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REVIEW ARTICLE

Rehabilitative interventions in pediatrics for neuromuscular disorders

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ABSTRACT

Introduction: pediatric neuromuscular diseases represent a clinical challenge due to their progressive nature and their impact on mobility and childhood autonomy.
Objective: to analyze the influence of rehabilitative interventions on functional outcomes and quality of life in children with neuromuscular disorders.
Methods: a systematic review of the scientific literature was conducted across several databases. The search was performed using an algorithm with keywords and Boolean operators, allowing the identification of relevant sources. Selected studies, after applying inclusion and exclusion criteria, were critically analyzed considering timeliness, methodological quality, and thematic relevance, and integrated into the final synthesis of the review.
Development: evidence shows that multidisciplinary rehabilitation, including physiotherapy, occupational therapy, and respiratory support, improves strength, mobility, and autonomy. Technologies such as electrical stimulation and assisted robotics enhance outcomes. Pain, scoliosis, and malnutrition are frequent complications requiring specific approaches. Family involvement is essential for adherence and emotional well-being. Although curative treatments do not exist, rehabilitation maximizes functional capacities and prevents complications. Genetic advances and neonatal screening enable earlier diagnoses, favoring personalized strategies.
Conclusions: pediatric rehabilitation in neuromuscular diseases is essential to preserve motor function and improve quality of life. An individualized, multidisciplinary approach, with family support and the use of emerging technologies, constitutes the most effective strategy to address these progressive conditions.

Keywords: Neuromuscular Diseases; Pediatrics; Rehabilitation; Physical Therapy Services.

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INTRODUCTION

Neuromuscular diseases (NMDs) represent one of the major challenges in pediatrics, with rehabilitation posing a significant clinical complexity due to their intricate nature. Clinical manifestations vary widely depending on the specific disease, often involving multiple systems, frequently leading to severe disability and even death.[1] In most cases, rehabilitation medicine interventions play a crucial role in preventing complications and enhancing quality of life, as available research indicates that pain is one of the frequent and significant problems in patients with NMDs, although studies enabling its identification remain limited.[2,3] Generating updated information on this highly specific topic through continuous literature reviews on NMDs—and how they have adversely affected numerous children—significantly contributes to exploring new rehabilitation methods aimed at improving quality of life.

NMDs constitute a group of hereditary or acquired neurological disorders whose main characteristic is progressive loss of muscle strength and, in some cases, degeneration of muscles and the nerves controlling them, which can manifest at any stage of life. Although progression varies considerably, the resulting deficits often combine to produce musculoskeletal damage, impairing daily activities. Given this diversity, treatment approaches are symptom-based, aiming to slow disease progression—a principle emphasized in professional consensus on essential guidelines and priority objectives.[4,5]

Physicians must educate patients and promote lifestyles emphasizing activities that maintain function. This remarkable expansion of knowledge will ultimately lead to treatments targeting the underlying causes of these pathologies. Such progress is evident in genetic and molecular biology fields, which have broadened awareness of the diverse mechanisms beyond classical findings involved in NMDs. Historically, electrodiagnostic studies were commonly used as an extension of the physical examination to better characterize these disorders.[6]

Advances in genomic medicine have greatly enhanced understanding and diagnostic support for NMDs. Annually, a list of pathologies is published, grouped into 16 categories encompassing over 300 diseases classified by identified genes. However, some NMDs present genomic variants that complicate diagnosis, highlighting the importance of recognizing that we face a group of diseases with significant underreporting and limited knowledge of clinical, diagnostic, and therapeutic aspects. Notably, during the COVID-19 pandemic, patients with NMDs experienced a substantial reduction in physical activity, negatively impacting their quality of life and accelerating muscle mass loss and disease progression.[2]

Although it is true that, to date, no curative treatment exists for many NMDs—particularly hereditary forms—it is incorrect to claim these diseases are untreatable, especially now with the emergence of novel therapies for some of them. These therapeutic strategies are increasingly complex and specific, requiring coordinated care across multiple levels of health and rehabilitation services. These patients should receive rehabilitation (RHB), preventive measures for severe or lethal cardiovascular events, and ventilatory and nutritional support, always considering the perspectives of both the patient and their family.[1,7]

Given the limited knowledge regarding rehabilitation treatments for pediatric NMDs, their varied symptoms, societal impact, patient well-being, and recent advances in therapeutic mechanisms, this review was conducted with the objective of analyzing the influence of rehabilitative interventions on the functional evolution and quality of life of children with neuromuscular disorders.

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METHODS

This study was designed as a systematic literature review following the PRISMA 2020 guidelines to ensure transparency, reproducibility, and methodological rigor. The search period was limited to 2020–2024 to identify the most relevant rehabilitative interventions applied in pediatrics for managing neuromuscular disorders.

Information sources included widely recognized academic databases: PubMed/MEDLINE, Scopus, SciELO, and Google Scholar. Additionally, secondary references from selected articles were reviewed to broaden the identification of relevant studies. Grey literature outside institutional repositories was excluded, as the aim was to prioritize indexed, peer-reviewed scientific publications.

The search strategy employed an algorithm combining keywords and Boolean operators. MeSH and DeCS terms such as "neuromuscular diseases," "rehabilitation," "pediatric," "physical therapy," and "occupational therapy" were combined using AND and OR operators to maximize sensitivity and specificity. Publications in Spanish and English were included to integrate evidence from diverse clinical and linguistic contexts.

Inclusion criteria encompassed original articles, clinical trials, cohort studies, and systematic reviews published within the defined timeframe that directly addressed rehabilitative interventions in children with NMDs. Duplicates, articles without full access, irrelevant documents, publications prior to 2020, as well as letters, editorials, clinical practice guidelines, and theses were excluded.

The selection process occurred in several phases: initial title and abstract screening to exclude non-relevant studies, followed by full-text evaluation of potentially eligible articles. Initially, 32 records were identified; after removing duplicates and applying exclusion criteria, 13 articles remained. Two additional articles were included through reference checking, resulting in a total of 15 sources. The procedure was documented using a PRISMA flow diagram (Figure 1), illustrating each selection stage.

Data extraction and analysis were performed systematically, collecting key variables such as author, publication year, methodological design, pediatric population characteristics, type of rehabilitative intervention, clinical outcomes, and main findings. Information was organized into comparative matrices to facilitate interpretation. A qualitative synthesis was conducted, as methodological and outcome heterogeneity precluded formal meta-analysis. This approach enabled integration of available evidence and provided a critical, up-to-date overview of rehabilitative interventions in pediatrics for neuromuscular disorders.

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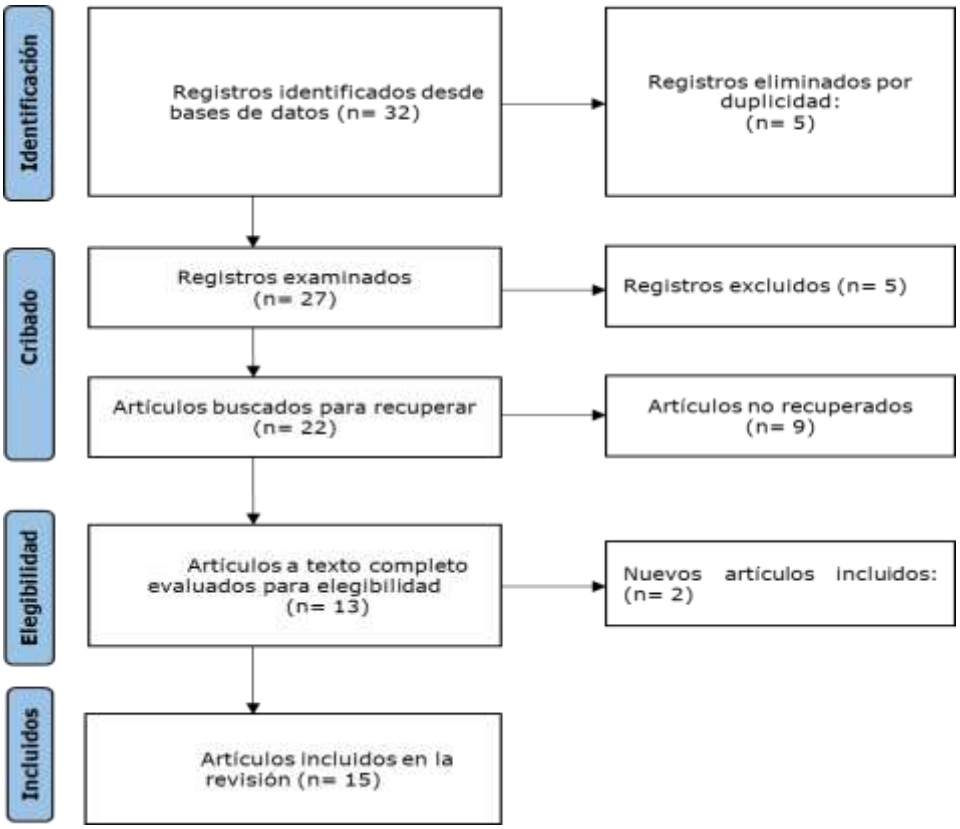

Fig. 1 PRISMA flow diagram.

DEVELOPMENT

This investigative process revealed that NMDs in pediatrics significantly impair motor and sensory functions, as they affect the peripheral nervous system and encompass a wide spectrum of distinct syndromes. They require diverse treatments or procedures for timely rehabilitation based on symptom monitoring, aiming to slow disease progression. The evolutionary analysis of this pathology has led to better understanding and progressively influenced rehabilitation approaches.[4,8]

Most NMDs lack etiological treatments and are incurable—but not untreatable. Therefore, rehabilitation is essential to maximize functional capacities and improve quality of life for both the patient and their family.[5] Despite advances, clinical history and physical examination remain the cornerstone of NMD diagnosis. These disorders can manifest at any age—from birth to adulthood—with highly variable clinical origins or syndromes.[1,6,9]

Assessing the extent of pain in NMDs is generally complex due to the multitude of conditions affecting children. Nevertheless, studies confirm that pain is a frequent and significant problem in NMDs.[3] It should be noted that NMDs are among the leading causes of pediatric mortality, presenting a wide variety of clinical presentations—as previously mentioned—which sometimes complicates initial diagnostic approaches. Since many of these disorders cause progressive disability in children, timely diagnosis is crucial. Traditionally, this group has been divided into muscle disorders (myopathies) and peripheral nerve disorders (neuropathies).[10,11]

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Thus, the sequence of diagnostic evaluations should follow prioritization guidelines, initially focusing on cardiopulmonary function due to vital necessity. Subsequently, factors affecting quality of life—such as pain and neuromuscular function—are assessed to help the child achieve minimal independence, followed by more analytical studies.[4]

In many cases, the orthopedic surgeon may be the first specialist consulted to evaluate complaints of weakness or lower limb deformities resulting from these conditions and must remain alert to the primary diagnosis. The pediatric orthopedic surgeon must be prepared to guide appropriate physiotherapy and perform surgeries to improve function, prevent deformity, or provide comfort when necessary. This specialist must be familiar not only with the musculoskeletal system but also with other domains that may influence treatment and require preoperative management.[8,12]

Despite the diverse primary mechanisms of these disorders, certain secondary musculoskeletal complications are shared. A clear example is scoliosis—the deviation from normal spinal alignment—which is almost always associated with sagittal plane anomalies such as kyphosis or lordosis, or with a rotational component. In general, idiopathic scoliosis represents the vast majority of pediatric scoliosis cases.[9]

Advances in DNA sequencing technologies have enabled identification of causative genes for these pathological groups; however, more than half of affected children and adults still lack a molecular diagnosis due to limited access to these technologies. This places neurological examination, histopathology, and biochemistry in a crucial diagnostic role in our setting. Consequently, multi-level health and rehabilitation strategies have been developed, emphasizing preventive management of cardiovascular events in this patient population.[2]

Among the disorders identified, sleep disturbances are present in more than 75 % of children with neuromuscular disorders. Multiple factors contribute to sleep disorders, including poor initiation and maintenance of naps, reduced bed mobility, body position discomfort, and hypoventilation due to contractures—requiring respiratory support such as bilevel positive airway pressure (BiPAP) masks—and central nervous system abnormalities.[13]

Another challenge is Duchenne muscular dystrophy, as previously mentioned—a disorder characterized by progressive neurological deterioration and worsening muscle weakness due to the absence of dystrophin protein in skeletal muscle. It affects only young boys due to its X-linked recessive inheritance pattern, resulting from spontaneous mutations in the dystrophin gene (producing an unstable dystrophin protein that degrades rapidly). Diagnosis typically requires a muscle biopsy revealing connective tissue infiltration.[14]

Among the disabilities caused by this pediatric condition, weakness is primary, impairing gait and functional activities in ambulatory children affected by these often degenerative pathologies. Gait speed is a key indicator of health and disability, representing a complex, multifaceted activity. Six-minute walk tests—widely used as endurance and ambulatory capacity assessments—have shown significant limitations in children with spinal muscular atrophy type 3, congenital muscular dystrophy, and older boys with Duchenne muscular dystrophy.[15]

Given their uniqueness and complexity, NMDs are often diagnosed after significant delays, by which time irreversible muscle damage may limit treatment efficacy when therapies are available. In this context, neonatal screening could offer a solution for early detection and intervention. The future of NMD detection in newborns lies in a global technological shift—from biochemical to genetic approaches—enabling rapid therapy development and flexible adaptation of treatable disease lists.[16]

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Collected evidence indicates that NMDs are poorly understood, with diverse—and often unknown—causes. They may be hereditary or acquired and manifest at any life stage. Their progressive nature gradually diminishes functional capacity, leading to loss of autonomy.[17]

Individual patient characteristics result in diverse clinical scenarios. Each patient should undergo a meticulous general physical evaluation to determine the most appropriate, individualized intervention—whether outpatient or inpatient—tailored to their specific context.[18]

According to Rodríguez-Núñez et al.,[19] evidence-based clinical practice underpins these recommendations, whose epistemological validity depends on the internal rigor of the scientific process generating the knowledge. Protecting the airway in NMD patients is critical, as it is severely compromised by disease-related limitations. Rodríguez Gómez et al.,[20] note that NMDs can impair the respiratory system, causing variable degrees and onset of respiratory morbidity depending on respiratory and swallowing muscle involvement, nutritional status, and ambulatory capacity.

Giménez et al.,[21] mention that, unfortunately, any patient requiring respiratory support—regardless of diagnosis—who is admitted to the intensive care unit typically needs mechanical ventilation and tracheostomy. However, in NMD patients, non-invasive procedures can be chosen, optimizing quality of life and reducing hospital costs. Cifuentes-Silva et al.,[22] conducted a study to describe respiratory muscle strength and endurance variables in NMD patients.

A neuromuscular patient is defined as ambulatory if they can walk independently for ten meters or more without assistance or support. Mozzoni,[23] described standardized protocols for timed performance tests in ambulatory NMD patients. These tests are reproducible, valid, and accessible tools to measure functional performance and track progression over time. To achieve optimal performance, each test may be repeated up to three times, with the shortest time (in seconds and milliseconds) recorded as the final result, and a maximum allowed time of 45 seconds per test.

On the other hand, children with NMDs face a high risk of malnutrition, primarily due to feeding disorders stemming from muscular impairment. In this context, Martínez Costa et al.,[24] discuss merosin deficiency—a severe autosomal recessive congenital muscular dystrophy in which muscle involvement prevents independent ambulation. Affected children have a high malnutrition risk due to digestive problems related to muscular impairment. Nutritional support should be implemented progressively based on clinical evolution, starting with assessment of current intake and estimated caloric requirements. Further research is needed to generate more information on this topic, given its social impact, effects on patient quality of life and health, and the need to raise awareness about these pathologies and the challenges faced by affected patients.

CONCLUSIONS

Rehabilitation in children with NMDs represents a significant challenge due to the progressive and heterogeneous nature of these conditions. However, rehabilitative interventions have proven essential for improving motor function, preventing secondary complications, and optimizing quality of life. An individualized approach—tailored to each child's specific needs and disease characteristics—is crucial to maximizing therapeutic benefits. The multidisciplinary model, incorporating physiotherapy, occupational therapy, respiratory and psychological support, has demonstrated effectiveness in addressing the multiple dimensions of these diseases.

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Furthermore, active family participation in the rehabilitation process is vital—not only to ensure treatment adherence but also to provide a supportive environment that facilitates the child's adaptation and emotional well-being.

BIBLIOGRAPHIC REFERENCES

1. Barros G, Moreira I, Ríos R. Treatment rehabilitation and management of neuromuscular diseases. Revista Medica Clinica Las Condes [Internet]. 2018 [Citado 20/05/2025]; 29(5):560–9. Disponible en: https://www.elsevier.es/es-revista-revista-medica-clinica-las-condes-202-articulo-tratamiento-rehabilitacion-manejo-global-S0716864018301032

2. Arriaga Rivera J. Tratamiento de rehabilitación en las enfermedades neuromusculares. Lux Médica [Internet]. 18 de enero de 2023 [Citado 20/05/2025];18(52). Disponible en: https://portal.amelica.org/ameli/journal/486/4863677010/html/

3. López M, Miró J. Dolor en niños y adolescentes con enfermedades neuromusculares. Rev Soc Esp Dolor [Internet]. 2013 [Citado 20/05/2025]; 20(3): 142-149. Disponible en: https://scielo.isciii.es/scielo.php?script=sci_arttext&pid=S1134-80462013000300007

4. Martin E, Renouil M, Lombard C, Ruzic J, Poinsot E. Enfermedades neuromusculares en el niño y el adolescente: detección, evaluaciones específicas, prioridades, tratamientos de rehabilitación y quirúrgicos. EMC - Kinesiterapia - Medicina Física [Internet]. noviembre de 2015 [Citado 20/05/2025]; 36(4):1–15. Disponible en: https://www.sciencedirect.com/science/article/abs/pii/S1293296515741447

5. Valdebenito R, Ruiz D. ASPECTOS RELEVANTES EN LA REHABILITACIÓN DE LOS NIÑOS CON ENFERMEDADES NEUROMUSCULARES. REV MED CLIN CONDES [Internet]. 2014 [Citado 20/05/2025]; 25(2):295–305. Disponible en: https://www.elsevier.es/es-revista-revista-medica-clinica-las-condes-202-articulo-aspectos-relevantes-rehabilitacion-los-ninos-S0716864014700410

6. Hornyak IV JE, Pangilinan PH. Rehabilitation of Children and Adults Who Have Neuromuscular Diseases. Phys Med Rehabil Clin N Am [Internet]. noviembre de 2007 [Citado 20/05/2025]; 18(4): 883–97. Disponible en: https://pubmed.ncbi.nlm.nih.gov/17967367/

7. Page MJ, McKenzie JE, Bossuyt PM, Boutron I, Hoffmann TC, Mulrow CD, et al. The PRISMA 2020 statement: An updated guideline for reporting systematic reviews. The BMJ [Internet]. 29 de marzo de 2021 [Citado 20/05/2025]; 372(71). Disponible en: https://pubmed.ncbi.nlm.nih.gov/33782057/

8. Mary P, Servais L, Vialle R. Neuromuscular diseases: Diagnosis and management. Orthop Traumatol Surg Res [Internet]. 1 de febrero de 2018 [Citado 20/05/2025]; 104(S1): S89–95. Disponible en: https://pubmed.ncbi.nlm.nih.gov/29196274/

9. Driscoll SW, Skinner J. Musculoskeletal Complications of Neuromuscular Disease in Children. Phys Med Rehabil Clin N Am [Internet]. febrero de 2008 [Citado 20/05/2025]; 19(1):163–94. Disponible en: https://pubmed.ncbi.nlm.nih.gov/18194756/

ISSN: 1561-3194 **RNPS:** 1877
Rev Ciencias Médicas. 2025; 29(S2): e7022

10. Kleinsteuber K, Castiglio C. Enfermedades Neuromusculares en Niños. Rev Médica Clínica Las Condes [Internet]. 2003 [Citado 20/05/2025];14(2). Disponible en: https://www.clinicalascondes.cl/clcprod/media/contenidos/pdf/MED_14_3/EnfermedadesNeuromuscularesNinos.pdf

11. Kleinsteuber K, Avaria M de los Á. Enfermedades Neuromusculares en Pediatría. Rev Ped Elec [Internet]. 2005 [Citado 20/05/2025]; 2(1): 52-61. Disponible en: https://www.revistapediatria.cl/volumenes/2005/vol2num1/pdf/enfermedades_neuromusculares.pdf

12. Birch JG. Orthopedic Management of Neuromuscular Disorders in Children. Semin Pediatr Neurol [Internet]. 1998 [Citado 20/05/2025]; 5(2): 78-91. Disponible en: https://pubmed.ncbi.nlm.nih.gov/9661242/

13. Skalsky AJ, Dalal PB. Common Complications of Pediatric Neuromuscular Disorders. Phys Med Rehabil Clin N Am [Internet]. el 1 de febrero de 2015 [Citado 20/05/2025]; 26(1):21–8. Disponible en: https://pubmed.ncbi.nlm.nih.gov/25479776/

14. Michel C, Collins C. Pediatric Neuromuscular Disorders. Pediatr Clin North Am [Internet]. 1 de febrero de 2020 [Citado 20/05/2025]; 67(1): 45–57. Disponible en: https://pubmed.ncbi.nlm.nih.gov/31779836/

15. Kennedy RA, Carroll K, McGinley JL, Paterson KL. Walking and weakness in children: A narrative review of gait and functional ambulation in paediatric neuromuscular disease. J Foot Ankle Res [Internet]. 2 de marzo de 2020 [Citado 20/05/2025]; 13(1): 10. Disponible en: https://pubmed.ncbi.nlm.nih.gov/32122377/

16. Dangouloff T, Boemer F, Servais L. Newborn screening of neuromuscular diseases. Neuromuscular Disorders [Internet]. 1 de octubre de 2021 [Citado 20/05/2025]; 31(10): 1070–80. Disponible en: https://pubmed.ncbi.nlm.nih.gov/34620514/

17. Bruno L. Guía de las enfermedades neuromusculares. Información y apoyo a las familias. ASEM[Internet]; 2008 [Citado 20/05/2025]. 217. https://sid-inico.usal.es/idocs/F8/FDO21378/guia_enfermedades_neuromusculares.pdf

18. Zenteno D, Torres G, Cox V. Indicaciones de rehabilitación respiratoria en niños y adolescentes con enfermedades respiratorias crónicas. Rev Chil Enferm Respir [Internet]. 2022 [Citado 20/05/2025]; 38(1):26–32. Disponible en: https://www.scielo.cl/scielo.php?script=sci_arttext&pid=S0717-73482022000100026

19. Rodríguez-Núñez I, Monsalve-Campos K, Bretti MJ, Otzen T, Zenteno D. La rehabilitación respiratoria infantil a la luz de la investigación cualitativa: Aquello que los números no pueden mostrar. Andes Pediatrica [Internet]. 1 de mayo de 2023 [Citado 20/05/2025]; 94(3): 392–400. Disponible en: https://www.scielo.cl/scielo.php?pid=S2452-60532023005000707&script=sci_abstract

20. Rodríguez Gómez Y, Romero Millares RL. Recomendaciones basadas en la evidencia para la permeabilización de las vías respiratorias de enfermos neuromusculares. Revista Cubana de Medicina Física y Rehabilitación [Internet]. 2020 [Citado 20/05/2025]; 12(2): 83-6. Disponible en: https://revrehabilitacion.sld.cu/index.php/reh/article/view/348

ISSN: 1561-3194 **RNPS:** 1877
Rev Ciencias Médicas. 2025; 29(S2): e7022

21. Giménez GC, Maidana E, Galeano S, Núñez D, Prado Antlagic F, Bach J. Decanulación en Paciente Pediátrica con Enfermedad Neuromuscular: reporte de caso. Medicina Clinica y Social [Internet]. 1 de mayo de 2021 [Citado 20/05/2025]; 5(2):106–10. Disponible en: https://www.medicinaclinicaysocial.org/index.php/MCS/es/article/view/167

22. Cifuentes-Silva E, Bueno-Buker D, Pastene-Maureira C, Pacheco-Valles A. Caracterización de fuerza, resistencia y función pulmonar en sujetos con enfermedades neuromusculares con el polimorfismo R577X del gen ACTN3. Andes Pediatrica [Internet]. 2023 [Citado 20/05/2025]; 94(4): 520–8. Disponible en: https://www.scielo.cl/scielo.php?script=sci_arttext&pid=S2452-60532023000400520

23. Mozzoni J. Pruebas cronometradas en pacientes deambuladores con enfermedades neuromusculares. AJRPT. [Internet]. 25 de octubre de 2022 [Citado 20/05/2025]; 4(3): 49-53. Disponible en: https://www.scielo.org.ar/scielo.php?script=sci_arttext&pid=S2618-40952022000300049&lng=es&nrm=iso

24. Martínez Costa C, Crehuá Gaudiza E. Nutrición en el niño con enfermedad neuromuscular. En: Torres Peral R, editor. Tratamiento nutricional en Pediatría [Internet]. Madrid, ERGON; 2023 [Citado 20/05/2025]. Disponible en: https://www.nutricionemocional.es/sites/default/files/libro_de_casos_clinicos_en_nutricion_enteral_pediatrica.pdf