# Familial hypercholesterolemia in Türkiye: A comprehensive narrative review of the literature

## Türkiye'de ailesel hiperkolesterolemi: Literatürün kapsamlı incelemesi

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**Abstract**

**Aim:** Familial Hypercholesterolemia (FH), a genetic disorder characterized by elevated LDL (low-density lipoprotein) cholesterol levels, is a significant risk factor for premature cardiovascular disease (CVD). Heterozygous FH (HeFH) is particularly underdiagnosed and undertreated worldwide, including in Turkey. This review aimed to evaluate the current status and characteristics of FH in Turkish population, highlighting potential differences from global trends.

**Methods:** To determine the current status of familial hypercholesterolemia in Turkey, PubMed (MEDLINE), Embase, Google Scholar, the Turkish Higher Education Council Thesis Center, and the Web of Science databases were searched.

**Results:** The literature review conducted to assess the current status of FH in Turkey revealed a lack of sufficient studies providing comprehensive data on the condition within the country. The results of studies indicate that the prevalence of FH in Turkey might be higher than the global average, possibly due to genetic and environmental factors unique to the region. Additionally, Turkish patients with FH were found to have a higher burden of untreated LDL cholesterol and an earlier onset of CVD symptoms compared to global reports. These findings underscore a substantial gap in early detection and treatment strategies for FH in Turkey.

**Conclusion:** This study suggests the urgent need for improved screening programs, public awareness campaigns, and tailored therapeutic interventions to address the FH burden in Turkey. By highlighting regional variations, this research contributes to the broader understanding of FH and emphasizes the importance of global collaboration in tackling this silent but impactful condition.

**Keywords:** Atherosclerosis; dyslipidemias; familial hypercholesterolemia; genetic; heterozygous; homozygous

**Öz**

**Amaç:** Yüksek LDL (düşük yoğunluklu lipoprotein) kolesterol düzeyleri ile karakterize genetik bir bozukluk olan Ailesel Hiperkolesterolemi (AH), erken yaşta kardiyovasküler hastalık (KVH) için önemli bir risk faktörüdür. Heterozigot AH (HeAH), Türkiye de dahil olmak üzere dünya çapında yeterince teşhis edilmemekte ve yeterince tedavi edilmemektedir. Bu derleme, Türkiye'de AH'nin mevcut durumunu ve özelliklerini değerlendirmeyi ve küresel eğilimlerden olası farklılıkları vurgulamayı amaçlamaktadır.

**Yöntem:** Trükiye'deki AH ile ilgili güncel durumun tespiti için PubMed (MEDLINE), Embase, Google Scholar, the Turkish Higher Education Council Thesis Center, and the Web of Science veri tabanlarında tarama yapıldı.

**Bulgular:** Türkiye'de AH'nin mevcut durumunu değerlendirmek için yürütülen literatür incelemesi, ülke içinde durum hakkında kapsamlı veri sağlayan yeterli çalışmanın eksikliğini ortaya koymuştur. Çalışmaların sonuçları, Türkiye'de AH yaygınlığının, muhtemelen bölgeye özgü genetik ve çevresel faktörler nedeniyle küresel ortalamadan daha yüksek olabileceğini göstermektedir. Ek olarak, AH'li Türk hastaların, küresel raporlara kıyasla tedavi edilmemiş LDL kolesterol yükünün daha yüksek olduğu ve KVH semptomlarının daha erken başladığı bulunmuştur.

**Sonuç:** Bu bulgular, Türkiye'de AH için erken teşhis ve tedavi stratejilerinde önemli bir boşluğun altını çiziyor. Bu çalışma, Türkiye'de AH yükünü ele almak için iyileştirilmiş tarama programlarına, kamuoyu farkındalık kampanyalarına ve özel terapötik müdahalelere acil ihtiyaç olduğunu öne sürüyor. Bölgesel farklılıkları vurgulayarak, bu araştırma AH'nin daha geniş bir şekilde anlaşılmasına katkıda bulunuyor ve bu sessiz ama etkili durumla başa çıkmada küresel iş birliğinin önemini vurguluyor.

**Anahtar Sözcükler:** Ailesel hiperkolesterolemi; ateroskleroz; dislipidemiler; genetik; heterozigot; homozigot

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## INTRODUCTION

Familial hypercholesterolemia (FH) is an inherited disorder of lipid metabolism that predisposes individuals to premature cardiovascular disease. Clinically, FH manifests in two forms: the milder heterozygous form (HeFH) and the more severe homozygous form (HoFH) (1). The condition arises from genetic mutations in the genes encoding the low-density lipoprotein receptor (LDLR), low-density lipoprotein receptor adaptor protein-1 (LDLRAP1), apolipoprotein B (ApoB), or proprotein convertase subtilisin/kexin type 9 (PCSK9), leading to defective LDL clearance and significantly elevated LDL cholesterol levels (2,3).

FH can present with dermatological manifestations such as tendon xanthomas, corneal arcus, and xanthelasmas (4). However, the primary concern in these patients is markedly elevated LDL-C levels and the consequent atherosclerosis. LDL-C concentrations are typically above 190 mg/dL, and due to the congenital nature of the disorder, significant atherosclerosis may develop by adulthood, particularly affecting the coronary arteries. This results in an increased risk of myocardial infarction (MI), valvular heart disease, stroke, and premature mortality, often before the ages of 50-60 (5).

Early diagnosis and timely intervention are crucial in managing FH. Implementing screening programs targeting the relatives of diagnosed individuals is crucial for identifying more cases and initiating preventive measures at an earlier stage (6). Despite this, global screening efforts for FH remain insufficient, with limited epidemiological studies conducted in most countries (7,8). Consequently, the majority of individuals with FH worldwide remain undiagnosed. Studies have also revealed that even among diagnosed patients, treatment goals are often unmet, and therapy is frequently suboptimal (9).

FH is a significant public health issue in Turkey, as it is globally. Unfortunately, no large-scale studies or systematic screening initiatives have been conducted to assess the prevalence of FH in Turkey. Recent meta-analyses indicate a lack of data estimating FH prevalence in the Turkish population (8,10). Therefore, there is an urgent need for comprehensive research and screening efforts to address this critical health challenge in Turkey.

This review aimed to evaluate the current status and characteristics of FH in Turkish population, highlighting potential differences from global trends.

## MATERIALS AND METHODS

Although this review follows a structured approach in its preparation, it is designed as a narrative literature review rather than a systematic review. As such, no specific methodological framework (e.g., PRISMA or similar protocols) was applied in its development.

This review aimed to review the existing research on FH conducted in Turkey and to summarize the current status of the country regarding screening, diagnosis, treatment, and follow-up of the disease. To achieve this, comprehensive searches were performed across several databases, including PubMed (MEDLINE), Embase, Google Scholar, the Turkish Higher Education Council Thesis Center, and the Web of Science, up to February 3, 2025. Keywords and Medical Subject Headings (MeSH) used in the search included combinations of "familial hypercholesterolemia" and "Turkey" or "familial hypercholesterolaemia," and "Türkiye." Additional searches were conducted using these terms in Turkish (ailesel hiperkolesterolemi, ailesel hiperlipidemi, ailevi hiperkolesterolemi, ailevi hiperlipidemi).

The selection process began with screening the titles of the retrieved articles to exclude irrelevant publications. Abstracts and full texts of the remaining articles were then thoroughly reviewed. Studies not conducted within the geographical boundaries of Turkey were excluded from the analysis. Furthermore, publications categorized as reviews, case reports, letters to the editor, or book chapters were omitted to ensure the focus remained on primary research and clinical relevance.

As a result, 43 research articles and 16 theses were evaluated (figure 1).

Among the included studies, 11 were classified as prevalence studies. The majority of these epidemiological investigations were retrospective, utilizing data obtained from hospital records or national health ministry databases. Thirteen studies focused on the treatment and long-term follow-up of FH patients, while four adopted a survey-based design to assess awareness. Additionally, eight studies evaluated comorbidi-

ties and cardiac function, and 17 involved genetic and molecular analyses. Notably, 31 studies were conducted in a prospective format (table 1).

## DIAGNOSIS AND CLINICAL MANIFESTATIONS

Although FH is a genetic condition, its diagnosis can often be made based on clinical findings. Several diagnostic criteria are utilized to identify FH, including the Simon Broome criteria from the United Kingdom, the Make Early Diagnosis to Prevent Early Death (MEDPED) criteria from the United States, and the American Heart Association (AHA) criteria, and the Dutch Lipid Clinic Network (DLCN) criteria from the Netherlands. (11-13).

Typically, FH is diagnosed after the occurrence of a premature cardiovascular event associated with LDL-C levels > 160 mg/dL or in individuals presenting with LDL-C levels > 190 mg/dL along with a family history of premature cardiovascular disease (CVD). However, atypical cases frequently remain undiagnosed and untreated unless systematic screening programs are implemented (8).

An accelerated progression of atherosclerosis serves as a critical indicator for suspecting FH. Patients with FH exhibit generalized atherosclerosis involving multiple vascular territories, with cholesterol deposits predominantly affecting the proximal segments of the arteries (9,14).

The European Atherosclerosis Society (EAS) recommends the following criteria for the clinical diagnosis of HoFH: genetic confirmation of two mutant alleles at the LDLR, APOB, PCSK9, or LDLRAP1 gene locus, or an untreated LDL-C level exceeding 500 mg/dL (13 mmol/L), or treated LDL-C levels ≥ 300 mg/dL (8 mmol/L) in combination with either cutaneous or tendon xanthomas. EAS recommends the use of the Dutch Lipid Clinic Network (DLCN) criteria to diagnose HeFH (figure 2) (12).

## SCREENING OF FH

Screening plays a crucial role in the early identification of FH. Various approaches are available, including universal, systematic, targeted, and opportunistic screening. Current guidelines advocate for cascade screening, a form of targeted screening, following the diagnosis of an index case (12). This method entails clinical or genetic screening of family members. However, clinicians must remain vigilant as negative genetic testing results do not definitively rule out FH.

Evidence from the United Kingdom indicates that cascade screening can reduce the age at diagnosis and improve the initiation rates of statin therapy among FH patients (15). The Netherlands pioneered the systematic implementation of cascade screening in 1994. In this program, screening 5442 relatives of 237 index cases identified 2039 individuals as heterozygous carriers of LDLR mutations (16).

## TREATMENT

Individuals with HeFH typically exhibit untreated cholesterol levels ranging from 250 to 300 mg/dL (6.5–7.8 mmol/L), which can result in cardiovascular events occurring between ages 30–50 in males and 40–60 in females (11). For those with HoFH, severe atherosclerotic events can manifest in early childhood. Without treatment, HoFH patients rarely survive beyond 30 years of age. Despite the significantly elevated risk of premature atherosclerosis in FH patients, they can potentially achieve a normal life expectancy if low LDL-C levels are attained early and maintained consistently. Consequently, prompt diagnosis and effective cholesterol-lowering therapy are vital for these individuals (14,16,17).

Recent advancements in treatment options have enabled patients to achieve their LDL-C targets (12). The main objective in treating FH is to minimize the risk of atherosclerotic cardiovascular disease. While lifestyle changes, including diet modifications and physical activity, are crucial, they are often insufficient. Research indicates that dietary interventions, although beneficial, do not provide adequate protection for these patients (18). Medication remains the cornerstone of treatment. Statins are still considered the first-line therapy for FH patients (12). If necessary, statin therapy can be increased to the maximum tolerated treatment (13). Additional medications such as ezetimibe, bile acid sequestrants, and novel PCSK9-targeting therapies are utilized, either independently or in conjunction with statins, to further reduce LDL-C levels. In cases unresponsive to

**Table 1.** Classification of included studies

| Study category | Number of studies | Design/Notes |
|---|---|---|
| Prevalence Studies | 12 | Mostly retrospective; hospital/national health data |
| Treatment & long-term follow-up | 13 | Includes LDL apheresis, pharmacotherapy |
| Awareness & survey studies | 6 | Patient/physician knowledge assessments |
| Comorbidity & cardiac function | 9 | CVD, aortic valve involvement, imaging |
| Genetic & molecular analyses | 19 | LDLR, APOB, PCSK9 variants, novel mutations |
| Total studies | 59 | Original research: 43 • Specialty theses: 16 |
| Prospective design | 31 | Subset of above categories |

LDL: Low density lipoprotein, CVD: Cardiovascular disease, LDLR: Low-density lipoprotein receptor, APOB: Apolipoprotein B,

**Table 2:** FH prevalence in different groups and regions (8)

| | Worldwide | USA | Europe | Middle East | Asia |
|---|---|---|---|---|---|
| | Prevalence (%) | Prevalence (%) | Prevalence (%) | Prevalence (%) | Prevalence (%) |
| General population | **0.32** | **0.32** | **0.32** | **0.28** | **0.19** |
| Ischemic heart disease | **3.2** | **4.38** | **2.26** | **1.69** | **3.57** |
| Premature ischemic heart disease | **6.7** | **2.83** | **8.04** | **12.16** | **5.43** |

FH: Familial hypercholesterolemia, USA: United States of America, %: Percent

maximal and combined medical therapy, lipoprotein apheresis or liver transplantation may be necessary (3). Lipoprotein apheresis can be used frequently, especially in HoFH cases resistant to drug therapies. (3,13). Although liver transplantation is recommended for cases refractory to lipoprotein apheresis, it has been utilized in only a limited number of instances.

Recommendations of European Society of Cardiology (ESC) and European Atherosclerosis Society (EAS) for the detection and treatment of patients with HeFH (12):

- It is recommended to consider the diagnosis of FH in patients with chronic heart diseases (CHD) aged <55 years for men and <60 years for women, in people with relatives with premature fatal or non-fatal CVD, in people with relatives who have tendon xanthomas, in people with severely elevated LDL-C (in adults > 5 mmol/L [>190 mg/dL], in children > 4 mmol/L [>150mg/dL]), and in first-degree relatives of FH patients.
- It is recommended that FH should be diagnosed using clinical criteria and confirmed, when available, with DNA analysis.
- Once the index case is diagnosed, family cascade screening is recommended.
- It is recommended to treat FH patients with atherosclerotic cardiovascular diseases (ASCVD) or who have another major risk factor as very-high-risk, and those with no prior ASCVD or other risk factors as high-risk.
- For FH patients with ASCVD who are at very-high risk, treatment to achieve at least a 50% reduction from baseline and an LDL-C < 1.4 mmol/L (<55 mg/dL) is recommended. If goals cannot be achieved, a drug combination is recommended.

We now have more effective tools than ever to manage FH. Statins, ezetimibe, and PCSK9 inhibitors can effectively regulate LDL-C levels in these patients. Controlling lipid levels significantly improves patient outcomes by preventing heart attacks, strokes, and premature deaths. However, data reveal that both the identification of these patients and their consistent treatment post-diagnosis encompass only a small fraction. A considerable number of patients remain either undiagnosed or, if diagnosed, do not receive regular treatment (9,14,19).

These observations indicate that FH may be underdiagnosed in numerous populations. Despite its prevalence, FH remains largely unrecognized and undertreated. In the Greek study, only 63.1% of patients were undergoing lipid-lowering drug treatment when enrolled in the registry, and the vast majority (87.9%) of treated patients did not reach LDL-C targets (14). Likewise, in the Russian study, only 63% of FH patients were prescribed statins, and merely 3% achieved the LDL-C goal based on 2019 ESC/EAS guidelines (19).

These findings underscore the necessity for enhanced awareness, early detection, and proper management of FH to decrease the risk of premature cardiovascular disease in affected individuals.

## CURRENT STATUS OF FAMILIAL HYPERCHOLESTEROLEMIA IN THE WORLD

Familial hypercholesterolemia (FH) is among the most common genetic disorders, with an estimated prevalence of approximately 1 in 200 to 500 heterozygotes in the world (2,8,14).

A comprehensive meta-analysis reported the prevalence of FH across different populations and clinical presentations as follows (8):

In the general population, the prevalence is approximately 1 in 313 individuals. Among individuals with ischemic heart disease (IHD), FH prevalence is 10 times higher (1 in 31). For those with premature IHD, the prevalence increases 20-fold (1 in 15). In cases of severe hypercholesterolemia (LDL-C≥190 mg/dl) the prevalence is 23-fold higher (1 in 14). Heterozygous FH prevalence is estimated at 1 in 313, while homozygous FH occurs at a rate of about 1 in 400,000 (table 2) (8).

In Greece, a study reported a median age of FH diagnosis of 42.2 years, with 47.8% of patients having a family history of cardiovascular disease (CVD) and 21.1% having a personal history of CVD (14).

Data on FH prevalence is available for only a small fraction of countries, with just 17 of 195 (9%) reporting prevalence estimates for the general population. Consequently, the true prevalence remains unknown for 91% of countries worldwide (8).

Recent studies suggest FH may be more common than previously believed. For instance, a study analyzing exome sequencing and electronic health records from 50,726 individuals in the Geisinger Health System estimated FH prevalence at 1 in 256 for unselected participants and 1 in 118 for participants undergoing cardiac catheterization (20). In Russia, a study across 11 regions found definite or probable heterozygous FH in 0.58% (1 in 173) of the population (19).

Over time, FH prevalence estimates have risen. A meta-analysis showed an increase from 1 in 588 (0.17%) in data up to 2012 to 1 in 333 (0.30%) by 2018. Intermediate estimates included 1 in 476 (0.21%) in 2015, 1 in 456 (0.22%) in 2016, and 1 in 323 (0.31%) in 2017 (8).

When the Simoon Broome diagnostic criteria were used, the prevalence was found to be approximately three times higher from the average prevalence, which is around 1%. The prevalence obtained with clinical findings, Dutch Lipid Clinic Network (DLCN), and genetic studies resulted in similar rates (0.32%, 0.31% and 0.34%, respectively) (8).

A meta-analysis of individuals with atherosclerotic cardiovascular disease (ASCVD) found FH prevalence to be significantly elevated, occurring in 1 in 17 (5.8%) of cases. Among ASCVD patients, the highest frequency was noted in those with ST-elevation myocardial infarction at 1 in 14 (7.1%). In stroke or transient ischemic attack (TIA) patients, the prevalence was 1 in 75 (1.3%) (21).

Globally, FH prevalence varies by ethnicity, with the highest frequency observed in individuals of Black descent (0.52%, 1 in 192) and the lowest among Asians (0.25%, 1 in 400) (10). In Asia, although FH prevalence is generally lower (0.19%), it rises to levels comparable with other regions (3.6% vs. 3.2% respectively) when considering individuals with IHD. A study in the Middle East reported a prevalence of 0.28% for the region (8).

No specific data on FH prevalence in Turkey is available, leaving the country among those with significant data gaps on this topic (8, 21).

## CURRENT STATUS OF FAMILIAL HYPERCHOLESTEROLEMIA IN TURKEY

Two of the most significant studies on FH in Turkey are the A-HIT1 and A-HIT2 studies, recognized as the first registry studies in this field. The A-HIT1 study aimed to identify homozygous FH (HoFH) patients undergoing lipid apheresis, evaluating their follow-up, treatment regimens, and physicians' approaches. Conversely, the A-HIT2 study focused on heterozygous FH (HeFH) patients attending outpatient clinics, assessing their diagnosis and treatment outcomes (22-24).

The A-HIT1 study included 88 HoFH patients from 19 lipid apheresis centers. Findings highlighted a lack of standardization in lipid apheresis practices across centers. The mean age of initiation for lipid apheresis was 21 ± 12 years, with a treatment frequency of 19.9 ± 14 days. Only 11.6% of patients underwent weekly apheresis. The mean diagnostic age was 12 ± 11 years, with earlier diagnosis positively influencing quality of life and emotional well-being. Early-onset

coronary artery disease (CAD) was noted in 57.8% of patients, while only 5.7% achieved target LDL-C levels (17,22,23).

The A-HIT2 study analyzed 1,071 patients (HoFH and HeFH), of whom 42.8% were on statin therapy. Despite 66% receiving intensive statin treatment, only 23 patients (2.1%) achieved LDL-C targets. FH awareness among patients was low (9.5%), with a mean diagnostic age of 47 ± 14 years. The first cardiovascular event occurred at an average age of 50 ± 10 years, suggesting most HeFH diagnoses were made post-event (17,22-24).

A national electronic health records analysis by Sonmez et al. evaluated data from 83,063,515 individuals. Probable or definite FH was identified in 0.63% of adults and 0.61% of the total population. Adults with LDL-C >4.9 mmol/L (190 mg/dL) accounted for 4.56%. The prevalence of FH in children and adolescents was 0.37%. Among adults and children with FH, lipid-lowering therapy (LLT) use was 32.1% and 1.5%, respectively, with LLT discontinuation rates of 65.8% and 77.9%. Few patients achieved LDL-C targets (25).

These findings suggest that FH prevalence in Turkey (0.61%) is approximately double the global average. However, as the study relied on records rather than direct patient evaluations, the results must be interpreted cautiously (25). Nevertheless, FH remains a significant public health issue in Turkey.

Screening programs in Turkey are limited. One study screened families of 51 pediatric HeFH patients, identifying 7 additional FH cases and initiating statin therapy for 25 individuals (26). Most patients with LDL-C >250 mg/dL, regardless of FH diagnosis, do not receive regular LLT, and only a few achieve LDL-C targets (17, 24, 27, 28).

The EPHESUS study revealed that 618 (41.7%) of 1,482 ASCVD patients had FH (10.9% probable-definite, 30.7% possible). These rates are notably high compared to other countries, though no genetic analysis was performed. FH diagnoses were based on clinical criteria in patients already under cardiology follow-up for ASCVD, likely contributing to elevated rates (28-32). Despite the high prevalence, it has been observed that treatment regimens for patients remain incomplete. Even when diagnosed, many patients exhibit misconceptions regarding their treatment. For instance, one study reported that 28% of patients vol-

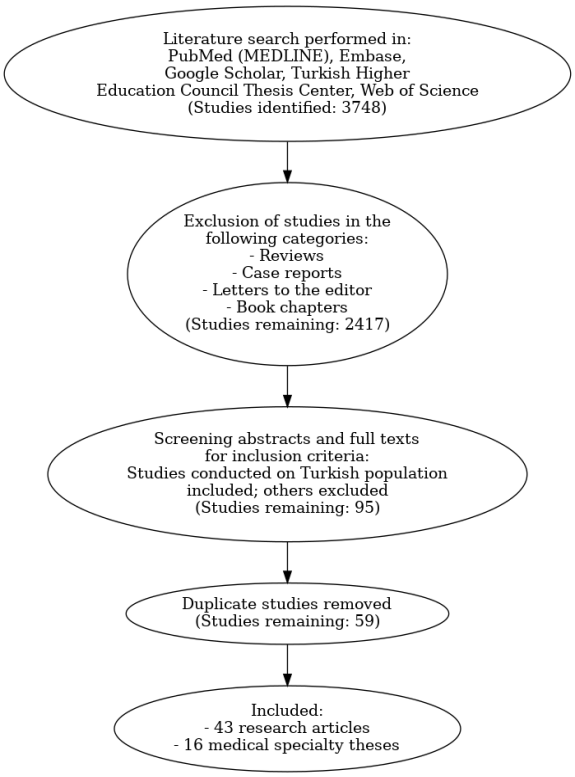


**Figure 1.** Selection process of included studies

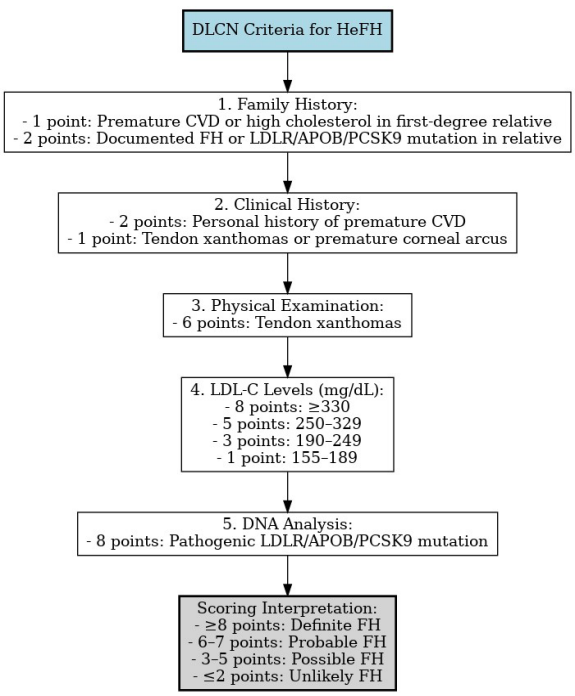


**Figure 2.** Dutch Lipid Clinic Network (DLCN) criteria for heterozygous familial hypercholesterolemia (HeFH) (12, 13)

untarily discontinued lipid-lowering therapy, 21% did not initiate any treatment, and consequently, nearly half of the patients (49%) were left untreated (30).

While no general population prevalence studies have been conducted in Turkey, analyses of hospital and ministry records report higher FH rates than in other countries. For instance, a 2019 university hospital study of 20,151 individuals identified an FH prevalence of 1.32%, significantly higher than the 0.32% rate reported in meta-analyses (32).

Other studies in Turkey primarily involve small cohorts, focusing on biochemical parameters, demographics, or treatment outcomes in diagnosed patients (33-64). The majority of these studies have focused on cardiac functions and clinical findings, genetic analyses, LDL apheresis, statin therapy, the efficacy of PCSK9 inhibitors, and the attitudes and behaviors of patients regarding the disease, primarily in small cohorts of individuals with a prior diagnosis. To date, no large-scale studies have comprehensively estimated the FH burden in Turkey.

## DISCUSSION

Familial hypercholesterolemia (FH) is one of the most prevalent genetic disorders, effecting a significant portion of the population. Elevated LDL-C levels present from birth contribute to premature atherosclerosis, cardiovascular disease (CVD), and early mortality. Although genetic testing provides a definitive diagnosis, due to its high cost and limited availability, it is recommended that cases be evaluated or referred to specialized centers for familial hypercholesterolemia (FH) based on non-genetic diagnostic criteria or even an isolated LDL-C level exceeding 190 mg/dL (65). Consequently, early diagnosis and treatment are crucial to preventing these adverse outcomes (5-7).

Community-based screening is among the most effective strategies for early diagnosis. However, such programs are implemented in only a few countries globally. In many nations, including Turkey, most patients remain undiagnosed, and even among those diagnosed, achieving treatment targets is uncommon (7-9). In a study including data from Turkey and many countries, only 32% of patients diagnosed with familial hypercholesterolemia (FH) achieved their target cholesterol levels. Statin therapies were frequently prescribed at low doses, and among those failing to reach treatment goals, only 13% were escalated to combination therapy with second-line agents (66).

In Turkey, the lack of comprehensive studies on FH presents a challenge. This compilation reviewed 43 research articles and 16 theses focusing on FH in Turkey. Notably, no studies have systematically assessed the prevalence and societal burden of FH through community-based screening. Although the study by Sonmez et al., utilizing Ministry of Health records, provides some insights, it lacks the precision required to draw definitive conclusions (25).

Both the study by Sonmez et al. and other large-scale investigations suggest a higher prevalence of FH in Turkey compared to global averages. For instance, FH prevalence in the general population is estimated at 0.61–1.32% in Turkey, approximately two to three times the global average. Furthermore, among individuals diagnosed with CVD, FH prevalence in Turkey is reported at 10.9%, significantly exceeding the global average of 3.2% (25-28, 32). While these findings may be influenced by the limitations of the studies, they underscore the urgency of addressing FH in Turkey. When compared to Mediterranean countries with similar dietary patterns and climatic conditions, such as Italy and Greece, such high prevalence rates have not been observed. This suggests that factors beyond diet, climate, or lifestyle habits may be contributing to these findings (14, 67). The relatively high incidence of consanguineous marriages further highlights the need for large-scale prevalence studies to provide more accurate data and clarify the true burden of FH in the population.

## CONCLUSION

FH represents a substantial public health concern, contributing to premature cardiovascular events and mortality worldwide. As with many other countries, Turkey lacks sufficient data on the societal burden and prevalence of FH. The markedly higher FH prevalence reported in Turkish studies compared to global data underscores the need for robust epidemiological research. Large-scale studies are essential to better understand the extent of FH in Turkey and to develop effective public health strategies to address this issue.

## Limitations

This narrative review has several limitations, including the scarcity of large-scale studies in Turkey and reliance on predominantly small-scale data from the available literature on familial hypercholesterolemia. As a non-systematic analysis, it may not capture the full spectrum of evidence, potentially affecting the generalizability of conclusions.

## Conflict-of-interest and financial disclosure

The authors declare that they have no conflict of interest to disclose. The authors also declare that they did not receive any financial support for the study.

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