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Assessment of Family Physicians' and Family Physician Assistants' Knowledge of Cystic Fibrosis Disease and Screening Tests

Aile Hekimlerinin ve Aile Hekimliği Asistanlarının Kistik Fibrozis Hastalığı ve Tarama Testi Hakkındaki Bilgi Düzeylerinin Araştırılması

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Abstract

Aim: Since January 1, 2015, our country's newborns have been screened for cystic fibrosis (CF) in primary health care services. Currently, CF is recognized as a childhood disease, yet more than 50% of patients are over the age of 18, and the median survival is 44.4 years. Therefore, family physicians play a role in managing the disease that extends beyond the neonatal period to include follow-up with adult patients. This study aimed to investigate primary care physicians' and family medicine residents' knowledge of CF and screening.

Matherial and Method: The study population consisted of family physicians and family medicine residents working in the city center of Konya. The online questionnaire included questions designed to measure the participants' sociodemographic characteristics and their level of knowledge about CF. A total of 230 volunteer physicians participated in the study.

Results: Family medicine specialists had a higher knowledge score than the other participants. The mean knowledge score on the questionnaire, which had a maximum score of 44 points, was 27.16±5.57. Family physicians demonstrated a high level of knowledge regarding screening tests and organ involvement in cystic fibrosis (CF). 95.7% of physicians knew that recurrent lung infections are a sign of CF, while 60.4% knew that regular salt use is necessary. Only 20% of participants were aware of cystic fibrosis-related diabetes.

Conclusion: Early diagnosis of cystic fibrosis (CF) is crucial for preserving lung function. Therefore, it would be beneficial for primary care physicians to receive regular training on CF diagnosis and follow-up, as well as information about CF-related complications in patients with prolonged life expectancy.

Keywords: Family medicine, cystic fibrosis, family physician

Öz

Amaç: Ülkemizde 1 Ocak 2015 tarihinden itibaren birinci basamak sağlık hizmetlerinde yenidoğanlarda kistik fibrozis (KF) tarama programı uygulanmaktadır. Günümüzde KF, medyan sağkalım süresi 44,4 yıl olan bir çocukluk çağı hastalığı olarak bilinmektedir ve hastaların %50'den fazlası 18 yaşın üzerindedir. Bu nedenle, aile hekimlerinin hastalık yönetimindeki rolü yenidoğan dönemi ile sınırlı olmayıp, yetişkin hastaların takibini de içermektedir. Bu çalışmanın amacı birinci basamak hekimlerinin ve aile hekimliği asistanlarının KF ve tarama konusundaki bilgilerini araştırmaktır.

Gereç ve Yöntem: Çalışmanın evrenini Konya il merkezinde görev yapan aile hekimleri ve aile hekimliği asistanları oluşturmuştur. Çevrimiçi anket formu, katılımcıların sosyodemografik özelliklerini ve KF hakkındaki bilgi düzeylerini ölçmek için tasarlanmış bilgi sorularını içermektedir. Çalışmaya toplam 230 gönüllü hekim dahil edilmiştir.

Bulgular: Aile hekimliği uzmanlarının bilgi puanı diğer katılımcılara göre daha yüksekti. Katılımcıların maksimum 44 puan alabileceği ankette ortalama bilgi puanı 27.16±5.57 idi. Aile hekimlerinin KF'de tarama testi ve organ tutulumu hakkında bilgi düzeyleri yüksekti. Tekrarlayan akciğer enfeksiyonlarının KF bulgusu olduğunu bilen hekimlerin oranı %95.7 iken, düzenli tuz kullanımının gerektiğini bilen hekim oranı %60.4'tü. Katılımcıların sadece %20'si kistik fibrozis ilişkili diyabet hakkında bilgi sahibiydi.

Sonuç: KF'nin erken tanısı akciğer fonksiyonlarının korunması için çok önemlidir. Bu nedenle birinci basamak hekimlerinin KF tanısı ve takibi konusunda düzenli eğitim almaları ve yaşam beklentisi uzamış KF hastalarının erişkin yaşamındaki komplikasyonlar konusunda bilgilendirilmeleri yararlı olacaktır.

Anahtar Kelimeler: Aile hekimliği, kistik fibrozis, aile hekimi

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INTRODUCTION

Cystic fibrosis (CF) is one of the most common autosomal recessive genetic disorders in Caucasians, with an incidence of one in 2500-3500 live births. The disease is caused by mutations in CFTR, a cystic fibrosis transmembrane conductance regulator gene encoding a chloride and bicarbonate channel expressed in the apical membrane of epithelial cells.[1] CF is a multisystem disease that most commonly affects the respiratory system (bronchiectasis, sinusitis), pancreas (CF-related diabetes with endocrine dysfunction, malabsorption with exocrine dysfunction), gastrointestinal system (distal bowel obstruction syndrome, biliary liver disease), and reproductive system (congenital absence of vas deferens in males, decreased fertility in females).[2,3]

CF screening is included in newborn screening programs in many countries. In our country, it has been included in the screening program of the Ministry of Health since January 1, 2015. The aim of the program is early diagnosis of the disease and initiation of treatment before clinical findings appear. In our country, Immune reactive trypsinogen (IRT) is checked in the heel blood as a screening method, the patient is recalled in case of values of 90 mmol and above and IRT is checked again in the heel blood and if it is 70 mmol and above, the screening test is considered positive and the patient is directed to sweat test.[4,5] In a patient with a positive IRT screening test, two positive sweat tests or genetic evidence of the disease is diagnostic, even if clinical findings have not yet occurred. Patients diagnosed early through the screening program have been shown to have more normal weight, height and body mass index, better pulmonary function tests and longer life expectancy.[6,7] Primary care physicians play an important role in the effective continuation of the CF screening program, which has a significant impact on quality of life and disease prognosis.

While CF was initially described as a fatal childhood disease in 1938,[8] the median projected survival has since increased to 44.4 years. Currently, more than 50% of individuals living with CF are aged 18 years or older, indicating that it is not only a childhood disease, but also a condition from which affected individuals transition into adulthood. Consequently, the role of primary care providers, such as family physicians, becomes pivotal not only during the screening program phase but also in subsequent follow-ups as patients progress through different stages of life.[9]

The objective of the present study was to assess the knowledge level of family physicians and family medicine assistants, who are in frequent contact with CF patients and newborns who constitute the screening population, regarding CF disease and screening.

MATERIAL AND METHOD

The study was carried out with the permission of Necmettin Erbakan University Pharmaceutical and Non-Medical Device Research Ethics Committee (Date: 09.07.2024, Decision No: 2024/5131). All procedures were carried out in accordance with the ethical rules and the principles of the Declaration of Helsinki.

The population of the cross-sectional and descriptive study consisted of family physicians and family physician assistants working in Türkiye, Konya city center. In the questionnaire form, in addition to questions regarding socio-demographic information such as age, year of graduation, title, etc., informational questions prepared by the researchers according to the literature were included to measure the basic level of knowledge about CF with multiple-choice questions. The online questionnaire form was shared on social media platforms used by primary care physicians in Konya a total of three times with an interval of one month. Between October 10, 2024 and December 12, 2024, the survey link was active and a total of 230 physicians voluntarily completed the survey. The identity information of the participants was kept confidential.

To assess the knowledge level of the participants about the diagnostic methods, screening test, clinical findings and treatment modalities of CF disease, 15 knowledge questions were prepared for the participants. Each correct answer by the participants was scored as 1 (one) point and the other answers were scored as 0 (zero) points. The knowledge scores for CF disease and screening test were calculated according to the answers to the information questions. Some questions had more than one correct answer. When the total knowledge scores were analyzed, the lowest score that participants could receive from the knowledge questions was zero and the highest score was 44.

Statistical Analysis

Data from the study were analyzed using the SPSS v.27 statistical program. Percentage distributions were used to examine descriptive characteristics, and measures of central tendency and prevalence (mean, standard deviation, median, etc.) of continuous variables were calculated. For further analysis, statistical significance was accepted as $p<0.05$ with a 95% confidence interval.

The Shapiro-Wilk test was used to assess the conformity of continuous variables to the normal distribution. Student t test was used to compare numerical data with two groups that fit the normal distribution, and one-way ANOVA test was used to compare more than two groups. After the ANOVA test, the Tukey test was used in the post hoc analysis to determine the group/groups from which the difference originated.

To test the agreement between continuous variables, Spearman correlation analysis was performed after assessing their conformity to the normal distribution.

RESULTS

A total of 230 volunteer family physicians and family physician assistants in the city center of Konya were part of the study.

The occupational distribution of participants is shown in **Figure 1**. The median age was 34 years (min:25; max:60). The mean time since graduation was 12.36±9.13 years (min:0; max:35). Of the participants in the residency program, 64.2% (n=86) reported receiving a pediatric rotation. Of the residents who received a pediatric rotation, 61.6% (n=53) were family medicine residents and 38.4% (n=33) were contracted family medicine residents.

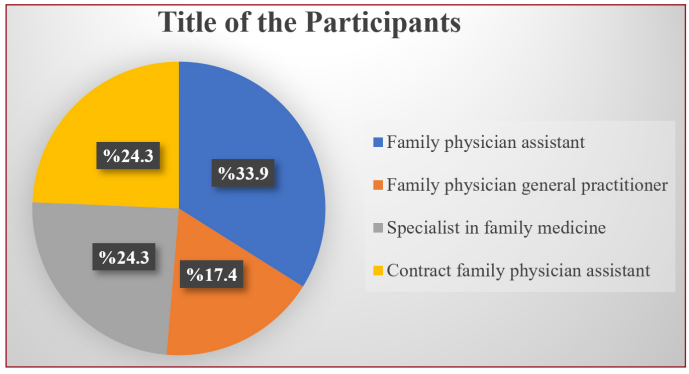

Figure 1.

The distribution of the responses of the family physicians and family physician assistants who participated in the study to the information questions about CF disease and screening test is shown in **Table 1** and **Table 2**.

When analyzing the total knowledge score, the lowest score was 13 (0.4%; n=1) and the highest score was 40 (1.7%; n=4). The mean knowledge score of the participants was 27.16±5.57 points.

There was no statistically significant difference between the title of the physician and the status of receiving education about CF, following patients diagnosed with CF and referring patients with a pre-diagnosis of CF (p>0.05). Specialists in family medicine were found to have a higher level of knowledge about CF than other physicians (general practitioner-family medicine assistant, p=0.035; general practitioner-family medicine specialist, p<0.001; general practitioner-family medicine assistant, p=0.007). The comparison of knowledge scores about CF disease and screening test according to some socio-demographic and professional background characteristics of the participants is shown in **Table 3**.

There is a weak and negative correlation between the age and length of study of the participants and their knowledge scores about CF disease and screening test (r=-0.052 and r=-0.022, respectively). However, this relationship was not statistically significant (p=0.44 and p=0.74, respectively). The correlation analysis between the age of the participating physicians and the time elapsed since graduation and their knowledge scores regarding CF disease and screening test is shown in **Table 4**.

Table 1. Distribution of responses to the questions about CF disease and screening tests (questions 1-7) by family physicians and family physician assistants who participated in the study.

1-Sample with newborn screening test for cystic fibrosis (n= 230)		
Heel prick test*	199	86.5
Sweat	30	13.1
Venous blood	1	0.4
2-Definitive diagnostic method(s) for cystic fibrosis (n=230) †		
Genetic mutation analysis *	199	86.5
Sweat test*	158	68.7
Measurement of the nasal potential difference *	46	20.0
3-Inheritance of cystic fibrosis (n= 226) ‡		
Autosomal recessive *	193	85.4
Dependent on X	12	5.3
Autosomal dominant	11	4.9
Sporadic	10	4.4
4-The system is not expected to be involved in cystic fibrosis (n=230)		
Neurological system *	216	93.9
Pancreas	10	4.4
Sweat glands	2	0.9
Respiratory system	1	0.4
Gastrointestinal system	1	0.4
5-Neonatal/infancy manifestations of cystic fibrosis (n=230) †		
Recurrent lung infection*	220	95.7
Growth retardation*	200	87.0
Wheezing*	182	79.1
Meconium ileus*	163	70.9
Diarrhea*	155	67.4
Nasal polyp	107	46.5
Skin manifestations	106	46.1
Diabetes mellitus	58	25.2
Edema*	48	20.9
Pseudo-bartter syndrome*	45	19.6
Osteoporosis	23	10.0
6-CF-suspicious microorganisms in sputum or throat cultures(n=230) †		
*P. aeroginosa**	158	68.7
K. pneumonia	107	46.5
M. cataralis	84	36.5
*S. aureus**	62	27.0
*S. maltophilia**	53	23.0
S. pneumonia	41	17.8
Respiratory syncytial virus	19	8.3
7-Gastrointestinal tract finding(s) of cystic fibrosis (n=230) †		
Meconium ileus*	174	75.7
Diarrhea*	171	74.3
Vomiting*	128	55.7
Constipation*	100	43.5
Jaundice*	95	41.3
Distal intestinal obstruction syndrome *	88	38.3
Rectal prolapse*	72	31.3

*Correct answer, †More than one option can be marked, ‡Four participants did not respond.

Tablo 2. Distribution of responses to the questions about CF disease and screening tests (questions 8-15) by family physicians and family physician assistants who participated in the study.

8-Respiratory manifestation(s) of cystic fibrosis (n=230) †		
Recurrent lung infection *	227	98.7
Wheezing*	206	89.6
Bronchiolitis *	168	73.0
Nasal polyp*	139	60.4
Sinusitis*	108	47.0
Barrel chest*	38	16.5
9-Evidence of late cystic fibrosis in patients with mild mutations (n=230)		
Infertility*	156	67.8
Nasal polyp	47	20.4
Sinusitis	20	8.7
Diarrhea	7	3.0
10-Late complication of cystic fibrosis (n=230)		
Bronchiectasis	105	45.7
Diabetes*	46	20
Clubbing	45	19.6
Osteoporosis	24	10.4
Hipersplenism	10	4.3
11-Treatment method(s) used in cystic fibrosis (n=230) †		
Pancreatic enzyme*	188	81.7
Bronchodilator*	165	71.7
Dornaz alpha*	146	63.5
Salt*	139	60.4
Multivitamin*	102	44.3
12-Sign(s) of pulmonary exacerbation in cystic fibrosis (n=230) †		
Cough, increased sputum, respiratory distress*	181	78.7
New-onset cough, shortness of breath, loss of appetite and wheezing*	148	64.3
New infiltration on chest radiography that was not present before*	141	61.3
New findings on auscultation (rales, rhonchi, etc.) *	112	48.7
Newly grown microorganism*	108	47.0
13-In cystic fibrosis, the first thing that comes to mind in the patient who receives pancreatic enzyme and presents with abdominal pain and gas (n=230)		
Inadequate enzyme intake*	159	69.1
A normal finding	48	20.9
Hunger	14	6.1
Vitamin deficiency	9	3.9
14-The organ most affected by cystic fibrosis (n=230)		
Lung*	228	99.1
Liver	2	0.9
Brain	-	-
Heart	-	-
15-Why salt is not recommended in patients with cystic fibrosis (n=230)		
They lose Na and Cl through sweat *	218	94.8
They lose Na and Cl in urine	5	2.2
They lose Na and Cl in feces	5	2.2
There is a lack of uptake	2	0.9

* Correct answer, † More than one option can be marked.

Table 3. Comparison of knowledge scores about cystic fibrosis disease and screening test according to some sociodemographic and occupational background characteristics of the participants

Feature	Knowledge Score		
	n	X̄±SS(Mean)	Test
Age			
34 years and below	115	27.49±5.71	t=0.870 p=0.385
35 years and older	110	26.84±5.50	
Time after graduation (years)			
9 years and below	118	27.15±5.72	t=-0.60 p=0.952
10 years and above	111	27.11±5.42	
Title			
Family physician assistant	78	26.88±5.13	F=7.613 p<0.001*
Family physician general practitioner	40	24.05±5.93	
Specialist in family medicine	56	29.25±4.94	
Contract family physician assistant	56	27.68±5.59	
Pediatrics rotation status of assistant physicians			
Yes	86	27.66±5.64	t=1.303 p=0.195
No	48	26.42±4.65	
Participation in any training on cystic fibrosis			
Yes	55	27.60±5.71	t=0.669 p=0.504
No	175	27.02±5.54	
Follow-up status of patients diagnosed with cystic fibrosis†			
Yes	42	28.38±5.64	t=1.681 p=0.094
No	160	26.78±5.48	
Referral of patients with a prediagnosis of cystic fibrosis‡			
Yes	48	28.25±5.36	t=1.528 p=0.128
No	166	26.87±5.54	

*Family physician general practitioner - Family physician assistant, p=0.035; Family physician general practitioner - Specialist in family medicine, p<0.001; Family physician general practitioner - Contract family physician assistant, p=0.007
†Participants who stated that they did not remember the follow-up of patients diagnosed with cystic fibrosis were excluded.
‡Participants who stated that they did not remember referring patients with a prediagnosis of cystic fibrosis were exclude

Table 4. Correlation analysis between the age and graduation period of the physicians participating in the study and their knowledge scores on CF disease and screening test

		Total Score
Age	r*	-0.052
	p	0.44
	n	225
Time after graduation	r*	-0.022
	p	0.74
	n	230

* Spearman correlation was performed due to skewed distribution.

DISCUSSION

Cystic fibrosis is a disease that is screened by primary care physicians, and the present study is one of the few studies to investigate the knowledge of primary care physicians and family medicine residents about CF. We believe that the results are important for raising awareness in primary care. It can be said that the knowledge of CF among the participating physicians is at an intermediate level. According to the National Cystic Fibrosis Registry System (NCRS) 2023 data, 336 (15.04%) of the 2234 CF patients living in Turkey were 18 years and older. The age at which patients are diagnosed

with CF can vary from 1 month to 41 years.[10] Since CF can be diagnosed at any age and patients can reach adulthood, it is important to determine the level of knowledge of family physicians about this disease. When the total knowledge score was analyzed in our study, the mean score of the participants in the knowledge questions was 27.16±5.57 points, which can be scored as high as 44 points. Accordingly, it can be said that the knowledge of the physicians participating in the present study about CF disease and screening is at a moderate level.

Since January 1, 2015, newborns in our country are screened for CF by heel-prick blood sampling. The newborn screening test is performed by measuring IRT. Babies with a positive screening test are referred to a higher center for sweat testing as soon as possible (available at: www.kistikfibrozisturkiye.org). Approximately 90% of the physicians who participated in our study were aware that heel-stick blood screening and genetic mutation analysis should be performed for definitive diagnosis. The other test that should be performed for definitive diagnosis was the sweat test, and about 70% of the physicians knew this information. These results suggest that physicians' awareness about the screening program and CF diagnostic tests is high in our country. The screening program allows infants to be diagnosed at an early stage, which makes treatment and follow-up more beneficial.

In our study, when the knowledge level of the participants about the early signs of CF was examined, almost all of the participants knew that frequent lung infections could be an early sign, while about 15% did not know that CF could cause growth retardation. In a study by Cesur et al, the frequency of CF was found to be 5.3% in children presenting to a pediatric clinic with the complaint of recurrent lung infection, while the frequency of CF was found to be 8.8% in children presenting with growth retardation.[11] In children with frequent lung infections, height and weight percentile values should be monitored in primary care clinics. If there is a growth pause, we think it may be useful to evaluate the patients for CF. The rate of those who responded nasal polyps and osteoporosis to the findings seen in the neonatal and infant periods was 46.5% and 10%, respectively. We believe that it would be appropriate to increase awareness of this issue since both findings are seen in the late period.

When renal tubular functions are normal in the presence of hyponatremia, hypochloremia, hypokalemia and metabolic alkalosis, this picture is called Pseudo-Bartter Syndrome (PBS) and can be observed especially in CF patients in infancy. In a study conducted by Eyuboglu et al. on CF patients in our country, the rate of PBS was found to be 10% and it was shown that PBS patients were diagnosed earlier.[13] Only about 20% of the physicians who participated in our study were informed about PBS. Considering the hot climate of our country and the insistence of families to dress their babies tightly, the possibility of CF patients presenting with PBS may have increased. Therefore, we think it is important to increase the awareness of PBS among primary care physicians. Infertility is an important presenting symptom of adult CF. This group of patients has rarer mutations and generally does not have pancreatic insufficiency. While 95% of male CF patients are infertile and this is related to the absence of vas deferens, fertility in female CF patients is lower compared to the normal population due to malnutrition and dark cervical mucus.[14] In our study, the awareness of infertility among the participants was found to be about 70%. We recommend a detailed history regarding CF in primary care, especially for male infertility.

The prevalence of cystic fibrosis-related diabetes (CFRD) increases markedly with age, with a prevalence of 2% in childhood, 19% in adolescence, and 40% to 50% in adulthood in people with CF.[15] CFRD is associated with increased morbidity and mortality. Adequate nutritional status is known to be critical for maintaining lung function and survival in CF. Deterioration of pulmonary and nutritional status begins 2 to 4 years before the actual diagnosis of CFRD.[16,17] In our study, only 20% of the physicians were aware that CFRD is a late complication. Because the nutritional approach to diagnosis, follow-up, and treatment of CFRD differs from that of type 1 and type 2 diabetes mellitus, there is a need for education to increase awareness of this issue among primary care physicians.

When the relationship between the age of the physicians in our study and the time elapsed since graduation and the total knowledge score on the test was examined, no statistically significant difference was found. On the other hand, the mean knowledge score of general practitioners was found to be statistically significantly lower than that of participants with other titles. Based on these results, we believe that physicians who continue or complete their specialty training in a tertiary hospital after medical school have a higher awareness of cystic fibrosis and that additional studies should be conducted to increase the knowledge level of general practitioners.

When the questions about CF treatment were analyzed in our study, 80% of the physicians knew that pancreatic enzyme was used in treatment, about 60% knew that inhaled dornase alfa and salt were used, and about 50% of the physicians knew that multivitamins were used. Awareness of the use of salt is still not at the desired level. Since CF-related PBS is common in our country, it may be useful to increase the level of physicians' knowledge about salt treatment in infants.

In Asseri's study, primary care physicians correctly answered 13.5% of the questions about CF treatment modalities.[18] In 2019, in a study conducted by Demirtaş et al. among primary care physicians in our country, the rate of physicians who knew about daily salt treatment was 23.5%, the rate of physicians who knew about inhaled dornase alfa use was 37.4%, and the rate of physicians who knew about pancreatic enzyme treatment was 34.8%.[19] We believe that the level of knowledge of our physicians about CF treatment has increased due to the increase in the number of CF centers

in the last 5 years, the fact that these patients have a longer life expectancy with new treatments, and the increase in the number of patients diagnosed through screening.

Limitations

The study has limitations in that it only included family physicians in the Konya province. The presence of a Department of Pediatric Chest Diseases in the province may have increased the physicians' level of knowledge. Studies with more participants from across the country may also be useful for determining primary care physicians' knowledge levels.

CONCLUSION

Life expectancy and quality of life have increased significantly with the increasing number of treatment options in CF. The level of knowledge of primary care physicians has also increased in recent years. We believe that the increasing number of pediatric pulmonologists in our country in recent years is also effective in this regard. Early diagnosis of CF is very important both for prevention of malnutrition and for preservation of lung function. Therefore, it would be beneficial for family physicians to be regularly trained in CF diagnosis and follow-up and informed about adult complications of CF.

Abbreviations

CF: Cystic fibrosis, **CFRD**: Cystic fibrosis-related diabetes, **CFTR**: Cystic fibrosis transmembrane conductance regulator, **IRT**: Immune reactive trypsinogen, **NCRS**: National Cystic Fibrosis Registry System, **PBS**: Pseudo-Bartter Syndrome

ETHICAL DECLARATIONS

Ethics Committee Approval: The study was carried out with the permission of Necmettin Erbakan University Pharmaceutical and Non-Medical Device Research Ethics Committee (Date: 09.07.2024, Decision No: 2024/5131).

Informed Consent: All patients signed the free and informed consent form.

Referee Evaluation Process: Externally peer-reviewed.

Conflict of Interest Statement: The authors have no conflicts of interest to declare.

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Author Contributions: All of the authors declare that they have all participated in the design, execution, and analysis of the paper, and that they have approved the final version.

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