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ORIGINAL ARTICLE

Investigation of Awareness Level of Spinal Muscular Atrophy Disease in Turkish Society

Ender Coşkunpınar, Yusuf Ordu, Burak İraz, Emine Yiğit, Buhari Yılmaz, Halime Yıldırım

Department of Medical Biology, University of Health Sciences, Faculty of Medicine, Istanbul, Türkiye

Abstract

Introduction: In this study, it was aimed to measure and investigate the level of awareness about SMA disease in individuals aged 18–65 in Turkish society. We believe that the obtained data will contribute to epidemiological studies and literature.

Methods: A total of 2920 individuals between the ages of 18–65 who were compos mentis were included in our study. In the study, the data collected through online applications were analyzed by using a questionnaire consisting of 2 parts and a total of 24 questions. Data analysis was performed using the SPSS version 25 package program.

Results: A decrease in the level of awareness about SMA disease is observed with increasing age (p=0.0001), individuals with higher education levels are more conscious about SMA disease (p=0.0001), individuals with knowledge about SMA types have received undergraduate and graduate education (p=0.0001), it was determined that 38% of the volunteers thought that SMA disease could be treated with this method. It was determined that 75% of the individuals participating in the study could not be diagnosed with SMA disease early, 20.12% thought that SMA disease could be treated with gene therapy, and that they were aware of the fact that Ministry of Health covered the treatment of certain types of SMA patients.

Discussion and Conclusion: Based on these results, it is understood that individuals who have consanguineous marriages do not know that consanguineous marriages have an effect on SMA disease. Awareness about SMA increases as you go east. We attribute this to the high number of patients in those regions. When the individuals with knowledge about gene therapy and SMA were compared, it was determined that 28,09% of the individuals had knowledge about both gene therapy and SMA disease. Today, the fact that news and social media posts about SMA disease and gene therapy are thought to have an impact on this result.

Keywords: Public awareness level; SMA; SMN1; SMN2.

Spinal muscular atrophy (SMA) is an autosomal recessive neuromuscular disease that occurs as a result of mutations in the survival motor neuron (SMN) gene[1,2]. The pathogenesis of the disease is not fully known. Nevertheless, SMA is a disease in which spinal cord anterior horn cells and brainstem motor nuclei are involved and whose pathology can be explained by rapidly progressive, programmed cell death. Therefore, SMA affects the life-long functional status of children[3]. Approximately 94% of SMA patients carry homozygous SMN1 exon 7 deletions, regardless of disease severity. SMA is the most common genetic cause of death in childhood and the second most common autosomal recessive disease in humans[4]. For type-1 SMA, the incidence has been reported as 1/25,000 and the prevalence as 1/80,000[5]. SMA is a neuromuscular disease caused by abnormally low expression levels of the SMN protein. Traditionally, low levels of SMN have been thought to cause selective death of lower motor neurons, leading to denervation and atrophy of skeletal muscles. However, numerous recent studies suggest that SMA may be a multisystem disorder rather than just a disease of lower motor neurons. There are some promising treatments for SMA, but targeting treatment effectively to all affected cells and tissues remains a major challenge[6]. In humans, besides SMN2 duplications, two SMN-independent protective modifier genes can greatly influence SMA severity. Asymptomatic individuals with homozygous SMN1 deletions and only three or four copies of SMN2 have recently been reported[7]. Identifying and characterizing the pathological changes occurring in all

Correspondence: Ender Coşkunpınar, M.D. Department of Medical Biology, University of Health Sciences, Faculty of Medicine, Istanbul, Türkiye
Phone: +90 532 240 71 57 **E-mail:** ecoskunpinar@gmail.com
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cell types and tissues affected by SMA is crucial for the successful development of new SMA therapeutics[8,9]. An alternative treatment strategy for SMA is gene replacement therapy. In 2019, the FDA approved intravenous administration for the treatment of patients with SMA in their first two years of life, regardless of disease severity[10]. In this study, it was aimed to measure and investigate the level of awareness about SMA (Spinal Muscular Atrophy) disease in individuals between the ages of 18-65.

Materials and Methods

Study Design

A total of 2920 volunteers between the ages of 18-65, living in the Republic of Türkiye, who are mentally stable, were included in the study. The study was approved by the T.C. Health Sciences University Hamidiye Scientific Research Ethics Committee on 29.01.2021 with the meeting number 2021/4 and the decision numbered 4/17, and was carried out in accordance with the Declaration of Helsinki. A questionnaire consisting of 2 parts and a total of 24 questions was used in the research. In the first part of the questionnaire, there were demographic questions consisting of 5 items, and in the second part, there were questions consisting of 19 items related to the level of awareness about SMA (Spinal Muscular Atrophy) disease in Turkish society. Research data were collected using online applications.

Statistical Analysis

The analysis of the data was carried out using the SPSS 25 package program. Percentage and frequency values for categorical variables, and arithmetic mean and standard deviation or median, minimum and maximum values according to their conformity with normal distribution for quantitative variables, were presented. Comparisons between two independent categorical variables were evaluated with the chi-square test. For the data conforming to the normal distribution, independent sample t-test was used to compare categorical variables with two categories and quantitative variables, and one-way analysis of variance (ANOVA) was used to compare variables with more than two categories and quantitative variables. In the analysis of the relationship between two quantitative variables, the data showing a normal distribution were analyzed with Pearson's correlation. Type I error rate (α) was taken as 0.05 in the study[11].

Results

As a result of the survey study, it was determined that the awareness about SMA disease was higher in individuals with higher education levels (p=0.0001) (Table 1), and the level of awareness about SMA disease decreased with increasing age (p=0.0001) (Table 2). It was determined that 38% of the volunteers thought that SMA disease can be treated with "gene therapy", regarding the level of awareness in the society about the treatment of SMA disease (Table 3). When individuals who had and did not have knowledge about both SMA disease and gene therapy were compared, 28.09% of the individuals had knowledge about both gene therapy and SMA disease, and 30.16% thought that SMA could be treated with gene therapy (Table 4). It was determined that 75% of the individuals participating in the study thought that SMA disease could not be diagnosed early, 20.12% thought that it could be treated with gene therapy and that the Ministry of Health covers the treatment of certain types of SMA patients (Table 5). As a result of the examination of the effect of geographical region and

Table 1. Comparison of education level and level of awareness about SMA disease

Education Level	Having Knowledge About SMA Disease				Total Number of Participants (N)	p
	Participant's Answer					
	Yes		No			
	(N)	%	(N)	%		
Primary school	7	70.00	3	30.00	10	*0.0001
Middle school	10	30.30	23	69.70	33	
High school	28	35.90	50	64.10	78	
Associate degree	264	51.26	251	48.74	515	
Bachelor degree	1450	68.95	653	31.05	2103	
Postgraduate	123	68.71	56	31.29	179	

In the evaluation made by applying the Pearson Chi-square and One-way ANOVA test, it is seen that the awareness about SMA disease is higher in individuals with higher education levels.

Table 2. Comparison of the relationship between age and awareness of SMA disease in the community

Age	Having Knowledge About SMA Disease					
	Participant's Answer				Total Number of Participants (N)	p
	Yes		No			
	(N)	%	(N)	%		
18-24	1338	66.80	665	33.20	2003	*0.0001
25-34	378	66.67	189	33.33	567	
35-44	94	49.47	96	50.53	190	
45-55	58	46.03	68	50.97	126	
56-65	13	41.93	18	58.06	31	

In the analysis made by applying the Pearson Chi-square test, it is observed that the level of awareness about SMA disease decreases with age.

Table 3. Level of Awareness in the Community Regarding the Treatment of SMA Disease with Gene Therapy

Distribution of individuals with knowledge about gene therapy			Individuals' thoughts on the treatment of SMA disease with gene therapy		
Participant's Answer	(N)	%	Participant's Answer	(N)	%
I have information	1223	41.9	Yes It is treatable	1111	38.1
			No It cannot be treated	73	2.5
I don't have information	1696	58.1	I have no idea	1735	59.4

* Frequencies.

Table 4. Comparison of Individuals with and Without Knowledge of SMA Disease and Gene Therapy

A) Having Knowledge About SMA Disease									B) Can SMA disease be treated with gene therapy?								
	Yes		No		I have no idea		Total	p		Yes		No		I have no idea		Total	p
	(N)	%	(N)	%	(N)	%	(N)			(N)	%	(N)	%	(N)	%	(N)	
Have information about gene therapy about SMA disease?									Do you have knowledge								
Yes	820	67.05	44	3.60	359	29.35	1223	*0.0001	Yes	880	46.76	56	2.97	946	50.27	1882	*0.0001
No	291	17.16	29	1.71	1376	81.13	1696		No	230	22.20	17	1.64	789	76.16	1036	

A) *According to the results of the evaluation using the Pearson Chi-Square test, when the individuals who had knowledge about gene therapy and SMA were compared, it was seen that 28.09% of the individuals had knowledge about both gene therapy and SMA disease; **B)** When the opinions of individuals about SMA disease and whether this disease can be treated with gene therapy were compared, it was seen that 30.16% thought that SMA could be treated with gene therapy.

Table 5. Comparison of individuals' knowledge that SMA disease can be treated with gene therapy and that the Ministry of Health covers the treatment of certain types of SMA patients

Do you know that the Ministry of Health covers the treatment of certain types of SMA patients?						
	Yes		No		Total	p
	(N)	%	(N)	%	(N)	
Can SMA disease be treated with gene therapy?						
Yes	587	52.88	523	47.12	1110	*0.0001
No	42	57.53	31	42.47	73	
I have no idea	494	28.47	1241	71.53	1735	

*According to the evaluation made by applying the Pearson Chi-Square test, it is seen that 20.12% of the individuals participating in the study both think that SMA disease can be treated with gene therapy and are aware that the Ministry of Health covers the treatment of certain types of SMA patients.

Table 6. Examination of the effect of geographical region and educational status on consanguineous marriage

	B	S.E.	Wald	df	Sig.	Exp(B)	95% C.I.for EXP(B)	
							Lower	Upper
Geographical Region								
Central Anatolia			30.790	6	0.0001			
Eastern Anatolia	1.069	.316	11.444	1	0.001	2.913	1.568	5.412
Southeastern Anatolia	.065	.353	.034	1	0.855	1.067	.534	2.133
Marmara	.557	.357	2.439	1	0.118	1.746	.868	3.514
Black Sea	.670	.305	4.813	1	0.028	1.954	1.074	3.554
Aegean	.419	.429	.957	1	0.328	1.521	.657	3.522
Other	-.678	.296	5.240	1	0.022	.508	.284	.907
Educational Status								
Primary school			95.584	5	0.0001			
Middle school	-2.271	.821	7.649	1	0.006	.103	.021	.516
High school	-2.302	.549	17.581	1	0.0001	.100	.034	.294
Associate degree	-1.919	.477	16.210	1	0.0001	.147	.058	.374
Licence	-.443	.433	1.048	1	0.306	.642	.275	1.499
Postgraduate	.409	.412	.984	1	0.321	1.505	.671	3.376

*Binary logistic regression.

educational status on the status of consanguineous marriage, it has been shown that Central Anatolia, Eastern Anatolia, Southeastern Anatolia, Black Sea and other regions are related with consanguineous marriages. However, it was determined that the low level of education also had an effect on consanguineous marriages (Table 6).

Discussion

A negative correlation was found between consanguineous marriages and the thought that consanguineous marriages had an effect on SMA disease. Based on these results, it is understood that individuals who had consanguineous marriages did not know that consanguineous marriages had an effect on SMA disease. Awareness of SMA increases as headed east. We attribute this to the high number of patients in those regions. When the individuals with knowledge about gene therapy and SMA were compared, it was determined that 28.09% of the individuals had knowledge about both gene therapy and SMA disease. Today, news and social media posts about SMA disease and gene therapy are thought to have an impact on this result. A positive correlation was found between a known genetic disease and the individual's knowledge of SMA. This is because individuals with genetic diseases are thought to be more interested in such diseases. Based on the determination that individuals who had knowledge about SMA types have received undergraduate and graduate education, it is necessary to train medical students in the early period, especially on the subtypes of the disease, its treatment and gene-therapy. It has been determined that as the level of education increases, there is a decrease in consanguineous marriages and an increase in individuals who think that consanguineous marriages have an effect on SMA disease.

Ethics Committee Approval: The study was approved by the T.C. Health Sciences University Hamidiye Scientific Research Ethics Committee on 29.01.2021 with the meeting number 2021/4 and the decision numbered 4/17, and was carried out in accordance with the Declaration of Helsinki.

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