Original Article

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Knowledge and Awareness of Familial Hypercholesterolaemia among Registered Medical Practitioners in Tamil Nadu: Are They Suboptimal?

Internal Medicine Section

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ABSTRACT

Introduction: Familial Hypercholesterolaemia (FH) is the most common monogenic disorder causing premature Coronary Artery Disease (CAD). However, the majority of people with FH are undiagnosed and under treated.

Aim: To determine awareness, knowledge and practices of registered medical practitioners regarding FH in India.

Materials and Methods: Physicians from a southern state of India (Tamil Nadu) who see the general cases were requested to complete a structured online survey questionnaire based on the outcomes on screening, diagnostic and service aspects of FH.

Results: A total of 133 physicians were surveyed, 27.9% perceived themselves to have above average familiarity with FH and 71.4% correctly described FH. 41.4% of physicians were unaware and unsure whether they had FH patients under their care. The awareness of specific aspects of FH were as follows: heritability 35.3%, prevalence 31.6%, typical lipid profile 34.6%, CVD relating to FH13.5%, genetic testing 33.1%, cascade screening 41.4%, preventive, management and referral services for FH 12.8%, 49.6% of them thought that the age for screening young people for FH should be 13 to 18 years. 84.2% selected GP's as the most effective health care provider for the early detection and care of FH as being useful. 69.2% selected interpretive commenting on lipid profile to highlight patients at risk of FH. 91.7% and 19.5% of physicians identified statins as monotherapy and statin with ezetimibe as combination therapy for FH, respectively.

Conclusion: The study identified substantial deficit in the awareness and knowledge of FH among primary care physicians in Tamil Nadu. Extensive and continuous medical education programs are required to close the gap in coronary prevention.

Keywords: Coronary artery disease, Models of care, Physicians

INTRODUCTION

Familial Hypercholesterolaemia (FH) is the most common monogenic lipid disorder causing premature Coronary Artery Disease (CAD). However, the majority of people with FH are undiagnosed and undertreated in India. A strategy for the prevention of CAD long before its onset will be more cost-effective than providing interventions at a stage when the disease is well established [1]. High burden of CAD in India is of particular concern as it also affects the productive workforce aged 35-65 years. It is estimated that 52% of CAD related deaths occur below the age of 50 years and around 25% of acute myocardial infarction happen below the age of 40 years in India [2]. The prevalence rates of CAD have been escalated by 11% in the last decade and are expected to continue with the same pattern if the current situation prevails. It was thought that CAD is prevalent only in high income countries, but, its burden is now being transferred to the developing countries as evident by its presence in India [3]. The high frequency of FH, with the predominance of the FH Gujarat mutation (47%) in South African Indians is suggestive of a founder effect. It is unlikely that a founder effect would have manifested itself in South Africa, due to the large initial founder population of about being 150000 Indian immigrants. It is therefore probably the mutation P664L that originated from India, strongly suggesting that the increased incidence of these CpG hotspot mutations indicate the high frequency of FH in India, and that the group in South Africa represents the incidence in the Indian subcontinent [4].

The prediction of two novel mutations in clinically diagnosed hyperlipidaemia patients and the absence of the transition mutations analysed confirm the mutational heterogeneity among Indians warrants a search for new LDL receptor gene defects. The genes involved in hyperlipidaemia should be the main target for the management of CAD [5].

According to The Million Death Study, CardioVascular Diseases (CVD) leads to 1.7 – 2.0 million deaths in India [6]. The epidemic of CVD in India needs an urgent policy. Cardiovascular events can delay the implementation of public health policies, decreasing the use of tertiary care to improve health economics [7,8]. Increased prevalence of dyslipidemia in 31-40 year males is suggestive that this group is at increased risk of developing CAD leading to young infarcts. The low percentage of adults with controlled lipid concentrations suggests that there is a need for awareness programs for the prevention and control of dyslipidemia [9].

Several international groups have developed diagnostic criteria based on a combination of clinical signs, family history and cholesterol measurements [10]. The evidence gap is acknowledged by an international guidance, which aims to make the best use of contemporary practice and technology to achieve the best outcomes for the care of FH [11]. It should accordingly be implemented for a better clinical judgement and adjusted according to the country-specific needs. Most people in the community have contact with their primary care physician or family doctor. The Prayas, a non-profit organization in India, currently has a network of 400 mentors and 5,500 mentees, enables Primary Care Physicians (PCP) (mentees) to receive training from key opinion leaders (mentors) updating the latest developments in medicine [12].

Knowledge and awareness of FH among PCPs regarding FH should be tested. The role of PCPs in the care of FH has not been adequately defined and there should be a significant shortfall in awareness, knowledge and practices among family doctors. The major barriers to FH management would be knowledge deficits regarding the international guidelines about the diagnosis and management of FH.

Owing to lack of adequate number of formally trained lipdologist, many patients with dyslipedemia are seen by General Practitioners (GPs). This study explores the knowledge of the GPs regarding identification of FH and its risk factors, and evaluation and management of FH.

Assessing current knowledge and practices is the starting point. The information obtained will be employed to design effective teaching and training modules for PCPs in the detection and management of FH, as improving the quality of care of FH, education and training of PCPs is essential [5].

MATERIALS AND METHODS

The study group included the PCPs and family physicians who take care of general cases from the southern state of India (Tamil Nadu). An advertisement was posted on websites and announcements were made in the national conference of Biochemistry and Endocrinology inviting the doctors to participate in the study if they are interested. The text used in the advertisement and announcement was approved by the Institutional Review Board. The prospective participants contacted us and completed the survey. The web link was sent to their friends and the sampling was mostly systematic random and snow ball a certain extent. Completion of the survey was purely voluntary.

Survey design and conduction of the study

The questionnaire based survey [13] using a web link was sent to the randomly selected study subjects by mail, if there was no response within two weeks, a reminder mail was sent, if the subject failed to respond after the second mail, they were reminded by phone calls. Participants were asked to fill in a 24-item survey. The survey comprised of two parts; part 1 covered about the familiarity, clinical features, screening, referral services and management of FH as well as the risk factors and premature CVD; and part 2 included the details about their area of practice, years of practice and the number of patients seen by them. The participants were asked to choose from the predefined key questions and answers; there were no opended questions. The final study protocol was reviewed and approved by the Institutional Ethical Committee of Sree Balaji Medical College & Hospital. (Ref No. 002/SBMC/IHEC/2014-151). Informed consent was not required.

DATA COLLECTION

Data were collected on a structured questionnaire based on the outcomes on screening, diagnostic and service aspects of FH between October 2014 and January 2015 using a unique survey web link. The survey questionnaire was developed based on the expert recommendations and guidelines [13]. The survey was voluntary, anonymous and completed without referring to any literature. The questionnaire employed made enquiries regarding general familiarity with FH, awareness of national and international guidelines for FH, the clinical description of FH, identification of the typical lipid profile, prevalence and inheritance of FH, extent of elevation of lipoproteins that is at risk of CVD, definition of premature CVD and clinical features in FH, whether the diagnosis requires genetic confirmation, methods for alerting PCPs about family screening, treatment and referral practices regarding patients with severely elevated cholesterol, type of health professional best placed to detect FH, number of patients with FH currently being treated, specific treatments, and practices. PCPs were asked to select one correct answer to questions from a list of options provided. There were no opened questions. De-identified demographic data were sought from the participants including, gender, qualifications and training status, years of practice, number of patients seen in clinic per month and location of practice as metropolitan, rural or other.

STATISTICAL ANALYSIS

The data was entered into Microsoft Excel 2003. The data was analysed using SPSS v16.0. Descriptive statistics are presented as percentages for the discrete variables and as mean and standard deviation for the continuous variables.

RESULTS

A total of 149 physicians completed the online survey during the data collection period, there were six duplicates and 10 of them were specialists, which reduced the eligible number to 133. The response rate was 77.37%.

Knowledge and awareness of FH

27.9% of GP's rated them selves as above average with familiarity of FH. Only 40.6% were aware of the international guidelines for the detection and management of FH. 71.4% correctly described FH, 35.3% were aware of the 1 in 2 heritability in first-degree relatives, 31.6% were aware of the prevalence, 34.6% identified the typical lipid profile, 13.5% were aware of CAD relating to FH, 33.1% were aware of genetic testing and 24.8% were aware of cascade screening, 12.8% were aware of preventive, management and referral services for FH. Total 41.4% of physicians were unaware and unsure whether they had FH patients under their care. Also, 49.6% of them thought that the age for screening young people for FH should be 13 to 18 years.

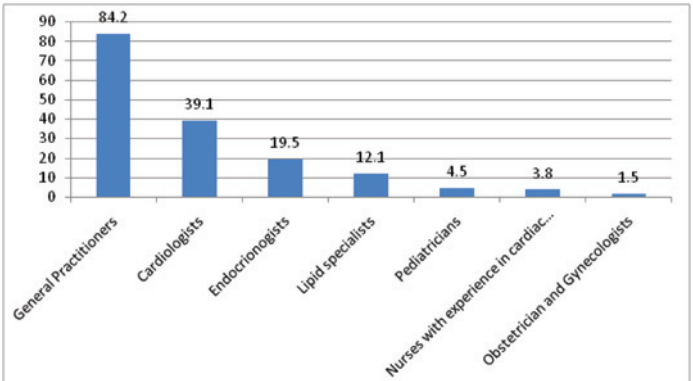
Detection of FH

82% of doctors selected the GPs as the most effective health care provider for the early detection of FH [Table/Fig-1] and 57.1% selected the routine clinical examination to document premature CAD [Table/Fig-2].

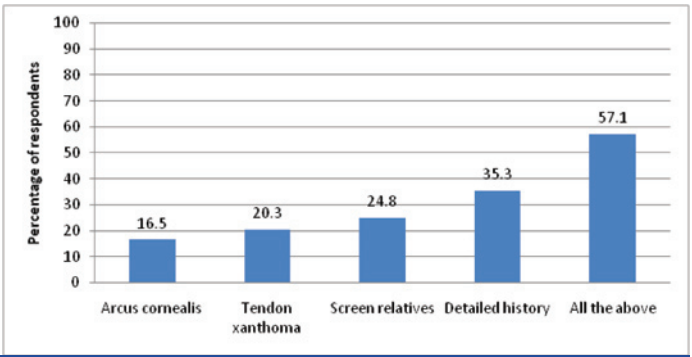
The majority of physicians were unaware of assessment of FH using Dutch Lipid Clinical Network Criteria [10] and the age at which heart disease is considered to be premature is significantly lower [Table/Fig-3].

AWARENESS	
Familiarity of FH rated as above average	27.9%
Awareness about FH guidelines	40.6%
Awareness about lipid specialists	12.8%
KNOWLEDGE	
Correctly described FH	71.4%
Correctly identified lipid profile	34.6%
Correctly identified prevalence of FH in the community	31.6%
Correctly identified the transmission rate of FH to first degree relatives	35.3%
Correctly identified the CVD risk in untreated FH patients	13.5%
Correctly identified that genetic testing was not required to accurately diagnose FH	33.1%
Selected statins to best treat Hypercholesterolaemia	91.7%
Selected a combination of statin and ezetimibe to treat severe Hypercholesterolaemia	19.5%
PRACTICE	
Screened patients with premature CVD for FH, including screening family members	24.8%
Unaware or unsure whether they had patients under their care	41.4%
Performed routine family screening of patients with FH	41.4%
The most prevalent age for screening young people in a kindred with FH was 13—18 years, which was selected by	49.6%
Referred patients to lipid specialists	12.8%
OPINIONS ON DETECTION	
Selected GP's as the most effective health care provider for the early detection of FH	84.2%
Selected interpretive commenting on lipid profiles to highlight patients at risk of FH	69.2%

[Table/Fig-1]: Summary of GP's responses to questions about fh awareness, knowledge and practice.



[Table/Fig-2]: Most appropriate health care provider to treat FH.

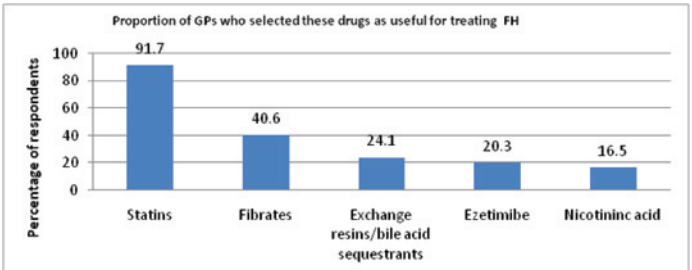


[Table/Fig-3]: Methods for detecting FH in Hypercholesterolaemia patients.

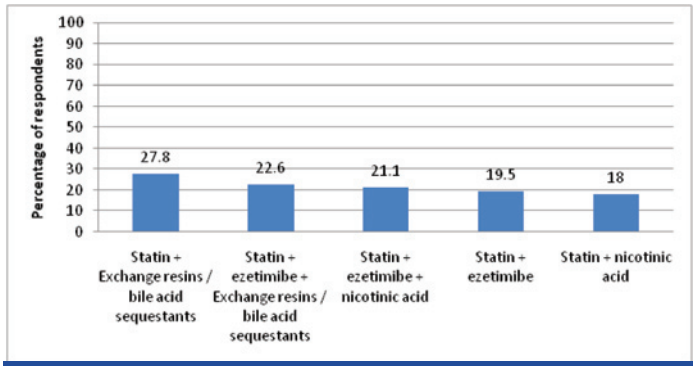
Management practices

66% of the participants selected the option that an alert from the lab when the lipid profile is abnormal to highlight patients at risk of FH would be useful. 83% and 22% of physicians identified statins as monotherapy and statin and ezetimibe as combination therapy for FH respectively. The medications used to treat FH are demonstrated in [Table/Fig-4,5].

49.6% of physicians selected 13–18 years as the age to test young individuals for hypercholesterolaemia in a family with premature CAD and 12.8% of them knew about the referral clinics, and 24.8% opted for screening the patient's children and other close relatives if they have patients with FH.



[Table/Fig-4]: Monotherapy prescribed by respondents to treat FH.



[Table/Fig-5]: Drug combinations prescribed by respondents to treat FH.

DISCUSSION

This pilot study is the first audit to determine the awareness of FH among physicians in India. Defining the role of PCPs in the care of FH is essential for developing multidisciplinary and integrated total quality management. Our study aims to assess the awareness, knowledge, and clinical practices on FH management. The knowledge regarding the familiarity, description, lipid profile, prevalence, inheritance, the availability and necessity of genetic diagnostic tests for FH were obtained.

The risk factors and description of premature CAD, cascade screening of relatives, the role of health care providers, specialist clinical services, and the drug treatment for FH were also assessed. The overall knowledge regarding FH among physicians in India is lower. Our study demonstrates the importance of conducting successful educational intervention and training for PCPs regarding FH. Successful evaluation of how FH contributes to the CVD epidemic in India is very important now [14].

Our results are consistent with the previous cross- sectional study conducted by Pang et al., showing a significant gap in knowledge and awareness of FH in Japan, South Korea and Taiwan [15]. Greater awareness of FH is also needed among cardiologists in a study by Foody et al., [16]. A study carried out by Bell et al., among GPs in Western Australia demonstrated the awareness of national guidelines, hereditability, prevalence and diagnostic features of FH was not optimal [13].

Dyslipidemia patients within the community utilising the existing primary care services is a viable target for the detection of FH. An integrated screening program, using interpretative comments on lipid profiles by PCPs in detection and management can be developed [10]. Further work and research is required to develop physician's education and clinical tools for FH detection in primary care and to determine the impact on the detection and management of FH in India. The information also will be employed to design effective teaching and training modules for physicians in the detection and management of FH. Most of our current knowledge on FH stems from studies conducted in the West. At present, research regarding FH in the Indian population is minimal. Therefore there is a tremendous need for further research to be done and to determine the true prevalence of FH in India.

Majority of people with FH are undiagnosed [10]. Finding undiagnosed patients with FH in clinical management would be a good approach [17]. Absolute cardiovascular risk assessments and identifying FH is possible by PCPs given their place in the community [18].

Management of well controlled and low complexity FH patients would be in primary care for long-term or for shared care, while specialist service is required for high complexity patients. There are both diagnosed and undiagnosed cases of FH in primary care not known to secondary care [19]. The gap in the detection of FH is due to inadequate documentation of premature CAD among first-degree relatives in public hospitals [20]. Specialists and primary care providers require education and awareness campaigns to optimize FH management and reduce the burden of premature vascular disease due to dyslipidaemias.

Education and training of primary care providers in lipid management is important for improving and maintaining the total quality of care. The majority of people with FH who are detected early may be managed in primary care. The role of primary care in the care of FH has not been adequately defined and our preliminary data suggest a significant shortfall in awareness, knowledge and practices among family doctors. Humphires et al., reports that GP's require a working knowledge of FH to carry out cascade testing, risk management, and identification of potential new probands of FH from the practice list [21]. The international guidance acknowledges evidence gaps, but aims to make the best

use of contemporary practice and technology to achieve the best outcomes for the care of FH. It should accordingly be employed to inform clinical judgement and be adjusted for country-specific and local health care needs and resources [11]. The guidance from the International FH Foundation, the National Lipid Association Expert Panel, the European Atherosclerosis Society, and the Japanese guidelines should provide a basis to establishing country-specific models of care in Asia. Gap in cardiovascular preventative medicine should be remedied by several approaches aimed at improving awareness of FH among physicians, and health care professionals [16]. The expert in the prevention of CVD should demonstrate knowledge and competence in existing patient-specific, systems approaches to prevention of CVD [21].

Among the study participants, 41.4% of them were unaware or unsure whether they had patients under their care is alarming as the prevalence of FH is now known to be approximately 1:200 globally. This reinforces that FH will need to be detected and managed in primary care 71.4% of the participants were able to clinically define FH, although only 40.6% of them were aware of the national guidelines and 34.6% of PCP's could correctly identify the typical lipid profile. Knowledge of the prevalence and heritability of FH were also around 30%. Only 24.8% of physicians declared that they currently had patients with FH in their practice, reflecting that FH is underdiagnosed in the community [22,23]. An 84.2% of the general physicians stated they were the most effective healthcare provider to detect FH, which is encouraging as general physicians are central to both systematic and opportunistic FH detection [24].

Among the GPs, 69.2% of them preferred interpretative comments on the laboratory forms to alert possible FH. Interpretative comments have previously been associated with greater LDL cholesterol reductions and increased specialist referral in individuals at high risk of FH. 91.7% of GPs identified the more effective cholesterol lowering treatments as statins but only 19.5% of them selected the combination therapy such as statin and ezetimibe for FH.

With the information obtained, the educational and training sessions will be conducted to improve the awareness and knowledge in the detection and care of FH in India in primary health care. Mutation analysis of LDL receptors can be performed in clinically identified FH patients aiming to select a cut off diagnostic level of plasma LDL cholesterol in the future. Campaigns can be planned to improve the public awareness. As FH is common among patients with premature CAD, detection of FH can be planned in coronary care units [25].

There are between 14 and 34 million individuals with FH Worldwide based on extrapolations from the estimated 1/500–1/200 prevalence. The theoretical frequency of heterozygous FH of 1/500 – 1/200 prevalence is estimated to be 3.6 to 9.0 million for Western Pacific region [10]. It has been estimated that over five million people with FH are living in the Asia-Pacific region [14]. With this assumption, it would be estimated that there will be between 3.5 and 5 million people with FH in India.

CVDs are a leading cause of death in India. The huge burden of CVD is a consequence of the high prevalence of cardiovascular risk factors like obesity, diabetes mellitus and hypertension [26]. In addition to these compounding factors, FH should also be detected early and managed to prevent CVD.

LIMITATION

There are several limitations to our study design. Firstly, as the study used a unique web link which requires the internet facilities to participate, this prevented those practicing in rural areas and senior practitioners who generally are not well versed in internet usage. Secondly, the overall study population was small, which reduced the statistical power of the study. Thirdly, the study was conducted for GPs from southern states of India only, but we are extending this FH knowledge and awareness assessment study in all parts of India.

CONCLUSION

Despite recognition that patients with FH are at risk for premature CAD, they are still under-diagnosed. This could be rectified by conducting the epidemiological studies for the community prevalence of LDL cholesterol levels correlating with the clinical features to determine the cut-off levels for Indian FH patients.

Our survey suggests that there is a substantial deficit in the awareness and knowledge regarding FH among the registered medical practitioners in Tamil Nadu. Our findings support the need for educational sessions on FH for the doctors in India. Further work and research is required to develop GP education and clinical tools for FH detection in primary care. There is a need for including FH topics in the medical college's curricula as well as having structured and more frequent CME activities at teaching hospitals, especially targeting the GPs. In addition, knowledge regarding FH can be improved by the use of posters and leaflets displayed in hospitals as well as by using the print (newspapers and journals) and the electronic media (radio and television) by campaigning a national FH day.

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