

# GUIDE FOR YOUNG MEN WITH DUCHENNE MUSCULAR DYSTROPHY (DMD)

*What you need to  
know about DMD and  
managing your condition*

**Empower yourself to make informed  
choices about your health**



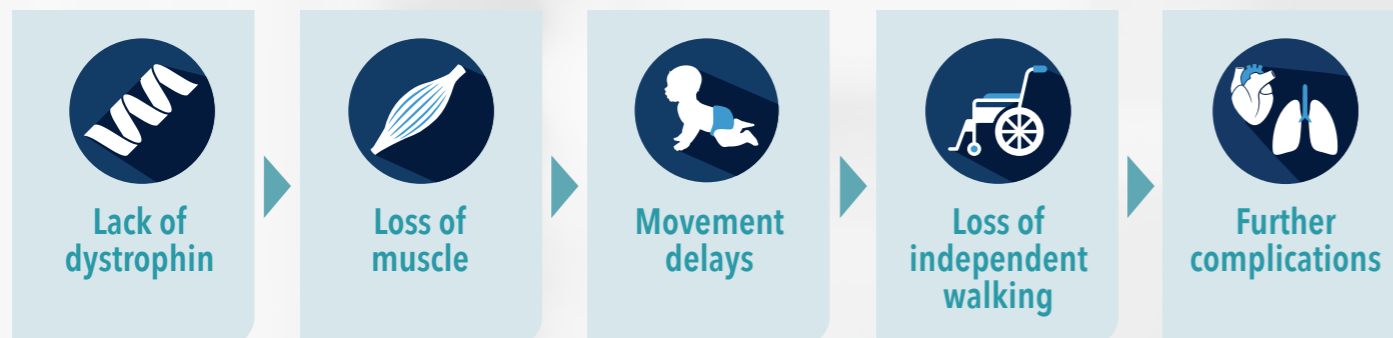
## DMD IS CAUSED BY MUTATIONS (CHANGES) IN THE GENE THAT MAKES DYSTROPHIN, A PROTEIN THAT KEEPS MUSCLES WORKING PROPERLY

In Duchenne muscular dystrophy (also called DMD or just Duchenne), there is a mutation in the gene coding for a protein called dystrophin. As a result of this mutation, the body cannot produce enough of this protein, making muscle cells more sensitive to damage. These muscle cells become replaced with scar tissue and fat, which causes the muscles to lose strength and results in difficulty moving.

### WITH DMD, A GENE MUTATION PREVENTS THE BODY FROM PRODUCING DYSTROPHIN



### DMD IS CHARACTERISED BY THE PROGRESSIVE LOSS OF MUSCLE FUNCTION



Muscle weakness starts in early childhood and gets worse over time. Over the years, heart and lung muscles become impacted.

## THE PROGRESSIVE NATURE OF DMD EXTENDS INTO ADULTHOOD

### LATE TEENS INTO ADULTHOOD



#### WEAKNESS OF MUSCLES IN THE RIB CAGE

- Breathing support required
- Greater likelihood for infections



#### WEAKNESS OF THE HEART MUSCLES

- Irregular heartbeats
- Impaired ability of heart to pump blood



#### IMPAIRED MUSCLES IN DIGESTIVE SYSTEM

- Constipation
- Diarrhoea



#### UPPER BODY FUNCTION LOSS

- Self-feeding difficulties

**STAYING ON YOUR PERSONALISED TREATMENT PLAN WILL BE THE BEST WAY TO DELAY DISEASE PROGRESSION.**

## TRANSITIONING TO ADULT DMD CARE REQUIRES ADULT SPECIALISTS

As you enter adulthood, there will be many changes in your life, including the care you receive for DMD. You will transition from paediatric to adult multidisciplinary care, where different adult specialists work together to provide comprehensive care. These specialists are better equipped to manage the progression of DMD as you grow into adulthood.

Transitioning to adult care begins with your Paediatric Neurologist, who will initiate and support this process until your transition is complete. Your new Adult Neurologist will be vital in providing you with adult DMD care. Adult Neurologists are experts in nervous system disorders and are well positioned to help you manage your neuromuscular disease as an adult.

### GET TO KNOW AND STAY IN TOUCH WITH YOUR ADULT CARE TEAM

The different adult specialists that provide care to you may not work in the same healthcare centre, so you may need to travel to different centres to receive multidisciplinary care. Even if your care as an adult feels less connected, it is important to remain actively engaged with your doctors during and after your transition to adult care to achieve independence and get the support you need.

If your Adult Neurologist does not work with a care coordinator (someone who can help facilitate communication among your healthcare team and help with referrals), ask for the name of the person to contact for questions, issues, or emergencies.

**AS YOU TRANSITION INTO ADULTHOOD, YOU WILL GAIN MORE RESPONSIBILITY FOR MANAGING YOUR OWN HEALTHCARE.**

## REGULAR VISITS TO ADULT SPECIALISTS ARE IMPORTANT FOR MANAGING YOUR HEALTH

As you grow into adulthood, you will have an adult treatment plan that includes seeing many different adult specialists. You may visit these specialists more frequently as well. Having them collaborate to create a treatment plan designed specifically for you will be the best way to maintain your health.

It is important to see these adult specialists at the following times:

Area of care	What this supports	Whom you should see and how often
Neuromuscular care	for nerve and muscle health	Adult Neurologist, your primary specialist for managing DMD, twice a year or as needed
Respiratory care	for lung health	Respiratory Specialist every 6 to 12 months or as needed
Cardiac care	for heart health	Cardiologist annually or as needed
Renal care	for kidney health	Nephrologist annually or as needed

Other important specialties may be part of your multidisciplinary care:

Area of care	What this supports	Whom you should see and how often
Musculoskeletal medicine and physiotherapy	preservation of movement	Orthopaedic Physician and/or Physiotherapist as needed
Nutrition and bowel health	for diet, weight, and digestive health	Nutritionist for diet and weight every 6 to 12 months; Gastroenterologist for digestive health as needed
Bone health	for bone weakness due to corticosteroid use	Bone Health Specialist annually if taking corticosteroids or as needed
Psychosocial care	for mental, emotional, social, and spiritual needs	Psychosocial Support Specialist as needed
Endocrinology	for hormones	Endocrinologist every 2 years to monitor testosterone levels or as needed
Speech and language therapy	for speech and/or swallowing difficulties	Speech Therapist as needed
Occupational therapy	for everyday activities	Occupational Therapist as needed
Clinical psychology	for mental health	Clinical Psychologist/Psychiatrist as needed
Social work or care coordination	for navigating social systems and/or care	Social Worker or Care Coordinator as needed

## TREATMENTS TO HELP MANAGE DMD PROGRESSION

While there is currently no cure for DMD, different therapeutic approaches can help manage symptoms. Treatments include:



- **Corticosteroids** for muscle strength and function



- **Mutation-specific therapies** or **gene replacement therapies** to restore dystrophin production



- **Angiotensin-converting enzyme (ACE) inhibitors** or **angiotensin receptor blockers (ARBs)** for heart disease



- **Physiotherapy** to help maintain muscles and joints

Regular evaluations with your healthcare team may include tests that assess your disease progression and determine whether additional action is needed.

Medications work effectively when the right doses are taken at the right time. Dosing may be based on body weight, so the dose may change as you continue to grow. Your healthcare team will check your body weight and tell you if dosing adjustments are needed with any medications you are taking.

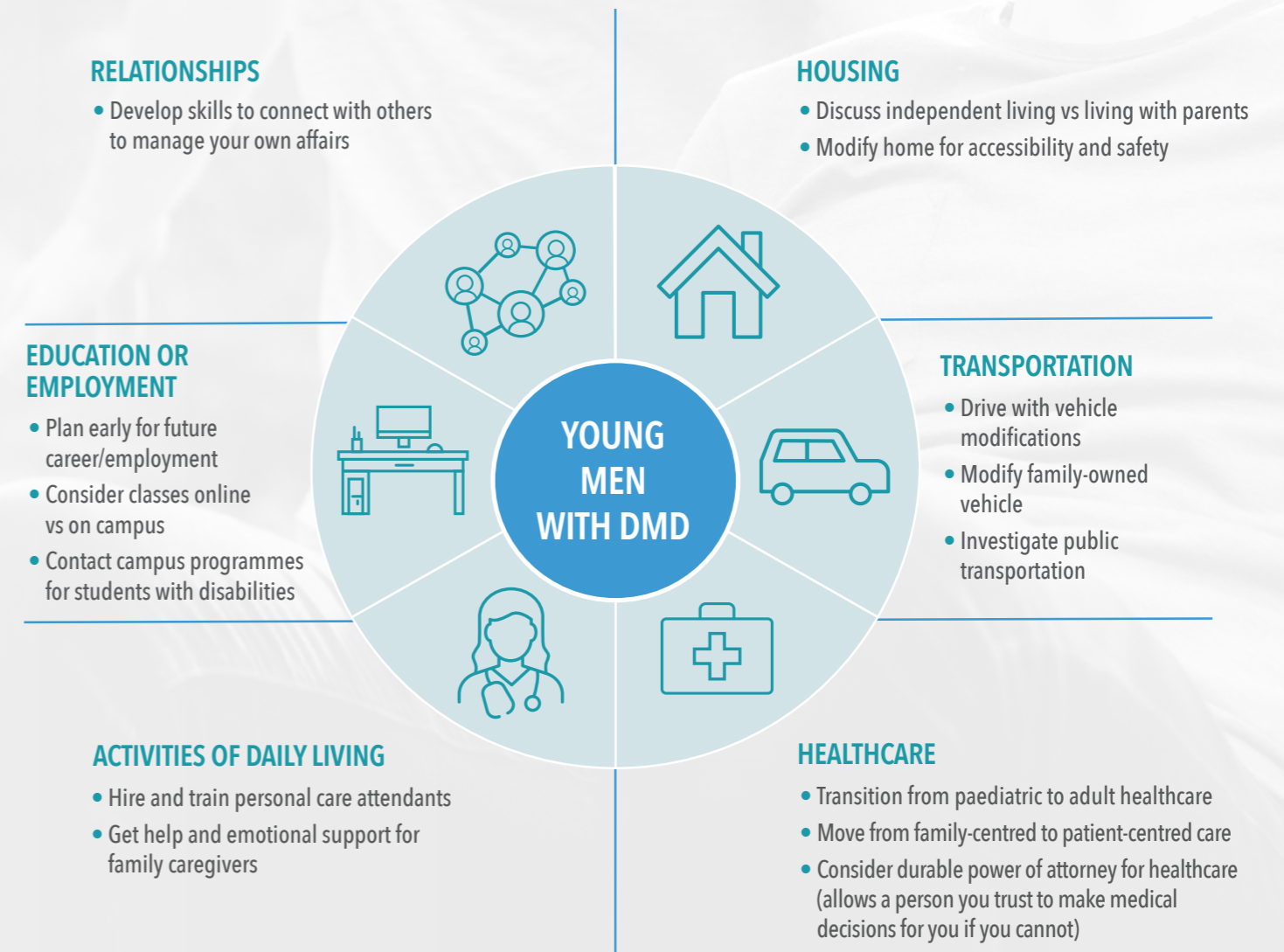
During and after the transition period, it is important to continue with specialist care and stay on treatment as prescribed by your adult healthcare team.

**IMPORTANT: DO NOT STOP OR MAKE A CHANGE TO ANY TREATMENT FOR YOUR DMD WITHOUT FIRST CHECKING WITH YOUR DOCTORS.**

## SUCCESSFULLY TRANSITIONING INTO ADULthood IS POSSIBLE WITH PLANNING AND COMMUNITY SUPPORT

Entering adulthood can be a rewarding time as you plan for a future that may include higher education, a job, or new relationships. As you prepare for new opportunities that require more independence, your transition plan should be built around your individual goals, as well as the community services and providers needed to achieve them. This may include social workers, educators, your adult care team, and family members.

### TRANSITION PLANNING INVOLVES MULTIPLE PARTS



Adapted from Birnkrant DJ et al. *Lancet Neurol.* 2018;17(5):445-455.

## DMD GLOSSARY

### ADULT CARE TEAM

Different adult specialists who work together to provide comprehensive care

### ADULT MULTIDISCIPLINARY CARE

A treatment plan that includes seeing many different adult specialists

### ADULT NEUROLOGIST

Medical expert in nervous system disorders; can help manage neuromuscular disease in adults

### ANGIOTENSIN-CONVERTING ENZYME (ACE) INHIBITORS

Treatments for heart disease

### ANGIOTENSIN RECEPTOR BLOCKERS (ARBs)

Treatments for heart disease

### CARDIAC CARE

Medical specialty focused on heart health

### CARE COORDINATOR

Someone who can facilitate communication among the healthcare team and help with referrals

### CLINICAL PSYCHOLOGY

Medical care focused on mental health

### CORTICOSTEROIDS

Treatments to help improve muscle strength and respiratory function

### DUCHENNE MUSCULAR DYSTROPHY (DMD)

A genetic disorder characterised by a progressive loss of muscle function

### DYSTROPHIN

A protein that strengthens muscles and protects them from injury

### ENDOCRINOLOGY

Medical specialty focused on hormonal monitoring

### FAMILY-CENTRED CARE

Families and healthcare providers working together as a team to develop a treatment plan that best addresses a patient's wants and needs

### GASTROENTEROLOGIST

Medical specialist focused on digestion and bowel health

### GENE REPLACEMENT THERAPIES FOR DMD

Treatments to help restore dystrophin production

### MUSCULOSKELETAL MEDICINE

Medical specialty focused on movement

### MUTATION-SPECIFIC THERAPIES FOR DMD

Treatments to help restore dystrophin production

### MUTATIONS

Changes in genes; DMD is caused by mutations in the dystrophin gene

### NEUROMUSCULAR CARE

Medical specialty focused on nerve and muscle health

### NUTRITIONIST

Medical specialist focused on proper eating and weight

### OCCUPATIONAL THERAPY

Medical specialty focused on facilitating everyday activities

### PAEDIATRIC NEUROLOGIST

Expert in nervous system disorders in children; can initiate and support the process of helping the transition to adult care and an adult treatment plan

### PATIENT-CENTRED CARE

A carefully planned treatment approach that focuses on a patient's individual goals and ability to independently manage healthcare

### PHYSIOTHERAPY

Medical specialty focused on muscle and joint function

### POWER OF ATTORNEY

Legal authority that allows a trusted person to make medical decisions when a patient cannot

### PSYCHOSOCIAL CARE

Medical care focused on mental, emotional, and spiritual needs

### RENAL CARE

Medical specialty focused on kidney health

### RESPIRATORY CARE

Medical specialty focused on lung health

### SOCIAL WORK

A profession that helps navigate social systems and coordination of care

### SPEECH AND LANGUAGE THERAPY

Medical specialty that addresses speech and/or swallowing difficulties

### TRANSITION FROM PAEDIATRIC TO ADULT HEALTHCARE

The process of preparing patients to successfully manage their own care and providing support as they transition into adulthood

**NOTES**

**ABOUT DMD**

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**TRANSITIONING TO ADULT DMD CARE**

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**PERSONALISED DMD TREATMENT PLAN**

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**QUESTIONS TO ASK YOUR HEALTHCARE TEAM**

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*Preparing for  
adulthood includes  
planning for your adult  
healthcare needs*

You are not alone in your transition to adult DMD care, but your active involvement is vital for achieving independence and getting the healthcare support you will need.

- DMD is characterised by the progressive loss of muscle function
- Adult multidisciplinary care is important to help you manage your DMD
- Staying on your personalised treatment plan will be the best way to delay disease progression
- Your transition plan should be built around your individual goals, as well as the community services and providers needed to achieve them