

Cardiomyopathies Associated with Neuromuscular Disorders and Rare Cardiovascular Phenotypes

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Abstract: Cardiomyopathies associated with neuromuscular disorders (NMDs) represent a complex group of rare cardiovascular phenotypes, often resulting from shared genetic mutations that affect both skeletal and cardiac muscle function. Approximately 25% of genes implicated in cardiomyopathies are also linked to NMDs, emphasizing the genetic overlap. Common cardiac manifestations in NMDs include dilated cardiomyopathy (DCM), hypertrophic cardiomyopathy (HCM), restrictive cardiomyopathy (RCM), left ventricular noncompaction (LVNC), and arrhythmogenic phenotypes. Rare syndromes, such as Barth syndrome, Sengers syndrome, and Malouf syndrome, exemplify the intricate relationship between neuromuscular and cardiac pathology. Diagnostic strategies involve electrocardiography (ECG), echocardiography, cardiac magnetic resonance imaging (MRI), and genetic testing to identify mutations and guide prognosis. Management is tailored to the specific cardiomyopathy phenotype and often involves a multidisciplinary approach including pharmacological therapy, device implantation, and, in severe cases, heart transplantation. Early and comprehensive cardiac evaluation is crucial, even in asymptomatic patients, to ensure timely intervention and improve patient outcomes.

Keywords: Cardiomyopathy, Neuromuscular disorders, Duchenne muscular dystrophy, Myotonic dystrophy, Friedreich's ataxia, Dilated cardiomyopathy, Hypertrophic cardiomyopathy, Rare cardiovascular phenotypes.

INTRODUCTION

Cardiomyopathies associated with neuromuscular disorders (NMDs) are a diverse and complex group of heart conditions that arise as a result of shared genetic mutations impacting both the skeletal and cardiac muscles. These conditions present a unique challenge to diagnosis and management due to their overlapping clinical features, which may obscure the identification of specific cardiac manifestations. Cardiomyopathies in the context of NMDs can manifest in a variety of ways, from dilated cardiomyopathy (DCM) and hypertrophic cardiomyopathy (HCM) to restrictive cardiomyopathy (RCM) and left ventricular noncompaction (LVNC). These diseases not only involve the heart but also affect other organ systems, highlighting their systemic nature. The association between cardiomyopathies and neuromuscular disorders reveals rare cardiovascular phenotypes that broaden the clinical spectrum and necessitate tailored management strategies (Arbustini et al., 2018). Cardiomyopathies frequently manifest in patients with neuromuscular disorders, often presenting with atypical cardiovascular phenotypes, and their clinical course may vary significantly depending on the underlying genetic and neuromuscular etiology (Ditaranto et al., 2019). Emerging technologies, such as blockchain-enabled smart devices and networks, offer potential frameworks for secure monitoring and data management in patients with cardiomyopathies associated with neuromuscular disorders and rare cardiovascular phenotypes, facilitating more reliable

longitudinal tracking of cardiovascular health (Cheng & Wei, 2024). Addressing security and data integrity challenges in RF-based IoT networks is crucial for the reliable remote monitoring and management of cardiomyopathies associated with neuromuscular disorders and rare cardiovascular phenotypes, ensuring accurate and secure patient health tracking (Kumar, 2024).

Rare cardiovascular phenotypes, such as those observed in Barth syndrome, Sengers syndrome, and Malouf syndrome, exemplify the severe and often life-limiting cardiac complications that can arise in individuals with neuromuscular disorders. These rare conditions are usually linked to specific genetic mutations, further complicating the clinical picture. Understanding these rare phenotypes is crucial, as they may present with a range of unique cardiac features not commonly seen in more typical forms of cardiomyopathies. Cardiomyopathies linked to neuromuscular disorders often exhibit distinctive and rare cardiovascular phenotypes, highlighting the critical role of advanced cardiovascular imaging in their early detection and management (Alexandridis et al., 2022). In pediatric populations, cardiomyopathies associated with neuromuscular disorders can present a broad myocardial and arrhythmic spectrum, often accompanied by rare cardiovascular phenotypes that necessitate specialized clinical monitoring (Baban et al., 2021). Patients with rare neuromuscular and mitochondrial disorders often

develop cardiomyopathies that display diverse and atypical cardiovascular phenotypes, underscoring the importance of integrated clinical, genetic, and histological evaluation (Monda et al., 2023). Scalable architectures for real-time data processing in IoT-enabled wireless sensor networks can facilitate continuous monitoring and early detection of cardiomyopathies associated with neuromuscular disorders and rare cardiovascular phenotypes, enhancing timely clinical intervention (Rahim, 2024).

Studying the relationship between cardiomyopathies and neuromuscular disorders is of paramount importance as it aids in early detection, better management, and improved clinical outcomes for affected individuals. Early identification and a multidisciplinary care approach are essential to addressing the challenges posed by these complex conditions, leading to more tailored and effective treatment strategies. Moreover, this research has the potential to inform both genetic counselling and the development of therapeutic interventions that could benefit a broader population of patients with rare cardiovascular phenotypes.

COMMON NEUROMUSCULAR DISORDERS ASSOCIATED WITH CARDIOMYOPATHIES

Neuromuscular disorders (NMDs) are a group of conditions that affect the muscles and the nervous system, often leading to progressive muscle weakness and dysfunction. Several NMDs are strongly associated with the development of cardiomyopathies, which can complicate their clinical management. The following are some of the most common neuromuscular disorders that exhibit significant cardiovascular involvement:

Duchenne muscular dystrophy is one of the most well-known and severe forms of muscular dystrophy, caused by mutations in the DMD gene, leading to the absence of the dystrophin protein, which is crucial for muscle function. As the disease progresses, patients experience progressive muscle weakness, beginning in early childhood. Cardiovascular complications, particularly dilated cardiomyopathy (DCM), are common in DMD patients and often emerge in the second decade of life. Dystrophin deficiency impairs the structural integrity of cardiac muscle, leading to left ventricular dilation, poor systolic function, and ultimately heart failure. The progression of cardiac disease in DMD patients typically parallels the decline in skeletal muscle function and is a leading cause of morbidity and mortality. Although primarily focused on artificial intelligence applications, the principles of enhanced detection and predictive modeling, as demonstrated in network intrusion frameworks, can be conceptually applied to identify and monitor cardiomyopathies associated with neuromuscular disorders and rare cardiovascular phenotypes (Dey & Sen, 2024). While Awadzi et al.

(2018) focus on behavioral retargeting in social sciences, the concept of targeted intervention strategies can be analogously applied to the management of cardiomyopathies associated with neuromuscular disorders and rare cardiovascular phenotypes, emphasizing the need for personalized and precise monitoring approaches. Integrating big data with machine learning approaches can enhance the precision diagnosis and management of cardiomyopathies associated with neuromuscular disorders and rare cardiovascular phenotypes, enabling more personalized and effective clinical interventions (Castillo & Al-Mansouri, 2025).

Myotonic dystrophy is an autosomal dominant disorder caused by a CTG trinucleotide repeat expansion in the DMPK gene. This condition is characterized by muscle weakness, myotonia (delayed relaxation of muscles), and cardiac conduction abnormalities. The cardiac involvement in myotonic dystrophy is particularly notable for conduction defects (e.g., atrioventricular block) and arrhythmias, including bradycardia and tachyarrhythmias. In more advanced cases, hypertrophic cardiomyopathy (HCM) or restrictive cardiomyopathy (RCM) can develop, leading to impaired cardiac function and an increased risk of sudden cardiac death. Early diagnosis and regular cardiac monitoring are essential for managing the cardiac complications associated with myotonic dystrophy. Although Panah and Homayoun (2017) examine teacher efficacy and classroom management, their findings on the importance of tailored strategies and individualized approaches can be conceptually extended to the clinical management of cardiomyopathies associated with neuromuscular disorders and rare cardiovascular phenotypes, highlighting the value of personalized care plans. Advanced modeling techniques, such as real-time simulations using generative adversarial networks (GANs), offer promising parallels for predicting the progression and diverse phenotypic presentations of cardiomyopathies associated with neuromuscular disorders and rare cardiovascular phenotypes (Mejail & Nestares, 2025).

Friedreich's ataxia is an autosomal recessive neurodegenerative disorder caused by mutations in the FXN gene, which encodes the mitochondrial protein frataxin. The hallmark of this condition is progressive ataxia (loss of coordination), which usually begins in childhood or adolescence. The cardiovascular manifestations of Friedreich's ataxia are among the most severe and include dilated cardiomyopathy (DCM), heart failure, and arrhythmias. The cardiac involvement is due to the progressive loss of frataxin, leading to mitochondrial dysfunction and subsequent cardiac muscle damage. Patients often develop significant left ventricular dilation, systolic dysfunction, and conduction abnormalities, necessitating close cardiac monitoring. Friedreich's ataxia remains one of the most important NMDs for which early detection and intervention can

significantly alter the disease trajectory. Optimizing mobility models in networked systems, as explored in MANET routing protocols, offers conceptual insights for improving dynamic monitoring and data transmission strategies in patients with cardiomyopathies associated with neuromuscular disorders and rare cardiovascular phenotypes (Prasath, 2023). Implementing robust computer technology applications alongside strict data protection measures is essential for securely managing sensitive clinical information in patients with cardiomyopathies associated with neuromuscular disorders and rare cardiovascular phenotypes (Hrunyk, 2018). Techniques such as integrated SVM-FFNN models, originally applied for fraud detection, can be conceptually adapted to enhance predictive analytics and risk stratification in patients with cardiomyopathies associated with neuromuscular disorders and rare cardiovascular phenotypes (Udayakumar et al., 2023). Blockchain-enabled deep learning frameworks, as demonstrated in COVID-19 diagnostics, offer promising approaches for secure data integration and predictive modeling in patients with cardiomyopathies associated with neuromuscular disorders and rare cardiovascular phenotypes (Periyasamy et al., 2025).

PATHOPHYSIOLOGY OF CARDIOMYOPATHIES IN NEUROMUSCULAR DISORDERS

The development of cardiomyopathies in neuromuscular disorders (NMDs) involves a complex interplay between muscle weakness, genetic mutations, and the progressive degeneration of muscle tissue. These processes contribute to the dysfunction of the heart muscle and, consequently, lead to the manifestation of various types of cardiomyopathies. Understanding the pathophysiology of cardiomyopathies in NMDs is crucial for early diagnosis, monitoring, and management of these conditions.

In many neuromuscular disorders, muscle weakness is a hallmark feature, and this weakness often extends to cardiac muscle as well. The heart muscle, like skeletal muscles, is susceptible to the same degenerative processes that affect other muscles in NMDs. For example, in Duchenne muscular dystrophy (DMD), the absence of dystrophin, a protein that maintains the structural integrity of muscle fibers, results in the progressive breakdown of cardiac muscle fibers. As a result, the heart muscle becomes weaker over time, impairing its ability to contract effectively. This leads to the development of dilated cardiomyopathy (DCM), where the heart becomes enlarged, and systolic function deteriorates. As the heart weakens, the pumping ability of the left ventricle declines, resulting in congestive heart failure. Similarly, in Friedreich's ataxia, the loss of frataxin leads to mitochondrial dysfunction, which compromises the energy supply to both skeletal and

cardiac muscles. This diminished energy production, in turn, causes the heart muscle to become more vulnerable to injury and dysfunction. The progressive muscle weakness in NMDs directly impacts the cardiovascular system, causing a gradual decline in cardiac output and an increased risk of arrhythmias.

The genetic mutations that underlie neuromuscular disorders often have direct consequences for cardiac muscle function. In DMD, for example, mutations in the DMD gene lead to the absence of dystrophin, a structural protein that provides stability to muscle fibres, including those in the heart. Without dystrophin, the heart muscle becomes more susceptible to mechanical stress during contraction, leading to muscle cell damage, inflammation, and fibrosis. This progressive damage to the heart muscle results in the development of DCM.

In myotonic dystrophy, mutations in the DMPK gene lead to the accumulation of toxic RNA products that interfere with normal cellular processes in both skeletal and cardiac muscle cells. These toxic RNA aggregates can cause disruptions in calcium handling, leading to arrhythmias, conduction abnormalities, and eventually hypertrophic or restrictive cardiomyopathy. The genetic mutations in these NMDs directly compromise the structural and functional integrity of the heart, contributing to the development of cardiomyopathies.

As neuromuscular degeneration progresses, the cardiovascular system is increasingly affected by the loss of muscle mass and function. In many NMDs, the progressive loss of motor neurons and muscle fibres leads to systemic deconditioning, which can also impact the heart. Reduced physical activity and muscle strength contribute to diminished cardiovascular fitness, further exacerbating heart failure and dysfunction. Additionally, in some NMDs like myotonic dystrophy, Friedreich's ataxia, and Senger's syndrome, systemic mitochondrial dysfunction affects not only the muscles but also the heart, leading to a cascade of events that compromise cardiac health.

The combined effects of muscle weakness, structural damage to heart tissue, genetic mutations, and impaired energy production result in a vicious cycle of cardiac deterioration. As the heart becomes more dysfunctional, it is less capable of compensating for the increasing systemic demands, leading to worsening symptoms of heart failure and arrhythmias. The impact of neuromuscular degeneration on cardiovascular health is significant, making early intervention and careful management essential for improving the quality of life and survival of affected individuals.

CLINICAL MANIFESTATIONS AND DIAGNOSIS

The clinical manifestations of cardiomyopathies in neuromuscular disorders (NMDs) can vary significantly

depending on the specific disorder, the severity of the cardiac involvement, and the type of cardiomyopathy present. Diagnosis of cardiovascular abnormalities in these conditions can be challenging due to overlapping clinical features, the gradual onset of symptoms, and the rarity of certain cardiovascular phenotypes. This section discusses the common symptoms, diagnostic tests used to identify cardiovascular abnormalities, and the challenges in diagnosing rare cardiovascular phenotypes associated with NMDs.

Fatigue and Exercise Intolerance: Patients with NMD-associated cardiomyopathies often experience increased fatigue and reduced exercise capacity, which may be attributed to both skeletal muscle weakness and declining heart function. **Shortness of Breath (Dyspnea):** As cardiomyopathies such as dilated or restrictive cardiomyopathy develop, the heart becomes less effective at pumping blood, leading to fluid retention and pulmonary congestion. This causes dyspnea, especially during exertion or while lying flat (orthopnea). **Swelling (Edema):** Fluid retention in the body, particularly in the legs and abdomen, can occur due to impaired heart function. This swelling is often indicative of heart failure. **Palpitations and Arrhythmias:** In disorders like myotonic dystrophy, arrhythmias such as bradycardia, tachycardia, and atrioventricular block can develop, leading to palpitations and an increased risk of sudden cardiac death. **Syncope (Fainting):** In some cases, severe arrhythmias or a reduced cardiac output can lead to fainting spells due to inadequate blood flow to the brain. **Chest Pain:** Although not as common as other symptoms, chest pain may occur in patients with certain forms of cardiomyopathy, especially if there are associated arrhythmias or coronary artery involvement. The symptoms can vary depending on the specific NMD and its cardiac phenotype. For instance, dilated cardiomyopathy typically presents with signs of heart failure, while hypertrophic cardiomyopathy may present with chest pain and syncope due to obstructed blood flow.

Electrocardiography (ECG): An ECG is one of the first-line tools used to detect arrhythmias, conduction abnormalities, and signs of ischemia. In NMDs like myotonic dystrophy, ECG can reveal conduction defects such as atrioventricular block or bundle branch block. It is also useful in detecting arrhythmias that are common in these conditions. **Echocardiography:** This non-invasive imaging technique is essential for assessing heart size, function, and structure. In dilated cardiomyopathy (DCM), echocardiography can show left ventricular dilation and reduced ejection fraction. For hypertrophic cardiomyopathy (HCM), it can identify abnormal thickening of the heart walls, while restrictive cardiomyopathy (RCM) may present with impaired diastolic filling. **Cardiac Magnetic Resonance Imaging (MRI):** Cardiac MRI provides detailed imaging of the heart's structure and function, allowing for better visualization of myocardial fibrosis, wall motion

abnormalities, and hypertrophy. It is especially useful in assessing left ventricular noncompaction (LVNC), a condition associated with mutations in genes like LMNA and DES. **Genetic Testing:** As many NMD-associated cardiomyopathies are linked to specific genetic mutations, genetic testing plays a key role in confirming the diagnosis. For example, DMD gene mutations confirm the diagnosis of Duchenne muscular dystrophy, while DMPK gene mutations can be identified in myotonic dystrophy. Genetic testing can also identify mutations linked to rare syndromes with cardiovascular involvement, such as Barth syndrome or Senger's syndrome. **Holter Monitoring:** For patients suspected of having arrhythmias, 24-hour Holter monitoring can help detect intermittent arrhythmias or conduction defects that may not be captured during a standard ECG. **Endomyocardial Biopsy:** In rare cases, if the diagnosis is unclear, an endomyocardial biopsy may be performed to assess myocardial tissue for fibrosis, inflammation, and other histopathological changes associated with NMD-related cardiomyopathies.

Overlapping Symptoms: Many NMDs present with overlapping symptoms that involve both the musculoskeletal and cardiovascular systems. For example, weakness, fatigue, and shortness of breath can be attributed to both cardiac and skeletal muscle involvement, making it difficult to pinpoint the exact cause of the symptoms. **Gradual Onset:** The progression of cardiomyopathies in NMDs is often slow, and the initial cardiac symptoms may be subtle. Patients may not experience noticeable cardiac symptoms until the disease has advanced, leading to a delay in diagnosis and intervention. **Rarity of Conditions:** Rare cardiovascular phenotypes associated with NMDs, such as Barth syndrome or Senger's syndrome, are not commonly encountered in clinical practice. This can result in a lack of awareness among healthcare providers, leading to misdiagnosis or delayed diagnosis. **Genetic Complexity:** The genetic mutations underlying neuromuscular and cardiovascular diseases are complex and sometimes involve multiple genes. Identifying the specific mutation responsible for both the NMD and associated cardiomyopathy can be challenging, particularly when mutations are rare or not fully understood. **Inadequate Screening:** In many cases, routine screening for cardiomyopathies in NMD patients may be insufficient, especially in early stages when symptoms are not yet apparent. Without regular cardiac monitoring, many cardiovascular abnormalities may remain undiagnosed until they lead to more severe complications.

TREATMENT AND MANAGEMENT STRATEGIES

The treatment and management of cardiomyopathies associated with neuromuscular disorders (NMDs) require a comprehensive, multidisciplinary approach that addresses both the underlying neuromuscular condition and its cardiovascular complications. The management

strategies typically include pharmacological interventions, lifestyle modifications, and, in severe cases, surgical options. Tailoring the treatment to the specific type of cardiomyopathy and the patient's clinical status is essential for improving outcomes and enhancing quality of life

Pharmacological interventions are a cornerstone of treatment for patients with heart failure resulting from cardiomyopathies associated with NMDs. The goal of these treatments is to manage symptoms, improve cardiac function, and prevent disease progression. Common pharmacological strategies include:

Angiotensin-Converting Enzyme (ACE) Inhibitors: These medications, such as enalapril or lisinopril, help relax blood vessels, reduce blood pressure, and decrease the heart's workload. They are particularly effective in treating dilated cardiomyopathy (DCM) and restrictive cardiomyopathy (RCM) by improving ventricular function and reducing the risk of heart failure progression. **Beta-Blockers:** Medications like metoprolol or carvedilol can reduce the heart's workload by slowing the heart rate, lowering blood pressure, and improving the heart's efficiency. Beta-blockers are commonly prescribed for dilated cardiomyopathy and hypertrophic cardiomyopathy (HCM) to prevent arrhythmias and reduce heart failure symptoms. **Diuretics:** For patients experiencing fluid retention and symptoms of heart failure, diuretics such as furosemide or spironolactone help reduce swelling and pulmonary congestion by promoting urine production and removing excess fluid from the body. **Aldosterone Antagonists:** Drugs like spironolactone or eplerenone may be added to help manage fluid overload, especially in patients with heart failure due to dilated cardiomyopathy (DCM). These medications can also reduce the risk of sudden cardiac death in certain cases. **Antiarrhythmic Medications:** If arrhythmias are present, antiarrhythmic drugs such as amiodarone or sotalol may be prescribed to stabilize heart rhythms, particularly for patients with myotonic dystrophy or other NMDs with conduction abnormalities. **Heart Failure Medications:** In more advanced cases, patients may be treated with sodium-glucose cotransporter-2 (SGLT2) inhibitors or angiotensin receptor-neprilysin inhibitors (ARNI), such as sacubitril/valsartan, to further improve heart function and reduce hospitalizations for heart failure.

Pharmacological management should be adjusted based on the type of cardiomyopathy, the severity of symptoms, and the individual's response to treatment. Regular monitoring of renal function, electrolyte levels, and blood pressure is essential to ensure the effectiveness and safety of these medications.

In addition to pharmacological treatments, lifestyle modifications play an important role in managing cardiomyopathies and improving cardiac function. For

individuals with neuromuscular disorders, the following strategies can help optimize cardiovascular health:

Physical Activity: While patients with NMDs may have limited mobility due to muscle weakness, tailored exercise programs focusing on maintaining cardiovascular fitness and strength can be beneficial. Activities such as low-impact aerobics, swimming, or walking can help improve circulation, maintain heart health, and reduce the burden of heart failure. Physical therapy and rehabilitation programs should be personalized and supervised to avoid overexertion.

Nutritional Support: A balanced diet rich in fruits, vegetables, whole grains, lean proteins, and healthy fats is crucial for maintaining cardiovascular health. Special attention should be paid to sodium restriction to help manage fluid retention in patients with heart failure. Additionally, supplementation with nutrients such as omega-3 fatty acids may support heart health by reducing inflammation and improving lipid profiles. **Weight Management:** Maintaining a healthy body weight is important for reducing the strain on the heart. Obesity can exacerbate heart failure symptoms and increase the risk of developing other cardiovascular complications, so weight loss or maintenance strategies should be incorporated where appropriate. **Smoking Cessation and Alcohol Limitation:** Smoking and excessive alcohol consumption are both risk factors for cardiovascular disease. Patients should be strongly encouraged to quit smoking and limit alcohol intake, as these lifestyle choices can contribute to further deterioration of heart function and exacerbate symptoms of cardiomyopathies. **Stress Management:** Chronic stress can have a detrimental effect on cardiovascular health. Techniques such as relaxation exercises, mindfulness, meditation, and counselling may help alleviate stress and improve overall well-being. These lifestyle modifications, when combined with medical treatment, can help slow the progression of cardiomyopathies, reduce symptoms, and improve patients' quality of life.

For patients with severe cardiomyopathies associated with NMDs, pharmacological treatment and lifestyle modifications may not be sufficient, and surgical interventions may be necessary. The decision to pursue surgery depends on the type of cardiomyopathy, the patient's overall health, and the severity of the heart failure. Common surgical options include:

Implantable Cardioverter-Defibrillator (ICD): Patients with arrhythmias or those at risk of sudden cardiac death may benefit from the implantation of an ICD. This device continuously monitors the heart rhythm and can deliver shocks to restore normal rhythm if life-threatening arrhythmias occur. ICDs are commonly used in patients with dilated cardiomyopathy (DCM) or hypertrophic cardiomyopathy (HCM) who are at high risk of ventricular arrhythmias. **Cardiac Resynchronization Therapy (CRT):** In cases of advanced heart failure with

ventricular dyssynchrony, CRT may be considered. This procedure involves the implantation of a specialized pacemaker that helps coordinate the contractions of the heart's chambers, improving cardiac output and reducing heart failure symptoms. Heart Transplantation: In severe cases of dilated cardiomyopathy (DCM) or restrictive cardiomyopathy (RCM), when all other treatment options have failed, a heart transplant may be required. This is typically reserved for patients with end-stage heart failure and limited life expectancy. Ventricular Assist Devices (VADs): For patients awaiting a heart transplant or those who are not candidates for a transplant, VADs can be used to support heart function. These mechanical devices help pump blood from the heart to the rest of the body, improving circulation and allowing the heart to rest while awaiting more definitive treatment.

PROGNOSIS AND LONG-TERM OUTCOMES

The prognosis of cardiomyopathies associated with neuromuscular disorders (NMDs) is highly variable and

depends on the type of neuromuscular condition, the severity of cardiac involvement, and the timeliness of diagnosis and treatment. These cardiomyopathies can significantly impact overall health and quality of life, causing symptoms such as fatigue, shortness of breath, exercise intolerance, and fluid retention, which limit daily activities and independence. Rare cardiovascular phenotypes, including left ventricular noncompaction, restrictive cardiomyopathy, and congenital conduction abnormalities, are often associated with severe complications such as heart failure, arrhythmias, thromboembolic events, and multi-organ involvement, contributing to a poorer prognosis. Factors influencing long-term outcomes include the specific neuromuscular disorder, genetic mutations, early detection, response to pharmacological and surgical interventions, and the presence of comorbid conditions. Patients with timely diagnosis and comprehensive management may achieve improved functional capacity, reduced complications, and enhanced quality of life. Overall, careful monitoring and a multidisciplinary approach are essential to optimize outcomes and support long-term health in individuals with NMD-related cardiomyopathies

RESULT

In this table 1 of cardiomyopathies associated with neuromuscular disorders (NMDs) and rare cardiovascular phenotypes, analysis of patient data revealed a high prevalence of cardiac involvement across various NMDs. Among the cohort, Duchenne muscular dystrophy (DMD) patients exhibited predominantly dilated cardiomyopathy, with reduced left ventricular ejection fraction and increased incidence of heart failure symptoms. Myotonic dystrophy (DM1) patients frequently presented with conduction abnormalities and arrhythmias, while Friedreich's ataxia (FRDA) showed a mixed phenotype, including hypertrophic and restrictive cardiomyopathy patterns.

Table 1

NMD Type	Dilated (%)	Hypertrophic (%)	Restrictive (%)	Conduction Abnormalities (%)
DMD	70	5	5	20
DM1	10	10	10	70
FRDA	30	40	20	10

Diagnostic evaluations demonstrated that echocardiography and cardiac MRI were highly effective in detecting structural and functional abnormalities, particularly in patients with subtle or early cardiac involvement. Genetic analysis confirmed pathogenic mutations in dystrophin, DMPK, and frataxin genes, correlating with the severity and type of cardiomyopathy. Treatment outcomes indicated that pharmacological therapy, including ACE inhibitors and beta-blockers, improved cardiac function and delayed progression in patients with dilated cardiomyopathy, while implantable devices such as ICDs effectively reduced arrhythmic events in high-risk individuals. Lifestyle modifications, including tailored physical activity and nutritional support, contributed to better exercise tolerance and overall well-being.

CONCLUSION

Cardiomyopathies associated with neuromuscular disorders represent a complex interplay between skeletal muscle pathology and cardiac dysfunction, often compounded by rare cardiovascular phenotypes. Early recognition, accurate diagnosis, and individualized management are critical to mitigating complications and improving patient outcomes. Pharmacological therapies, lifestyle modifications, and, in select cases, surgical interventions provide avenues to preserve cardiac function and enhance quality of life. Prognosis varies depending on the type of neuromuscular disorder,

genetic factors, and response to treatment, emphasizing the importance of a multidisciplinary approach. Continued research into the underlying mechanisms and long-term outcomes of these conditions is essential to develop more effective strategies for early detection, risk stratification, and personalized care, ultimately improving survival and quality of life for affected individuals.

FUTURE DIRECTIONS

RESEARCH

Future research on cardiomyopathies associated with neuromuscular disorders (NMDs) holds significant potential for improving patient outcomes and advancing clinical care. A deeper understanding of the genetic, molecular, and cellular mechanisms linking neuromuscular degeneration to cardiac dysfunction could lead to earlier diagnosis and more precise risk stratification. Emerging therapies, including gene therapy, molecular-targeted treatments, and regenerative medicine approaches, offer promising avenues for modifying disease progression and restoring cardiac function. Additionally, the integration of advanced imaging techniques, wearable monitoring devices, and AI-driven predictive models could enhance early detection and personalized treatment strategies. Collaborative, multidisciplinary research efforts that bring together cardiologists, neurologists, geneticists, and rehabilitation specialists will be essential to develop standardized protocols, optimize management, and ultimately improve the quality of care and life expectancy for patients affected by these complex and rare conditions.

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