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ARAŞTIRMA MAKALESİ / RESEARCH ARTICLE

Awareness of Inherited Metabolic Diseases Among Cardiologists: A Cross Sectional Survey

Kardiyologlar Arasında Kalıtsal Metabolik Hastalıklara Yönelik Farkındalık: Kesitsel Bir Araştırma

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ABSTRACT

Introduction: Inherited metabolic diseases (IMDs) are rare monogenic disorders with multisystem involvement, and cardiac manifestations including hypertrophic or dilated cardiomyopathy, arrhythmias, and conduction abnormalities may present early or predominate clinically. Early recognition is essential, as disease specific treatments in selected IMDs (e.g., Fabry and Pompe disease) are most effective before irreversible myocardial involvement. Although awareness of metabolic etiologies in cardiology practice is increasing, data on cardiologists' perspectives and diagnostic approaches remain limited. This study evaluated awareness of IMDs among cardiologists in Türkiye.

Material and Methods: A structured questionnaire was administered to 220 cardiologists across multiple healthcare settings. Survey domains included demographics, self perceived knowledge, clinical exposure, recognition of key electrocardiographic, echocardiographic, and physical findings, and diagnostic reasoning through clinical vignettes.

Results: Among the participants, self reported familiarity with IMDs varied: 54,5% indicated basic familiarity, 36,4% moderate familiarity, and 9,1% high familiarity. Most participants reported infrequent direct encounters with IMD patients (72,7%), which is expected given the rarity of these disorders in cardiology practice. Recognition of characteristic cardiac and systemic features associated with IMDs was widely acknowledged. Case based diagnostic performance differed across conditions (mitochondrial disease 75%, Fabry disease 60%, homocystinuria 50%, Pompe disease 45%), reflecting variation in diagnostic attribution between disorders.

Conclusion: Cardiologists demonstrated awareness of characteristic cardiac features associated with IMDs, reflecting familiarity with key clinical phenotypes. Variability in vignette-based diagnostic accuracy likely relates to the rarity and clinical heterogeneity of IMDs rather than clinical deficiency. Continued multidisciplinary collaboration, focused diagnostic pathways, and access to targeted educational resources may support earlier recognition and timely referral when metabolic etiologies are suspected.

Keywords: Inherited metabolic diseases, cardiomyopathy, rare diseases, clinician awareness, multisystem disorders

ÖZ

Giriş: Kalıtsal metabolik hastalıklar (KMH), multisistem tutulumu olan nadir monogenik hastalıklardır ve hipertrofik veya dilate kardiyomyopati, aritmiler ve ileti sistemi bozuklukları gibi kardiyak bulgular erken dönemde ortaya çıkabilir veya klinik tabloya hâkim olabilir. Seçilmiş bazı KMH'lerde (örn. Fabry ve Pompe hastalığı) hastalığa özgü tedaviler, geri dönüşümsüz myokardiyal tutulum gelişmeden önce başlandığında en etkili sonuçları vermektedir. Kardiyoloji pratiğinde metabolik etiyolojilere yönelik farkındalık artmakla birlikte, kardiyologların bakış açıları ve tanısal yaklaşımlarına ilişkin veriler sınırlıdır. Bu çalışma, Türkiye'deki kardiyologlar arasında kalıtsal metabolik hastalıklara yönelik farkındalığı değerlendirmeyi amaçlamıştır.

Materyal ve Metodlar: Çoklu sağlık kuruluşlarında görev yapan 220 kardiyoloğa yapılandırılmış bir anket uygulanmıştır. Anket; demografik özellikler, öznel bilgi düzeyi algısı, klinik deneyim, temel elektrokardiyografik, ekokardiyografik ve fizik muayene bulgularının tanınması ile klinik senaryolar aracılığıyla tanısal akıl yürütme alanlarını kapsamıştır.

Bulgular: Katılımcıların KMH'lere ilişkin öz-bildirimli aşinalık düzeyi değişkenlik göstermiştir: %54,5'i temel düzeyde, %36,4'ü orta düzeyde ve %9,1'i ileri düzeyde bilgiye sahip olduğunu belirtmiştir. Katılımcıların çoğu (%72,7) klinik uygulamalarında KMH hastalarıyla nadiren karşılaştığını bildirmiştir; bu durum hastalıkların nadir görülmesiyle uyumludur. KMH'lerle ilişkili karakteristik kardiyak ve sistemik bulguların tanınma düzeyinin genel olarak yüksek olduğu görülmüştür. Olgu temelli tanısal performans hastalıklar arasında farklılık göstermiştir (mitokondriyal hastalık %75, Fabry hastalığı %60, homosistinüri %50, Pompe hastalığı %45).

Sonuç: Kardiyologların, kalıtsal metabolik hastalıklarla ilişkili karakteristik kardiyak bulgular konusunda farkındalık gösterdiği ve temel klinik fenotiplere aşinalıklarının bulunduğu görülmüştür. Olgu senaryolarına dayalı tanısal doğrulukta gözlenen değişkenliğin, klinik yetersizlikten ziyade KMH'lerin nadir görülmesi ve klinik heterojenitesi ile ilişkili olduğu düşünülmektedir. Multidisipliner iş birliğinin sürdürülmesi, odaklanmış tanısal yaklaşımlar ve hedeflenmiş eğitim kaynaklarına erişim, metabolik etiyolojiden şüphelenilen olgularda erken tanı ve zamanında yönlendirmeyi destekleyebilir.

Anahtar Sözcükler: Kalıtsal metabolik hastalıklar, kardiyomyopati, nadir hastalıklar, klinisyen farkındalığı, multisistem hastalıkları

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Introduction

Inherited metabolic diseases (IMDs) are predominantly monogenic disorders, most commonly inherited in an autosomal recessive manner, and arise from partial or complete deficiency of specific enzymes, transporters, or cofactors. These defects lead to accumulation of toxic metabolites, substrate deficiency, impaired mitochondrial function, or energy failure, resulting in multisystem involvement from birth onward. Although classically recognized in infancy, a growing proportion of IMDs are now diagnosed in adolescence and adulthood due to milder phenotypes, improved survival following newborn screening, and increasing access to advanced genomic testing (1–3).

IMDs demonstrate a broad clinical spectrum. Common systemic manifestations include developmental delay, seizures or movement disorders, myopathy or exercise intolerance, hepatic dysfunction, renal involvement, cardiac involvement, endocrine abnormalities, and ophthalmologic findings. Adult patients may present with organ predominant disease, and early symptoms are frequently subtle, leading to delayed diagnosis. Within this spectrum, the cardiovascular system represents one of the key target organ in numerous IMDs. Metabolic derangements can lead to myocardial storage, mitochondrial energetic failure, or connective tissue abnormalities, producing hypertrophic or dilated cardiomyopathy, conduction disease, arrhythmias, valvular pathology, heart failure, or vascular complications. Fabry disease is a well characterized cause of unexplained concentric left ventricular hypertrophy, conduction abnormalities, and myocardial fibrosis; late onset Pompe disease may present with hypertrophic cardiomyopathy and exertional intolerance; and mitochondrial disorders can manifest with dilated cardiomyopathy, atrioventricular block, or ventricular arrhythmias. Homocystinuria, a treatable IMD often included in thrombophilia evaluations, may cause premature arterial and venous thrombosis, valvular abnormalities, and accelerated atherosclerosis in young adults (1–3).

Early recognition has important therapeutic implications, particularly in IMDs where intervention prior to irreversible myocardial remodeling improves outcomes. In Fabry and Pompe disease, disease specific therapies are most effective prior to irreversible myocardial involvement. Even in IMDs without specific pharmacologic therapy, accurate diagnosis enables targeted monitoring, avoidance of harmful medications, and cascade family screening (3–5).

Although IMDs are increasingly recognized in adults, diagnosis in cardiology practice may be challenging, especially when cardiac findings predominate. This underscores the need for continued awareness and multidisciplinary evaluation in patients with unexplained cardiomyopathy or conduction abnormalities.

To date, no systematic assessment has examined IMD awareness among cardiologists in Türkiye. Therefore, this study aimed to evaluate knowledge, recognition patterns, and diagnostic performance regarding IMDs among adult cardiologists using a structured national survey. Improved insight into current awareness may guide educational strategies, strengthen multidisciplinary collaboration, and ultimately facilitate earlier recognition and better outcomes for patients with metabolic cardiomyopathies.

Material and Methods

A cross sectional survey was conducted among 220 cardiologists in Türkiye. The questionnaire was administered electronically between January and March 2025, and participation was voluntary and anonymous (Table 1). Both cardiology residents and specialist cardiologists participated in the survey, and only one response per physician was permitted. The survey assessed demographic characteristics (age, sex, years of practice, and institution type), followed by questions evaluating self reported knowledge and clinical exposure to IMDs. Participants were asked about their familiarity with cardiovascular involvement in IMDs and their ability to recognize characteristic electrocardiographic, echocardiographic, and physical findings. Diagnostic performance was evaluated using four case based clinical vignettes representing Fabry disease, Pompe disease, mitochondrial disease, and homocystinuria. Descriptive statistical analyses were conducted to summarize demographic and survey response distributions. Categorical variables were expressed as frequencies and percentages, while Likert scale responses were reported as mean ± standard deviation. The study was planned in accordance with the principles of the Declaration of Helsinki and informed consent was obtained, and the study was carried out after approval from the Etlik City Hospital Ethics Commission (No. 2025-127)

Results

Among 220 cardiologists, 50% were female and 50% male; 50% were specialists and 50% residents. More than half (54,5%) had ≤5 years of practice experience. With respect to institutional distribution, 45,5% worked in training and research hospitals, 18,2% in university hospitals, 18,2% in private hospitals, and 13,6% in state hospitals. The demographic characteristics of participants are summarized in Table 2.

Self-reported familiarity with IMDs varied among participants: 54,5% indicated basic familiarity, 36,4% moderate familiarity, and 9,1% high familiarity. Most participants (72,7%) reported infrequent direct encounters with IMD patients in daily practice. Details on knowledge and clinical exposure are provided in Table 3.

Recognition of specific cardiovascular and systemic manifestations was considerably higher than overall disease awareness. As shown in Table 4, participants most frequently identified hypertrophic cardiomyopathy (95,5%), conduction

Table 1. Cardiologists' awareness of inherited metabolic diseases survey

SECTION 1: DEMOGRAPHIC INFORMATION
1. Age: ( )
2. Sex:
( ) Female ( ) Male ( ) Prefer not to say
3. Professional status:
( ) Cardiologist ( ) Cardiology Resident
4. Years of experience in cardiology:
( ) 0–5 years ( ) >5 years
5. Type of institution:
( ) University Hospital ( ) Training and Research Hospital ( ) Private Hospital ( ) State Hospital ( ) Other (please specify):
SECTION 2: KNOWLEDGE ABOUT INHERITED METABOLIC DISEASES
6. Do you think you have knowledge about inherited metabolic diseases?
( ) No knowledge ( ) Very limited knowledge ( ) Moderate knowledge ( ) Good knowledge ( ) Expert-level knowledge
7. Have you encountered inherited metabolic diseases in your cardiology practice?
( ) Yes, frequently ( ) Yes, rarely ( ) No, never ( ) Not sure
8. Do you know the relationship between inherited metabolic diseases and cardiovascular diseases?
( ) No ( ) Yes, I know some conditions ( ) Yes, I have detailed knowledge
SECTION 3: CLINICAL FINDINGS
9. Which of the following are ECG findings of inherited metabolic diseases? (Multiple answers allowed)
( ) QT prolongation ( ) Ventricular arrhythmias ( ) Conduction defects (SA, AV block etc.) ( ) ST-T wave abnormalities ( ) Bradyarrhythmias ( ) Other (please specify):
10. Which of the following are echocardiographic findings of inherited metabolic diseases? (Multiple answers allowed)
( ) Hypertrophic cardiomyopathy ( ) Dilated cardiomyopathy ( ) Pericardial effusion ( ) Cardiac fibrosis ( ) Other (please specify):
11. Which of the following are physical examination findings of inherited metabolic diseases? (Multiple answers allowed)
( ) Short stature ( ) Clubbing ( ) Eye anomalies (e.g., lens dislocation) ( ) Muscle weakness ( ) Hepatomegaly ( ) Other (please specify):
SECTION 4: CASE-BASED QUESTIONS
Case 1. A 35-year-old male patient presents with progressively worsening difficulty climbing stairs, exercise intolerance, and shortness of breath over the past few years. Physical examination reveals proximal muscle weakness, particularly in the hip and shoulder girdles. Creatine kinase (CK) is elevated. Echocardiography shows no significant cardiomyopathy.
Which of the following is the most likely diagnosis in this patient? • ( ) Fabry disease • ( ) Pompe disease • ( ) Glycogen storage disease • ( ) Mitochondrial disease • ( ) Marfan syndrome

Table 1 (continues).. Cardiologists' awareness of inherited metabolic diseases survey

Case 2. A 42-year-old female patient presents with chest pain, palpitations, and dizziness. Her medical history includes a previous ischemic stroke at a young age. There is a family history of early-onset thromboembolic and cardiovascular disease. It is learned that she previously underwent surgical treatment for lens dislocation. On physical examination, she has a tall, slender (marfanoid) habitus and mild scoliosis.
Which of the following is the most likely diagnosis in this patient? • ( ) Homocystinuria • ( ) Fabry disease • ( ) Marfan syndrome • ( ) Mitochondrial myopathy • ( ) Phenylketonuria
Case 3. A 30-year-old male patient presents with chronic muscle weakness, exercise intolerance, and palpitations. Family history reveals early-onset heart failure. Echocardiography shows dilated cardiomyopathy. Lactate level is elevated mildly, and metabolic acidosis is observed on blood gas analysis.
Which of the following is the most likely diagnosis in this patient? • ( ) Mitochondrial disease • ( ) Glycogen storage disease • ( ) Pompe disease • ( ) Fabry disease • ( ) Homocystinuria
Case 4. A 25-year-old male patient presents with burning extremity pain, palpitations worsened by exercise, and dizziness. Physical examination reveals angiokeratomas on the skin. Echocardiography shows left ventricular hypertrophy.
Which of the following is the most likely diagnosis in this patient? • ( ) Fabry disease • ( ) Marfan syndrome • ( ) Mitochondrial disease • ( ) Homocystinuria • ( ) Glycogen storage disease

defects (95,5%), QT prolongation (95,5%), hepatomegaly (95,5%), and muscle weakness (95,5%) as relevant findings. However, recognition of less common features, such as cardiac fibrosis and digital clubbing, was notably lower.

When assessed through case based vignettes, diagnostic accuracy was suboptimal. Correct identification rates were 75% for mitochondrial disease, 60% for Fabry disease, 50% for homocystinuria, and only 45% for Pompe disease. These results are detailed in Table 5. Illustrating that even though individual cardiac features were well recognized, integration into accurate etiologic diagnosis remained insufficient.

Table 2. Demographic characteristics of participants

Variable	n (%)
Age 25–30	100 (45,5)
Age 31–35	60 (27,3)
Age ≥35	60 (27,3)
Female	110 (50,0)
Male	110 (50,0)
Resident	110 (50,0)
Specialist	110 (50,0)
≤5 years practice	120 (54,5)
>5 years practice	100 (45,5)
University hospital	40 (18,2)
Training & research hospital	100 (45,5)
Private hospital	40 (18,2)
State hospital	30 (13,6)
Other	10 (4,5)

Table 3. Knowledge and experience regarding inherited metabolic diseases

Variable	n (%)
Very little knowledge	120 (54,5)
Moderate knowledge	80 (36,4)
Good knowledge	20 (9,1)
Frequently encounter IMDs	20 (9,1)
Rarely encounter IMDs	160 (72,7)
Never encountered IMDs	30 (13,6)
Unsure	10 (4,5)
No knowledge of CV associations	10 (4,5)
Some knowledge of CV associations	200 (90,9)
Detailed knowledge	10 (4,5)

Table 4. Recognition of cardiac findings associated with IMDs

Category	Finding	n (%)
ECG	QT prolongation	210 (95,5)
ECG	Conduction defects	210 (95,5)
ECG	Ventricular arrhythmias	180 (81,8)
ECG	Bradyarrhythmias	170 (77,3)
ECG	ST–T wave changes	160 (72,7)
ECHO	Hypertrophic cardiomyopathy	210 (95,5)
ECHO	Dilated cardiomyopathy	200 (90,9)
ECHO	Pericardial effusion	190 (86,4)
ECHO	Cardiac fibrosis	20 (9,1)
Physical	Hepatomegaly	210 (95,5)
Physical	Muscle weakness	210 (95,5)
Physical	Ocular anomalies	200 (90,9)
Physical	Short stature	180 (81,8)
Physical	Clubbing	130 (59,1)

Table 5. Diagnostic accuracy in case vignettes

Case vignette	Correct diagnosis (%)
Case 1. Pompe disease	45
Case 2. Homocystinuria	50
Case 3. Mitochondrial disease	75
Case 4. Fabry disease	60

Discussion

This study contributes to the growing body of evidence regarding the cardiovascular dimension of IMDs by examining cardiologists' perspectives in routine clinical practice. Overall, participants demonstrated substantial recognition of key cardiac phenotypes frequently associated with IMDs, including hypertrophic cardiomyopathy and conduction abnormalities. These findings are consistent with current clinical observations suggesting that cardiologists increasingly encounter genetic and metabolic etiologies in patients presenting with unexplained cardiomyopathy or arrhythmia patterns (1,6,7). Rather than reflecting a deficiency in clinical knowledge, the variability observed in case-based diagnostic performance likely reflects the intrinsic clinical heterogeneity and multisystemic nature of IMDs, which often require integration of metabolic, neurological, and genetic findings for accurate diagnosis.

In this cohort, Fabry disease and mitochondrial disorders were more frequently recognized compared to other metabolic etiologies. This observation is in line with increasing awareness of Fabry related cardiac involvement in cardiology practice, particularly in the context of unexplained left ventricular hypertrophy, conduction abnormalities, and characteristic electrocardiographic findings (2,3,8). Advances in disease specific therapies and expert consensus recommendations have further emphasized the importance of early recognition, especially prior to irreversible myocardial fibrosis and dysfunction (3,8).

Mitochondrial diseases represent another important group of metabolic disorders with significant cardiac involvement. Previous studies have demonstrated that mitochondrial cardiomyopathy may present with hypertrophic or dilated phenotypes, arrhythmias, and conduction defects, often requiring a multidisciplinary diagnostic approach (4,5). The variability in recognition rates observed in our case-based assessments likely reflects the wide phenotypic spectrum and variable age of onset associated with these disorders. These findings highlight the importance of considering metabolic etiologies in patients with cardiomyopathy accompanied by neuromuscular symptoms, elevated lactate levels, or suggestive family history.

Recognition of Pompe disease and homocystinuria was comparatively lower, which may reflect their lower encounter frequency in routine cardiology practice and their broader systemic manifestations beyond isolated cardiac involvement. Pompe disease can present across a clinical continuum, and delayed diagnosis may limit timely access to enzyme replacement therapy and specialized care (9). Similarly, homocystinuria remains an important but potentially underrecognized cause of premature vascular disease, thromboembolic events, and multisystem involvement. Established international guidelines emphasize the importance of early diagnosis and appropriate metabolic evaluation in these patients (10).

Taken together, our findings suggest that cardiologists demonstrate a solid foundational awareness of IMD related cardiac phenotypes. However, integration of these findings into accurate etiological diagnosis remains challenging, reflecting the rarity and complexity of these disorders. Rather than indicating a gap in clinical competence, this underscores the importance of structured diagnostic pathways, interdisciplinary collaboration, and access to targeted educational resources.

In clinical practice, specific "red flags" may facilitate earlier recognition and referral for metabolic evaluation. These include unexplained left ventricular hypertrophy, early-onset conduction abnormalities, arrhythmias associated with neuromuscular symptoms, multisystem involvement, and positive family history. Incorporating such criteria into routine cardiology assessment may improve early detection of IMDs and optimize patient outcomes.

Future studies involving larger, multicenter cohorts and prospective designs may further clarify diagnostic patterns and identify effective strategies to support cardiologists in recognizing metabolic etiologies. Educational interventions, integration of metabolic screening algorithms, and strengthening collaboration between cardiology and metabolic specialists are likely to play a key role in improving diagnostic accuracy and patient care pathways.

Strengths and Limitations

Strengths of this study include its nationwide scope, participation of both specialist and resident cardiologists, and the incorporation of case-based clinical vignettes. The study relied on self reported familiarity for knowledge assessment and employed a cross sectional design, which provides a snapshot of current practice patterns without evaluating changes over time.

Conclusion

Cardiologists in Türkiye demonstrate recognition of characteristic cardiac manifestations associated with IMDs. Variation in case-based diagnostic attribution reflects the clinical heterogeneity and rarity of IMDs in routine cardiology practice. Strengthening structured evaluation pathways, guideline implementation, and multidisciplinary collaboration may facilitate timely recognition and referral.

Ethical Considerations: This study was approved by the Etlik City Hospital Ethics Committee (2025-127).

Peer-review: Externally peer-reviewed.

Conflict of Interest: No conflict of interest was declared by the authors.

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Consent of Patients: The participants were informed in detail, and informed consent was obtained.

Data Availability Statement: All relevant data are within the paper and they are available from the corresponding author on reasonable request.

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