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Gaps in knowledge and practice for familial hypercholesterolemia among physicians caring for children in Saudi Arabia

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Abstract. – OBJECTIVE: Familial hypercholesterolemia (FH) is an inherited genetic disorder that can lead to an early-onset cardiovascular disease if left untreated. Proper awareness of FH among physicians, particularly those taking care of children can facilitate early detection, diagnosis, and treatment. However, data regarding the knowledge, awareness, and practices of physicians in relation to FH are limited worldwide, so we aimed to explore this aspect.

MATERIALS AND METHODS: This cross-sectional study was conducted among physicians who would potentially treat pediatric patients at governmental hospitals and primary health care centers in the Qassim region of Saudi Arabia. A self-administered questionnaire consisting of 28 questions that assessed the physicians' knowledge, awareness, and practices related to FH was distributed among the participants.

RESULTS: In total, 148 physicians were recruited, including pediatricians, family physicians, and primary care physicians (males *vs.* females, 52.7% *vs.* 47.3%; most common age group, 31-40 years [44.6%]), of which 46.6% were familiar with FH. The total mean knowledge score was 5.54 (standard deviation, 3.12) out of 15 points, with 53.4%, 36.5%, and 10.1% of the participants showing inadequate, moderate, and adequate knowledge levels, respectively. The knowledge score was significantly better in the older age group (H=16.155; *p*=0.01) and among pediatric endocrinologists (H=16.155; *p*=0.001), while it was significantly lower among resident physicians (H=6.575; *p*=0.037) and those who had been practicing for 1-5 years (H=5.329; *p*=0.021).

CONCLUSIONS: The current study established significant gaps in the awareness, knowledge, and practices related to FH. Pediatric endocrinologists had the best levels of knowledge of FH. These findings emphasize the need for more extensive educational programs regarding the screening, diagnosis, and management of FH.

Key Words:
Familial hypercholesterolemia, Knowledge, Children, Saudi Arabia.

Introduction

Familial hypercholesterolemia (FH) is an inherited autosomal dominant disorder caused by multiple gene defects that hinder the recycling of low-density lipoprotein (LDL). If left without proper management, it results in long-standing high levels of LDL cholesterol, causing early onset of atherosclerotic cardiovascular disease and accelerating the development of such diseases by 10-20 years[1-6]. FH should be considered in children showing persistent LDL levels >160 mg/dL, especially those with a family history of premature cardiovascular diseases[7,8]. Furthermore, there are four different diagnostic systems that can be used to diagnose FH[4,7,8].

FH is one of today's most prevalent genetic disorders, it remains largely undiagnosed and undertreated, especially in the pediatric age group[3,4,6,7]. Globally, the prevalence of FH is about 1 in 200-300 individuals[3,4,6,9,10]. In Europe, the estimated prevalence of FH is approximately 1 in 300 individuals, whereas in the United States and Denmark, the prevalence is approximately 1 in 200-250 individuals[4,11-13]. In South Africa, the prevalence is higher at 1 in 72 individuals[14]. Thus far, there have been no epidemiological studies on FH in Saudi Arabia, even though it is not uncommon. However, one study on the prevalence of FH in Arabian Gulf countries reported that the condition was found in 1 in every 112 individuals[15]. Nevertheless, based on the global prevalence rate, the approximate number of people with FH in Saudi Arabia is between 63,000 and 158,000, and

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further research on this condition is essential[16].

After conducting a review of the literature, we found at least three studies on physician awareness regarding this condition in Saudi Arabia. The studies[17-19] demonstrated a significant lack of awareness regarding FH among family physicians and internists. Moreover, this appears to be a global trend[20-24]. The responsibility of early diagnosis of FH relies heavily on pediatricians, family physicians, and general practitioners in primary health care centers, and they have an important role in improving the detection and management of FH[8]. The National Heart, Lung and Blood Institute and other societies recommend universal lipid profile screening between the ages of 9 and 11 years and then between the ages of 17 and 21 years. However, earlier and more frequent screening is recommended in patients with underlying risk factors or a family history of premature cardiovascular disease[9]. Cascade screening can start as early as two years of age in patients with a family history of premature cardiovascular disease or FH. In such cases, other family members should also be screened[8,25,26].

Despite the importance of appropriate awareness of FH among physicians who are responsible for treating pediatric patients, there are no published national- or international-level studies on the knowledge, awareness, and practices related to FH among these physicians of different specialties. Thus, our study aimed to assess FH awareness and knowledge among pediatricians, family physicians, and general practitioners in Qassim, Saudi Arabia.

Materials and Methods

Study Design, Setting, and Institutional Review Board Approval

This cross-sectional study was conducted among general pediatricians, Pediatric Endocrinologists, Cardiologists, family physicians, and primary care providers by using a self-administered online questionnaire. This study was conducted between June and August 2021 at 51 primary health care centers and five general hospitals in Qassim region, Saudi Arabia. Each participant was interviewed by the data collectors and provided informed consent before receiving the questionnaire, which was assigned in electronic form and prepared in English. This study was approved by the institutional review board of the Regional Ethics Committees in the Qassim region.

Questionnaire

The questionnaire has been used in two different previous studies conducted in Saudi Arabia[17,19], and it was originally developed by Bell et al[21]. However, it has been modified and validated by expert pediatric lipid physicians to target physicians caring for children. The changes are as follows: physicians' specialties were changed to target physicians who treat the pediatric age group, including family physicians. Smoking as a risk to increase CVD choice was removed since we target pediatric patients. National Lipid association criteria (NLA) for diagnosis of FH has been added. We also added questions to measure the awareness of universal lipid screening in pediatrics, the importance of testing lipid profile even in a non-fasting state and the initial treatment of FH in children. LDL cut-off for intervention and target of children with FH were changed according to the pediatric age levels. Moreover, therapy choices were modified to the FDA-approved treatment for pediatrics and awareness of lines of treatment of homozygous FH were added besides heterozygous FH.

The questionnaire was divided into three parts: the first part evaluated demographic characteristics, including age, sex, level of training, and years of practice. The second part assessed the physicians' knowledge of FH and was composed of 15 questions, and the participants asked to select the most correct option. The third part consisted of 10 items.

Statistical Analysis

Categorical and continuous variables had been summarized by using numbers, percentages, means and standard deviations. The knowledge toward hypercholesterolemia has been assessed using 15-item questions (Table I). Perceived correct responses were coded as 1 and incorrect responses as 0. The total score was calculated by adding all 15 items. Scores ranging from 0 to 15 points were obtained, indicating that the higher the score, the higher the level of knowledge regarding FH. The total knowledge score was divided into three categories to represents the level of knowledge: inadequate knowledge (0 to 5 points), moderate knowledge (6-10 points), and adequate knowledge (>10 points). Non-parametric tests (Mann Whitney Z-test and Kruskal Wallis H-test) were used to compare the participants' total knowledge score according to their socio-demographic characteristics. Statistical collinearity was determined by using the Shapiro Wilk test and Kolmogorov-Smirnov test. The knowledge score

Table I. Assessment of the knowledge of hypercholesterolemia (FH)[(n=148)].

Statement	N (%)
1. Correctly described FH	104 (70.3%)
2. Correctly identified the prevalence of heterozygous FH in the general population	34 (23.0%)
3. Correctly identified the transmission rate to first degree relatives	34 (23.0%)
4. Correctly identified the CHD risk in untreated FH	11 (07.4%)
5. Correctly identified the age threshold for premature CHD in males and females	24 (16.2%)
6. Correctly identified that genetic testing was not required to accurately diagnosis FH	68 (45.9%)
7. Correctly recognized Elevated Lipoprotein (a) to further increase the cardiovascular risk of someone with FH	31 (20.9%)
8. Aware of the clinical algorithms to diagnose patients with FH	
• The Simon Broome criteria	106 (71.6%)
• The Dutch Lipid Clinic Network DLCN criteria	125 (84.5%)
• The US MedPed Program	113 (76.4%)
• Montreal-FH-Score	131 (88.5%)
• NLA criteria	126 (85.1%)
9. The most prevalent age for screening young people for FH in a family with premature CHD and elevated LDL in the family was 0-6 years	14 (09.5%)
10. The most prevalent age for screening young people with FH was 9-11 years	39 (26.4%)
11 Aware of the cascade screening for patients with FH	106 (71.6%)
12. Aware of the untreated fasting lipid levels at which FH may be suspected in children and adolescents was >4.9 mmol/L (> 190 mg/dL)	31 (20.9%)
13. Aware that <3.4 mmol/L (< 130 mg/dL) was the LDL target for children with FH	27 (18.2%)
14. Aware that lifestyle modification was the initial intervention for children with FH	110 (74.3%)
15. Aware that patients with FH should receive statin medications as first-line treatment	112 (75.7%)
Knowledge score (mean ± SD)	5.54 ± 3.12
Level of knowledge	
• Inadequate knowledge	79 (53.4%)
• Moderate knowledge	54 (36.5%)
• Adequate knowledge	15 (10.1%)

FH – Familial hypercholesterolemia.

was deemed non-normally distributed. Therefore, non-parametric tests were applied. Furthermore, the participants' level of familiarity with FH was compared according to their socio-demographic characteristics by using Chi-square test. A *p*-value of 0.05 was considered statistically significant. All statistical data were analyzed using Statistical Packages for Social Sciences (SPSS) version 26 (SPSS Copr., Armonk, NY, USA).

Results

Sample Characteristics and Familiarity with FH

In total, 148 physicians participated in the study. Table II describes the socio-demographic characteristics of the physicians. The most common age group was 31-40 years (44.6%), and more than half of the participants (52.7%) were males. The majority of the participants were pediatricians (38.5%). Resident physicians constituted 42.6% of the study population, while consultants constituted 30.4%. Furthermore, 27.7% of the participants had been practicing for 1-5 years, while 27% had been practicing for 11-15 years.

Knowledge of FH Screening and Management

Table I presents the findings of the 15-item questionnaire used to assess physicians' knowledge of FH. The mean knowledge score was 5.54 (SD, 3.12), with 53.4%, 36.5%, and 10.1% of the participants being classified into the inadequate, moderate, and adequate knowledge categories, respectively. The details regarding each knowledge element and the correct answers are shown in Table I. The proportion of physicians who were aware of the Simon Broome criteria, the Dutch Lipid Clinic Network DLCN criteria, the US MedPed Program, Montreal-FH-Score, and the NLA criteria as clinical algorithms used to diagnose patients with FH were 71.6%, 84.5%, 76.4%, 88.5%, and 85.1%, respectively.

As shown in Table III, the knowledge score was significantly higher among physicians who were

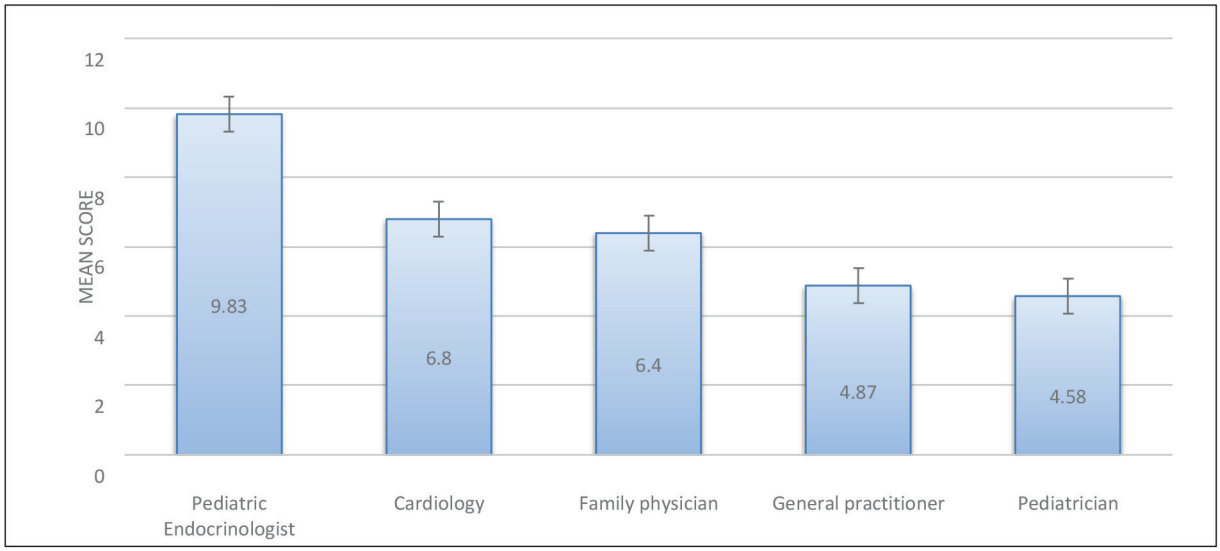

Figure 1. Knowledge score among specialties.

aged >40 years (H = 8.288; $p = 0.016$), those who were pediatric endocrinologists (H = 16.155; $p = 0.001$), and those who were familiar with FH (Z = -2.309; $p = 0.021$). On the other hand, the knowledge score was significantly lower among physicians who were in resident training (H = 6.575; $p = 0.037$) and those who had been practicing for 1-5 years (H = 5.329; $p = 0.021$). Pediatric endocrinologists had significantly higher knowledge scores than other specialists ($p = 0.001$) (Figure 1).

Practices Related to FH Management

In Table IV, for patients with premature coronary artery disease (CAD), physicians routinely performed examinations for arcus cornealis and tendon xanthomata, obtained a detailed family history of CAD, and screened close relatives for hypercholesterolemia (82.4%). More than two-thirds of the physicians (66.9%) indicated family medicine physicians as the healthcare providers that could effectively detect FH at an early stage. The proportion of physicians who had previously diagnosed patients with FH was 33.1%, and 38.5% had been conducting follow-up visits for FH patients. The proposed method for the detection of FH in current practice is through laboratory comments on the lipid profile alerting physicians to the possibility of FH (37.8%). Furthermore, approximately 42.6% of the physicians would routinely screen patients and their close relatives to check their lipid profile. Likewise, only 31.8% of physicians were aware of a specialist to refer patients for examination of lipid disorders. In addition, most physicians (72.3%) considered fasting to be a right practice before lipid profile screening (72.3%). The most common combination of drugs administered to patients with severe hypercholesterolemia was statin + ezetimibe (43.9%).

Familiarity with FH

When asked about their familiarity with FH, 53.4% of the physicians responded that they were not familiar, while 46.6% considered themselves familiar with this condition (Table II). In assessing the relationship between familiarity with FH and the socio-demographic characteristics of the patients, we observed that unfamiliarity with FH was more common among pediatricians ($\chi^2 = 10.878$; $p = 0.012$), those in the resident level of training ($\chi^2 = 11.778$; $p = 0.003$), and those who had been practicing for 1-5 years ($\chi^2 = 8.142$; $p = 0.043$), while familiarity with FH was more common among participants aged 31-40 years ($\chi^2 = 6.194$; $p = 0.045$; (Table V).

Awareness of Treatment Options and Cardiovascular Risks

Figure 2 characterizes the awareness of medications/interventions for pediatric heterozygous and homozygous FH patients. The most common medication/intervention was liver transplant (41.9%), followed by PCSK9 inhibitors (26.4%) and lipid apheresis (23.6%). Figure 3 presents a summary of physician responses to the most important factors that increase the cardiovascular risk in patients with FH.

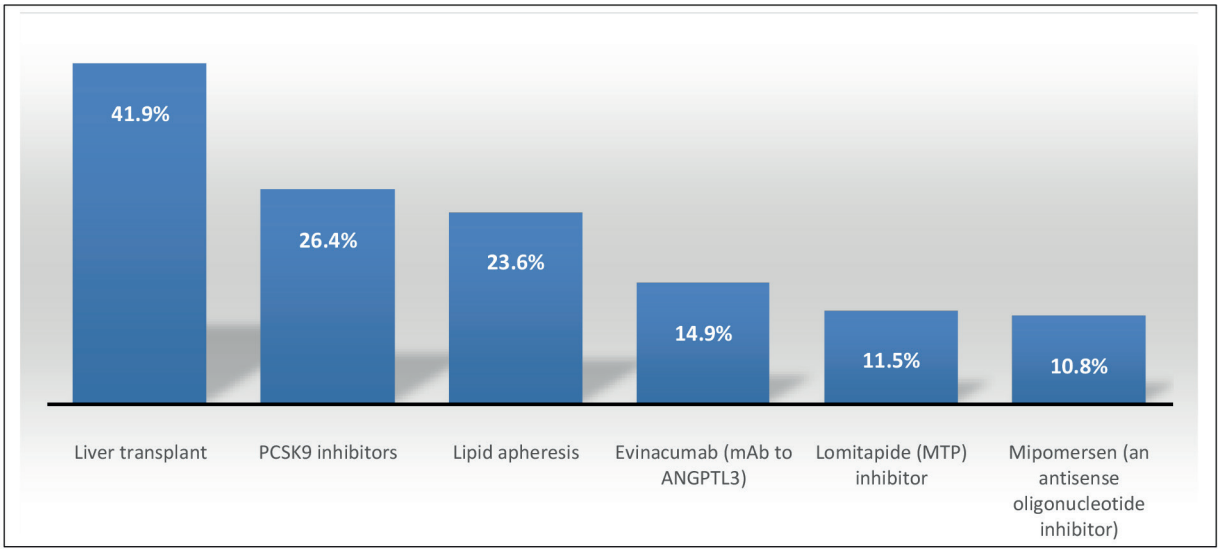

Figure 2. Awareness of medications/interventions other than statins for pediatric heterozygous and homozygous FH patients.

Table II. Socio-demographic characteristics of the physicians[n=148].

Statement	N (%)
Age group	
• 21-30 years	40 (27.0%)
• 31-40 years	66 (44.6%)
• >40 years	42 (28.4%)
Gender	
• Male	78 (52.7%)
• Female	70 (47.3%)
Primary medical specialty	
• Pediatric Endocrinologist	06 (04.1%)
• Pediatrician	57 (38.5%)
• Family physician	50 (33.8%)
• Cardiology	05 (03.4%)
• General practitioner	30 (20.3%)
Level of training	
• Resident	63 (42.6%)
• Registrar	40 (27.0%)
• Consultant	45 (30.4%)
Years in practice	
• 1-5 years	41 (27.7%)
• 6-10 years	33 (22.3%)
• 11-15 years	40 (27.0%)
• >15 years	34 (23.0%)
How familiar are you with familial hypercholesterolemia? *	
• Not familiar (Score 1-4)	79 (53.4%)
• Familiar (Score >4)	69 (46.6%)

*Response has a range from "Not all familiar" coded as 1 to "Extremely familiar" coded as 7.

Table III. Assessment of the Practice of hypercholesterolemia (FH)[n=148].

Statement	N (%)
In patients with documented premature coronary artery disease which one of the following do you routinely carry out?*	
• Examine for arcus cornealis	04 (02.7%)
• Examine for tendon xanthomata	09 (06.1%)
• Take a detailed family history of coronary artery disease	13 (08.8%)
• Screen close relatives for hypercholesterolemia	10 (06.8%)
• All of the above	122 (82.4%)
• None of the above	03 (02.0%)
In your view, which healthcare providers would be most effective at early detection of familial hypercholesterolemia and screening first-degree relatives?*	
• Lipid specialists	08 (05.4%)
• Primary care physician	70 (47.3%)
• Cardiologists	15 (10.1%)
• Pediatrician	43 (29.1%)
• Endocrinologists	35 (23.6%)
• Family medicine	99 (66.9%)
Have you ever diagnosed a patient with FH?	
• Yes	49 (33.1%)
• No	99 (66.9%)
Have you ever followed a patient with FH?	
• Yes	57 (38.5%)
• No	74 (50.0%)
• Don't remember	17 (11.5%)
Which one of the following choices could help you in the detection of FH in your practice?	
• Laboratory comment on lipid profile alerting to consider FH	56 (37.8%)
• Alert by the clinical software system in your practice	12 (08.1%)
• Direct telephone call from the laboratory	02 (01.4%)
• All of the above	64 (43.2%)
• None of the above	02 (01.4%)
• Other	12 (08.1%)
If you have patients with FH under your care, do you routinely screen their close relatives for this condition with lipid profile?	
• Yes, patient's children only	26 (17.6%)
• Yes, patient's children and other close relatives	63 (42.6%)
• No	16 (10.8%)
• Not applicable	43 (29.1%)
Are you aware of any specialist clinical services for lipid disorders to whom you can refer patients?	
• Yes	47 (31.8%)
• No	101 (68.2%)
What is your practice in the Lipid profile screening	
• Fasting	107 (72.3%)
• Non-fasting	18 (12.2%)
• I do not screen lipid profile in children	23 (15.5%)
Which drug combinations do you use to treat severe hypercholesterolemia?	
• Statin + Exchange resins / bile acid sequestrates	23 (15.5%)
• Statin + Ezetimibe	65 (43.9%)
• Statin + Ezetimibe + Exchange resins / bile acid sequestrates	40 (27.0%)
• Other	02 (01.4%)
• None of the above	18 (12.2%)

Table IV. Differences in the knowledge score in regards with the socio-demographic characteristics of the physicians[(n=148)].

Factor	Knowledge Total score (15) Mean ± SD	H/Z-test	*p*-value
Age group[a]			
• 21-30 years	4.27 ± 2.09	H=8.288	0.016**
• 31-40 years	5.98 ± 3.12		
• >40 years	6.05 ± 3.63		
Gender[b]			
• Male	5.77 ± 3.19	Z=-0.852	0.394
• Female	5.29 ± 3.04		
Primary medical specialty[a]			
• Pediatric Endocrinologist	9.83 ± 4.12	H=16.155	0.001**
• Pediatrician	4.58 ± 2.35		
• Family physician/Cardiology	6.44 ± 3.08		
• General practitioner	4.87 ± 3.22		
Level of training a			
• Resident	4.76 ± 2.83	H=6.575	0.037**
• Registrar	6.12 ± 2.96		
• Consultant	6.11 ± 3.45		
Years in practice a			
• 1-5 years	4.09 ± 2.19	H=5.329	0.021**
• 6-10 years	5.64 ± 3.29		
• 11-15 years	6.32 ± 3.03		
• >15 years	6.26 ± 3.50		
How familiar are you with familial hypercholesterolemia?[b]			
• Not familiar (1-4)	4.99 ± 2.71	Z=-2.309	0.021**
• Familiar (>4)	6.17 ± 3.44		
If you have patients with FH under your care, do you routinely screen their close relatives for this condition with lipid profile?			
• Yes, patient's children only	26 (17.6%)		
• Yes, patient's children and other close relatives	63 (42.6%)		
• No	16 (10.8%)		
• Not applicable	43 (29.1%)		
Are you aware of any specialist clinical services for lipid disorders to whom you can refer patients?			
• Yes	47 (31.8%)		
• No	101 (68.2%)		
What is your practice in the Lipid profile screening			
• Fasting	107 (72.3%)		
• Non-fasting	18 (12.2%)		
• I do not screen lipid profile in children	23 (15.5%)		
Which drug combinations do you use to treat severe hypercholesterolemia?			
• Statin + Exchange resins/bile acid sequestrates	23 (15.5%)		
• Statin + Ezetimibe	65 (43.9%)		
• Statin + Ezetimibe + Exchange resins/bile acid sequestrates	40 (27.0%)		
• Other	02 (01.4%)		
• None of the above	18 (12.2%)		

[a]*p*-value has been calculated using Kruskal Wallis H-test. [b]*p*-value has been calculated using Mann Whitney Z-test. **Significant at $p<0.05$ level.

Table V. Relationship between familiarity about familial hypercholesterolemia and the socio-demographic characteristics of the physicians(n=148).

Factor	Knowledge Total score (15) Mean ± SD	H/Z-test	*p*-value
Age group[a]			
• 21-30 years	4.27 ± 2.09	H=8.288	0.016**
• 31-40 years	5.98 ± 3.12		
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Primary medical specialty[a]			
• Pediatric Endocrinologist	9.83 ± 4.12	H=16.155	0.001**
• Pediatrician	4.58 ± 2.35		
• Family physician/Cardiology	6.44 ± 3.08		
• General practitioner	4.87 ± 3.22		
Level of training a			
• Resident	4.76 ± 2.83	H=6.575	0.037**
• Registrar	6.12 ± 2.96		
• Consultant	6.11 ± 3.45		
Years in practice a			
• 1-5 years	4.09 ± 2.19	H=5.329	0.021**
• 6-10 years	5.64 ± 3.29		
• 11-15 years	6.32 ± 3.03		
• >15 years	6.26 ± 3.50		
How familiar are you with familial hypercholesterolemia?[b]			
• Not familiar (1-4)	4.99 ± 2.71	Z=-2.309	0.021**
• Familiar (>4)	6.17 ± 3.44		
If you have patients with FH under your care, do you routinely screen their close relatives for this condition with lipid profile?			
• Yes, patient's children only	26 (17.6%)		
• Yes, patient's children and other close relatives	63 (42.6%)		
• No	16 (10.8%)		
• Not applicable	43 (29.1%)		
Are you aware of any specialist clinical services for lipid disorders to whom you can refer patients?			
• Yes	47 (31.8%)		
• No	101 (68.2%)		
What is your practice in the Lipid profile screening			
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• Statin + Exchange resins/bile acid sequestrates	23 (15.5%)		
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• Statin + Ezetimibe + Exchange resins/bile acid sequestrates	40 (27.0%)		
• Other	02 (01.4%)		
• None of the above	18 (12.2%)		

[a]*p*-value has been calculated using Kruskal Wallis H-test. [b]*p*-value has been calculated using Mann Whitney Z-test. **Significant at $p<0.05$ level.

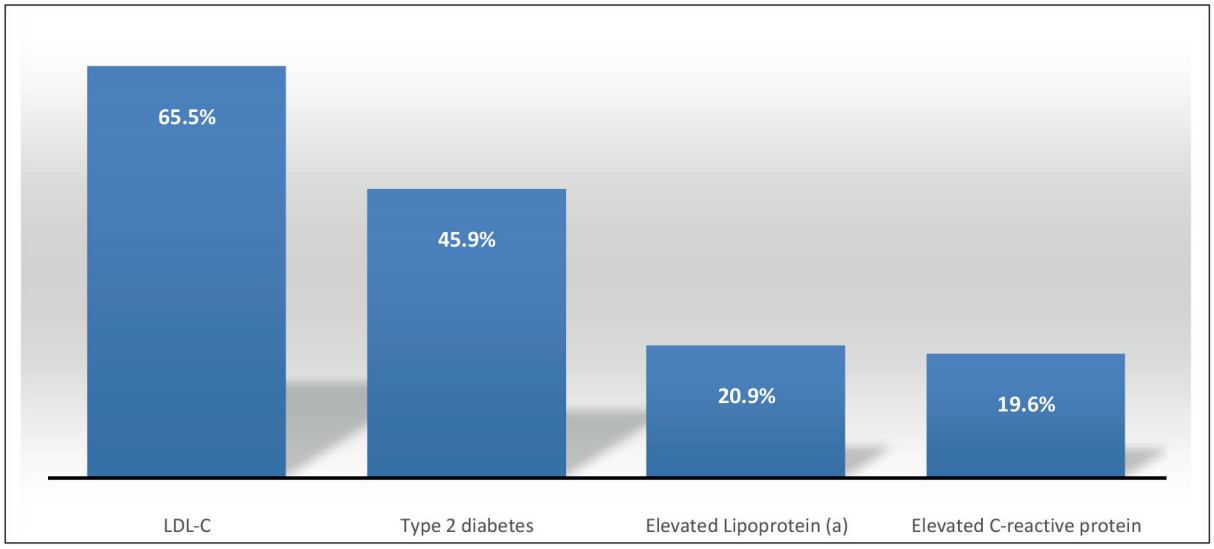

Figure 3. Cardiovascular risk factors associated with familial hypercholesterolemia.

Discussion

To our knowledge, our study is the first validated study to assess FH knowledge, awareness, and clinical practices among pediatricians, family physicians, and general practitioners in Saudi Arabia. In this study, more than half (53.4%) of the physicians were not familiar with FH. This finding is consistent with the result reported by Rangarajan et al[23], who found that 72.1% of the physicians were not familiar with FH and only 27.9% considered themselves to have above-average familiarity. However, several other studies[17,19,21,22] indicated average or above-average familiarity related to FH among physicians, with the rates of familiarity varying from 54% to 72%, which were higher than those in our report.

These low levels of knowledge and familiarity with FH might explain why it remains underdiagnosed to the extent that its prevalence in Saudi Arabia remains unknown. A major area of focus in clinical practice at present is to increase FH awareness among physicians, which may necessitate increased efforts to identify and treat these patients properly.

When we further assessed their knowledge, more than half of the physicians were able to correctly describe FH (73%) yet provided incorrect responses for questions regarding CAD risk (93%) and prevalence of FH (77%). In this regard, our results showed similarities with other findings. Two studies[20,21] conducted in the UK and Western Australia found that 89% and 80% of the physicians could describe FH, 30% and 27% could identify the prevalence of FH in the community, and only 14% and 29% were aware of the cardiovascular risks in untreated FH cases, respectively. Similarly, in other studies conducted in Saudi Arabia[17-19], 67%, 56%, and 76% of the physicians correctly defined FH; however, only 22%, 19%, and 21% were aware of the prevalence of FH. Furthermore, only 5.1%, 12%, and 5.8% knew of the cardiovascular risks. In addition, our study identified that most physicians, regardless of specialty, were unaware of the screening age for young children in a family with a history of premature CAD and elevated LDL (91%).

Despite the high prevalence of FH, most of the participants in our study had never diagnosed or followed up a patient with FH. This finding may be related to the poor awareness of the FH screening period in children (20% gave correct answers) and failure of 75% of the respondents to select the appropriate answer for the LDL cut-off level for FH. Regrettably, more than half of the physicians (68.2%) were unaware of any lipid specialist to whom they could refer patients when necessary. These findings are similar to those obtained in Riyadh by Maha et al[17], who reported that 70% of the participants had never diagnosed FH patients, 68% had never followed up on FH cases, and only 31% of the physicians were aware of the specific screening age[17]. Similarly, in the study by Batais et al[19], only 33% of the physicians had diagnosed a patient with FH, 35% had FH patients under their care, and only 41% were aware of the screening

age. These findings were in contrast with those from the UK study, where 45% of the physicians were aware of the screening age, 50% knew about lipid specialists, and 72% referred patients to lipid specialists[20]. In the study conducted in Western Australia, 62% knew about lipid specialists, and 52% were aware of the prevalent screening age[21]. Among the physicians participating in the present study, 55% thought that FH diagnosis required genetic testing, which is unavailable at most hospitals and primary health care facilities, potentially contributing to the low rate of diagnosis of this disease. Genetic testing is not required for diagnosis of FH in most cases[7,8].

A thorough awareness of different treatment options, interventions, and new medications proved to be highly effective in lowering LDL levels is crucial to facilitate referrals to a lipid specialist; furthermore, other lines of treatment besides statin treatment are essential to reach the LDL level goal in patients with heterozygous and homozygous FH[7,8]. However, the physicians in this study showed a significant lack of awareness of other options (Figure 2). Unfortunately, one of the most important aspects of FH is that it is associated with a 20-fold higher risk of cardiovascular diseases in the patients, compared with the general population[8,27], and only 7% of the study participants could provide correct answers regarding CAD risk in untreated FH. The corresponding results for physicians in other studies were similarly low: 9% in Asia[22], 14% in the UK[20], 29% in Western Australia[21], and only 5.1%, 5.8%, and 7.2% in Saudi Arabia[17-19]. Lipoprotein level (a) has been identified as an additional risk factor for CVS, necessitating aggressive treatment in adult and pediatric age groups[28,29]. However, only 20% of the physicians in this study selected lipoprotein level (a) as a treatment target, while most of them selected LDL level (65%).

Regarding lipid screening, most of the physicians in this study recommended screening in the fasting state (72%) or did not recommend lipid screening at all. However, non-fasting lipid profile screening is a useful initial screening test in children, and subsequent fasting lipid screening should then be conducted if the results are abnormal[7,30]. Since parents/guardians may find it difficult to let their children fast for 12 hours, the emphasis on lipid screening in the fasting state may have led to a low rate of lipid screening overall.

In comparing the mean knowledge scores among participants belonging to different specialties, family physicians (6.44 ± 3.08) and pediatric endocrinologist (9.83 ± 4.12) scored higher than general pediatricians (4.58 ± 2.35) and general practitioners (4.87 ± 3.22). This difference may be attributed to the fact that the family physicians have been exposed to adult patients with lipid abnormalities, including FH, in addition to pediatric patients, while pediatric endocrinologists are usually referred to for advanced lipid treatment[8]. Additionally, a sizable difference might be explained by the level of training and duration in practice. In this study, older participants (>40 years of age, 6.05 ± 3.63) and those with higher levels of experience in practice (>15 years, 6.26 ± 3.50) with an advanced level of training (consultants and registrars, 6.26 and 6.32) scored higher than younger doctors (20-30 years of age, 4.27) and junior residents (4.76 ± 2.19), consistent with the findings of the study conducted by Batais et al[19], which showed higher knowledge scores for participants with longer durations of practice. However, the scores among pediatricians were still lower than those among family physicians. Furthermore, most of the participants selected either family physicians (66%) or primary care practitioners (47%) as the most effective healthcare providers for the early detection of FH and for screening first-degree relatives. These findings are similar to those of previous studies[17,20,22] conducted in Saudi Arabia, Asia, and the UK. As per the NLA, primary care physicians are the responsible for the diagnosis and treatment of heterozygous FH, not the homozygous type, which needs to be managed by lipid experts, such as pediatric endocrinologist, cardiologist, or a health care provider with specialized lipidology training[8]. Treatment should be performed either through consultation with or by referral to a lipid specialist, pediatric endocrinologist, cardiologist or a health care worker with specialized lipidology training.

Our study results are consistent with the results of other studies that indicate the need for ongoing FH educational programs for all physicians caring for children in Saudi Arabia, as well as the importance of establishing models of care for FH management and registry in the country. This will enhance the detection rate and care for FH and ultimately decrease the rates of premature cardiovascular diseases. Such educational programs have been implemented in the UK, and significant improvements were reported in physicians' overall knowledge[20], management, and screening. Furthermore, the present study revealed that junior residents in pediatric and family medicine and primary care physicians have a significant knowledge gap, compared

with registrars and consultants with more years of practice and a higher level of training, which indicates the need to include more FH-related topics in the curricula for residency training in Saudi Arabia. Moreover, to increase awareness and knowledge among newly graduated physicians at the beginning of clinical practice, we suggest that FH topics are essential in the curricula of medical schools.

The main limitations of this study were the survey-based model, which can only highlight relationships without identifying causal associations; thus, the results cannot confirm or rule out casualty. Moreover, the COVID-19 pandemic affected the functioning of primary care centers, some of which were closed or changed to COVID-19 vaccine centers. The results' generalizability may have been limited due to the small sample size. However, our study was the first of its kind to be conducted among all potential specialties caring for children.

Conclusions

FH remains almost universally undiagnosed and untreated. General practitioners, family physicians, and pediatricians play an important role in the early detection and diagnosis of FH in children. However, this study highlighted significant deficits in FH knowledge, awareness, and clinical practices among physicians taking care of children in Saudi Arabia, indicating the need for more extensive educational programs on screening, diagnosis, and management of FH.

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Conflicts of Interest

The authors declare no conflicts of interest.

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