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ARAŞTIRMA MAKALESİ **RESEARCH ARTICLE**

Assessment of Knowledge, Attitudes and Practices Regarding SMA Carrier Screening and Newborn Screening Programs Among Physicians in Tertiary Healthcare Institutions

Duygu İlke Yıldırım*, Elif Yılgör
Department of Family Medicine, Medical Faculty, Selçuk University, Konya, Turkiye

Abstract

Aim: This study aims to evaluate the knowledge, attitudes, and behaviors of physicians in tertiary healthcare institutions regarding SMA carrier and newborn screening programs.

Materials and Methods: This descriptive, cross-sectional study was conducted between July 17 and August 16, 2024, following approval from the relevant institutional ethics committee. The study included 80% of physicians working in the internal medicine departments of tertiary healthcare institutions. Data were collected through both online and face-to-face methods. The online survey was administered via the online forms, while the face-to-face data collection was carried out using a structured questionnaire developed by the researchers. The questionnaire was designed to evaluate physicians' knowledge, attitudes, and practices concerning Spinal Muscular Atrophy (SMA) carrier screening and newborn screening programs. In addition, it included items on participants' sociodemographic characteristics.

Results: In this study, the mean age of physicians was 27.92 ±3.63 years, with 61.2% being female. Physicians with chronic diseases have significant knowledge about NSP and have significant scores of 50 and above on the SMA knowledge scale. Married physicians, physicians who have children, and physicians who have information about SMA are significantly more likely to source information about NSP from physicians and healthcare professionals. Participants whose source of information about SMA is a physician and physicians who have information about NSP know the majority of NSP tests at a significant rate. It was determined that the knowledge level of physicians, especially those with chronic diseases, was high about NSP and SMA, which can be diagnosed early.

Conclusion: We believe that it would be beneficial to focus on NSP training in medical school education, to organize courses on this subject for general practitioners, and to increase the quality of NSP training for internal medicine physicians in order to reach a sufficient level of NSP and SMA carrier screening in the society.

Keywords: *Newborn Screening, Spinal Muscular Atrophy, Physicians*

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***Sorumlu Yazar / Corresponding Author:** E-mail (DİY): azrailla@hotmail.com. ORCID: 0000-0002-3893-5173
E-mail (EY): elifyilgor@hotmail.com. ORCID: 0009-0007-7287-8049

INTRODUCTION

The Newborn Screening Program (NSP) for endocrine and metabolic diseases is conducted for all newborns to detect serious but treatable conditions. Its goal is to improve the prognosis of newborns diagnosed with these conditions, prevent or mitigate long-term complications, and reduce morbidity and mortality rates (1).

In phenylketonuria, phenylalanine accumulates in the blood, leading to irreversible brain damage. Congenital hypothyroidism, characterized by a deficiency of thyroid hormone, is commonly observed in our country due to endemic iodine deficiency. In individuals with biotinidase deficiency, biotin, a B vitamin, cannot be properly utilized. Cystic fibrosis, a genetic disorder primarily affecting the exocrine glands, impacts the digestive system and lungs. Congenital Adrenal Hyperplasia (CAH) can cause electrolyte imbalances and intellectual disabilities in infants, often accompanied by gender development anomalies, particularly in female infants (2). For newborn screenings, the first heel-prick blood sample is taken from every newborn within 48 hours after the first oral feeding. If screening results are suspicious, the following steps are recommended: "Measure T4 and TSH levels in serum" for congenital hypothyroidism, "Refer to the Sweat Test Center" for cystic fibrosis, "Refer to the Nutrition and Metabolism Clinic" for phenylketonuria and biotinidase deficiency, "Refer to the Endocrinology Clinic" for congenital adrenal hyperplasia, and "Refer to the Relevant Clinic" for spinal muscular atrophy (SMA) (3). Spinal Muscular Atrophy (SMA) is an autosomal recessive neuromuscular disease characterized by progressive muscle weakness and atrophy, resulting from the irreversible loss of motor neurons in the anterior horn of the spinal cord and the cranial nerve motor nuclei. SMA is classified into five types based on the severity of symptoms and the age of disease onset (4).

Prenatal diagnosis for SMA is recommended for couples who are both carriers. If identified, families receive information and genetic counseling. Since 2021, the SMA Carrier Screening Program has been implemented nationwide, offered to couples before marriage and to those already married upon request. The newborn screening program for SMA has been in place since 2022 (5-7).

Current treatments for SMA improve disease progression and prevent complications, but a complete cure has not yet been found. Physicians have encountered SMA prevention efforts during their education and practice. With SMA frequently covered in the media, there is often confusion about the information, and reliable sources are hard to find online. Accurate information provided by knowledgeable physicians is crucial for early diagnosis and treatment. This study aims to evaluate the knowledge, attitudes, and behaviors of physicians in tertiary healthcare institutions regarding SMA carrier and newborn screening programs.

MATERIALS AND METHODS

This study is cross-sectional and descriptive in nature. After obtaining ethical approval, it was conducted between 17.07.2024 and 16.08.2024 with the participation of 80% of physicians working in the internal medicine departments. The sample size was calculated using simple random sampling from the study population, with a power of 80% at an alpha level of 0.05, resulting in the inclusion of 387 physicians."

Data Collection Forms

A three-part data collection form, prepared based on the literature, was used to obtain the data:

1. **Sociodemographic Data Form**: The first section of the form included various sociodemographic characteristics of the participants(age, gender, marital status, parenthood, consanguinity) and medical history (presence of chronic disease). Seven questions were provided, and participants were asked to answer them.
2. **Newborn Screening Tests Knowledge Form**: The scale developed by Newcomb was subjected to validity and reliability testing by Erbay and Yıldız in 2020 (8). This section contained questions related to the knowledge, attitudes, and behaviors of the participants regarding newborn screenings. Eight questions were provided, and physicians were asked to answer them.

3. **SMA Knowledge Scale**: This scale was adapted based on the "Knowledge, Attitudes, and Practices Regarding SMA Carrier Screening" survey conducted by Şahin and colleagues in 2024 (9). A 21-item scale, prepared based on the literature, was used to assess the knowledge level of volunteer participants about SMA. Participants were asked to mark the most appropriate option (I don't know', 'False', or 'True) for each item. Correct answers were scored as 1 point, and incorrect answers were scored as 0 points. The total score obtained was converted into a 100-point grading system. Based on the 25th, 50th, and 75th percentile values of the total scores, knowledge levels were classified as "low," "medium," and "high." The Cronbach's alpha coefficient of the scale was found to be 0.906.

Some physicians working in internal medicine disciplines were given a face-to-face survey, while others were unable to be reached in person, so the survey was conducted online, and their responses were recorded. The identity information of the participating physicians was kept confidential, and the survey duration was approximately 2 minutes. After the participants were verbally informed in detail, their consent was obtained.

Statistical Analysis

Statistical analyses were conducted using the SPSS (Statistical Package for the Social Sciences) version 21.0 software. Descriptive statistical methods, such as frequency(n), percentage (%), mean ± standard deviation, and minimum and maximum values, were used to evaluate the data obtained from the study. For statistical analysis, descriptive data were expressed as numbers and percentages, and comparative analysis was performed using the Chi-square test. All analyses were conducted with a 95% confidence interval. A p-value of less than 0.05 was considered statistically significant.
The data, which were converted into tables and graphs, were included in the findings section.

RESULTS

The study included 387 physicians with a mean age of 27.92 ± 3.63 years, of whom 61.2% were female. Among participants, 59.4% were single, 15.7% had children, and 87.9% reported no chronic illnesses. Additionally, 94.1% were aware of NSP screening tests, with 74.6% obtaining information from healthcare professionals and 19.3% from the media.

Physicians were familiar with an mean of 4.17±1.24 out of five newborn screening tests, with 91.7% reporting knowledge of three or more tests. Awareness levels were as follows: 92.2% for Phenylketonuria, 91.2% for Hypothyroidism, 87.9% for Cystic Fibrosis, 77.3% for Biotinidase, 68.2% for SMA, and 68% for Congenital Adrenal Hyperplasia.

Table 1: SMA Knowledge Scale

	True		False/I Don't Know	
Premises of the SMA Knowledge Scale	n	%	n	%
It is genetically inherited	387	100	0	0
There are different types	381	98.4	6	1.6
SMA can be seen in the babies of carrier couples.	375	96.9	12	3.1
Breathing is affected.	374	96.6	13	3.4
The babies of carrier couples may be carriers of the disease.	372	96.1	15	3.9
Genetic analysis is necessary for diagnosis	372	96.1	15	3.9
History and physical examination are necessary for diagnosis.	364	94.1	23	5.9
Swallowing is affected.	363	93.8	24	6.2
Carrier couples can have healty babies.	355	91.7	32	83
There is bilateral muscle weakness	351	90.7	36	9.3

Nerve-muscle assessment is necessary for diagnosis.	346	89.4	41	10.6
Radiology and laboratory tests are necessary for diagnosis.	341	88.1	46	11.9
Being a carrier does not prevent having children.	340	87.9	47	12.1
Being a carrier does not prevent marriage.	336	86.8	51	13.2
There are symptomatic treatments available for relief.	329	85.0	58	15.0
Intelligence is not affected.	278	71.8	109	28.2
There are treatment options available.	262	67.7	125	32.3
Carrier status is common in the population.	245	63.3	142	36.7
Hearing is not affected.	216	55.8	171	44.2
There is no sensory loss.	186	48.1	201	51.9
Total		**387**		**100**

SMA:Spinal Muscular Atrophy

In this study, 97.2% of physicians believed they had knowledge of SMA, with 24.5% obtaining information from media and 72.7% from healthcare workers. All knew SMA is genetically inherited, 98.4% were aware of its different types, and 96.9% knew it occurs in babies of carrier couples. However, 51.9% incorrectly thought SMA involves sensory loss. Overall, 96.1% scored 50 or above on the knowledge scale, with an mean score of 84.58 ± 16.12. Responses to the SMA Knowledge Scale are detailed in Table 1.

There was a significant difference between those who knew about NSP tests and whose source of information on SMA was doctors-healthcare workers and those whose source of information was media-other (p=0.012). There was a significant difference between those who knew that there were three or more NSP tests and those who knew there were fewer than three among those whose source of information on SMA was doctors(p=0.001). This was analyzed using Yates' Chi-square test.

Table 2: Correlation of NSP Knowledge with NSP Knowledge Level, SMA Knowledge and Sociodemographic Indicators

Are you knowledgeable about NSP?								
Category	Yes		No		Total			
Variable	n	%	n	%	n	%	x^2	*p*
NSP Test Knowledge Level								
Below 3	25	6.5	7	1.8	32	8.3	15.8	**0.001**
3 and Above	339	87.6	16	4.1	355	91.7		
Is SMA a Screening Test?								
Yes	251	64.9	13	3.4	264	68.2	1.02	0.31
No	113	29.2	10	2.6	123	31.8		
Chronic Illness								
Present	39	10.1	8	2.1	47	12.1	11.7	**0.003**

Absent	325	84.0	15	3.9	340	87.9		
Has Children								
Yes	56	14.5	5	1.3	61	15.8	0.65	0.38
No	308	79.6	18	4.7	326	84.2		
Source of SMA Information								
Physicians	270	74.2	11	2.8	281	72.6	6.2	**0.012**
Press Media/Other	94	24.3	12	3.1	106	27.4		
SMA Knowledge Scale Score								
Below 50	9	2.3	6	1.6	15	3.9	32.3	**<0.0001**
50 and Above	355	91.7	17	4.4	372	96.1		
Total	364	94.1	23	5.9	387	100		

Chi-Square.
SMA: Spinal Muscular Atrophy; NSP: Newborn Screening Program

There was a significant difference between physicians with chronic illnesses who scored 50 or above on the SMA Knowledge Scale and those who scored below 50 (p=0.005). Among the physicians who had knowledge of Newborn Screening Tests, there was a significant difference between those who scored 50 or above on the SMA Knowledge Scale and those who scored below 50 (p<0.001). This was analyzed using Fisher's Exact Test (Table 2).

There was a significant difference between physicians with chronic illnesses who had knowledge of Newborn Screening Tests and those who did not (p=0.003). Among the physicians who had knowledge of Newborn Screening Tests, there was a significant difference between those who knew there were three or more tests and those who thought there were fewer than three (p=0.001). This was analyzed using Fisher's Exact Test.

Table 3: Correlation Between NSP Information Sources, SMA Knowledge and Sociodemographic Indicators

	Source of NSP Information							
Category	Physicians		Press Media Other		Total			
Variable	n	%	n	%	n	%	x^2	*p*
Are you knowledgeable about SMA?								
Yes	284	73.4	92	23.8	376	97.2	5.11	**0.03**
No	5	1.3	6	1.6	11	2.8		
Is SMA Screening a Test?								
Yes	196	50.6	68	17.6	264	68.2	0.08	0.43

No	93	24.0	30	7.8	123	31.8		
Marital Status								
Married	106	27.4	51	13.2	157	40.6	7.16	**0.005**
Single	183	47.3	47	12.1	230	59.4		
Has Children								
Yes	35	9.0	26	6.7	61	15.8	10.4	**0.001**
No	254	65.6	72	18.6	326	84.2		
Total	289	74.7	98	25.3	387	100		

Among physicians who were married, there was a significant difference between those whose source of NSP knowledge was doctors-healthcare workers and those whose source was media-other (p=0.005). This was analyzed using Pearson's Chi-square test. Among physicians with children, there was a significant difference between those whose source of NSP knowledge was doctors-healthcare workers and those whose source was media-other (p=0.001). This was analyzed using Yates' Chi-square test. Among physicians who had knowledge about SMA, there was a significant difference between those whose source of NSP knowledge was doctors and those whose source was media-other(p=0.034). This was analyzed using Fisher's Exact Test (Table 3).

Although 91.8% of physicians with children had a knowledge level of 3 or higher out of 5 on NSP tests, no significant difference was found compared to those without children. The majority of participants, whose source of information about NSP was physicians and healthcare workers, knew that SMA is a screening test, but not to a significant degree. Most of the participants with children had knowledge about NSP, but not to a significant degree. The majority of those whose source of information about SMA was physicians scored 50 or above out of 100 on the SMA Knowledge Scale, but not to a significant degree.

DISCUSSION

In our country, blood samples are routinely taken from the heels of all newborns free of charge to screen for diseases such as Phenylketonuria, Congenital Hypothyroidism, Biotinidase Deficiency, Cystic Fibrosis, Congenital Adrenal Hyperplasia, and Spinal Muscular Atrophy (10). Early diagnosis through the Newborn Screening Program (NSP) is critical to prevent irreversible complications. For example, untreated Congenital Hypothyroidism inevitably leads to permanent intellectual disability, and untreated Biotinidase Deficiency can cause muscle weakness, hearing loss, vision problems, and developmental delays. Similarly, Cystic Fibrosis manifests with frequent infections, malabsorption, and poor weight gain, while Congenital Adrenal Hyperplasia can lead to life-threatening adrenal insufficiency if undetected (10).

Spinal Muscular Atrophy (SMA), a preventable public health concern, is notably prevalent in our country. It is characterized by progressive muscle weakness and loss and is inherited genetically. Importantly, SMA does not preclude marriage or having children, and early diagnosis allows for timely intervention to improve prognosis (11).

This study assessed the knowledge, attitudes, and behaviors of physicians working in tertiary healthcare institutions concerning SMA carrier screening and the NSP. The majority of participants demonstrated a high level of knowledge about SMA and the NSP. However, a notable misconception was identified: despite the majority correctly understanding the genetic basis of SMA, over half erroneously believed that

sensory loss is a feature of SMA (12,13). This finding underscores the persistence of certain misunderstandings even among healthcare professionals with direct access to medical information.

Additionally, physicians with chronic illnesses showed significantly higher knowledge about the NSP and scored better on the SMA knowledge scale. Married physicians with children primarily reported other physicians as their main source of information. Interestingly, while most participants who sourced their NSP information from healthcare professionals recognized SMA as a screening test, this was not statistically significant. In contrast, those obtaining information from media or other non-professional sources exhibited lower knowledge levels regarding both NSP and SMA.
The literature predominantly focuses on mothers' knowledge and attitudes towards newborn screening. Bakırlıoğlu and Çetinkaya, for example, found that mothers often acquire information from non-professional sources and that younger mothers are at risk for inadequate knowledge and attitudes. Their better-informed participants, compared to previous studies, were mostly mothers of at least two children, suggesting experience contributes to knowledge (14). Fitzgerald et al. similarly identified maternal age under 25, primiparity, and low education as risk factors for poor knowledge (15). Şahin and colleagues assessed premarital individuals' knowledge on SMA carrier screening, concluding that education, employment, and income influence knowledge levels, and that their participants had moderate to high knowledge (16,17).

Our study offers a novel perspective by focusing on physicians rather than the general public or mothers. The critical insight here is that even healthcare providers with access to current medical information possess limited knowledge on this sensitive and widely debated topic. This raises important questions: Are physicians acquiring knowledge on SMA and NSP similarly to the general public? If not, what factors contribute to these gaps? Possible explanations include insufficient emphasis during medical education, inadequate continuing professional development, or limited exposure to updated guidelines and resources. These issues merit further investigation.

Moreover, our finding that personal experiences such as parenthood do not significantly affect physicians' views on screening contrasts with broader societal data, where parenthood often increases awareness and knowledge. This discrepancy highlights unique professional or cultural dynamics influencing physician attitudes and behaviors, which deserve more detailed exploration.

The limitations of this study include the possibility of hurried or inattentive responses due to the high number of preliminary questions, and its single-center design. Larger, multicenter studies with higher participation rates could strengthen generalizability. A major strength of our study lies in its focus on tertiary care physicians working in specialties not directly involved with primary care screening programs. Evaluating their knowledge of a primary care-related screening program provides valuable insights into information dissemination and awareness gaps within the healthcare system. This perspective adds significant value to the existing literature.

CONCLUSION

According to the results of this study, most physicians working in internal medicine departments at tertiary healthcare institutions demonstrated a high level of knowledge about both the Newborn Screening Program (NSP) and Spinal Muscular Atrophy (SMA). In particular, physicians with chronic illnesses showed significantly higher scores on the SMA knowledge scale, as well as greater awareness of newborn screening tests.

Physicians who reported healthcare professionals as their main source of information had significantly higher knowledge levels compared to those who obtained their information from the media or other sources. This suggests that professional medical communication channels are more effective in conveying accurate and comprehensive information about screening programs. Although a majority of physicians

with children had good knowledge about NSP, this finding was not statistically significant. Similarly, knowledge about SMA as a screening test was more prevalent among those informed by healthcare professionals, though this also did not reach statistical significance. While these findings indicate a generally high level of awareness, they also reveal specific misconceptions—for example, over half of the participants mistakenly believed that SMA involves sensory loss. This highlights the need for further targeted education. Based on these results, it may be beneficial to strengthen educational content related to NSP and SMA within medical training programs. Enhancing awareness through continuing medical education—particularly emphasizing accurate information dissemination among physicians—may improve knowledge levels and reduce misconceptions. Further multi-centered and larger-scale studies are recommended to validate these findings and support the development of more effective educational strategies.

No studies in the literature have focused on physicians' knowledge and attitudes towards NSP and SMA carrier screenings. Therefore, we believe our study will lead future research and help disseminate NSP screenings in newborns, especially SMA carrier screenings, which have become a public health problem, significantly improving prognosis through early diagnosis.

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