# GUIDE FOR ADULT NEUROLOGISTS

## Managing Young Adults with Duchenne Muscular Dystrophy (DMD)

**As an Adult Neurologist, you have a key role in providing care for these patients**

# DMD IS A RARE GENETIC DISORDER THAT CAUSES PROGRESSIVE MUSCLE DAMAGE AND DEGENERATION

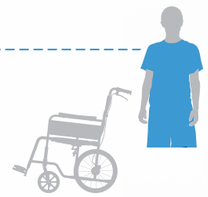
- Affects 1 out of every 3500 to 5000 newborn males worldwide[1-4]
- Caused by mutations in the dystrophin gene, resulting in absent or insufficient functional dystrophin[4,5]
- Characterised by progressive decline in muscle function, leading to loss of ambulation, respiratory and cardiac failure, and subsequent early death; once muscle is lost, it cannot be restored[2,6-8]
- Requires the need for adult multidisciplinary management, as improvements in the standard of care have led to patients living longer[9,10]

## DMD IS A CONTINUUM, WITH A SUCCESSION OF DISEASE MILESTONES

### INFANCY
Delayed motor milestones such as walking, delayed speech[2,6,11,12]

### EARLY CHILDHOOD
Unable to keep up with peers or climb steps[13]

### AROUND 2–3 YEARS
Rises from floor using Gowers' manoeuvre; does not walk smoothly (tip-toe walking), abnormal gait[6,14]



### UP TO 13 YEARS
Loss of independent ambulation (increasing use of wheelchair)[2,13,15]

### FROM 7 YEARS
Walking becomes increasingly difficult; may need a scooter or wheelchair after walking long distances[13]

### AFTER LOSS OF AMBULATION
Loss of self-feeding; orthopaedic, respiratory, and cardiac complications; respiratory support required[13]

### BY AROUND 20 YEARS
Upper body function is lost; patient becomes almost entirely dependent on a carer[13,16,17]

### 30 YEARS AND BEYOND
Respiratory and cardiac failure lead to early death[2,6,18,19]

## ADHERENCE IS CRITICAL TO DELAY DISEASE PROGRESSION

While there is no cure for DMD, it is important that patients stay on their current treatment regimens, including corticosteroids, before and after the transition of care.[9,19,20]



# ASSESSMENTS AND TREATMENTS TO MANAGE DMD PROGRESSION

## DMD ASSESSMENTS

As an Adult Neurologist, these assessments will help you manage disease progression and inform treatment decisions.

**AT LEAST EVERY 6 MONTHS**

### 6-MINUTE WALK TEST (6MWT)

Outcomes related to walking are highly relevant endpoints in ambulatory DMD patients because they measure the function of multiple muscle groups as well as cardiovascular activity.[21,22]

- 6MWT evaluates functional capacity in neuromuscular diseases and is a key outcome measure in clinical trials with ambulant patients with DMD

**AT LEAST EVERY 6 MONTHS**

### TIMED FUNCTION TESTS (TFTs)

TFTs reflect important milestones in patients with DMD, can help predict the likelihood of further disease progression, and have documented sensitivity to intervention with corticosteroids.[21,23]

- TFTs include time taken to climb or descend 4 stairs, stand from a supine position, and run/walk 10 metres

**AT LEAST EVERY 6 MONTHS**

### PERFORMANCE OF UPPER LIMB (PUL) SCALE

Assessment of upper limb strength is especially meaningful for non-ambulant young adults with DMD because they can perform important functional activities with limited distal motor function.[24-26]

- The PUL scale is sensitive to changes in upper limb strength over time and to differences in corticosteroid therapy

**AT EVERY VISIT**

### PATIENT QUALITY OF LIFE (QoL) ASSESSMENTS

QoL scales examine disease management in the context of participation in real-life situations and provide a more complete assessment of patient functioning beyond the medical aspects of DMD.[9,27]

- Various measures—including MAPA, ACTIVLIM, SF-36, WHOQOL-BREF, and DMD-QoL—assess ability to perform and frequency of participation in activities of daily life, as well as degree of meaningfulness experienced

ACTIVLIM=activity limitations for patients with upper and/or lower limb impairments; DMD-QoL=Duchenne Muscular Dystrophy-Quality of Life Measure; MAPA=Meaningful Activity Participation Assessment; SF-36=36-Item Short Form Health Survey; WHOQOL-BREF=abbreviated World Health Organisation Quality of Life Assessment.

## DMD TREATMENTS

**Corticosteroids** are the standard of care for preserving muscle strength and respiratory function.[6,9,28]

- Corticosteroid side effects should be monitored regularly; treatment should be continued unless side effects and risk outweigh evidence for benefit (or the patient chooses to discontinue)

Long-term high-dose corticosteroid use may lead to complications that include obesity, bone fractures, adrenal suppression and crisis, and hypertension. All patients taking steroids should be aware of the risk of an adrenal crisis and when to seek medical attention.[9]

**Mutation-specific therapies or gene replacement therapies** may restore dystrophin production.

**Symptomatic therapies** include those that attempt to[29]:

- Stabilise the muscle membrane and/or upregulate compensatory proteins
- Reduce the inflammatory cascade and/or enhance muscle regeneration

Some of these treatments have been approved by regulators, while others are in development, in regulatory review, or in clinical trials and might become available in the future.[6]

# YOUNG MEN WITH DMD REQUIRE MULTIDISCIPLINARY CARE TO MANAGE THEIR DISEASE

The Adult North Star Network (ANSN) notes that adults with DMD have complex medical needs that require a consensus-based standard of care that places more emphasis on nonambulant patients with advanced disease.[9]

## RESPIRATORY CARE

**EVERY 6 TO 12 MONTHS**

### ROUTINE ASSESSMENTS

- Monitoring should include measurement of forced vital capacity (FVC) and peak cough flow[9]
  - If FVC is <50% of predicted, referral to a specialist home ventilation clinic is recommended, even in asymptomatic patients
  - If there are symptoms of nocturnal hypoventilation, referral should be made regardless of FVC

### COMMONLY USED TREATMENTS

- Non-invasive ventilatory support or tracheostomy ventilation[9]

## CARDIOLOGY CARE

**ANNUALLY OR AS NEEDED**

### ROUTINE ASSESSMENTS

- Monitoring of cardiac function should include annual assessment by a cardiologist with imaging such as MRI or echocardiography[9]

### COMMONLY USED TREATMENTS

- Angiotensin-converting enzyme (ACE) inhibitors, mineralocorticoid inhibitors (eplerenone or spironolactone), and beta blockers[9]

## RENAL CARE

**ANNUALLY OR AS NEEDED**

### ROUTINE ASSESSMENTS

- Monitoring of renal function should include urea, electrolytes, and cystatin C[9]
  - If an abnormal result is found, a glomerular filtration rate (GFR) scan is recommended, with an urgent referral to a consultant nephrologist

### COMMONLY USED TREATMENTS

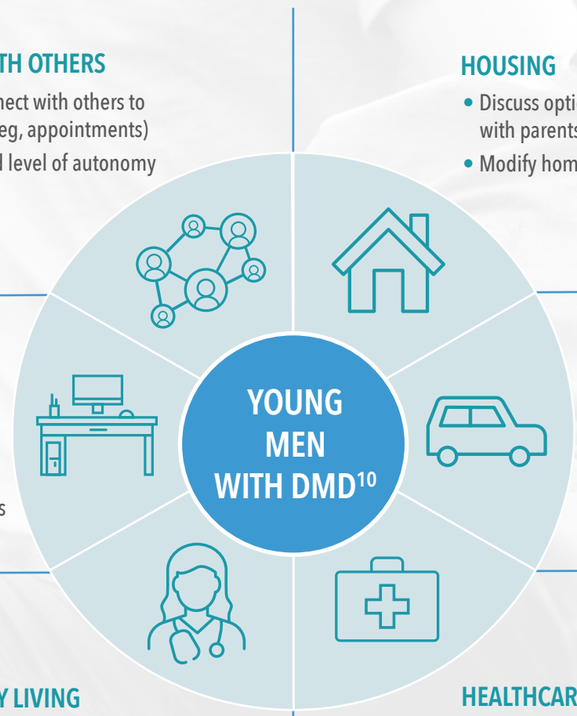
- Oxybutynin or tolterodine (may be considered for symptoms of sphincter dyssynergia)[9]



# YOUNG MEN WITH DMD REQUIRE ADDITIONAL SUPPORT AS THEY TRANSITION TO ADULTHOOD

Adults with DMD may have difficulty participating in the wider community and may be at risk of social isolation. Overcoming everyday barriers (such as physical access and transportation) may be challenging and require additional community support.[10,30,31]

## TRANSITION PLANNING FOR YOUNG MEN WITH DMD INVOLVES MULTIPLE COMPONENTS




### RELATIONSHIPS WITH OTHERS

- Develop skills to connect with others to manage own affairs (eg, appointments)
- Work towards desired level of autonomy and independence

### HOUSING

- Discuss options—independent living, living with parents, etc
- Modify home for accessibility and safety

### EDUCATION OR EMPLOYMENT

- Plan early for future vocation
- Consider classes online vs on campus
- Contact campus programmes for students with disabilities

### TRANSPORTATION

- Consider driving with vehicle modifications
- Modify family-owned vehicle
- Investigate accessible public transportation options

### ACTIVITIES OF DAILY LIVING

- Learn to hire and train personal care attendants
- Ensure respite for family caregivers

### HEALTHCARE

- Transition from paediatric to adult healthcare
- Move from family-centred to patient-centred provider interactions
- Assess the need for durable power of attorney for healthcare

Adapted from Birnkrant DJ et al. Lancet Neurol. 2018;17(4):347–361.

# As an Adult Neurologist

## Your knowledge and support are crucial as young men with DMD transition to adult care

## YOUNG MEN WITH DMD REQUIRE SPECIALISED CARE

- DMD is characterised by progressive decline in muscle function, leading to loss of ambulation, respiratory and cardiac failure, and subsequent early death; once muscle is lost, it cannot be restored[2,6-8]
- Patients should stay on their current treatment before and after the transition of care[9,20]
- The following assessments can help manage disease progression and inform treatment decisions: 6MWT, TFTs, PUL scale, patient QoL, and corticosteroid monitoring[6,9,21-28]
- Adult patients continue to require multidisciplinary care to manage their complex, progressive disease[9]
- As patients transition to adulthood, they require additional support with healthcare, housing, transportation, relationships, education/employment, and activities of daily living[10,30,31]

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