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Evaluation of Physicians' Knowledge and Anxiety Levels About Inherited Metabolic Diseases: Survey Study

Ece Öge Enver[1], Bilal Yılmaz[2]

[1]Department of Pediatrics, Dr. Lütfi Kırdar City Hospital, İstanbul, Türkiye
[2]Department of Pediatrics, Dr. Lütfi Kırdar City Hospital, İstanbul, Türkiye

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What does this study add to this topic?

- *The level of concern among physicians regarding congenital metabolic disorders has been revealed.*

Abstract

Objective: Inherited metabolic disorders (IMDs) are rare genetic disorders with complex clinicalpresentations. Due to their rarity and limited presence in medical education, there is a lack of knowledge and increased anxiety among physicians in the management of these diseases. This study aims to evaluate the knowledge and anxiety levels of physicians about IMDs and to determine the factors affecting their professional competence.

Method: A cross-sectional survey was conducted with the participation of 83 physicians across Türkiye. Participants completed an online questionnaire assessing demographic data, knowledge of IMDs, and anxiety about these diseases. The data were analyzed using statistical methods such as *t*-tests and chi-square tests.

Results: The mean knowledge level of physicians was 5.6 ± 2.1 on a 10-point scale, indicating that they had basic knowledge but lacked advanced knowledge. The mean anxiety score was 5.3 ± 2.4, and the most common cause of anxiety was reported as lack of knowledge by 60.2%. It was observed that physicians who saw IMD patients less frequently perceived their level of knowledge to be higher than it was due to their inability to adequately recognize the complexity of these diseases. Although the presence of a metabolic specialist in the institution increased self-confidence, it did not have a significant effect on knowledge and anxiety levels.

Conclusion: This study highlights the importance of systematic educational programs in IMD management by revealing physicians' lack of knowledge and increased anxiety levels. Interactive case-based learning, preparation of national guidelines, and interdisciplinary collaboration are critical to increasing knowledge and improving patient outcomes.

Keywords: inherited metabolic disease, survey, anxiety

Introduction

Inherited metabolic disorders (IMDs) are a complex group of disorders that develop due to genetically based enzyme deficiencies in metabolic processes.[1] These diseases usually present with severe clinical manifestations early in life and can lead to serious consequences such as permanent organ damage, neurological disorders, and death if undiagnosed or untreated. However, depending on the degree of enzyme deficiency, they can also be diagnosed at older ages, which makes metabolic diseases necessary in the differential diagnosis of every age group from 7 to 77.[2]

The difficulties in diagnosing IMDs arise from the fact that they have a broad clinical spectrum. Symptoms are often non-specific and can overlap with many other diseases, such as vomiting, lethargy, seizures, growth retardation, etc. This makes early recognition of IMD difficult.[3] Furthermore, limited access to specific laboratory tests and the lack of widespread genetic analysis are also factors that negatively affect the diagnostic process. In the literature, it has been noted that it takes an average of 5-7 years to diagnose IMD, which worsens patient prognosis.[4]

Early diagnosis and management of IMDs is of significant importance in modern medicine; nevertheless, the wide spectrum, complexity, and rarity of the disease cause knowledge gaps and challenges in diagnostic processes, especially for general physicians and health professionals in other

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Corresponding author: Ece Öge Enver, Department of Pediatrics, Dr Lütfi Kırdar City Hospital, İstanbul, Türkiye **e-mail:** eceoge@gmail.com
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specialties. The limited exposure to these diseases in medical education and their rarity in clinical practice present one of the main difficulties in the diagnosis and treatment of IMDs. These deficiencies indicate that, particularly in the field of rare diseases, there are significant gaps in education. At the same time, the complexity of the clinical, biochemical, and genetic characteristics of metabolic diseases increases doctors' anxiety and causes confusion in managing IMD.[5,6]

In countries like Türkiye, where genetic diseases are quite common, the importance of awareness-raising events for the diagnosis and management of IMDs increases even more. Particularly in societies where consanguineous marriages are common, indicating a higher prevalence of genetically inherited diseases, increasing knowledge and skills of physicians in these diseases is necessary to improve diagnosis and treatment processes[7,8] Still, there is not enough information addressing doctors' training requirements in this field within national health systems.

This study aims to evaluate doctors' level of IMD knowledge, their worries about working with certain diseases, and how this circumstance affects their professional performance. Furthermore, in accordance with the data collected by the questionnaire approach, it aims to identify methods to raise doctors' degrees of awareness and training.

The study is a rare attempt to understand the approach of healthcare professionals to this important disease group and aims to fill the gap in the literature. In this context, the results obtained are expected to contribute to the development of health policies and the creation of specific training programs for IMD management. There is no study on the level of knowledge about metabolic diseases among physicians. This study is important to reveal the concerns of physicians about inherited metabolic diseases and to draw attention to the preparation of educational materials on this subject.

Method

This is a cross-sectional survey study designed to assess physicians' knowledge and anxiety levels about inherited metabolic disorders (IMDs). The presented study was approved by the Clinical Research Ethics Committee, approval number 2024/010.99/10/53.

Participants and Inclusion Criteria

Physicians working in health institutions throughout Türkiye were included in the study. The inclusion criteria were determined as follows:

- providing active clinical service as a physician,
- to have clinical experience or basic knowledge about IMD, and
- accepting participation in the study voluntarily.

Metabolic specialists were excluded from the study to avoid biasing the data.

Questionnaire Structure and Application Process

Participants were administered a 20-question online questionnaire, which was prepared in line with the literature review and expert opinions and tested and validated on a pilot group. The questionnaire consists of 3 main sections:

1. **Demographic Data:** Six questions include demographic data about the respondents such as age, gender, and the hospital they are working in.
2. **Level of Knowledge:** Seven questions ask about the general level of knowledge about congenital metabolic diseases.
3. **Level of Anxiety:**Seven questions assessing the difficulties experienced by physicians when working with IMD and their occupational anxiety.

The questionnaire was designed in a Likert scale format (1: Strongly disagree to 10: Strongly agree) and distributed via an online platform. Prior to accessing the survey, participants were provided with a comprehensive explanation of the study's purpose, data confidentiality, and the voluntary nature of participation. Informed consent was obtained electronically from all respondents. Only after confirming consent were participants granted access to complete the survey. The questionnaire also included open-ended questions to capture individual opinions and suggestions.

Statistical Analysis

The data obtained were analyzed using IBM SPSS (IBM SPSS Corp.; Armonk, NY, USA) 27.0 software. The following methods were applied in data analysis:

- Mean, standard deviation, median, minimum, maximum, frequency, and ratio values were used in the descriptive statistics of the data.
- The distribution of variables was measured by the Kolmogorov-Smirnov and Shapiro-Wilk tests. An independent sample t-test was used to analyze quantitative independent data with a normal distribution.
- Kruskal-Wallis and Mann-Whitney U tests were used to analyze quantitative independent data with non-normal distribution.
- Chi-square test was used in the analysis of qualitative independent data, and Fisher test was used when chi-square test conditions were not met.

Ethical Approval and Disclosure

The study was approved by the ethics committee and conducted in accordance with the Declaration of Helsinki. (Dr. Lütfi Kırdar City Hospital Ethics Committee Approval Date: November 29, 2024/Approval Number: 2024/010.99/10/53).

Results

A total of 83 physicians participated in the study; 68.7% of the participants were female and 31.3% were male. The mean age of the participants was 37.1 ± 7.4 years, 47% were 35 years of age or younger, and 53% were over 35 years of age. The institutions where the participants worked were distributed as follows: 34.9% city hospitals, 25.3% training and research hospitals, 13.3% private hospitals, 10.8% university hospitals, 9.6% state hospitals, 3.6% medical practices, and 2.4% other. The proportion of those who had a specialist in congenital metabolic diseases in their institution was 55.4%, while the proportion of those who did not was 44.6%.

About 67.5% of the participants were pediatricians and 33.8% were working in. other specialties (e.g., pediatric gastroenterology, pediatric nephrology, and family medicine). The rate of those who have received minor training is 32.5%, while the rate of those who have not is 67.5% (Tables 1 and 2).

Level of Knowledge

The knowledge level of the participants about inherited metabolic disorders (IMDs) was evaluated on a scale of 1-10, and the mean knowledge level was found to be 5.6 ± 2.1. No significant

Table 1. Distribution of Demographic Data of Physicians

		Min-Max	Median	Mean ± ss/n-%
Age		25.0 - 60.0	36.0	37.1 ± 7.4
Age	≤ 35			39 47.0
	>35			44 53.0
Gender	Female			57 68.7
	Male			26 31.3
Institution you work for	City Hospital			29 34.9
	Training and Research Hospital			21 25.3
	Private Hospital			11 13.3
	University Hospital			9 10.8
	State Hospital			8 9.6
	Medical Practice			3 3.6
	Other			2 2.4
Is there an inherited metabolic disease specialist at your institution?	No			37 44.6
	Yes			46 55.4
Specialization branch				
Pediatrics				56 67.5
Side branch				
Pediatric gastroenterology				3 3.6
Pediatric nephrology				2 2.4
Pediatric chest diseases				2 2.4
Pediatric emergency				2 2.4
Pediatric endocrinology				2 2.4
Pediatric cardiology				2 2.4
Pediatric intensive care				2 2.4
Gastroenterology				2 2.4
Neonatology				2 2.4
Pediatric assistant				1 1.2
Child neurology				2 2.4
Pediatric hematology				1 1.2
Other				
Family medicine				2 2.4
General practitioner				1 1.2
No information				1 1.2

difference was found between pediatricians and participants from other specialties in terms of knowledge level ($P > .05$). The rate of those who stated that they listened to inherited metabolic disorders lectures during education was 96.4%, but the contribution of these lectures to the level of knowledge was not statistically significant ($P > .05$).

Participants had a high rate of stating that they had difficulty in treatment planning when a patient diagnosed with IMDs presented to the emergency room; when this situation was evaluated on a scale of 1-10, the average score was found to be 5.5 ± 2.4. There was no significant relationship between the level of knowledge about IMDs and the field of study, age group, or subspecialty training status.

Table 2. Score for Feeling Knowledgeable about Inherited Metabolic Disorders

		Min-Max	Median	Mean ± SS	*P*	
Age	≤ 35	2.0 - 10.0	5.0	5.5 ± 2.0	.442	m
	> 35	1.0 - 9.0	6.0	5.7 ± 2.2		
Gender	Female	1.0 - 10.0	5.0	5.6 ± 2.1	.627	m
	Male	1.0 - 9.0	6.0	5.7 ± 2.1		
Working in a university-city-training and research hospital	(-)	1.0 - 9.0	5.0	5.3 ± 2.2	.057	t
	(+)	3.0 - 10.0	6.0	6.3 ± 1.8		
Congenital metabolic diseases specialist at the institution	(-)	1.0 - 9.0	5.0	5.6 ± 1.9	.939	m
	(+)	1.0 - 10.0	6.0	5.6 ± 2.3		
Specialization	Minor Subspecialty	1.0 - 10.0	6.0	5.8 ± 2.4	.355	t
	Pediatrics	1.0 - 9.0	5.0	5.3 ± 1.9		
Completing specialization	(-)	1.0 - 9.0	4.0	5.2 ± 2.8	.328	m
	(+)	1.0 - 10.0	6.0	5.7 ± 1.9		
Minor education	(-)	1.0 - 9.0	6.0	5.7 ± 2.0	.577	m
	(+)	1.0 - 10.0	5.0	5.4 ± 2.3		
Taking metabolic diseases course	(-)	1.0 - 9.0	5.0	5.0 ± 5.7	.926	m
	(+)	1.0 - 10.0	5.0	5.6 ± 2.0		
Frequency of seeing patients diagnosed with inherited metabolic disorder	[1]1-2 per week	2.0 - 9.0	5.0[3]	4.9 ± 1.8	***.002***	K
	1-2 per month	1.0 - 9.0	5.0[3]	5.2 ± 2.0		
	[3]-1-2 per year	1.0 - 10.0	7.0	6.7 ± 2.2		
Inherited metabolic disorders cannot be identified only in the neonatal and infancy period	(-)	1.0 - 10.0	5.0	5.5 ± 2.1	.070	m
	(+)	5.0 - 9.0	7.0	7.2 ± 1.5		
No life expectancy in patients with inherited metabolic disease	Wrong	1.0 - 10.0	6.0	5.8 ± 2.0	***.012***	m
	That's right	1.0 - 5.0	3.0	3.0 ± 2.0		

[K]Kruskal-Wallis (Mann-Whitney *U* test)/[m]Mann-Whitney *U* test/[t]Independent sample *t* test
[3]Difference with 1-2 per year $P < .05$.

Anxiety Level

The anxiety levels of the participants about IMDs were evaluated in the range of 1-10, and the mean anxiety score was calculated as 5.3 ± 2.4. No significant difference was observed between male and female participants in terms of anxiety levels ($P > .05$). The most common causes of anxiety were lack of information (60.2%), complexity of the topics (24.1%), and low frequency of seeing patients (13.3%).

There was no significant difference between the level of anxiety and the presence or absence of a metabolic specialist in the institution where the participant worked ($P > .05$). However, it was found that the anxiety levels of the participants who had listened to a course on metabolic diseases were significantly lower than those who had not ($P < .05$).

Education and Management Competence

Considering IMDs, 51.8% of the participants considered themselves competent in planning examinations. Participants who had a metabolic specialist at their institution of employment reported higher self-confidence than those who did not, but this difference was not statistically significant ($P > .05$).

Statistical Evaluations

The frequency of seeing patients with IMDs showed a positive correlation with the level of knowledge. In particular, it was found that the knowledge level scores of the participants who saw 1-2 patients per year were significantly higher than those who saw patients weekly or monthly ($P < .05$).

Discussion

In this study, physicians' knowledge and anxiety levels about inherited metabolic disorders (IMD) were evaluated. The findings show that physicians experience a lack of knowledge due to the rarity and complexity of IMD and that this deficiency leads to professional concerns. This situation is in parallel with the necessity of increasing education and awareness about rare diseases as stated in the literature.[9]

The mean knowledge level score of the physicians was 5.6 ± 2.1, indicating that they had basic knowledge about IMD but lacked

advanced knowledge. The fact that physicians who see IMD patients less frequently think that their level of knowledge is higher ($P < .05$) may indicate that experience in this field does not directly contribute to knowledge. In addition, this may also be due to the fact that physicians who see metabolic diseases realize how complex this patient group is and do not feel competent, while physicians who see fewer patients do not realize how complex this patient group is and underestimate these diseases. However, this situation also shows that the acquisition of knowledge about rare diseases should be supported not only by clinical experience but also by systematic training.

Due to the rarity of IMDs, the limited coverage of these diseases in medical school and residency training programs, and their rare prevalence in clinical practice are among the main reasons for the lack of knowledge. The fact that autosomal recessive diseases are common in countries like Türkiye, where the rate of consanguineous marriage is high, makes the need for information and education on IMDs even more important. The fact that the rate of consanguineous marriage is as high as 20-25% in our country increases the prevalence of metabolic diseases and requires healthcare professionals to encounter these diseases more frequently.[4]

In terms of education, interactive case-based learning methods should be disseminated, and continuing medical education (CME) programs should be increased. As seen in international examples, the establishment of national guidelines and registry systems for rare diseases can reduce delays in diagnosis and improve patient outcomes. In Türkiye, academic institutions and healthcare professionals need to work in collaboration to address the shortcomings in this area.

Level of Anxiety and Lack of Knowledge

The mean anxiety score of physicians was 5.3 ± 2.4, indicating that difficulties are common in the diagnosis and treatment processes of IMD. Factors such as lack of information, infrequent patient encounters, and complexity of diseases contribute to high anxiety levels. The fact that the most common cause of anxiety is lack of information (60.2%) clearly reveals the need for training in this field. Lack of information may cause clinical insecurity in physicians, leading to delays in diagnostic processes and adversely affecting patients. In particular, the development of special guidelines and decision support systems for the diagnosis and treatment of IMD may contribute to reducing physicians' anxiety levels.[10]

Training and Perception of Competence

The fact that the rate of those who considered themselves competent in planning investigations when IMD was diagnosed was limited to 51.8% shows the physicians' perception of inadequacy in this area. The fact that there was no significant difference between the knowledge and anxiety levels of physicians with subspecialty training and other physicians ($P > .05$) suggests that having or not having subspecialty training other than metabolic diseases does not change the confidence felt by physicians. In terms of education, the dissemination of interactive case-based learning methods, the inclusion of more topics related to metabolic diseases in continuing medical education (CME) programs, and the establishment of national guidelines and registry systems can reduce delays in the diagnostic process.

Institution Factor and Self-confidence

The fact that physicians with a metabolic specialist in their institution had higher levels of self-confidence suggests that a multidisciplinary approach may increase clinical confidence. However, the fact that this difference was not statistically significant may be related to the fact that the presence of a specialist alone does not significantly affect knowledge and anxiety, and that the presence of a specialist who can be consulted creates confidence, as well as the physicians' desire to be competent enough to make decisions on their own. In this case, educational materials and digital handbooks that can be accessed quickly may enable physicians to manage patients on their own without having to find a specialist.

Conclusion

This study evaluated physicians' knowledge and anxiety levels regarding inherited metabolic disorders (IMDs) and identified the main gaps in this field. The results indicated that insufficient knowledge and inadequate guidelines contributed to delays in diagnostic procedures and clinical uncertainties. The fact that there was no significant correlation between the frequency of seeing patients and the level of knowledge emphasizes the importance of educational programs. In nations characterized by elevated rates of consanguineous marriages, such as Türkiye, the significant prevalence of autosomal recessive diseases underscores the necessity of enhancing awareness regarding inherited metabolic disorders (IMDs). It is advisable to organize training programs comprehensively, establish national guidelines, and expand multidisciplinary teamwork. This study highlights the necessity of innovative educational strategies and policy modifications to remedy the knowledge deficit in the management of IMDs.

Data Availability Statement: The data that support the findings of this study are available on request from the corresponding author.

Ethics Committee Approval: Ethics committee approval was received for this study from the Ethics Committee of Kartal Dr.Lutfi Kırdar City Hospital (Date: 29.11.2024, Number: 2024/0l0.99/l0/53).

Informed Consent: Informed consent was obtained electronically from all respondents who participated in this study.

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