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Knowledge, clinical practices, and educational needs regarding cystic fibrosis among general pediatricians in Türkiye: A cross-sectional survey

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Abstract

Although general pediatricians play a key role in the early recognition of cystic fibrosis and timely referrals, real-world data on cystic fibrosis-related knowledge and practice patterns are limited in settings with nationwide newborn screening. This study aimed to evaluate general pediatricians' knowledge, awareness, and clinical practices related to cystic fibrosis, and to identify potential gaps that may affect early diagnosis and management. We conducted a cross-sectional, questionnaire-based online survey of general pediatricians actively practicing in Türkiye (November–December 2025). The survey included demographics, a 13-item cystic fibrosis knowledge assessment (score range 0–13), clinical practice items (reported descriptively), and education/training needs. A total of 412 pediatricians were included (mean age 37.81±5.63 years; 54.1% female). The mean knowledge score was 10.11±1.32. Clinically relevant uncertainty was observed for newborn screening interpretation: 35.0% selected “not sure” for the statement “cystic fibrosis cannot be present if newborn screening is negative.” Referral frequency for suspected cystic fibrosis was low (frequently 1.5%, rarely 88.3%, never 10.2%). Self-perceived adequacy in diagnosing/referring suspected CF was moderate (2.94±0.82) and strongly correlated with knowledge score ($r=0.647$, $p<0.001$). Only 19.2% reported adequate cystic fibrosis education during medical school; 78.4% reported a need for additional training, with face-to-face courses and online webinars being the most preferred formats. In conclusion, general pediatricians demonstrated moderate-to-high cystic fibrosis knowledge overall, but substantial uncertainty in newborn screening interpretation and very low referral frequency for suspected cystic fibrosis. Targeted, practice-oriented educational interventions and streamlined referral pathways may help reduce delays in diagnosis and improve linkage to specialist cystic fibrosis care.

Keywords: Cystic fibrosis, medical education, knowledge assessment

Introduction

Cystic fibrosis (CF) is a multisystem autosomal recessive genetic disorder caused by mutations in the cystic fibrosis transmembrane conductance regulator (CFTR) gene. These mutations lead to abnormal chloride transport across epithelial cells. CF primarily affects the respiratory and gastrointestinal systems, and it is one of the most common life-limiting inherited conditions among children in Europe and North America [1,2]. Advances in diagnosis, nutritional support, multidisciplinary care, and the introduction of CFTR modulator therapies have substantially improved the survival rate and quality of life of patients with CF in recent decades [3].

Early diagnosis is a key factor in determining long-term outcomes in CF. Newborn screening programs enable the earlier identification of affected infants, allowing for the prompt initiation of treatment and specialized follow-up, which are associated with improved growth parameters, better pulmonary function, and reduced morbidity [4,5]. However, the effectiveness of these programs depends heavily on physicians' ability to correctly interpret screening results, recognize early clinical manifestations, and promptly refer patients to specialized CF centers.

General pediatricians play a pivotal role in the early detection and long-term care of children with CF, particularly in primary and

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secondary healthcare settings. They are often the first physicians to evaluate infants and children presenting with nonspecific symptoms such as failure to thrive, recurrent respiratory infections, or steatorrhea. Despite their critical role, previous studies have suggested that pediatricians may have knowledge gaps and exhibit variability in clinical practice regarding CF. This variability is particularly evident in their interpretation of newborn screening results and referral pathways [6-8].

Despite the growing body of literature evaluating CF awareness among primary care physicians, data focusing specifically on general pediatricians remains limited, particularly in countries where newborn screening programs have been implemented nationwide. It is essential to understand pediatricians' knowledge levels, clinical approaches, perceived barriers, and educational needs to optimize early diagnosis and ensure timely referral to specialized CF centers. Therefore, this study aimed to evaluate general pediatricians' knowledge, awareness, and clinical practices related to CF, and to identify potential gaps that may affect early diagnosis and management. The findings of this study may contribute to the development of targeted educational strategies and support the optimization of CF care pathways.

Material and Methods

Study design and participants

This cross-sectional, questionnaire-based study aimed to evaluate the knowledge, awareness, and clinical practices related to CF among general pediatricians. The study was conducted via an online survey platform from November to December 2025. Necmettin Erbakan University, School of Medicine ethical committee approval was obtained before starting the study (Decision no. [2025]/5959). All procedures were conducted in accordance with the principles of the Declaration of Helsinki. Participation was voluntary, and the confidentiality of responses was strictly maintained.

Eligible participants were general pediatricians who were actively practicing in Türkiye at the time of the study. Participation was voluntary, and no incentives were offered. Physicians who did not complete all mandatory sections of the questionnaire were excluded from the final analysis.

Survey Instrument

Data were collected using a structured questionnaire developed by the authors. The questionnaire was based on a review of existing literature and previously published surveys that assessed CF-related knowledge among physicians. Prior to dissemination, the questionnaire was pilot-tested with a small group of general pediatricians to evaluate its clarity, relevance, and the time required for completion. Minor revisions were made accordingly.

The final survey consisted of four sections: (i) demographic and professional characteristics (age, sex, years of practice, type of healthcare institution, and prior experience with CF patients, including the approximate number of patients followed); (ii) a knowledge assessment comprising 13 items covering CF genetics, clinical manifestations, diagnostic approaches,

complications, prognosis, and current treatment options; (iii) clinical practices and approaches addressing diagnostic strategies for suspected CF, symptom recognition, referral behaviors, perceived barriers to appropriate referral, and self-reported confidence in managing/referring suspected CF; and (iv) education and training needs exploring prior CF education during medical school and residency, information sources used to update CF knowledge, perceived need for additional education, and preferred educational formats.

Knowledge items were scored by assigning one point to each correct response, while incorrect and "not sure" responses were scored as zero. A total CF knowledge score (range: 0-13) was calculated by summing correct responses, with higher scores indicating greater CF-related knowledge. Self-perceived competence in diagnosing and referring patients with suspected CF was assessed using a 5-point Likert scale (1=not adequate, 2=slightly adequate, 3=moderately adequate, 4=adequate, 5=very adequate), with higher scores indicating greater perceived adequacy.

For selected items assessing clinical practice and care pathways, response options were not scored as correct/incorrect; instead, they were reported descriptively. Where interpretive grouping was used (e.g., "core CF multidisciplinary team" vs "additional/consulted specialties"), this was based on established standards-of-care documents [4-5,9-11].

The questionnaire was administered electronically via Google Forms, and the responses were anonymized. Participants were required to provide electronic informed consent before accessing the survey.

The minimum required sample size was calculated based on estimation of a proportion in a cross-sectional study. Assuming an expected adequate knowledge prevalence of 50%, a 95% confidence level, and a margin of error of 5%, the minimum sample size was calculated as 384 participants. To account for potential non-response and incomplete questionnaires, the target sample size was increased by approximately 20%.

Statistical Analysis

Statistical analyses were performed using IBM SPSS Statistics for Windows, Version 26.0 (IBM Corp., Armonk, NY, USA). Continuous variables were assessed for normality and are presented as mean±standard deviation or median (interquartile range), as appropriate. Categorical variables are reported as frequencies and percentages. For multiple-response items, each option was reported as the number and percentage of participants selecting that option (percentages may exceed 100%).

The total CF knowledge score was analyzed as a continuous variable. Associations between knowledge score and demographic or professional characteristics were evaluated using the Student's t test for two-group comparisons. For comparisons across more than two groups, one-way ANOVA was performed followed by Bonferroni-adjusted post-hoc tests where appropriate. The association between the total knowledge score and self-perceived adequacy score was assessed using

Spearman’s correlation coefficient. A two-sided p-value of <0.05 was considered statistically significant.

Result

Participant characteristics

A total of 412 general pediatricians were included in the study. The mean age was 37.81±5.63 years, and 54.1% were female. Most participants had completed their residencies at training and research hospitals (59.5%), and most were currently employed at training and research hospitals (60.4%). Prior CF follow-up experience was common: 71.4% had followed one to five CF patients, while 7.0% had no prior experience following CF patients (Table 1).

Table 1. Demographic and professional characteristics of the participants	
Characteristic	Value (n [%] or mean±SD)
Age (years), mean±SD	37.81±5.63
Gender, n (%)	
Male	189 (45.9)
Female	223 (54.1)
Residency training institution, n (%)	
University hospital	167 (40.5)
Training and research hospital	245 (59.5)
How many years have you been working as a pediatrician?, n (%)	
0-5 years	177 (43)
6-10 years	88 (21.4)
11-15 years	118 (28.6)
>16 years	29 (7)
Type of current hospital, n (%)	
Training and research (Tertiary) hospital	249 (60.4)
Secondary state hospital	130 (31.6)
University hospital	24 (5.8)
Private hospital	9 (2.2)
Have you ever followed a patient diagnosed with cystic fibrosis?, n (%)	
0 patients	29 (7)
1-5 patients	294 (71.4)
6-20 patients	59 (14.3)
>20 patients	30 (7.3)

Responses to CF knowledge items

Participants demonstrated high accuracy on several core knowledge statements, including CF inheritance (95.4% correct), ΔF508 as the most common CFTR mutation (90.0% correct), and sweat testing as a reliable diagnostic tool (87.9% correct).

However, notable uncertainty emerged in clinically relevant areas. For the statement "CF cannot be present if newborn screening is negative," 61.9% selected the correct response, while 35.0%

answered "not sure." Similarly, for the statement "All patients are followed in CF centers after diagnosis," responses were dispersed, with 38.1% selecting "not sure," indicating uncertainty regarding post-diagnosis care pathways. Table 2 summarizes the responses to CF knowledge.

Knowledge score and factors associated with knowledge level

The mean total knowledge score was 10.11±1.32. There was no difference in knowledge scores by gender (p=0.644). However, scores were higher among pediatricians who completed residency at a university hospital than at a training and research hospital (10.22±1.27 vs. [9].94±1.37; p=0.035).

Scores differed across years of practice (p<0.001), with the lowest mean score observed in the group with more than 16 years of experience (9.21±1.01). Post hoc analysis revealed that the>16 years group differed significantly from the other experience groups.

Knowledge scores also varied by current workplace (p=0.008). The lowest score was among pediatricians working in private hospitals (9.00±1.22). Post hoc comparisons showed that the private hospital group differed significantly from the university and training-and-research hospital groups.

Higher knowledge scores were associated with greater CF patient follow-up exposure (p<0.001), with a graded increase from those reporting 0 patients (9.69±0.93) to those reporting>20 patients (10.70±1.02). Post hoc testing revealed significant differences between the groups with lower exposure (0 and 1-5 patients) and higher exposure (6-20 and>20 patients). Table 3 shows the comparison of knowledge scores according to demographics.

Clinical practices, perceived barriers, self-efficacy, and educational needs

When asked about the initial approach when CF is suspected, 65.5% selected a referral based on symptoms. Meanwhile, 31.3% selected sweat testing and 3.2% selected genetic analysis as the initial step.

The most frequently selected suggestive symptoms were poor weight gain (96.3%), steatorrhea (94.9%), and chronic cough (93.4%). However, 25.9% also selected allergic rhinitis, which was a nonspecific option. This suggests a potential over-attribution of nonspecific symptoms.

In the context of a CF-diagnosed patient presenting with a complaint, 78.2% indicated that they would refer to pediatric pulmonology.

Although a multidisciplinary approach was broadly endorsed (e.g., pediatric pulmonology 99.5%, gastroenterology 91.2%, child psychiatry 89.5%), a substantial proportion also selected disciplines labeled as “not essential but trained and trusted referrals” such as pediatric allergy/immunology (69.9%).

Regarding referral behavior for suspected CF, only 1.5% reported frequent referral, while 88.3% reported rare referral and 10.2% reported never referring.

Table 2. Responses to cystic fibrosis knowledge assessment

Knowledge statement / Item	True	False	Not sure
CF is an autosomal recessive genetic disease.	393 (95.4)*	15 (3.6)	4 (1)
ΔF508 is the most common CFTR mutation in CF.	371 (90)*	3 (0.7)	38 (9.2)
Sweat test is a reliable diagnostic tool for CF.	362 (87.9)*	14 (3.4)	36 (8.7)
CF cannot be present if newborn screening is negative.	13 (3.2)	255 (61.9)*	144 (35)
Meconium ileus may be seen in CF.	337 (81.8)*	10 (2.4)	65 (15.8)
In all CF patients, respiratory symptoms start early in life.	34 (8.3)	271 (65.8)*	107 (26)
Pancreatic enzyme deficiency increases stool fat content in CF.	321 (77.9)*	8 (1.9)	83 (20.1)
CF must be ruled out in children with recurrent bronchiolitis attacks.	329 (79.9)*	43 (10.4)	40 (9.7)
The rate of infertility in CF is low.	13 (3.2)	366 (88.8)*	33 (8)
CFTR modulator therapies can change the natural course of CF.	375 (91)*	3 (0.7)	34 (8.3)
All patients are followed in CF centers after diagnosis.	119 (28.9)	136 (33)*	157 (38.1)
CF is a disease affecting only childhood.	7 (1.7)	396 (96.1)*	9 (2.2)
CF-related diabetes is a common complication.	252 (61.2)*	75 (18.2)	85 (20.6)
Data were presented as n (%). *: correct answer, CF: cystic fibrosis, CFTR: cystic fibrosis transmembrane conductance regulator			

Table 3. Responses to cystic fibrosis knowledge assessment

Variable	Score	p value
All participants	10.11±1.32	
Gender		
Male	10.07±1.33	0.644
Female	10.13±1.30	
Residency training institution		
University hospital	10.22±1.27	0.035
Training and research (Tertiary) hospital	9.94±1.37	
How many years have you been working as a pediatrician?		
a. 0-5 years	10.34±1.29	
b. 6-10 years	10.10±1.16	<0.001*
c. 11-15 years	9.98±1.42	
d.>16 years	9.21±1.01	
Type of current hospital		
a. Training and research hospital	10.20±1.30	
b. Secondary state hospital	9.93±1.27	0.008**
c. University hospital	10.46±1.44	
d. Private hospital	9.00±1.22	
As a pediatrician, have you ever followed up with a patient diagnosed with cystic fibrosis?		
a. 0 patient	9.69±0.93	
b. 1-5 patients	10.01±1.36	<0.001***
c. 6-20 patients	10.51±1.23	
d.>20 patients	10.70±1.02	
*: Bonferroni: d<a,b,c (p<0.001, p=0.008, and p=0.024 respectively), **: Bonferroni: d < a,c (p=0.041 and p=0.027 respectively), ***:Bonferroni: a<c,d (p=0.034 and p=0.018 respectively); b<c,d (p=0.042 and p = 0.033 respectively). Data were presented as mean±standard deviation		

The most commonly reported challenge in the diagnostic approach was recognizing symptoms (67.0%), followed by follow-up and treatment planning (18.7%).

Most participants believed that CF patients could be appropriately followed in their region (93.4%). The most frequently reported barrier complicating referral was family socioeconomic barriers (58.2%), followed by lack of a CF center in the region (26.9%).

The self-perceived adequacy in diagnosing/referring suspected CF was moderate (2.94 ± 0.82 on a 1-5 Likert scale). This score

was significantly correlated with the CF knowledge score ($r=0.647$, $p<0.001$).

Only 19.2% reported adequate CF education during medical school, while 78.4% reported a need for additional education.

A total of 33.2% reported not updating their CF knowledge; the most commonly used resources included UpToDate (18.4%) and colleagues (15.7%). Preferred educational formats were face-to-face courses (36.7%) and online webinars (28.6%) (Table 4).

Table 4. Clinical practices, perceived barriers, self-efficacy, and educational needs related to cystic fibrosis among general pediatricians

Item / Question	p value
Initial approach when CF is suspected, n (%)	
Sweat test	129 (31.3)
Genetic analysis	13 (3.2)
Referral based on symptoms	270 (65.5)
Symptoms considered suggestive of CF , n (%)	
Poor weight gain	397 (96.3)
Steatorrhea	391 (94.9)
Chronic cough	385 (93.4)
Rectal prolapse	308 (74.7)
Meconium ileus	329 (79.8)
Allergic rhinitis*	107 (25.9)
Prolonged jaundice	224 (54.3)
If a CF-diagnosed patient presents with a complaint, n (%)	
Continue follow-up	90 (21.8)
Refer to pediatric pulmonology	322 (78.2)
Which disciplines are essential for CF care?, n (%)	
Pediatric pulmonology	410 (99.5)
Pediatric gastroenterology	376 (91.2)
Pediatric infectious diseases	317 (76.9)
Child psychiatry	369 (89.5)
Pediatric endocrinology	86 (20.9)
Otolaryngology	71 (17.2)
Physical therapy and rehabilitation	68 (16.5)
Pediatric allergy/immunology**	288 (69.9)
Pediatric neurology**	77 (18.7)
Pediatric cardiology**	69 (16.7)
Referral frequency for suspected CF, n (%)	
Frequently	6 (1.5)
Rarely	364 (88.3)
Never	42 (10.2)

Table 4. Continued

Item / Question	p value
Most challenging step in CF diagnostic approach, n (%)	
Recognizing symptoms	276 (67)
Requesting & interpreting sweat test	23 (5.6)
Appropriate referral	18 (4.4)
Informing family	14 (3.4)
Follow-up & treatment planning	77 (18.7)
None	4 (1)
Belief that CF patients can be appropriately followed in your region, n (%)	
Yes	385 (93.4)
No	3 (0.7)
Partly	21 (5.1)
No idea	3 (0.7)
Main factor complicating referral, n (%)	
Hospital referral chain / appointment system	42 (10.2)
Lack of CF center in region	111 (26.9)
Family socioeconomic barriers	240 (58.2)
Own knowledge gap	19 (4.6)
Self-perceived adequacy in diagnosing/referring suspected CF (Likert scale)	2.94 ± 0.82
Adequate CF education during medical school, n (%)	
Yes	79 (19.2)
No	333 (80.8)
Followed CF patients during residency training, n (%)	
Yes	217 (52.7)
No	195 (47.3)
Sources used to update CF knowledge, n (%)	
Congress/symposium	54 (13.1)
Journals/books	33 (8)
UpToDate	76 (18.4)
Colleagues	65 (15.7)
Social media	47 (11.4)
I do not update	137 (33.2)
Need additional education on CF, n (%)	
Yes	323 (78.4)
No	89 (21.6)
Preferred educational format, n (%)	
Face-to-face course	151 (36.7)
Online webinar	118 (28.6)
Short video–podcast	85 (20.6)
Written guideline and its summary	58 (14.1)
CF: cystic fibrosis, *: Allergic rhinitis is not a specific symptom for CF (5,9). **: These disciplines are not essential in standard CF care, but they are trained and trusted referrals (10)	

Discussion

In this cross-sectional online survey of general pediatricians in Türkiye, we observed a moderate-to-high overall level of CF knowledge, accompanied by clinically relevant gaps that may directly affect early diagnosis, timely referral, and family counseling. The mean knowledge score (10.11 ± 1.32 on a 13-item scale) indicates that most respondents recognized core CF concepts such as autosomal recessive inheritance, the diagnostic role of sweat testing, and the impact of CFTR modulators. Nevertheless, uncertainty clustered around practical, high-stakes areas—particularly the interpretation of newborn screening (NBS) results and post-diagnostic care pathways—where delayed or incomplete action can translate into preventable morbidity.

A prominent finding was the degree of uncertainty related to NBS interpretation. Although many participants correctly rejected the statement that CF cannot be present with a negative NBS, a substantial proportion selected “not sure,” indicating hesitation rather than confident understanding. This is important because NBS is a risk-identification strategy, not a diagnostic test; symptomatic children can still have CF despite a negative screen, and abnormal screening results require structured confirmatory steps. [12,13] Standards of care emphasize that the benefits of NBS depend on timely confirmatory testing, consistent communication with families, and rapid linkage to specialist services [13-16]. Therefore, even modest knowledge gaps at the primary care level may lead to missed or delayed diagnoses, inconsistent counseling, or unnecessary parental distress.

In parallel with the identified knowledge gaps, our findings related to practice suggest a knowledge-implementation gap. Although most respondents indicated that CF patients could be treated in their region, and many selected referral-based approaches when CF was suspected, the reported referral frequency for suspected cases was strikingly low. Most participants described referrals as rare or nonexistent. This discrepancy implies that the barriers extend beyond knowledge and may involve system constraints, workflow issues, or uncertainty about the urgency and logistics of referrals. Notably, respondents frequently cited socioeconomic barriers and limited center availability as reasons for referral difficulty, underscoring the structural determinants of access. Previous studies have shown that CF patient outcomes improve when care is delivered in high-performing, specialized centers where multidisciplinary teams provide structured monitoring and escalation [9,17-19]. Center-based analyses have also demonstrated variability in outcomes across care sites, showing that organizational and delivery structures influence longitudinal health trajectories. [14] From a public health standpoint, strengthening referral pathways and minimizing logistical friction during the initial diagnostic period could have significant long-term benefits.

We also found that knowledge scores varied among professional subgroups. Pediatricians with more experience treating CF patients demonstrated higher knowledge scores, which is

consistent with experiential learning and repeated engagement with guideline-based care. Conversely, the lowest scores were observed among clinicians with more than 16 years of experience and among those working in private hospitals. While cross-sectional data cannot establish causality, these patterns align with the “knowledge obsolescence” phenomenon observed in rapidly evolving fields. Modern CF care has substantially changed due to advances in early detection, chronic therapies, and mutation-targeted treatments [3,20-22]. As CF management becomes more specialized and dynamic, targeted continuing medical education for clinicians with less routine exposure may be valuable, especially for those in settings where CF is rarely encountered.

Several response patterns suggest areas where recognition of symptoms could be improved. Classic red-flag symptoms, such as poor weight gain, steatorrhea, chronic cough, and meconium ileus, were frequently selected. However, a quarter of respondents also identified allergic rhinitis as a symptom of CF. This may reflect a tendency to attribute nonspecific respiratory symptoms to CF or to expand the CF phenotype beyond the features that are most predictive of the disease. In practice, improving recognition at the front lines requires emphasizing symptom clusters and context (e.g., chronic wet cough, recurrent lower respiratory infections, signs of malabsorption, and growth faltering). It is also important to reinforce that a NBS does not eliminate the need for a symptom-driven evaluation when clinical suspicion is high [13,23]. Therefore, practical decision aids focused on “what to do today” when CF is suspected may be more impactful than purely theoretical content.

With respect to multidisciplinary care, respondents strongly endorsed a team-based approach. This aligns with contemporary standards that emphasize multidisciplinary CF centers as the backbone of comprehensive care [9-11,15]. However, the pattern of selected disciplines indicates variability in how pediatricians conceptualize “essential” CF teams versus consultative specialties. It is important to note that contemporary care models increasingly recognize that CF teams can be adapted to local resources and that additional specialties can become relevant depending on comorbidities, complications, or local service structures [10]. Therefore, rather than labeling selections as “incorrect,” it is more scientifically defensible to interpret these patterns as reflecting diverse perceptions of care coordination needs. This perspective is also consistent with modern efforts to redefine and clarify team roles within CF care delivery. In the context of survey research, such findings support interventions that clarify role delineation and referral triggers, such as when input from endocrinology, allergy/immunology, ear, nose, and throat, psychiatry, or rehabilitation is indicated, rather than simply testing for a single “correct” configuration.

A clinically meaningful finding was the strong correlation between self-perceived competence and knowledge score. While confidence is not a surrogate for competence, it can influence behavior, particularly in time-sensitive processes

such as communicating abnormal screening results or arranging confirmatory testing. Previous studies have shown that primary care providers have difficulty notifying families of CF NBS results and navigating uncertainty. These studies also suggest that communication support could reduce distress and improve follow-through [24,25]. Taken together, our data suggest that interventions targeting both knowledge and self-efficacy (e.g., structured scripts, short counseling checklists, and rapid-access consultation pathways) may be more effective than knowledge-focused education alone.

This cohort had substantial educational needs. Only a minority reported adequate CF education during medical school, and many participants reported not regularly updating their CF knowledge despite acknowledging a need for additional training. Given the growing complexity of CF care, which ranges from chronic pulmonary maintenance therapies to modulator-era disease trajectories, educational strategies must be accessible and clinically actionable [3,20-22]. Notably, respondents favored face-to-face courses and online webinars, suggesting that blended, case-based continuing medical education modules could be well received. A high-yield curriculum for general pediatricians might prioritize the following: (i) NBS interpretation and confirmatory testing; (ii) evaluating symptoms despite negative NBS; (iii) urgent referrals and interim management while awaiting specialist review; (iv) counseling language balancing reassurance and urgency; and (v) local referral logistics and shared care responsibilities after diagnosis.

This study has several strengths, including its large sample size and its structured instrument, which covers knowledge, practice patterns, perceived barriers, and educational preferences. However, limitations should also be considered. First, self-reported data may not accurately reflect observed clinical behavior and can be influenced by social desirability bias. Second, voluntary online participation introduces selection bias, potentially enriching the sample with clinicians who are more interested in CF. Third, although the questionnaire was developed with expert input and piloted for clarity, further psychometric validation (e.g., test-retest reliability and construct validity) would strengthen its application in the future. Finally, heterogeneity in healthcare systems across cities may influence referral feasibility independently of individual knowledge, which limits causal interpretations of practice patterns.

Conclusion

In conclusion, although pediatricians demonstrated strong foundational awareness of CF, they had notable gaps in NBS interpretation, counseling, and system navigation. Practice patterns suggest that structural barriers and workflow uncertainties may contribute to under-referral despite perceived regional care availability. These findings support targeted, practical educational interventions and the optimization of clear, low-friction referral pathways-particularly at the earliest points of the CF care continuum where primary pediatrics has the greatest impact.

Conflict of Interests

The authors declare that there is no conflict of interest in the study.

Financial Disclosure

The authors declare that they have received no financial support for the study.

Ethical Approval

This study was approved by the Necmettin Erbakan University, School of Medicine ethical committee (Decision no. [2025]/5959), (12.09.2025).

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