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IMPACT OF DUCHENNE MUSCULAR DYSTROPHY ON PATIENTS' DAILY LIVES: A QUALITATIVE REVIEW OF FUNCTIONAL STATUS, QUALITY OF LIFE, AND PSYCHOSOCIAL ADJUSTMENT

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ABSTRACT

Duchenne Muscular Dystrophy (DMD) is a debilitating X-linked genetic disorder characterized by progressive muscle degeneration due to dystrophin deficiency, predominantly affecting males with an incidence of 1 in 3,500 live births. This qualitative review synthesizes literature and patient perspectives to explore the profound impact of DMD on daily life, encompassing functional decline, diminished quality of life (QoL), and psychosocial challenges. Through a pharmacy-focused lens, it examines therapeutic interventions—such as corticosteroids, exon-skipping drugs (e.g., eteplirsen), and emerging gene therapies—and their effects on symptom management, adherence, and long-term outcomes. Key findings highlight relentless physical deterioration leading to loss of ambulation, respiratory complications, and cardiac issues, often compounded by emotional distress, social isolation, and family burden. Pharmacotherapeutic strategies offer hope by mitigating progression and enhancing functionality, yet barriers like side effects, high costs, and access disparities persist. Drawing from clinical studies and lived experiences, this review underscores the need for integrated pharmacy care, including patient counselling and multidisciplinary support, to improve psychosocial adjustment and overall well-being. Ultimately, while DMD profoundly disrupts patient life, targeted therapies and holistic interventions can foster resilience and dignity.

KEYWORDS: Duchenne Muscular Dystrophy (DMD), Muscular Dystrophy, Quality of Life, Functional Status, Psychosocial Adjustment, Pharmacotherapy, Therapeutic Interventions, Patient Perspectives.

INTRODUCTION

Duchenne Muscular Dystrophy (DMD) is a severe, progressive genetic disorder caused by mutations in the DMD gene on the X chromosome, leading to the absence of dystrophin protein essential for muscle fibre integrity. This results in relentless muscle degeneration and weakness, typically manifesting in early childhood with symptoms like delayed motor milestones, frequent falls, and difficulty climbing stairs. DMD predominantly affects males, with an incidence of approximately 1 in 3,500 live male births worldwide, though carrier females may exhibit milder symptoms. Without intervention, patients often lose ambulation by adolescence and face life-threatening complications such as respiratory failure or cardiomyopathy by their 20s or 30s. Current therapeutic approaches, including corticosteroids, exon-skipping therapies, and emerging gene-based treatments, aim to slow progression and improve quality of life, underscoring the critical role of pharmacotherapy in managing this debilitating condition.

The clinical landscape of DMD has undergone a fundamental transformation over the past several decades, evolving from an acutely terminal pediatric condition into a chronic, multi-systemic disorder that spans into adulthood.[1,2,3] This shift is primarily attributed to significant advancements in standardized care protocols, including the widespread use of corticosteroids, improved cardiac management, and the implementation of home mechanical ventilation. As life expectancy has extended—with many patients now surviving into their third, fourth, or even fifth decades—the focus of clinical research and care has expanded beyond survival to encompass the complex, day-to-day reality of living with progressive muscle degeneration.[2,4] This review explores the day-to-day realities of DMD through a pharmacy lens, focusing on functional status, psychosocial adjustment, and therapeutic impacts.

IMPACT OF DUCHENNE MUSCULAR DYSTROPHY ON PATIENTS' DAILY LIVES

Duchenne Muscular Dystrophy (DMD) imposes profound, progressive challenges on patients' daily functioning, evolving from childhood mobility issues to severe, life-limiting impairments. Clinically, DMD results from dystrophin deficiency, leading to muscle fiber

necrosis, fibrosis, and weakness, with symptoms intensifying over time (Emery, 2002; Bushby et al., 2010).

In early stages, patients experience fatigue from morning to night, hindering routine activities like walking or climbing stairs. By adolescence, ambulation loss necessitates wheelchairs, exacerbating exhaustion and limiting independence. After 10 years of progression, DMD often advances to advanced stages where patients face relentless fatigue, respiratory decline, and cardiac complications, making even basic tasks arduous (Manzur et al., 2008).

Work and self-care become problematic: Hand stiffness and contractures impair fine motor skills, requiring assistance for dressing, eating, or writing. Dental issues, such as loose teeth and jaw weakness, complicate chewing and nutrition, increasing risks of aspiration or malnutrition (van den Engel-Hoek et al., 2014). Psychosocially, these burdens foster isolation, depression, and reliance on caregivers, significantly diminishing quality of life (QoL) as measured by tools like the Pediatric Quality of Life Inventory (Varni et al., 2005).

Therapeutically, corticosteroids and emerging therapies (e.g., exon-skipping drugs) may delay some declines, but side effects like weight gain or immunosuppression add layers of complexity (McDonald et al., 2018). Holistic pharmacy interventions, including counselling on adherence and supportive devices, are essential to mitigate these impacts and enhance adjustment. This progression underscores DMD's holistic toll, necessitating multidisciplinary care to preserve dignity and functionality.

FUNCTIONAL STATUS AND THE LONGITUDINAL PROGRESSION OF MOBILITY

The functional decline in DMD is characterized by several punctuated transitions that redefine a patient's autonomy. The hallmark of DMD is the relentless, progressive deterioration of skeletal, cardiac, and respiratory muscles due to the absence of the dystrophin protein.[2,5] This functional decline is not a uniform descent but a series of punctuated transitions that redefine the patient's relationship with their environment and their sense of autonomy. The disease typically progresses through four recognized clinical stages, each characterized by specific motor impairments and increasing levels of care dependency.[2,5]

The Early and Late Ambulatory Phases

In the early ambulatory stage, which generally encompasses children aged 5 to 7, the functional impact is often subtle yet pervasive. Muscle weakness manifests as a lack of strength and increased fatigue, cited as primary symptoms by 100% and 85.7% of ambulatory patient/caregiver dyads, respectively.[5,6] Children in this phase often adopt compensatory motor patterns, such as the Gowers' manoeuvre to stand from supine or walking on their toes due to calf pseudohypertrophy and tendon tightening.[6] Qualitative reports indicate that while these children can walk unaided, they are slower than their peers and frequently require intermittent wheelchair use during outings to manage exhaustion.[6] This early fatigue often creates the first social barriers, as children struggle to keep up with friends during play, a phenomenon described as an "emotional journey shaped by inevitable progression".[7,8]

As patients transition to the late ambulatory phase (ages 8 to 11), the focus shifts toward the imminent loss of gait. During this period, climbing stairs becomes a significant challenge for 57.1% of patients, and running remains problematic for 46.4%.[5,9] The physical effort required to maintain ambulation often comes at the cost of extreme fatigue, which begins to impact activities of daily living (ADLs).[5,6] Transfers, such as getting off the floor or in and out of a car, become increasingly difficult for approximately 35.7% of this group.[5]

The Non-Ambulatory Phase and Upper Limb Function

The loss of ambulation, typically occurring between the ages of 10 and 14, represents the most profound functional milestone in the DMD lifecycle.[5,10] In the non-ambulatory phase, the clinical focus pivots from lower extremity mobility to the preservation of upper limb function, which becomes the primary determinant of independence and quality of life.[4,11] Early-stage non-ambulatory patients often retain significant use of their hands and fingers, enabling them to use computers, touchpads, and mice.[4,12] However, as the disease progresses, weakness moves from proximal muscles (shoulders and elbows) to distal muscles (wrists and fingers).[11]

Upper limb impairment significantly impacts the patient's ability to perform essential tasks. Research using the Performance of the Upper Limb (PUL) assessment indicates that 77.8% of non-ambulatory patients face challenges in reaching for objects, 61.1% struggle with fine motor skills like turning pages or using utensils, and 55.6% cannot lift their arms above their heads.[5,11,13] The functional consequences are severe, with 83.3% of non-ambulatory individuals requiring total assistance for ADLs such as dressing and bathing.[5] Transfers also

become more complex, with 72.2% of non-ambulatory patients requiring mechanical aid or total caregiver assistance to get in and out of bed.[5]

Table 1: Functional milestones and reported impact in ambulatory vs. non-ambulatory DMD patients.

Functional Milestone	Ambulatory (Reported Impact)	Non-Ambulatory (Reported Impact)
Lack of Physical Strength	100.0%[5]	94.4%[5]
Fatigue/Tiredness	85.7%[5]	77.8%[5]
Climbing Stairs	57.1%[5]	Total Loss
Running/Fast Walking	46.4%[5]	Total Loss
Upper Body Reaching	42.9%[5]	77.8%[5]
Fine Motor Skills	60.7%[5]	61.1%[5]
Bed Transfers	35.7%[5]	72.2%[5]
Dressing Independence	53.6% (Requires Help)[5]	83.3% (Requires Help)[5]

Cardiorespiratory Involvement and Systemic Decline

Functional status in DMD is also defined by late-stage cardiorespiratory complications. Respiratory function begins to decline in the second decade, necessitating nocturnal non-invasive ventilation and, eventually, daytime support.[1,14] Cardiac involvement is nearly universal, with dilated cardiomyopathy and arrhythmias becoming more prevalent as patients reach adulthood.[4,14] Interestingly, prospective studies have found a negative correlation between upper limb movement and cardiac parameters such as septal MAPSE, suggesting that motor function and cardiac health are deeply intertwined.[4] Patients taking heart failure therapies, such as ACE inhibitors, have shown better overall mobility than those without such treatments, highlighting the importance of systemic management in preserving functional status.[4]

DIMENSIONS OF QUALITY OF LIFE: DIVERGENT PERSPECTIVES

Quality of life (QoL) in DMD is a multidimensional construct that encompasses physical health, psychological state, level of independence, and social relationships.[4,15] Assessing QoL in this population is challenging because traditional generic instruments often overemphasize physical mobility, which may lead to inappropriately low scores for individuals who have adapted psychologically to their condition.[2,3,15]

The Disability Paradox and Adaptation

A consistent finding across the literature is the "disability paradox": while physical health scores are significantly lower in DMD patients compared to the general population, mental health and overall life satisfaction scores are often comparable or even higher.[1,16] For example, a study of Dutch adult men with DMD found that while they scored significantly lower on SF-36 physical domains, their scores for mental health and role limitations due to emotional problems were equal to or higher than general population reference values.[1] Approximately 73% of these men stated their overall QoL was "(very) good," indicating a successful psychological adaptation to severe physical disability.[1]

This adaptation is often facilitated by a recalibration of personal goals and expectations.[3,4] Patients frequently report that they do not define themselves by their illness; instead, they focus on their remaining competencies, such as digital social interactions or academic achievements.[16,17] The "hammock lifestyle" theory suggests that life satisfaction may even improve after the loss of ambulation, as the constant anxiety and physical strain associated with trying to walk and the fear of falling are removed.[15]

Discrepancies in Proxy Reporting

One of the most critical issues in QoL assessment is the persistent discrepancy between patient self-reports and parent proxy-reports.[10,15] Parents and caregivers consistently rate the child's QoL lower than the child rates themselves.[10,15] This gap is particularly evident in the psychological and social domains, where parents may project their own grief, caregiver burden, and fears for the future onto the patient.[15,18] This underestimation by parents can have significant clinical implications, potentially influencing decisions regarding life-prolonging treatments or palliative care.[1,15]

Disease-Specific vs. Generic Assessment Tools

Generic tools like the EQ-5D are criticized for their inadequacy in capturing the nuanced experience of DMD, as their utility-based scores are heavily weighted toward physical mobility.[2,18] In contrast, disease-specific tools like the PedsQL 3.0 Neuromuscular Module and the newly developed DMD-QoL offer a more granular view of the challenges faced by this population.[3,4,18]

Table 2: Summary of key quality of life instruments used in DMD research.

QoL Instrument	Domains Assessed	Key Findings in DMD
SF-36 (Adults)	Physical and mental health, vitality, social functioning	Low physical scores; high mental health and vitality association with QoL[1]
WHOQOL-BREF	Physical, psychological, social, environment	Lower scores in social relationships, transport, and employability[1]
PedsQL 3.0	Neuromuscular disease, communication, family resources	Negative correlation between upper limb movement and total mobility scores[3,4]
DMD-QoL (14-item)	Physical, psychological, social impact	Adults score lower on physical subscales but higher on psychological subscales than children[18]
Ferrans PRQoL Model	Health, social/economic, psychological, family	Non-Hispanic patients reported significantly better PRQoL than Hispanic patients[19]

Socioeconomic and Cultural Factors

Recent quantitative analysis has highlighted that QoL is not solely determined by disease progression. The Ferrans et al. Patient-Reported Quality of Life (PRQoL) model emphasizes individual and environmental factors, such as education and ethnicity.[19] Research has revealed that non-Hispanic DMD male patients often report a statistically significant better PRQoL than Hispanic patients, suggesting that cultural barriers, language, and access to specific healthcare resources may play a hidden role in the lived experience of the disease.[19] Furthermore, financial impacts and the availability of healthcare services are cited as major themes in the "Other" domain of QoL, which are frequently missed by standard assessment tools.[15]

NEUROPSYCHIATRIC COMORBIDITIES AND THE DYSTROPHIN-BRAIN AXIS

The absence of dystrophin has profound implications for the central nervous system, leading to a spectrum of neurodevelopmental and neuropsychiatric challenges that significantly impact psychosocial adjustment.[20] Dystrophin is expressed in the brain in several isoforms (Dp427, Dp140, and Dp71), which are critical for neuroplasticity and synaptic function in the cerebral cortex, hippocampus, and amygdala.[20,21]

Oromotor Dysfunction and Dental Manifestations

The progressive nature of DMD significantly impacts orofacial and masticatory muscles. As the disease advances, specifically during the second decade of life, patients experience a notable decline in oromotor function. Dystrophin deficiency leads to a marked reduction in bite force and neuromuscular coordination of the jaw. Clinical studies indicate that patients often suffer from macroglossia (enlarged tongue) and reduced tongue motility, which interferes with the ability to fragment food into a safe bolus. This "day-to-day reality" of chewing difficulty increases the risk of choking and nutritional deficiencies.

The weakening of the perioral muscles causes a shift in the structural balance of the mouth, often resulting in lateral and anterior open bites. This skeletal change can lead to perceived tooth mobility (loose teeth) and dental crowding. Chronic open-mouth posture, often a result of respiratory weakness, further exacerbates gingival inflammation and poor oral hygiene.[20,22]

Prevalence of ASD, ADHD, and Cognitive Impairment

The prevalence of neurodevelopmental disorders in the DMD population is significantly higher than in the general population.[20,22,23] A retrospective chart review indicated an ADHD prevalence of 18.4% and an ASD prevalence of 12.7%, while other studies have reported even higher rates, with some cohorts showing up to 34% for ADHD and 31% for ASD-related symptoms.[22,24] These conditions are often comorbid; 63% of DMD patients with mental health concerns experience more than one diagnosis.[22]

Cognitive deficits are also prevalent, particularly in executive function, short-term memory, and processing speed.[20] These issues are not progressive but are stable developmental traits that complicate the management of the physical disease.[10,20] For example, a child with ADHD may find it difficult to comply with the necessary stretching exercises or the use of orthotic devices, leading to faster contracture development.[22]

Table 3: Prevalence and clinical impact of neuropsychiatric comorbidities in DMD.

Neuropsychiatric Condition	Estimated Prevalence in DMD	Clinical Impact
ADHD	18.0% - 44.0%[22, 24]	Impaired school functioning, poor treatment adherence[22]
ASD / Autistic Features	7.0% - 31.0%[21, 22, 24]	Difficulties with communication and

		social reciprocity[21,25]
Anxiety Disorders	24.0% - 29.0%[22, 26]	Fears of deterioration, social withdrawal[26, 27]
Depression	6.7% - 27.0%[22, 27]	Reduced engagement in rehabilitation, fatigue exacerbation[26, 27]
Intellectual Disability	~25.0%[22, 23]	Limitations in adaptive functioning and autonomy[23, 28]

The Behavioural Impact of Corticosteroids

While corticosteroids are the standard of care for prolonging muscle function and delaying the loss of ambulation, they carry a significant behavioural burden.[11,22] Approximately 70% of mental health concerns in DMD patients are reported to start after the initiation of steroid use.[22] The most common behavioural side effects include increased anger, aggression, and irritability, which affect 52% of the population.[22,27] These behaviours place an immense strain on the family unit and can lead to social isolation as parents struggle to manage the child's outbursts in public or school settings.[17,27]

Despite the high prevalence of mental health concerns, psychosocial services remain underutilized.[22,27] One study found that while 86% of patients had identified mental health needs, only 26% received counselling or therapy, and 50% were neither referred nor seen by a specialist.[22] This gap in care is often due to limited appointment times, a lack of specific guidelines for mental health in neuromuscular clinics, and cultural barriers.[22]

PSYCHOSOCIAL ADJUSTMENT: RESILIENCE AND SOCIAL PARTICIPATION

Psychosocial adjustment in DMD is the process by which patients and their families navigate the emotional and social demands of the illness.[7,20] This journey is characterized by both significant stressors and remarkable examples of resilience.[7,16]

Peer Relationships and Social Isolation

Social interaction is a primary area of concern throughout the DMD lifespan.[29] In childhood, physical limitations and fatigue make it difficult for boys to keep up with peers, often leading to social exclusion and bullying.[8,29] As they move into adolescence, social networks tend to shrink; the loss of independent mobility and the logistical challenges of wheelchair use make spontaneous socializing difficult.[20,27] Many patients report feeling "invisible" or being treated differently by others, which can lead to low self-esteem and social withdrawal.[30,31]

However, interactive situations with other young people and adult role models with DMD have been shown to have a positive impact on social skill development and resilience.[27]

Coping Strategies and the Family Context

Coping mechanisms are essential for successful adjustment to disease progression.[30,32] Adaptive strategies include acceptance, positive reframing, and problem-focused coping, such as independently seeking information about the disease.[26,33,34] In contrast, maladaptive strategies like avoidance, denial, or emotional suppression are linked to poorer mental health outcomes and reduced participation in care.[26,32]

The family environment is a powerful moderator of the patient's adjustment.[16,20] Close parental relationships, adequate family income, and a supportive connection to the community act as protective factors.[16] Conversely, high levels of caregiver stress and maladaptive parental coping can increase the risk of emotional and behavioural problems in the child.[20,26] Siblings also play a critical role; research indicates that having more siblings may actually improve transition readiness for young adults with DMD, perhaps by providing a broader social support network and fostering a sense of normalcy.[35]

The Emotional Journey of Inevitable Progression

The knowledge of DMD's natural history creates an "emotional journey" for patients and families.[7,36] Younger subjects are often more concerned with the physical implications of their disease, while older subjects voice greater concern about psychosocial issues and their shortened life span.[37] Despite these existential concerns, many older individuals maintain a "can-do" attitude and a sense of hope, often taking life "day by day".[7,38]

ENVIRONMENTAL FACILITATORS: ASSISTIVE TECHNOLOGY AND HOME MODIFICATIONS

Assistive technology (AT) and environmental modifications are the primary facilitators of autonomy for individuals with DMD.[39,40] These tools are not merely aids but "catalysts for independent living," allowing patients to transcend their physical limitations.[40,41]

Mobility Devices and Daily Autonomy

The transition to mobility aids is a major milestone. Manual wheelchairs and power scooters provide independence for those with sufficient upper body strength, while standing wheelchairs offer the added benefit of stretching hips, knees, and ankles, which can prevent

contractures.[40] Seat elevators in power chairs allow users to maintain eye-to-eye contact with peers, which is vital for self-esteem and social interaction.[40] Home modifications are equally essential. Accessible bathrooms with transfer seats, grab bars, and lowered toilet seats are critical for maintaining independence in personal hygiene.[42] Other modifications, such as lowered light switches, doorknob adapters, and the use of zippers instead of buttons, allow patients to perform daily tasks that would otherwise require caregiver intervention.[40,42]

Digital AT and Social Connectivity

For individuals with severe muscle weakness, digital assistive technology is a lifeline. Sip-and-Puff (SNP) devices, which use air pressure signals to control computer mice or keyboards, allow patients to engage in education, employment, and social networking.[40] Emerging technologies like brain-computer interfaces (BCIs), virtual reality (VR), and AI-driven devices are opening new avenues for communication and immersion in simulated real-life contexts, potentially reducing the sense of isolation.[39] As digital care becomes more integrated into the lives of those with disabilities, ethical considerations regarding privacy and human contact must be carefully addressed through user-inclusive design approaches.[43]

Table 4: Assistive technology categories and their impact on daily life in DMD patients.

Assistive Category	Technology/Modifications	Impact on Daily Life
Mobility	Power wheelchairs, seat elevators, standing chairs	Facilitates eye-level social interaction and community access[40]
Independence in ADLs	Grab bars, doorknob adapters, zipper clothing	Reduces caregiver burden for hygiene and dressing[40,42]
Communication	Sip-and-Puff devices, eye-trackers, social robots	Bridges the gap to education and remote social interaction[39,40]
Orthotics	Ankle-foot orthoses (AFOs), night splints	Prevents contractures and extends independent walking time[12, 40]
Home Environment	Ramps, widened doorways, lowered light switches	Enhances autonomy and safety within the living space[42]

NAVIGATING THE TRANSITION TO ADULTHOOD

The transition from adolescence to adulthood is a particularly challenging period for DMD patients, marked by shifts in medical care, education, and living arrangements.[30,35] This

transition is often focused on the move from pediatric to adult medical teams, but qualitative evidence suggests that achieving personal independence is of equal concern to patients.[14,35]

Education, Employment, and the Barrier of Care

Many young adults with DMD desire to pursue higher education and meaningful careers.[1,17] However, the transition domains where participants need the most help are education and employment (52.5%) and activities of daily living (57.0%).[35] The need for constant personal care services can be a significant barrier to employment, as many workplace environments are not equipped to accommodate the level of assistance required.[1] Transport and leisure activities also remain problematic, with limited accessible options restricting the social lives of adult men with DMD.[1]

Parental Anxiety and the Shift in Roles

For parents, the transition to independent living is a mixture of "anxiety and excitement".[44] Moving out of the family home to a college dorm or independent living facility requires the parent to relinquish the role of primary caregiver and trust third-party personal care assistants.[44] This transition is often fraught with fears about the quality of care and the potential for staff abandonment, yet it is also seen as a necessary step for the patient's individual growth and happiness.[44]

Palliative Care and End-of-Life Discussions

In the late stages of the disease, palliative care becomes increasingly important, focusing on symptom control and quality of life rather than just life extension.[14,45] Discussions about end-of-life (EoL) care, including the use of ventilators and the patient's wishes for their final days, are often avoided until a crisis occurs.[45] Qualitative research emphasizes the importance of initiating these discussions proactively and sensitively, respecting the wishes of those who may choose not to discuss EoL at all.[45] Palliative care should be integrated into the multidisciplinary team early on, ensuring that patients and families are informed about all available assistance and potential difficulties.[45]

TREATMENT GOALS: ALIGNING CLINICAL OUTCOMES WITH PATIENT NEEDS

As new molecular therapies and gene treatments are developed, it is crucial to ensure that the outcomes measured in clinical trials align with the goals of the patients.[9,11] The treatment goals of DMD patients shift significantly as the disease progresses.[5] Ambulatory patients

and their caregivers prioritized maintaining current functioning (71.4%), improving muscle strength (25.9%), and reducing fatigue (22.2%).[5,9] Their primary desire is "stabilization"—keeping the child as close to their current functional level as possible for as long as possible.[8]

In contrast, non-ambulatory patients and caregivers seek increased upper body strength (42.1%) and greater independence in ADLs (31.6%).[5,9] For these individuals, the ability to reach for a cup, use a computer, or adjust themselves in bed is more meaningful than the theoretical possibility of walking.[11] This divergence underscores the need for patient-centred measurement strategies, such as the PUL-PROM, which can capture small but significant changes in upper limb function that directly impact daily life.[11]

Table 5: Treatment goal categories for ambulatory vs. non-ambulatory DMD patients.

Treatment Goal Category	Ambulatory Focus	Non-Ambulatory Focus
Primary Physical Goal	Maintaining ambulation/strength[5]	Improving upper body reaching/fine motor[5]
Symptom Management	Reducing fatigue and falls[5,42]	Improving respiratory/cardiac stability[4,11]
Autonomy Goal	Keeping up with peers[8]	Independence in ADLs and transfers[5,9]
Expectation from Therapy	Stabilization of motor decline[8]	Slowing the loss of finger/hand function[11]

CONCLUSION

The day-to-day reality of Duchenne muscular dystrophy is a complex landscape defined by the interplay between progressive physical loss and psychological resilience. This qualitative review highlights several key areas for clinical improvement and future research.

First, the management of DMD must transcend the purely physical. The high prevalence of neurodevelopmental comorbidities and the significant impact of corticosteroids on behaviour require that neuropsychological screening and mental health services be integrated into standard multidisciplinary care.[20,22,27] Addressing these "invisible burdens" is critical for improving both patient QoL and caregiver resilience.[26]

Second, quality of life must be assessed using tools that are sensitive to the adaptation that occurs in chronic illness.[2,18] Clinical decisions should prioritize the patient's self-reported perspective, recognizing that their perception of well-being often remains high despite severe physical disability.[1,15] Clinicians must also be aware of the "disability paradox" and work to help parents align their perceptions with those of the patient to ensure patient-centred care.[10,15]

Third, functional outcomes in clinical research should be stage-appropriate. While delaying the loss of ambulation remains a primary goal, the preservation of upper limb function is equally important for those in the non-ambulatory stage.[9,11] Instruments like the PUL-PROM provide a valuable framework for evaluating treatments in later stages where traditional tests like the 6MWT are no longer applicable.[11]

Finally, the transition to adulthood requires a comprehensive, life-course approach that includes vocational, social, and palliative support.[14,35] By fostering independence through assistive technology and providing proactive, sensitive palliative care, the medical community can support DMD patients in living full, meaningful lives well into their adult years.[40,45] The "day-to-day reality" for these patients is not just one of decline, but of constant adaptation, strength, and the pursuit of a future where they are neither defined nor limited by their diagnosis.[7,17]

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