Increasing awareness toward the early diagnosis and treatment of genetic diseases can prevent the spread of these conditions and significantly improve individuals' quality of life in the future. Health professionals play a central role in the prevention and management of genetic diseases(14). They must accurately inform patients and emphasize the importance of genetic screenings so that patients view these programs not as obligations but as necessities.

As expected, the desire to undergo testing increased with the participants' level of education. Studies in the literature indicate that the level of education is a significant factor in the desire to undergo genetic testing, with a higher level of education often associated with a greater understanding and a more cautious approach towards genetic tests(15, 16). This situation leads to implications for the need for targeted educational programs to facilitate informed decision-making across different educational strata and public health policy.

The absence of disabled individuals among close relatives and disbelief in the genetic transmission of SMA were seen to increase the willingness to undergo screening. This could be a phenomenon known as "fear of the unknown" or "anxiety about a bad outcome." People may want to detect and manage a potential health issue in advance, thinking that genetic diseases not seen in their families could pose a "hidden" risk to themselves. This trend is reinforced by increasing public awareness of the carrier status and transmission of genetic diseases and the growing accessibility of genetic tests.

Research indicates that individuals' beliefs about the accuracy of health screenings and the validity of these tests can influence their willingness to participate in screening tests(17, 18). The belief in the accuracy of SMA screenings, the perception that a negative screening result could impede marriage, and the belief that screenings cannot be conducted after marriage are significantly associated with the desire to undergo testing. Such beliefs can influence individuals' participation in screening tests and their preference for the timing of these tests. Notably, the belief in the importance of pre-marital screening tests can lead to a higher willingness to undergo such tests before marriage. Health professionals and policymakers should consider these beliefs and perceptions when developing and implementing screening programs.

Study limitations

Limitations of this study include its execution in only three family health centers in the central district of Erzincan, which may limit the generalizability of the findings to the broader population of Erzincan or other regions. Additionally, the use of a researcher-developed questionnaire due to the lack of a standardized measurement tool in the literature may limit the validity and reliability of the survey. Lastly, the assumption that every individual applying for a pre-marital health certificate may potentially have children does not take into account fertility and the desire to have children as independent variables. However, this methodological choice is due to the fact that women's fertility status cannot be biochemically verified and would only be based on self-report. Moreover, even a young woman may not be fertile. Nonetheless, the relatively low average age of the participants included in our study minimizes this limitation.

CONCLUSION

In conclusion, the study conducted among individuals applying for a marriage certificate in the central district of Erzincan reveals a notable gap in awareness and understanding of SMA and the importance of SMA screening. While a majority of the participants are aware of SMA, their knowledge often stems from less formal sources such as TV and social media, which may not provide comprehensive or accurate information. This superficial awareness, coupled with the mandatory nature of pre-marital screening programs, suggests that individuals might not fully grasp the significance of these screenings beyond a bureaucratic requirement. The study underscores the crucial role of health professionals in bridging this knowledge gap, emphasizing the necessity for targeted educational campaigns and personal counseling to foster a deeper understanding of genetic diseases, their prevention, and management.

Moreover, the study highlights the influence of education level, familial history, and personal beliefs on the willingness to undergo SMA screening, pointing to the need for public health strategies that cater to diverse segments of the population. By

enhancing public awareness and understanding of genetic conditions and the benefits of early detection, health authorities can encourage more informed participation in screening programs, thus improving the overall effectiveness of genetic disease prevention efforts.

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