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RESEARCH ARTICLE

FAMILIAL HYPERCHOLESTEROLAEMIA IN GREEK FEMALES. AN EPIDEMIOLOGICAL STUDY

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Abstract

Background: Cardiovascular diseases are the leading cause of death worldwide. One of the risk factors for cardiovascular diseases is dyslipidemia, defined as the imbalance of lipids in the blood. Familial hypercholesterolemia (FH) is a genetic disorder of lipoprotein metabolism characterized by high levels of total cholesterol in plasma with harmful cardiovascular consequences starting in childhood.
Aim: The aim of the present study was to explore the frequency of familial hypercholesterolemia in the female population of Greece.
Method and Material: This is a cross-sectional observational study, in which individuals of Greek origin, residing in Greece, were selected. The study sample consisted of 2995 individuals. Data analysis was performed with IBM SPSS 21.0.
Results: The mean age of the women was 48.3 years. The prevalence of FH was 1.9%, with 1.8% of women having a probable diagnosis of FH and 0.1% having a definite diagnosis of FH. Familial hypercholesterolemia was more common in women with artery hypertension ($p<0.001$), diabetes mellitus ($p=0.01$) and coronary artery disease ($p<0.001$). Familial hypercholesterolemia was more common in older women ($p<0.001$) and with a higher body mass index ($p=0.02$).
Conclusions: Optimal screening and diagnosis of FH are of primary importance for the prevention of premature cardiovascular events. Management of patients with FH requires an interprofessional approach, including primary care providers, cardiologists, endocrinologists, dietitians, pharmacists, and nurses, to improve outcomes.

Keywords: Familial hypercholesterolemia, women, incidence.

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INTRODUCTION

Cardiovascular diseases are the leading cause of death worldwide. One of the risk factors for cardiovascular diseases is dyslipidemia, presented as a lipids' imbalance in the blood. The most common form of dyslipidemia is cholesterolemia, in which LDL cholesterol blood levels are elevated. Greece is a moderate-risk country in terms of the risk of cardiovascular disease, while 27% of deaths due to cardiovascular disease are related to hypercholesterolemia.[1-3]

Familial hypercholesterolemia (FH) is a genetic disorder of lipoprotein metabolism characterized by high levels of total plasma cholesterol with harmful cardiovascular consequences that begin in childhood. It is mainly caused by mutations in the Low Density Lipoprotein Receptor (LDLR) gene. More than 1,800 mutations of the above receptor have been described in the international literature.[1-3]

In order to diagnose the disease, criteria such as the Simon Broome and Dutch criteria are used. Clinical diagnosis is made based on high plasma LDL-C levels, family history of hypercholesterolemia, history of early cardiovascular event and the presence of tendon xanthomas.[4]

A very important manifestation of familial hypercholesterolemia is atherosclerosis, which presents itself mainly in adulthood, but has an early onset as early as the 1st decade of the individual's life. Subclinical atherosclerosis has been observed in children with heterozygous and homozygous forms of familial hypercholesterolemia.[5]

In adulthood, the first manifestation is usually acute myocardial infarction, which can occur as early as the third decade of life and on average 20 years earlier than in individuals without familial hypercholesterolemia.[4]

Regarding the prevalence of the disease, studies show that it varies between countries. The following results are indicative: 1 in 217 people in Denmark, 1 in 150 among French Canadians in Quebec and 1 in 70 among Africans in South Africa.[6-8]

Early diagnosis and treatment of the disease is vital. However, there seems to be a distinction between the two sexes, with women being less likely to be diagnosed and to start lipid-lowering treatment. In addition, it is difficult to continue treatment without interruption, such as for childbearing purposes, resulting in failure to achieve the desired LDL levels. Early diagnosis is of fundamental importance for the primary prevention of the disease, while it is crucial to perform a genetic test so that if treatment is deemed necessary, it can start as soon as possible. The ideal strategy for controlling familial hypercholesterolemia is targeted testing of individuals (mainly children) who have risk factors for the onset of the disease. Another strategy is that of genetic testing, which involves searching for common genes that cause the disease in children and their close relatives.[9,10]

Representation of women in trials studying FH and lipid-lowering drugs is critical to improving outcomes and reducing sex disparities. Many studies have had low rates of female representation, with the exception of the CASCADEFH (Cascade Screening for Awareness and Detection of Familial Hypercholesterolemia) registry, in which 60% were women.[11]

AIM

The aim of the present study was to explore the frequency of familial hypercholesterolemia in the female population.

Sub-objective

To investigate the clinical characteristics and risk factors of women in relation to the disease.

MATERIAL – METHODS

Research design

This is a cross-sectional observational study, in which individuals of Greek origin, residing in the country of Greece, were selected. For the diagnosis of FH in adults the Dutch Lipid Clinic Network (DLCN) criteria were used.

Study sample

The studied sample consisted of 2995 Greek women. The selection of individuals was random, in order to increase the representativeness of the sample. Data collection took place between 2020 and 2022.

Inclusion criteria for study participants

Individuals must:

- Be female
- Be of Greek origin
- Be between 18 to 95 years of age

Exclusion criteria for study participants

- Severe cognitive impairment or taking psychiatric medications
- Age less than 18
- One or both parents not of Greek origin
- Incomplete data from laboratory tests or history

Data Collection – Measurements

A closed-ended questionnaire was used to collect data. The information was obtained and recorded after a personal interview. Data collection was carried out by completing a questionnaire that was specifically designed for the needs of this study. The questions concerned demographic and anthropometric data of the study participants, their individual habits such as physical activity, smoking, and dietary habits.

Ethics

Written informed consent was obtained from all study participants to participate in the research. Study participants were informed about the purpose of the study, the confidentiality of the data and the voluntary nature of participation. The duration of the personal interview was 10-15 minutes. Data collection was carried out after written permission from the competent authority.

STATISTICAL ANALYSIS

Categorical variables are presented as absolute (n) and relative (%) frequencies, while quantitative variables are presented as mean and standard deviation. The Kolmogorov-Smirnov criterion was used to test the distributions of quantitative variables for normality. Quantitative variables followed a normal distribution. To explore relationships between two categorical variables for independent samples, the x^2 test and Fisher's exact test were used. To investigate the existence of a relationship between a categorical variable and an ordinal variable, the x^2 test for trend was used. To investigate the existence of a relationship between a quantitative variable that followed a normal distribution and a dichotomous variable, the t test was used. The two-sided statistical significance level was set at 0.05. Data analysis was performed with IBM SPSS 21.0 (Statistical Package for Social Sciences).

RESULTS

Demographic characteristics of the women are presented in Table 1. The study sample included 2995 women >18 years of age. The mean age of the women was 48.3 years. The mean body mass index was 25.5 kg/m2, while 49.5% of the women had a normal body mass index, 29.8% were overweight, 17% were obese, and 3.8% were underweight.

Table 2 presents the individual habits of the study participants. 55.2% consumed a small amount of alcohol, 36.2% did not consume any alcohol, 7.7% consumed a moderate amount of alcohol and 0.5% consumed a large amount of alcohol. 80.8% exercised at least one day per week and 90.7% consumed fish at least one day per week. 26.9% of the women were current smokers, 6.8% were former smokers and 66.3% had never smoked.

Clinical characteristics of the women are presented in Table 3. 47.7% of women reported taking medication, while 47% reported having a medical condition. 21.3% had hypertension, 17.5% had hypothyroidism, 8.7% had diabetes, 4.1% had a stroke, 3% had coronary artery disease, 2.1% had hyperthyroidism, and 1.2% had myocardial infarction. 5.4% of women had xanthelasma, 2.3% had senile plaques, and 1.2% had tendon xanthomas. 3.6% of women had cholesterol >290mg/dl, and 2.4% had LDL >190mg/dl. 18.5% were receiving lipid-lowering therapy.

The criteria used for the diagnosis of familial hypercholesterolemia in the studied sample were those of Simon Broome. Those who had the information and did not meet the conditions were classified as not having the disease. According to these, 98.1% (n=2939) of the participants have not developed the disease, 1.8% (n=53) were diagnosed with a possible occurrence of the disease, while 0.1% (n=3) have a confirmed diagnosis (Table 4).

Relationships between categorical variables and familial hypercholesterolemia

The relationships between categorical variables and familial hypercholesterolemia are presented in Table 5. The statistically significant relationships found were the following:

- Increased body mass index was associated with an increased likelihood of familial hypercholesterolemia (p=0.03).

- Familial hypercholesterolemia was more common in women taking medications (p<0.001).
- Familial hypercholesterolemia was more common in women suffering from a disease (p<0.001).
- Familial hypercholesterolemia was more common in women suffering from arterial hypertension (p<0.001).
- Familial hypercholesterolemia was more common in women suffering from diabetes (p=0.01).
- Familial hypercholesterolemia was more common in women who suffered from coronary heart disease (p<0.001).
- Familial hypercholesterolemia was more common in women who suffered from myocardial infarction (p=0.005).
- Familial hypercholesterolemia was more common in women who suffered from a stroke (p=0.01).
- Familial hypercholesterolemia was more common in women who were on lipid-lowering therapy (p<0.001).
- Familial hypercholesterolemia was more common in women with a family history of coronary heart disease (p<0.001).

Relationships between quantitative variables and familial hypercholesterolemia

The relationships between quantitative variables and familial hypercholesterolemia are presented in Table 6. The statistically significant relationships found were the following:

- Familial hypercholesterolemia was more common in older women (p<0.001).
- Familial hypercholesterolemia was more common in women with a higher body mass index (p=0.02).
- Familial hypercholesterolemia was more common in women with higher systolic pressure (p<0.001).
- Familial hypercholesterolemia was more common in women with higher diastolic pressure (p=0.03).
- Familial hypercholesterolemia was more common in women with a higher total cholesterol number (p<0.001).
- Familial hypercholesterolemia was more common in women with higher triglycerides levels (p<0.001).
- Familial hypercholesterolemia was more common in women with higher LDL values (p<0.001).

DISCUSSION

The present study presents very important results regarding the incidence of familial hypercholesterolemia in the female population. According to the diagnostic criteria for familial hypercholesterolemia used by the Simon Broome organization, 98.1% (n=2939) of the participants had not developed the disease, 1.8% (n=53) were diagnosed with probable onset of the disease, while 0.1% (n=3) had a confirmed diagnosis. Based on the results of the study, hypercholesterolemia was more common in women who had arterial hypertension, diabetes mellitus, coronary artery disease, had a history of acute myocardial infarction or stroke. It was also more common in older women and with a high BMI.

The present study showed that FH was more common in older women. FH is a severely underdiagnosed and undertreated genetic disorder. Little is known about the variation in the prevalence of FH, and information for Central and Eastern Europe is scarce. The meta-analysis by Pajak et al.,[12] of six population-based studies in Poland showed that the prevalence of probable FH was 404/100,000 persons (95% CI = 277–531/100,000). In agreement with the findings of the present study, the meta-analysis showed that familial hypercholesterolemia was more prevalent in women than in men, and the prevalence was highest in the age group 45–54 years in men and 55–64 years in women. A possible explanation for this finding is that women, according to a recent meta-analysis of 3,022 publications, had a 10% increased chance of non-adherence to statin therapy compared to men.[13] Another important finding of the study was that hypercholesterolemia was more common in women with higher BMI and high blood pressure. According to the literature, FH and obesity are established risk factors for atherosclerotic cardiovascular disease. The Greek registry of Familial Hypercholesterolemia showed that obese individuals had higher odds of established CHD as well as early onset CHD compared to those with normal BMI. More than half of adults with FH are overweight or obese. Obesity was associated with an increased prevalence of CHD in this population.[14] A study by Bertsias et al.,[15] in which Greek medical students were studied, found that male students had higher rates of obesity than females. Also, in the same study, it was found that both BMI and the waist-to-hip ratio were strong predictors of dyslipidemia.

Arterial hypertension is an important risk factor in people with FH. As can be seen from the results of the study, most women in the sample had high values and were under treatment. A similar study by Zingg et al.,[16] highlights the importance of systematic blood pressure control in people with familial hypercholesterolemia, in order to prevent cardiological consequences.

Based on the results, hypercholesterolemia was more common in women with diabetes mellitus, which is a very important risk factor in people with FH. More generally, in the international literature there are studies that report the close relationship between risk factors (such as diabetes mellitus, arterial hypertension) and endothelial dysfunction, vascular inflammation and the formation of atherosclerotic plaques.[17]

The present study was conducted exclusively with a female sample as women with FH face specific care challenges, including underrepresentation in research, significant underestimation of risk and discontinuation of treatment during childbearing. Data from national and international registries and clinical trials show significant differences in healthcare for women with FH.

Inequalities in care are more pronounced in women than in men. Furthermore, FH affects women differently throughout their lives, in different ways, and at different times. Specific factors that are likely to affect women include the concomitant use of contraceptives with lipid-lowering therapy, the timing of pregnancy, breastfeeding, and finally, the decision about treatment goals at menopause. Early recognition and appropriate treatment before stopping treatment for childbearing can lead to a significant reduction in morbidity and mortality. Women have different access to care than men, and increasing awareness among all health care providers can improve diagnostic rates. Understanding the unique challenges faced by women with FH is crucial to addressing the gaps in care they face.[18]

Limitations of the study

• The sample of this study consists exclusively of women.

• The most important limitation of the study was the correct assessment of the women's individual habits such as smoking, physical activity, diet, alcohol consumption, as well as family history.

• The above variables depend on the reliability of the person answering and therefore valid tools for assessing the characteristics were used, resulting in the reduction of the above methodological issue.

CONCLUSIONS

In conclusion and according to the results of the study:

• The high incidence of familial hypercholesterolemia is evident in the Greek female population. The disease was more common in women who suffered from arterial hypertension, diabetes mellitus, coronary heart disease and had a history of acute myocardial infarction or stroke. It was also more common in older women and with a high BMI.

• The prevention of cardiovascular diseases is an urgent need because on the one hand they continue to be the first cause of death in the Western world and on the other hand their treatment is costly, painful and unfortunately often ineffective. The maximum efficiency of their prevention can be achieved through the understanding, early detection and management of risk factors, such as familial hypercholesterolemia.[19]

• Familial hypercholesterolemia is a serious genetic disease with devastating consequences in the life of the patient. Although the need for a strategy to develop a pre-symptomatic screening for the early detection of the disease is widely accepted, there is still no correct strategy for the optimal timing of such screening.

• Early initiation of lipid-lowering therapy, modification of risk factors, as well as changes in the lifestyle of patients, are of primary importance in order to improve the clinical outcome of the disease and reduce the high cardiovascular risk that these patients run.

• Patient registries are a powerful tool for recording, monitoring the disease, and promoting clinical practices, contributing to improving clinical outcomes and reducing healthcare costs.

• The management of patients with familial hypercholesterolemia requires a multidisciplinary approach, including primary care providers, pathologists, cardiologists, endocrinologists, dietitians, pharmacists, and nurses. Extensive analysis of treatment strategies at diagnosis and close monitoring of response to treatment are required.

• Women are much less likely to receive adequate primary prevention, counseling, and preventive treatment than men. Most

research on the causes, risk factors, and treatment of cardiovascular disease focuses on men.[20]

• Although female participation has increased significantly due to specific studies such as the Women's Health Initiative, women still represent less than ⅓ of the sample of studies in which participants belong to both sexes.[21]

• The representation of women in studies needs to be improved, so that there is an equitable and more effective treatment.

• The resulting lack of data on the female population hinders the making of sound clinical decisions for 51% of the world's population.[20]

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ANNEX

TABLE 1. Demographic characteristics of women.

Characteristics	**Mean**	**Standard Deviation**
Age	48.3	16.9
Height (centimeters)	163.7	6.6
Weight (kilograms)	68.3	13.8
Body mass index (kg/meter2)	25.5	5.1
BMI categories		
Underweight	109	3.8
Normal	1439	49.5
Overweight	865	29.8
Obese	493	17.0

TABLE 2. Individual habits of the studied sample.

Individual habits	**N**	**%**
Alcohol consumption [α]		
None	1081	36.2
Little	1656	55.5
Moderate	231	7.7
Heavy	15	0.5
Days of exercise per week [α]		
0	574	19.2
1	859	28.8
2	629	21.1
3	473	15.9
4	448	15.0
Days of fish consumption per week [α]		
0	278	9.3
1	1674	56.0
2	767	25.7
3	192	6.4
4	77	2.6
Smoking habits [α]		
Never smokers	1976	66.3
Former smokers	203	6.8
Current smokers	802	26.9
Electronic cigarettes [α]		
No	2882	97.0
Yes	90	3.0
Pack-years of smoking	44.2	452.1

[α] Values are expressed as absolute frequencies (n) and relative frequencies (%).

TABLE 3. Clinical characteristics of women.

Characteristics	n	%
Taking medications		
No	1559	52.3
Yes	1423	47.7
Xanthelasma		
No	2832	94.6
Yes	161	5.4
Symptoms		
No	2922	97.7
Yes	70	2.3
Symptoms on tendons		
No	2955	98.8
Yes	37	1.2
Presence of disease		
No	1587	53.0
Yes	1408	47.0
High blood pressure		
No	2356	78.7
Yes	637	21.3
Cholesterol >290mg/dl		
No	2666	96.4
Yes	99	3.6
LDL >190mg/dl		
No	2482	97.6
Yes	61	2.4
Diabetes mellitus		
No	2733	91.3
Yes	260	8.7
Lipid-lowering therapy		
No	2436	81.5
Yes	553	18.5
Coronary artery disease		
No	2901	97.0
Yes	89	3.0
Myocardial infarction		
No	2954	98.8
Yes	37	1.2
Ceremorrhage		

No	2865	95.9
Yes	123	4.1
Hypothyroidism		
No	2404	92.5
Yes	509	17.5
Hyperthyroidism		
No	2851	97.9
Yes	62	2.1
Systolic pressure[a]	120.1	17.7
Diastolic pressure[a]	73.9	19.4
Blood glucose[a]	97.1	33.2
Total cholesterol [α]	198.9	42.8
Triglyceride [α]	130.9	75.9
HDL[a]	57.4	29.1
LDL[a]	121.9	43.3

[α] Values are expressed as mean and standard deviation

TABLE 4. Classification of the sample for diagnosis of familial hypercholesterolemia according to the Simon Broome criteria.

	N	**%**
Familial Hypercholesterolemia		
Absence of disease	2939	98.1%
Possible diagnosis	53	1.8%
Definite diagnosis	3	0.1%

TABLE 5. Relationships between categorical variables and familial hypercholesterolemia.

Categorical variables	**Familial hypercholesterolemia**				**p-value**
	Absence of disease		Probable/definite disease		
	N	**%**	**N**	**%**	
Categories according to body mass index					0.03α
Underweight	108	99.1	1	0.9	
Normal	1421	98.7	18	1.3	
Overweight	848	98	17	2	
Obese	480	97.4	13	2.6	
Alcohol consumption					0.051α
None	1051	97.2	30	2.8	
Low	1634	98.7	22	1.3	
Moderate	227	98.3	4	1.7	
High	15	100	0	0	
Days of exercise in a week					0.84α
0	565	98.4	9	1.6	
1	840	97.8	19	2.2	
2	617	98.1	12	1.9	
3	464	98.1	9	1.9	
4	441	98.4	7	1.6	
Days of fish consumption in a week					0.45α
0	272	97.8	6	2.2	
1	1641	98	33	2	
2	755	98.4	12	1.6	
3	187	97.4	5	2.6	
4	77	100	0	0	
Smoking habit					0.35α
Never smokers	1943	98.3	33	1.7	
Former smokers	197	97	6	3	
Current smokers	785	97.9	17	2.1	
Electronic cigarette					0.78β
No	2829	98.2	53	1.8	

Yes	88	97.8	2	2.2	
Taking medication					<0.001β
No	1551	99.5	8	0.5	
Yes	1375	96.6	48	3.4	
Presence of disease					<0.001β
No	1580	99.6	7	0.4	
Yes	1359	96.5	49	3.5	
Arterial hypertension					<0.001β
No	2327	98.8	29	1.2	
Yes	610	95.8	27	4.2	
Diabetes mellitus					0.01β
No	2687	98.3	46	1.7	
Yes	250	96.2	10	3.8	
Lipid-lowering therapy					<0.001β
No	2422	99.4	14	0.6	
Yes	511	92.4	42	7.6	
Coronary artery disease					<0.001β
No	2853	98.3	48	1.7	
Yes	81	91	8	9	
Myocardial infarction					0.005γ
No	2901	98.2	53	1.8	
Yes	34	91.9	3	8.1	
Vascular stroke					0.01γ
No	2816	98.3	49	1.7	
Yes	117	95.1	6	4.9	
Family history of coronary artery disease					<0.001β
No	2007	100	1	0	
Yes	927	94.4	55	5.6	
Family history of stroke					0.14β
No	2012	98.4	32	1.6	
Yes	919	97.7	22	2.3	
Relatives over 90 years of age					0.52β
No	1931	98.3	34	1.7	

Yes	998	97.9	21	2.1	
Hypothyroidism					0.62β
No	2360	98.2	44	1.8	
Yes	498	97.8	11	2.2	
Hyperthyroidism					0.43β
No	2798	98.1	53	1.9	
Yes	60	96.8	2	3.2	

α test x2
β test x2
γ Exact test Fisher

TABLE 6. Relationships between quantitative variables and familial hypercholesterolemia.

Quantitative variables	**Familial hypercholesterolemia**						**p - value**
	Absence of disease			Probable/definite disease			
	N	**Mean**	**SD**	**N**	**Mean**	**SD**	
Age	2927	48.1	16.9	49	57.7	13.2	<0.001
Body mass index (kg/m2)	2857	25.5	5.1	49	27.2	5.2	0.02
Pack-years of smoking	598	44.7	45.5	12	21.2	12.5	0.86
Systolic pressure	2533	119.8	17.6	47	132.0	21.8	<0.001
Diastolic pressure	2521	73.8	19.5	47	80.0	12.3	0.03
Blood glucose	2233	96.9	33.3	44	105.9	27.2	0.08
Total cholesterol	2710	196.7	39.6	55	308.5	49.6	<0.001
Triglycerides	2689	129.6	74.3	49	198.5	119.5	<0.001
HDL	2556	57.3	29.1	42	61.9	30.8	0.31
LDL	2500	121.0	42.6	43	173.4	52.3	<0.001

α t test