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HYPERTROPHIC CARDIOMYOPATHY IN ATHLETES - A LITERATURE REVIEW

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ABSTRACT

Background: Hypertrophic cardiomyopathy (HCM) is one of the most common hereditary heart muscle disorders. It is characterised by unexplained left ventricular hypertrophy (LVH). In competitive sports, HCM can be particularly dangerous. It is a well-recognised leading cause of sudden cardiac death (SCD) among young athletes.
Aim: This study aimed to provide a comprehensive review of the current understanding of HCM within athletic populations, with a focus on the differences in pathophysiology compared to normal physiological adaptation.
Material and methods: Between April and July 2025, we searched the PubMed, Google Scholar, and PMC databases. We used both Medical Subject Headings (MeSH) and keywords in the free text. The search was limited to studies on humans. Due to the heterogeneity of the materials used, no statistical analyses or quantitative pooling were performed. The narrative was based on PRISMA principles.
Research results: HCM is a genetic disorder caused by mutations in sarcomere-affecting genes. These abnormalities result in abnormal energy metabolism, disturbances in calcium homeostasis and oxidative stress. This generates an energy deficit, which is exacerbated further during competitive sports. This mechanism can lead to life-threatening complications. Distinguishing between the heart's physiological adaptation to intense physical exertion and HCM requires an assessment of ventricular geometry, diastolic and systolic performance, coronary perfusion and the body's response to exercise testing.
Conclusion: HCM presents many challenges to sports medicine and diagnosis requires advanced tests. Thanks to extensive research on HCM, the range of therapeutic options in both pharmacotherapy and surgical treatment is constantly expanding.

KEYWORDS

Competitive Sport, Hypertrophic Cardiomyopathy, Sudden Cardiac Death, Athlete's Heart, Ventricular Hypertrophy

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Introduction.

Hypertrophic cardiomyopathy (HCM) stands out as one of the most prevalent inherited heart muscle disorders, characterized by unexplained left ventricular hypertrophy (LVH). This condition is typically defined by a maximal wall thickness of 15 mm or greater, occurring without obvious loading conditions such as high blood pressure or valve disease [1]. Current epidemiological data suggest that HCM affects approximately 1 in 500 people, though when we include individuals who carry the genetic mutation but haven't yet developed visible signs of the disease, the prevalence may be as high as 1 in 200 [2].

Within the world of competitive sports, HCM takes on particularly concerning significance. It has long been recognized as the primary underlying cause of sudden cardiac death (SCD) among young athletes [1][2]. Studies examining autopsy-confirmed cases reveal that HCM accounts for nearly 40% of sudden cardiac deaths in athletes, with the vast majority of these individuals having no symptoms prior to their fatal event [3]. Certain demographic groups, particularly young male and African American athletes, appear to face disproportionately higher risks [4].

One of the most significant challenges facing sports for cardiologists is distinguishing between pathological LVH and the normal heart muscle thickening that results from intensive athletic training, commonly referred to as "athlete's heart". Athletes diagnosed with HCM often present less severe LVH, larger heart chamber dimensions, and better preserved filling function compared to inactive HCM patients. This overlap makes diagnosis particularly difficult using echocardiography alone, especially when wall thickness falls into the "grey zone" between 13 and 16 mm [3][5]. Research on young athletes with HCM has shown that fewer than 15% display the concentric hypertrophy pattern that typically mimics normal physiological adaptation, highlighting how traditional heart function measurements alone are insufficient for accurate diagnosis [6].

Clinical decision-making regarding sports participation for athletes with HCM has traditionally favored caution, with most individuals being excluded from competitive activities. However, growing evidence suggests that moderate intensity exercise may actually improve functional capacity without significantly increasing adverse outcomes. Additionally, participation among athletes with implantable cardioverter defibrillators (ICDs) appears safer than previously thought [7]. In response to this evolving understanding, contemporary guidelines from both American Heart Association (AHA), American College of Cardiology (ACC) and European Society of Cardiology (ESC) have adopted more flexible, shared decision making approaches that may allow carefully selected individuals with HCM to engage in physical activity under supervised conditions [8].

This review aims to synthesize our current understanding of HCM within athletic populations, focusing on the pathophysiological differences from normal physiological adaptation, diagnostic challenges, epidemiological considerations, risk stratification strategies, and evolving management paradigms.

Material and Methods

Between April and July 2025, we performed a comprehensive research of the PubMed, PMC, and Google Scholar databases, using both MeSH (Medical Subject Headings) and free-text keywords (e.g., ‘hypertrophic cardiomyopathy,’ ‘left ventricular hypertrophy,’ ‘sudden cardiac death,’ ‘athlete's heart,’ ‘competitive sports’). The search was limited to human studies. Animal studies and in vitro studies were excluded. Meta-analyses, systematic reviews, randomised trials, non-randomised interventions, cohort studies, cross-sectional studies and case reports were included. Due to the heterogeneity of the used materials, no statistical analyses or quantitative synthesis were performed. A structured narrative synthesis based on PRISMA principles was presented.

AI was used to help refine the academic English of the manuscript, ensuring clarity, consistency, grammatical correctness and compliance with scientific writing standards. All AI instruments were used strictly as assistance tools under human supervision.

Research Results

The Molecular Basis of Hypertrophic Cardiomyopathy

HCM primarily follows an autosomal dominant inheritance pattern, caused by mutations in genes which code the components of the cardiac sarcomere, the basic contractile unit of heart muscle cells. The most frequently affected genes include: MYH7 (which encodes β-myosin heavy chain), MYBPC3 (encoding cardiac myosin-binding protein C), TNNT2 (encoding cardiac troponin T), and TNNI3 (encoding cardiac troponin I). Together, these genes are responsible for approximately 60-70% of HCM cases, with MYH7 and MYBPC3 mutations being the most common. Scientists have identified over 1,400 distinct mutations, predominantly missense mutations that result in the production of dysfunctional proteins that get incorporated into the sarcomere [9]. These mutations disrupt normal sarcomeric function by altering force generation, calcium sensitivity, or contractile efficiency. For example, MYH7 mutations often lead to increased ATPase activity, resulting in heightened energy demands, while MYBPC3 mutations may reducing the availability of functional myosin-binding protein C. Less commonly, mutations in non-sarcomeric genes, such as those involved in mitochondrial function (MT-TI, MT-TG) or lysosomal storage (LAMP2, PRKAG2), contribute to HCM-like phenotypes, particularly in metabolic forms of cardiomyopathy [10].

The genetic mutations underlying HCM have profound effects on heart muscle cell metabolism, particularly mitochondrial function. Mitochondria play a crucial role in meeting the high energy demands of cardiac muscle by producing ATP through oxidative phosphorylation. In HCM, sarcomeric mutations disrupt the delicate balance between energy supply and demand, leading to impaired mitochondrial energy production [11]. Sarcomeric mutations increase the energy cost of muscle contraction due to inefficient cross-bridge cycling or heightened calcium sensitivity. This creates a state of chronic energy deprivation, often described as an "energy-starved" heart muscle. Studies have demonstrated reduced phosphocreatine-to-ATP ratios in HCM hearts, indicating compromised energy reserves. Mitochondrial dysfunction becomes further exacerbated by structural abnormalities and impaired activity of electron transport chain complexes, particularly complexes I and IV [12][13]. In cases where HCM results from mutations in mitochondrial DNA (mtDNA), such as MT-TI or MT-TG, the impact on mitochondrial function is direct. These mutations impair mitochondrial protein synthesis, reducing the efficiency of oxidative phosphorylation and leading to ATP depletion. This becomes particularly relevant in athletes, where increased cardiac workload amplifies the energy deficit, potentially contributing to disease progression [13].

Mitochondrial dysfunction in HCM is closely linked to increased oxidative stress. Impaired electron transport chain function leads to excessive production of reactive oxygen species (ROS), which overwhelm cellular antioxidant defenses. Elevated ROS levels cause oxidative damage to mitochondrial DNA, proteins, and lipids, further impairing mitochondrial function in a destructive cycle. Oxidative stress also activates pro-hypertrophic signaling pathways, such as the calcineurin-NFAT pathway, promoting heart muscle hypertrophy and scarring [14]. Moreover, ROS-mediated damage contributes to inflammation and programmed cell death in heart muscle cells, exacerbating the characteristic muscle fiber disorder seen in HCM. Studies have demonstrated elevated markers of oxidative stress, such as malondialdehyde and 8-hydroxydeoxyguanosine, in HCM patients, with levels correlating with disease severity. In athletes, the high metabolic demands of exercise may amplify oxidative stress, potentially accelerating disease progression and increasing the risk of dangerous heart rhythms or sudden cardiac death [14][15].

The complex interact between genetic mutations, mitochondrial dysfunction, and oxidative stress activates several molecular pathways that drive HCM development. For instance, increased energy demand and reduced ATP availability activate AMP-activated protein kinase (AMPK), which attempts to restore energy balance but may also promote maladaptive hypertrophy. Similarly, oxidative stress-induced activation of mitogen-activated protein kinases (MAPKs) and nuclear factor kappa B (NF-κB) contributes to inflammation and scarring [16].

Calcium mishandling represents another critical consequence of HCM mutations. Sarcomeric mutations often increase myofilament calcium sensitivity, leading to prolonged calcium transients and elevated intracellular calcium levels. This disrupts mitochondrial calcium uptake, further impairing ATP production and increasing ROS generation. These metabolic and ionic disturbances contribute to diastolic dysfunction a hallmark of HCM and predispose individuals to dangerous heart rhythms [11][17][18].

In athletes with HCM, the metabolic and oxidative stress-related consequences of genetic mutations become particularly significant. Intense physical activity increases heart muscle energy demand and ROS production, potentially worsening mitochondrial dysfunction and oxidative damage. This may accelerate disease progression, particularly in individuals with high-risk mutations or advanced hypertrophy. The

combination of metabolic stress and structural abnormalities increases the risk of dangerous heart rhythms, making pre-participation screening and genetic counseling critical for this population [18].

Comparison of Cardiac Mechanics and Hemodynamics: healthy hearts and hearts with hypertrophic cardiomyopathy

The physiological LVH observed in an healthy athlete's heart represents a beneficial adaptation to the chronic volume and pressure loads imposed by intense physical training. This remodeling is characterized by a proportionate increase in both ventricular wall thickness and size, often termed balanced eccentric hypertrophy in endurance athletes. This adaptation allows for increased stroke volume (SV), which combined with a training-induced decrease in resting heart rate, maintains or increases cardiac output while reducing myocardial oxygen consumption per beat [19]-[22]. A characteristic feature of the healthy athlete's heart is preserved and often enhanced, diastolic function. The ventricles remain highly compliant and capable of rapid and complete filling. This is reflected in normal or elevated early diastolic filling velocity (E) and a low ratio of early diastolic mitral inflow velocity to mitral annular velocity (E/e′), indicating low LV filling pressures. The increased compliance and rapid relaxation allow for greater end-diastolic volume (EDV), which, through the Frank-Starling mechanism, prepare the ventricle for more forceful contraction and larger SV [19][20]. Systolic function typically remains normal or becomes increased. The hypertrophied muscle generates strong, effective contractions, leading to normal or increased ejection fraction (EF). The systolic work primarily focuses on generating high SV with low afterload, as systemic vascular resistance is often reduced in well-trained athletes [19][21]. The growth of heart muscle in the athlete's heart is accompanied by a proportionate increase in capillary density and coronary blood flow. This ensures that the increased metabolic demand of the hypertrophied muscle is met, preventing ischemia even during peak exertion [19][22].

In contrast, HCM represents a genetic disorder of the sarcomere, leading to chaotic heart muscle cell disorder, interstitial fibrosis, and asymmetrical LVH, most commonly affecting the interventricular septum. These structural abnormalities result in a profoundly different and pathological hemodynamic profile [23][24][25][26]. Diastolic dysfunction is the most prevalent and earliest hemodynamic abnormality in HCM. The hypertrophied, stiff, and fibrotic heart muscle has reduced compliance, resisting ventricular filling. This leads to decreased EDV and elevated LV end-diastolic pressure (LVEDP), which can deteriorate during exercise. The impaired relaxation and reduced compliance result in a restrictive filling pattern, characterized by blunted early diastolic filling velocity (E) and high E/e′ ratio, signifying increased LV filling pressures and a tendency toward exertional shortness of breath [23][24]. While overall EF may appear normal or even increased at rest, the hypertrophied septum can impede blood flow from the left ventricle into the aorta This phenomenon is known as left ventricular outflow tract obstruction (LVOTO). This obstruction creates a pressure gradient that increases afterload, reducing effective forward SV and increasing myocardial wall tension. The systolic work becomes inefficient, as a significant portion of the contraction force is wasted on overcoming the obstruction rather than propelling blood forward. The characteristic "dagger-shaped" Doppler signal serves as a classic sign of this dynamic obstruction [23][24][25]. Despite the absence of obstructive coronary artery disease, patients with HCM are highly susceptible to heart muscle ischemia. The hypertrophy and muscle cell disarray lead to a mismatch between myocardial oxygen supply and demand. The small-caliber intramural coronary arteries become compressed by the thickened muscle, particularly during systole, further limiting blood flow. The increased LVEDP also reduces the pressure gradient for coronary perfusion, which predominantly occurs during diastole. This chronic ischemia can contribute to ongoing fibrosis and heightened risk of dangerous heart rhythms and sudden cardiac death, particularly during exertion [23][26].

Table. 1 The fundamental differences between the athlete's heart and the HCM heart

Parameter	Healthy Athlete's Heart	Hypertrophic Cardiomyopathic Heart (HCM)
Ventricular Geometry	Symmetric, proportionate hypertrophy with preserved or increased chamber dimensions	Asymmetric hypertrophy with normal or reduced chamber size and muscle cell disarray
Diastolic Performance	Superior diastolic function with enhanced compliance and rapid relaxation	Progressive diastolic dysfunction with reduced compliance and impaired relaxation
Systolic Efficiency	High cardiac output through efficient contractions against low afterload	Inefficient work often against dynamic outflow obstruction
Coronary Perfusion	Proportionate increases in coronary flow capacity	Microvascular dysfunction and ischemia susceptibility
Exercise Response	Appropriate increases in cardiac output with maintained efficiency	Potential worsening obstruction, ischemia, and dangerous heart rhythms during physical activity

Described above differences explain why the athlete's heart represents beneficial adaptation while HCM poses significant health risks, particularly during intense physical activity. Understanding these distinctions proves crucial for accurate diagnosis and appropriate management decisions in athletic populations [24].

Contemporary risk stratification incorporates sophisticated tools including cardiovascular magnetic resonance imaging for scarring detection, genetic testing for mutation identification, exercise stress testing for hemodynamic assessment, and ambulatory monitoring for heart rhythm detection. The integration of these modalities enables more precise risk prediction than previously possible, though uncertainty remains inherent in predicting individual outcomes [27].

Discussion

The prevalence of HCM in competitive athletes mirrors that of the general population, affecting approximately 0.2% of participants in organized sports. However, the clinical significance of this prevalence extends far beyond simple numbers, as HCM represents the leading identifiable cause of sudden cardiac death in young athletes [1][2][28]. Demographic analysis reveals striking patterns in HCM-related sudden cardiac death among athletes. Male athletes demonstrate dramatically higher incidence compared to females, with ratios approaching 9:1 in some studies. This disparity likely reflects multiple factors, including higher male participation rates in competitive athletics, potential sex-related differences in disease expression, and varying susceptibility to exercise-induced dangerous heart rhythms [3][29]. Racial considerations add complexity to HCM epidemiology in sports medicine. African American athletes naturally develop greater degrees of left ventricular hypertrophy in response to training compared to Caucasian athletes, a physiological variation that can complicate the differential diagnosis between normal training adaptation and pathological HCM. This diagnostic challenge may contribute to both missed diagnoses and false-positive screening results within this population [4]. Specific sport risk patterns have emerged from large scale epidemiological studies. High-intensity sports with significant dynamic and static components, including basketball, American football, soccer, and rowing appear to confer greater risk for sudden cardiac events in athletes with underlying HCM. The hemodynamic demands of these activities, including rapid changes in venous return, afterload, and myocardial oxygen consumption, may serve as triggers for fatal heart rhythms in susceptible individuals [30].

The pathophysiological abnormalities underlying HCM create multiple barriers to optimal athletic performance while simultaneously increasing the risk of catastrophic events. Left ventricular outflow tract obstruction, present in approximately 70% of HCM patients, poses particular challenges during athletic activities. Exercise-induced increases in contractility and decreases in preload can dramatically worsen this obstruction, leading to reduced cardiac output despite increased metabolic demands [31]. The hemodynamic consequences of LVOTO extend beyond simple flow limitation. The increased afterload forces the left ventricle to generate higher pressures to maintain forward flow, dramatically increasing myocardial oxygen consumption. This creates a mismatch between oxygen supply and demand that can precipitate ischemia, dangerous heart rhythms, or hemodynamic collapse during intense exercise [11].

Diastolic dysfunction, almost universal in HCM, represents another fundamental limitation to athletic performance. The stiff, non-compliant ventricle cannot adequately fill during the shortened diastolic periods associated with exercise tachycardia. This filling impairment limits stroke volume augmentation and cardiac output response to exercise, manifesting as reduced exercise tolerance, early fatigue, and exertional shortness of breath [23][24]. The heart muscle architecture characteristic of HCM, including muscle cell disorder, interstitial fibrosis, and microvascular dysfunction creates a pro-arrhythmic substrate that becomes particularly vulnerable to exercise-related triggers. The combination of structural abnormalities, metabolic stress, and autonomic activation during competitive athletics creates ideal conditions for both atrial and ventricular arrhythmias [11][18][23]-[26].

The diagnosis of HCM in athletes extends far beyond medical considerations to encompass profound psychological and social implications that can persist throughout life. Many athletes experience grief processes similar to those associated with major losses, progressing through stages of denial, anger, bargaining, depression, and eventual acceptance. The identity crisis associated with HCM diagnosis can prove particularly devastating for athletes whose self-concept and social identity are intimately connected to their athletic performance. The abrupt cessation of competitive sports can leave athletes feeling lost, purposeless, and disconnected from their peer groups and support systems [32]. Family dynamics become significantly affected by HCM diagnosis in athletes, particularly given the genetic nature of the condition. Parents may experience guilt regarding their role in transmitting the condition or their previous encouragement of athletic participation. Siblings require screening and may face their own difficult decisions regarding sports participation. These

family stresses can strain relationships and create ongoing tension around medical management decisions [33]. The social isolation that may accompany withdrawal from team sports compounds these psychological challenges. Athletes often lose their primary social networks, coaching relationships, and the structured environment that organized sports provide. This social disruption can exacerbate depression and anxiety while complicating the transition to new activities and identities [33][34].

The economic implications of HCM diagnosis in athletic populations are substantial and multifaceted. For professional athletes, the immediate loss of current income can be devastating, particularly given the typically short duration of athletic careers and limited transferability of athletic skills to other professions. The loss of future earning potential may amount to millions of dollars for elite athletes in high-profile sports [32][35]. Educational opportunities represent another critical concern, as athletic scholarships may be discontinued following HCM diagnosis. The loss of educational funding can have profound long-term implications for career development and economic stability, particularly for athletes from socioeconomically disadvantaged backgrounds who depend on athletic scholarships for educational access [32]. Insurance considerations become critically important following HCM diagnosis. Life insurance premiums may increase substantially, or coverage may be denied altogether. Disability insurance becomes essential but may be difficult to obtain or prohibitively expensive. These insurance challenges can create significant financial vulnerability for affected athletes and their families [35].

The economic impact extends beyond the affected athlete to include family members who may require genetic testing and ongoing medical surveillance. The costs of comprehensive cardiac evaluation, genetic counseling, and long-term monitoring can create substantial financial burdens, particularly for families without adequate health insurance coverage [32].

The landscape of HCM management in athletes has undergone revolutionary changes in recent years, moving away from universal exercise restriction toward individualized risk assessment and shared decision-making approaches. The 2024 AHA/ACC/Multisociety Guidelines represented a paradigmatic shift that acknowledges the heterogeneity of HCM and the potential for safe exercise participation in carefully selected individuals The shared decision-making framework requires comprehensive discussion of risks, benefits, and alternatives between healthcare providers, athletes, and families. This approach respects athlete autonomy while ensuring appropriate medical guidance and emphasizes the importance of transparent communication about uncertainties in risk prediction Implementation of shared decision-making requires significant expertise in HCM management and careful consideration of medicolegal implications. Healthcare providers must be prepared to conduct sophisticated risk stratification, communicate complex medical information effectively, and support patients and families through difficult decision-making processes [8].

Recent therapeutic advances have expanded treatment options for athletes with HCM, potentially influencing sports participation decisions. Septal reduction therapies, including surgical septal myectomy and alcohol septal ablation, can effectively reduce outflow tract obstruction and improve functional capacity in selected patients. For athletes with significant LVOTO, these interventions may improve exercise tolerance and potentially reduce dangerous heart rhythm risk [18]. The introduction of mavacamten, a first-in-class cardiac myosin inhibitor, represents a paradigm shift in HCM drug therapy. By directly targeting the underlying sarcomeric dysfunction, mavacamten can reduce LVOTO, improve symptoms, and potentially modify disease progression. For athletes with obstructive HCM, mavacamten therapy might enable continued sports participation under appropriate monitoring, though long-term safety data in athletic populations remain limited [36]. Implantable cardioverter-defibrillators represent another therapeutic option for high-risk athletes with HCM. While ICDs can effectively terminate life-threatening heart rhythms, their use in athletic populations raises complex questions about appropriate sports participation and device-related complications. The psychological impact of ICD implantation and the potential for inappropriate device activation during athletic activities require careful consideration [7][37]. Advanced monitoring technologies, including wearable devices capable of continuous rhythm monitoring and activity tracking, offer new opportunities for real-time assessment of athletes with HCM. These technologies may enable early detection of concerning changes and prompt intervention, though their clinical validation and integration into care pathways require ongoing research [38].

The future of HCM management in athletes likely lies in precision medicine approaches that integrate genetic, phenotypic, and biomarker data to provide truly individualized risk prediction and therapeutic guidance. Polygenic risk scores, advanced imaging techniques, and circulating biomarkers may enhance risk stratification beyond current capabilities, enabling more confident decision-making regarding sports participation [8][39].

Conclusions

Hypertrophic cardiomyopathy in athletes represents one of the most challenging intersections in contemporary sports cardiology, requiring sophisticated integration of genetic medicine, advanced cardiac imaging, exercise physiology, and patient-centered care principles. The condition's significance extends far beyond its prevalence of 1 in 500 individuals, as it serves as the leading cause of sudden cardiac death in young competitive athletes, accounting for 30-40% of exercise-related fatalities in individuals under 35 years of age [2, 3].

The molecular foundation of HCM lies in mutations affecting sarcomeric proteins, predominantly involving the MYH7 and MYBPC3 genes. These genetic defects disrupt normal cardiac muscle function through multiple pathways, including impaired energy metabolism, oxidative stress, and calcium handling abnormalities. The resulting "energy-starved heart muscle" becomes particularly vulnerable during the metabolic demands of competitive athletics, where increased oxygen consumption and oxidative stress can precipitate life-threatening complications [9, 11].

The fundamental distinction between physiological "athlete's heart" adaptation and pathological HCM remodeling centers on contrasting hemodynamic profiles. While the athlete's heart demonstrates beneficial symmetric hypertrophy with preserved or enhanced diastolic function and optimal supply-demand matching, HCM hearts exhibit asymmetric hypertrophy, diastolic dysfunction, potential outflow tract obstruction, and microvascular dysfunction that predisposes to ischemia and dangerous heart rhythms [23]-[26][41].

The clinical management of athletes with HCM has undergone revolutionary transformation with the introduction of shared decision-making frameworks that emphasize individualized risk assessment over universal exercise restriction. This paradigm shift recognizes the tremendous variation in HCM presentation and acknowledges that carefully selected low-risk individuals may be able to continue competitive athletics under appropriate monitoring protocols [8].

Therapeutic advances continue to expand options for athletes with HCM. Septal reduction therapies can alleviate obstruction in selected patients, while the mavacamten, cardiac myosin inhibitor offers novel pharmaceutical intervention targeting the underlying sarcomeric dysfunction. Implantable cardioverter-defibrillators provide protection against sudden death for high-risk individuals, though their use in athletic populations requires careful consideration of device-related complications and psychological impact [8][37][38].

The management of athletes with hypertrophic cardiomyopathy exemplifies the complexity of modern sports cardiology, where genetic medicine, advanced imaging, exercise physiology, and patient-centered care converge to address one of the most challenging clinical scenarios in sports medicine. Success in this arena requires not only medical expertise but also sensitivity to the profound personal implications of these decisions for athletes whose identities and aspirations are intimately connected to their athletic participation [34][35][36][42].

Disclosure

Supplementary Materials

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