https://doi.org/10.4038/jccp.v54i1.7994

A study on knowledge, awareness, and practice of familial hypercholesterolemia among doctors at two tertiary care hospitals in Sri Lanka

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Journal of the Ceylon College of Physicians, 2023, **54**, 22-27

Abstract

Introduction: Familial Hypercholesterolemia (FH) is an inherited disorder affecting low-density lipoprotein cholesterol (LDL-C) metabolism. Individuals with FH have a substantially increased risk of cardiovascular disease (CVD). South Asia is burdened by premature CVD. Early detection and management of FH can reduce the burden of CVD. Despite growing awareness, FH remains underdiagnosed and therefore untreated, and assessing the present level of knowledge among doctors would help to implement educational activities.

Methods: Doctors from two large teaching hospitals in Sri Lanka were requested to complete a survey questionnaire comprising questions on awareness, clinical features, prevalence, inheritance, and treatment of FH.

Results: Of the 269 doctors surveyed, 168 (66.42%) of them correctly described FH and only 44 (16.66.%) knew about its global prevalence. 12 (4.55%) were aware of the risk of CVD associated with FH. Only 01 (0.75%) and 02 (1.5%) correctly identified the age threshold for premature CVD in men and women, respectively. 228 (87.36%) and 136 (54.84%) identified statins as monotherapy and statin and ezetimibe as combination therapy for FH, respectively. Furthermore, in practice 117 (44.66%) routinely screened family members for FH, and only half the doctors correctly identified the transmission rate to first-degree relatives (38.78%).

Conclusion: Substantial defects in the knowledge of FH were seen among doctors. Immediate steps have to be taken to increase awareness and knowledge about FH in doctors practicing in Sri Lanka.

Key words: familial hypercholesterolemia, knowledge, awareness, management practices, hypercholesterolemia, dyslipidemia, high LDL

Introduction

Familial hypercholesterolemia (FH) is a genetic disorder of cholesterol metabolism and can be caused by mutations in the LDLR, APOB, and PCSK9 genes. It is known to cause premature coronary artery disease (CAD).[1] FH is characterized by increased low-density lipoprotein (LDL)-cholesterol. There is also the presence of xanthomata and premature atherosclerotic cardiovascular disease (CVD). The risk of premature CAD is preventable or reversible through early detection and treatment of hypercholesterolemia. FH approximately affects 1 in 220 individuals globally.[2] Of the globally estimated 15.4 million people with FH, around 50% of FH patients live in Asia.[2] Patients with FH have a 30-50% risk of a cardiac event before the age of 60 years.

As the risk of CAD is very high in these patients, they should be identified at a younger age and started on treatment to control LDL cholesterol. Very few countries have implemented screening programs for FH.[3] The vast majority of people with FH in Sri Lanka are currently undiagnosed as there is no screening program for FH. Therefore, FH is often both underdiagnosed and undertreated.[4] To diagnose FH, sound

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Received 13 December 2022, accepted 10 February 2023.

knowledge about diagnostic criteria is required. Guidelines recommend prompt action to identify and manage patients affected by FH. Early management of patients with FH can prevent CAD. Once a patient is diagnosed, knowledge about the management options is required for optimal management.

Due to lack of awareness among doctors, FH is underdiagnosed and not treated adequately. Several studies done in the UK, Australia, and Asia Pacific regions have demonstrated inadequate knowledge about FH.[5] This study was carried out to test the knowledge, awareness, and practice among doctors about FH in Sri Lanka as diagnosing FH will help in reducing the high cardiovascular disease burden in South Asia especially in Sri Lanka.

Methods

This cross-sectional study was conducted at two large tertiary care hospitals in Sri Lanka; Colombo South Teaching Hospital (CSTH) and the National Hospital of Sri Lanka (NHSL). Both hospitals are tertiary care hospitals that provide care to a large number of patients annually. All doctors attending to 50 or more patients per month working in medical, medical subspecialties, paediatric, and surgical wards were recruited. This included specialists and non-specialists. The estimated sample size was 300. Due to the COVID epidemic, the sampling method was changed and all doctors who were working in the wards within two weeks in March 2021 and who were willing to participate in the survey were recruited. The link to the questionnaire was given out to all consenting doctors. Due to COVID-19, the rosters of doctors have been changed to reduce doctors working at a given time to minimize exposure to COVID-19. Two weeks were taken to collect data, as the authors postulated that within two weeks all doctors fulfilling the inclusion criteria would have worked during the period specified.

The study used a questionnaire based on the Familial Hypercholesterolemia Knowledge, Attitude, and Practice (FH-KAP) questionnaire which was initially made and utilized by Bell et al.[5] This questionnaire was used as it is a validated questionnaire and has been published widely in other countries and it will facilitate to compare the knowledge between countries.[5,6] It contained 20 items covering wide items of FH-KAP, 10 knowledge, 5 awareness, and 5 practice items. These items are found in many designs such as single best answer questions, 7-point Likert scale, dichotomous answers of 'Yes / No / Don't know, multiple answer questions, and make-out answer statements. The participants were asked to choose the right answer. They could select more than one answer in many questions. The FH questions evaluated the participants' knowledge of FH as clinical features, diagnostic lipid profile, mode of inheritance, prevalence, awareness of genetic cause for the diagnosis, and relation of FH to CAD. Permission was obtained from the original authors and the permission letter (obtained via email) can be produced on request. Ethical clearance for the study was obtained from the Ethics Review Committees of the CSTH and NHSL. Filling the questionnaire was taken as informed consent. Previous studies done with the same study instrument have also taken the filling form to be adequate as informed consent.

The statistical analysis including demographical analysis was conducted using Microsoft Excel 2013, R software (version 4.0.3), and R-Studio (version 1.4.1106). Graphical analysis was also carried out using Excel 2013 and the proportions of the response rates concerning each factor were assessed under separate sections according to the questionnaire.

Results

Out of the 300 doctors who were asked to complete an anonymous internet-based survey, 269 responses were received, which thus resulted in a response rate of 89.67%.

Demographics details

Among the 269 doctors, there were 160 females (59.7%) and 106 males (39.55%) with 2 (0.75%) responding as 'prefer not to say'. The majority of them (46.56%) were in the age category of 21 to 30 years with a gradual decrease in the number of respondents with the increase of age having the least from 51 to 60 years (8.4%). Over 70% (185) have been graduated less than 10 years ago. The median number of years in practice after graduation was found to be 5. There were only 5 doctors (1.89%) with over 30 years of post-graduation practice among the 264 who responded.

Awareness

As seen in Table 1, out of the 267 respondents (99.26%), 168 (62.92%) have rated their awareness of FH as average or above (4 and above). Moreover, 135 (51.53%) among the 262 who responded (97.4%) were found to be aware of the guidelines for the detection and management of FH. Awareness of the cascade screening for patients with FH was also found to be low among the respondents as only 35 (13.78%) knew about it.

Knowledge

Table 1 depicts a detailed summary of how the respondents answered the questions related to the assessment of their knowledge regarding FH. The combination of statins plus exchange resin/bile acid sequestrants was selected by 33.06% (82/248) of doctors to treat severe hypercholesterolaemia while 25.4% (63/248) preferred the statins with ezetimibe combination. Furthermore, statins were selected as the drug used to treat hypercholesterolaemia by the majority (228, 87.36%) of the 261 doctors who responded.

Table 1. Summary of the doctor's responses to questions on knowledge, practice, and attitudes

	Number (%)
Awareness	
Familiarity with FH rated as average or above	168 (62.92%)
Aware of FH guidelines	135 (51.53%)
Aware of lipid specialists	155 (60.31%)
Knowledge	
Correctly described FH	178 (66.42%)
Correctly identified the lipid profile	86 (39.81%)
Correctly identified the global prevalence of FH	44 (16.60%)
Correctly identified the transmission rate to first-degree relatives	102 (38.78%)
Correctly identified the CVD risk in untreated FH	12 (4.55%)
Correctly identified the age threshold for premature CVD	
• Men	01 (0.75%)
• Women	02 (1.5%)
Correctly identified that genetic testing was not required to accurately diagnosis FH	149 (56.02%)
Selected statins to treat hypercholesterolaemia	228 (87.36%)
Selected a combination of statin and ezetimibe to treat severe hypercholesterolaemia	136 (54.84%)
Practice	
Screened patients with premature CVD for FH, including screening family members	133 (50.19%)
Unaware or unsure whether they had FH patients under their care	02 (1.20%)
Performed routine family screening of patients with FH	117 (44.66%)
The most prevalent age for screening young people in a kindred with FH was 13-18 years	126 (48.65%)
Referred patients to lipid specialists	155 (60.31%)

Management practice and opinions on detection

Out of the 263 doctors who responded (97.77%), over 70% had never diagnosed a patient with FH. However, 98.8% were aware of having FH patients under their care. 164 (62.6%) indicated that they would examine for corneal arcus, and tendon xanthomata, take a detailed family history, and screen close relatives in patients with premature CVD.

The majority stated that they would screen the patient's children and other close relatives (117, 44.66%). 13.36% (35/262) of the respondents had indicated that they would not perform any routine family screening. The most frequent age category the doctors would theoretically screen children of patients with premature CVD (between 13 and 18 years old) was selected by 48.65% (126/259).

Discussion

The prevalence of FH is highest in Asia.[7] FH prevalence in Sri Lanka is not known. Despite the absence of national registries and screening programs, it is likely that FH prevalence in Sri Lanka might be on par with other South Asian countries.[2] There are very few small studies done in Sri Lanka on FH which focused mainly on the genetic mutation analysis[8]. The under recognition of FH in Sri Lanka is a grave problem as CVD mortality in Sri Lanka is high. The present study helps in creating awareness about FH. This study highlights important deficits in knowledge and awareness among doctors. Although the doctors believed they were familiar with FH, their knowledge regarding FH was not adequate. Acceptable knowledge was reported in FH description (66.42%) and lipid profile that diagnosed FH (39.81%) but lower when compared to other countries. The FH description and identification of lipid profiles that correctly describe FH among Malaysian (61.6% and 77.7%)[9], Western Australia (80% and 68%), United Kingdom (89% and 74%),[6] and Asia Pacific areas (86% and 65%)[10] were better than in the present study. The identification of description of FH and lipid profile of FH is similar to Southern India been 71% and 35%.[11] The low level of knowledge concerning FH in South Asia is a concern as South Asian regions are burdened with premature CVD risk and FH is one reason behind the higher risk.[12] Further educational activities need to be carried out in the form of continuous medical education activities to help doctors identify the lipid profile indicative of FH. Unless the lipid profile is identified, patients with possible FH will be missed in routine practice.

The knowledge on the prevalence of FH in the society was suboptimal as it was known to only 16.60%. Knowledge about heritability was also suboptimal with 38.78% identifying it. There is an underdiagnosis of FH worldwide. The diagnosis of FH could be improved if doctors understood the true prevalence of FH.

Overall, 81% of respondents identified type 2 diabetes and smoking as risk factors that further increased CVD risk in FH. The recognition of these factors was better in doctors from other Asian countries: Japanese (91% and 96%, respectively) than South Korean (80% and 86%, respectively) and Taiwanese (73% and 62%, respectively).[10] The prevalence of diabetes mellitus is rising in South Asian countries including Sri Lanka, hence identification of diabetes as a risk factor of CVD is important.

Elevated Lipoprotein (Lp(a)) as a significant risk factor was identified by 67% of participants. Lp(a) as a risk factor was frequently recognized by Korean physicians (83%) than Taiwanese (66%) and Japanese physicians (51%).[11] Lp(a) is a risk factor for atherosclerosis, in which Lp(a) concentration of >50 mg/dl has been suggested as elevated.[13] Similarly, Lp(a) concentration of 25 mg/dL has been suggested as the cut-off for reflecting the risk of CAD based on a South Indian study.[14] In concurrence, the estimation of Lp(a) levels is strongly recommended for ASCVD risk stratification in all patients at least once in their lifetime and especially if there is a history of premature CVD.[15] The recognition of Lp(a) as a risk factor by Sri Lankan doctors is noteworthy.

FH is a major public health problem as it is associated with an increased risk of premature CVD. Premature CVD is CVD occurring in men below 55 and women below 60 years. Only 01 (0.75%) and 02 (1.5%), doctors correctly identified the age cut off for premature CVD in men and women, respectively. Therefore, premature CVD will not be considered premature and therefore screening or FH would not occur. CVD is the leading cause of premature death from non-communicable diseases (NCDs) in Sri Lanka. The hospitalization trend from 2010 to 2017 shows that admissions due to IHD have increased over this decade in Sri Lanka.[16] Although FH is a major risk factor for CVD, only 12 (4.55%) doctors identified that the risk of CVD was increased 5 to 20 fold. This indicates that the majority did not have a good understanding that the risk of CVD is very high among patients with FH. Greater emphasis needs to be made during educational activities on the identification of premature CVD and screening them for FH.

The optimal LDL-C goal must be <2.5 mmol/L or <1.8 mmol/L with proven atherosclerotic CVD or at least a 50% decrement in LDL-C values.[17] In this study,

only 39.81% identified the lipid profile and 4.55% understood the CVD risk associated with FH. More educational activities are needed to educate about lipid levels in FH and the need for tight control of lipid levels.

Only half the doctors correctly identified the transmission rate to first-degree relatives and carried out family screening (38.78 % and 44.66%). Cascade screening is sighted as the most cost-effective FH screening method, and first-degree relatives have a 50% chance of having FH. Therefore, cascade screening is an effective screening tool[18] and only 35 (13.78%) knew about it. It is said that more than half of the estimated population of 10 million FH patients are living in the Asia Pacific region and the majority of patients are not aware of it. A case-finding program can be carried out to find undiagnosed patients.[15]

Sixty-eight percent of doctors selected the correct clinical description of FH in three selected countries, Japan, Korea, and Taiwan, while in the present study, it was 66.42%.[10] The prevalence was identified by only 16.60% in the present study, and 27% identified the prevalence of FH as 1:500. Thirteen percent of physicians identified the CVD risk in untreated FH patients as 20 times greater than the general population, while in the present study, the percentage of physicians who identified it was even lower as only 4.55% identified the correct risk.[10] It was evident that Asian counters with established FH registries have demonstrated a greater knowledge when compared to Sri Lanka.[10]

India is a South Asian country with high CVD morbidity and mortality. In a study done in India, the knowledge of the typical lipid profile was 34.6% and the risk of CVD relating to FH was identified by 13.5%. This level of knowledge is low when compared to other Asian countries but higher than the present study. This shows that there needs to be a significant improvement in the knowledge in Sri Lankan doctors.

Limitations

The study was carried out among the doctors working in the wards during the specified period to get a cross-section of the doctors as they were working on a rota. As only those interested in participating were given the questionnaire, the results of the study may be an overestimation of their knowledge because only those who were interested took part in the study. The baseline knowledge of all doctors may be even lower.

Conclusions

There is very limited data on FH in Sri Lanka and there are gaps in knowledge and care of FH among the doctors. The knowledge deficits identified would be useful when designing teaching activities and training programs for doctors. By creating registries and management guidelines, more attention can be focused to increase awareness among doctors about FH. Unless the awareness and knowledge to identify exist with the doctors, cases of FH may go unnoticed. Learning modules should be developed on identifying and management aspects of FH to improve the care of FH in Sri Lanka.

Author declarations

Ethics approval and consent to participate

Ethics approval was obtained from the Ethical Review Committees of CSTH and NHSL of Sri Lanka. Filling out the questionnaire was taken as informed consent by the participants.

Availability of data and materials

The datasets used during the current study are available from the corresponding author upon reasonable request.

Competing interests

No conflict of interest is associated with this study.

Funding

No funding.

Authors' contribution

ATM conceptualized the study. ATM and MDTCG collected data. ATM analyzed data, interpreted results and was responsible for manuscript writing. ATM and BGGKS analyzed the data. ATM, MDTCG, BGGKS read and approved the final version of the manuscript.

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