

Duchenne Muscular Dystrophy (DMD)

Processing a new diagnosis

A diagnosis of DMD can be a huge shock for parents, siblings, extended family members and friends. It is normal to feel an overwhelming mix of grief, confusion, anxiety, loneliness and helplessness as your family's life has been forever changed. Everyone will have different ways of getting through this time but it's important to know that help and support are available for you, your child and family. Hear advice on processing your diagnosis from a community member with shared experience.

The video is available at <https://youtu.be/A9XM71knDVw>.

Getting help processing a new diagnosis

There are many ways you can seek support as you process a new diagnosis:

- A psychologist is a university-qualified health professional who can help you talk about your thoughts and feelings to understand and cope with the challenges you and your family are facing. Visit our page on **Psychology** for more information.
- A counsellor is a trained professional who can help you talk about and work through problems. Visit our page on **Counselling** for more information.
- A social worker can provide information and support to people experiencing a range of issues including family problems, anxiety, depression, crisis and trauma. Visit our page on **Social Work** for more information.
- A genetic counsellor can help you understand how DMD is inherited and whether there are any implications for other members of your family. Your specialist will be able to advise whether a genetic counsellor is required. Visit our page on **Genetic Counselling** for more information.
- Your state or territory neuromuscular organisation can provide support, advice and information about living with DMD. Visit our page on **state and territory neuromuscular organisations** for more information.
- Your GP can talk to you about a mental health plan and how you can use this to help you access the support and services you need. Visit our page on **Wellbeing** for more information.
- Other people and families living with DMD have also experienced the emotional rollercoaster that comes with a diagnosis of DMD and are able to understand exactly how you're feeling. You are not alone. Connect with other people and families living with DMD in The Loop Community on our **Forum**.

How to talk to your child and others about a new diagnosis

Having a conversation with your child about their diagnosis is not easy. In fact it will probably be one of the hardest talks you will have. But, like all difficult conversations, it is important and necessary.

There are a lot of reasons why parents may not want to have this conversation, mostly centred on wanting to protect their child.

But avoiding the topic with your child is not helpful. Children are often aware of the differences between themselves and their siblings and/or peers and may hear their condition being discussed during medical appointments.

There is no right time to have this conversation. Research suggests the earlier you talk to your child about their condition, the more natural the conversation will become. The important part is to pick a time and commit to it.

So it is crucial that you have a role in your child learning about their diagnosis. This will enable you to support your child as they process the information and to be on hand to answer any questions your child may have. Together, you can learn about

the journey ahead.

It will also allow you to:

- provide answers to questions in an age appropriate way
- correct any misinformation your child has heard or read
- show your child that you have faith in their ability to handle difficult conversations.

Ultimately, these conversations are an important step in providing your child with the tools to succeed, navigate their world and develop independence and self-advocacy skills.

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Some important do's and don'ts:

- Do be positive but also realistic
- Do reassure them that they have done nothing wrong and this is not their fault
- Do tell them they will do many wonderful things in their lives – they may just do them differently
- Do tell the truth. Answer all their questions.
- Do consider your language. Try to avoid negative or emotive words, such as 'suffering from a condition'.
- Do use daily living examples that they can relate to.
- Don't avoid answering questions and don't shut the conversation down.

If you are having difficulty starting the conversation, try asking a couple of questions such as 'How are you feeling today?' or 'Do you know why you are tired?'

Remember, it is okay to be upset during the conversation but try to avoid breaking down as this will only cause your child greater distress. Children are resilient and they generally handle information of this nature far better than adults.

Understanding DMD and how it's Diagnosed

About DMD

DMD is the most common form of childhood muscular dystrophy (a group of diseases that cause your muscles to become progressively weaker). The symptoms of DMD are usually first noticed in early childhood, often between the ages of two and five years. DMD mostly affects boys but, in rare cases, can also affect females. About 1 in 5,000 newborn boys are affected by DMD.

What causes DMD?

DMD is caused by a change (also called a mutation) in a gene. The gene affected in DMD helps produce a protein (proteins are the building blocks of our muscles, organs and other tissues) called dystrophin. Dystrophin is usually found in our muscle cells. It works together with other proteins to strengthen muscle fibres and is important for forming healthy muscles. It also protects muscles from injury as they contract and relax and helps repair damaged muscle cells. People with DMD don't produce dystrophin. Without dystrophin, muscle cells are more easily damaged and can't work properly, causing weakness of skeletal, respiratory and cardiac muscles.

Skeletal muscles enable us to move our body and to stand and sit. Initially a child with DMD may have difficulty getting up from the floor, running, jumping and going up stairs. Respiratory muscles are those that are used for breathing and coughing. As DMD progresses physiotherapy or a cough assist machine can be used to assist in clearing mucus from a chest infection. Medications and scans of the heart are implemented to monitor cardiac muscle weakness.

How is DMD diagnosed?

Diagnosing DMD can be complex and involve a number of steps. Children can be diagnosed at any age but are usually diagnosed between the ages of three to five years.

Parents are usually the first to notice signs of DMD in their child, including:

- Experiencing delays in their overall motor development such as being slower to sit, stand or walk

- having a waddling type of walk
- being clumsy or falling over often
- finding it hard to run, jump or climb stairs
- needing help to get up from the floor and may “walk” their hands up their thighs in order to stand up (known as Gower’s sign)
- walking on their toes
- having larger than normal calf muscles (muscles at the back of the lower leg)
- showing signs of learning or communication difficulties, such as a delay in learning to speak.

A child showing signs of DMD should be assessed by their doctor and referred to a paediatrician (a doctor who specialises in health and diseases in children) for further tests.

The doctor may request blood tests to check the amount of an enzyme called creatine kinase (CK) in your child’s blood. CK often leaks from damaged muscle cells into the blood. High levels of CK in the blood can suggest a muscle problem but does not confirm DMD.

If creatine kinase levels are high, your child will be referred to a paediatric neurologist (a doctor who specialises in childhood brain and nerve conditions) at one of the major children’s hospitals. A geneticist (a doctor who specialises in genes) may also be involved for genetic testing. DMD is diagnosed with a further blood test to check whether there is a fault in the dystrophin gene. This test is usually arranged by a Neurologist or Geneticist. This can be a 2 stage test with initial results taking 8 – 12 weeks and a further 16 weeks for more detailed testing. This test can be done through the public hospital system at no cost but will involve a fee if done privately. Further information about genetic testing can be found on the [Healthdirect website](#). During this time your child may be asked to see a specialist physiotherapist to test their movement. Visit our page on [Physiotherapy](#) for more information.

The genetics of DMD

DMD is a genetic disease, meaning it is caused by an error in one of our genes – the dystrophin gene. In DMD, some of these mistakes are inherited (passed down from parents to their children) and there may often be a family member who has the condition. But gene changes can also happen randomly in the child (spontaneous mutations) with no family history.

DMD is inherited in an X-linked pattern because the dystrophin gene that can carry a DMD-causing mutation is on the X chromosome. Every boy inherits an X chromosome from his mother and a Y chromosome from his father, which is what makes him male. Girls get two X chromosomes, one from each parent. Each son born to a woman with a dystrophin mutation on one of her two X chromosomes has a 50 percent chance of inheriting the flawed gene and having DMD. Each of her daughters has a 50 percent chance of inheriting the mutation and being a *carrier*. Carriers may not have any disease symptoms but can pass the gene on to their future children. Most carriers have no symptoms of DMD however some carriers will experience symptoms such as muscle pain or weakness and are at a higher than average risk of developing heart problems. Carriers should undergo a complete cardiac evaluation in late adolescence or early adulthood, or sooner if symptoms occur, and should be evaluated every 5 years.

You can read more about the [genetics of DMD](#) by Save Our Sons Duchenne Foundation.

For more detailed information about how DMD is inherited visit [Your Genes, Your Health](#).

Long-term outlook

Living with DMD usually means life, and the future, looks different from what you had planned. However children and adults with DMD can lead fulfilling, rewarding lives just like everyone else. School, friendships, sports and recreation, arts, university, work and travel are all possible when you have DMD.

Inevitably the lifespans of people with DMD are shortened, due to breathing complications and heart muscle weakness that develops over time. The good news is that due to greater understanding of the condition and international consensus on standards of care, many people are surviving well into their third decade of life (mid to late 20’s). Increasing numbers of Australian men with DMD are living into their fourth and even fifth decade (40 – 50 years) and have completed school, studied at university, worked, loved, bought a house and/or travelled.

For more information about living with DMD, overcoming some of the day-to-day challenges and where to get the right support:

- [Living Life](#)
- [Counselling](#)
- [Social Work](#)

Understanding and planning for changes

Learning about DMD and surrounding yourself with the right healthcare providers, services and support can help you feel more in control of what lies ahead. Although every person's journey with DMD will be unique, the information below will help you understand this journey and how to prepare and plan for changes in the future.

What are the main symptoms of DMD?

The main symptom of DMD is muscle weakness and loss of muscle bulk that develops very slowly over time and can cause a range of issues, including:

- Movement
 - Difficulty moving, walking, running, climbing stairs, eventually requiring a manual wheelchair, then a power wheelchair and help with everyday activities such as getting in and out of bed, showering and eating a meal
- Muscle tiredness and cramps
- Tightened muscles (contractures)
- Posture
 - Weakness of the muscles that keep our backs straight and upright, leading to lordosis ('sway back' or increased curve in the lower back), scoliosis (curved spine) and the need for specialised seating for extra support
- Breathing
 - Breathing muscles gradually get weaker
 - Unable to cough effectively
 - Increased risk of serious lung or chest infections
- Heart
 - Heart enlargement (cardiomyopathy)
 - Irregular heart rate

How does DMD progress?

Doctors often use key stages or milestones to describe the progression of DMD, which are outlined below. Further information about the stages of DMD and how it progresses is available in [A Guide for Families-The Diagnosis and Management of Duchenne Muscular Dystrophy](#).

The stages can overlap and might be somewhat blurred when your child moves from one stage into the next. However it is useful to use the stages to identify the kind of care and interventions recommended at any particular time and what to expect of your care team.

Transitioning between stages and adapting to new milestones or challenges can be stressful. Seeking support for your child or yourself from a psychologist, counsellor or social worker can be useful at these times. These professionals can be incredibly helpful if you, your child or other members of the family are having a tough time or struggling with negative thoughts and feelings. Learn more about what services are available and how to access them in our [Psychology](#), [Counselling](#), [Social Work](#) and [Wellbeing](#) pages.

1. Ambulation Stage (Early Ambulation):

Refers to the period of time when your child remains walking and gains motor skills such as climbing stairs. They may have some difficulty getting up off the floor and may use furniture to help themselves up. You may also notice that your child walks on their toes. During this time the child's large calves (pseudohypertrophy) will become noticeable. However, your child can and should be encouraged to participate in activities with their peers being mindful that they should not exert themselves to the point of fatigue. During this early phase, steroids may be started to improve tiredness, decrease falls, maintain upper body strength and ability to walk, and preserve lung function.

2. Plateau Phase (Late Ambulatory):

Refers to the period of time when your child does not appear to be gaining any new motor skills such as being able to pedal the wheels of a bicycle. They may have trouble keeping up with their classmates, particularly during play and sport. They may become more easily tired after a busy day at kindergarten or school which leads to falls. This is a good time to speak with the school about how your child can remain involved in all school activities.

3. Transition Phase (Early Non Ambulatory):

Refers to the period of time when your child/teenager begins to lose the ability to walk independently and starts to use a manual wheelchair or electric scooter for longer walks and excursions, due to muscle fatigue and extreme tiredness. It is not recommended that a child pushes themselves in a manual wheelchair as the energy demand for this is high and upper limb strength is starting to decline during this phase. Increased falls may become apparent. You may also notice that your child begins to withdraw from their friends as everyday activities become a little harder. It is important at this stage that family, friends and educators work together to maintain social inclusion. Visit our page on [Friendships](#) for more information. For resources for teachers see our page for [Educators](#).

4. Non-Ambulation Phase (Late Non Ambulatory)

Refers to the period of time when a youth becomes dependent on others for activities of daily living and uses a power wheelchair for mobility. It is important that the wheelchair provided is correctly prescribed by an allied health professional to provide correct sitting position and posture support to prevent future issues. Though many activities of daily living can become difficult, it is important that independence is maintained, as much as possible. Working with an occupational therapist can assist with this. Visit our page on [Occupational Therapy](#) for more information.

Community Advice

Hear from a community member who has walked the path before you.

The video is available at https://youtu.be/_6oJ9Mc0UO8.

Life stuff

To find out more about living life with a neuromuscular condition and to access stories and peer-advice from the community, visit our [Living Life](#) section.

Where to find more information about living with DMD

[Save our Sons Duchenne Foundation](#)

This organisation's website provides information on Duchenne genetics, what to do after diagnosis, living with Duchenne, education and behaviour and steroids and endocrine issues.

[Duchenne Muscular Dystrophy fact sheet](#)

From Muscular Dystrophy New South Wales, 2017. This fact sheet includes considerations for future planning.

[What is Duchenne Muscular Dystrophy? A Guide for Parents and Families](#)

From Muscular Dystrophy Association, United States, February 2019. This guide includes what are the signs and symptoms of DMD, what should I know about DMD, how is DMD treated and a glossary of terms.

[Learn to Live with Neuromuscular Disorders \(NMD\): A Message for Parents](#)

The Duchess of Kent Children's Hospital at Sandy Bay, August 2017

[A Guide for Families - The Diagnosis and Management of Duchenne Muscular Dystrophy](#)

This guide is based on the care considerations for DMD developed and endorsed by international experts. It summarises the medical care guidelines in plain language for people and families affected by DMD. The family guide is available in other languages through the [TREAT-NMD Neuromuscular Network](#).

Care considerations for Duchenne Muscular Dystrophy

Below are links to the more detailed care considerations on which the family guide above are based. Although these are

written in medical terms they may be useful for discussions with specialists, doctors, nurses and allied health professionals.

- **Part 1: diagnosis, and neuromuscular, rehabilitation, endocrine, and gastrointestinal and nutritional management**
- **Part 2: respiratory, cardiac, bone health, and orthopaedic management**
- **Part 3: primary care, emergency management, psychosocial care, and transitions of care across the lifespan**
- **Care Considerations *Pediatrics* Supplement** The *Pediatrics* supplement, funded by the Centers for Disease Control and Prevention, United States, is a set of 13 articles that expands on the clinical care for each area, outlined in the *Lancet Neurology* articles. The supplement provides new resources, including a toolkit to help individuals with DMD transition into adult medical care. These articles and resources can help families and providers manage DMD care. Also included is an article on evaluation of the care considerations, with key elements of care that clinics can measure.

International organisations

Parent Project Muscular Dystrophy (US)

You can find an excellent range of resources and information about Duchenne muscular dystrophy, including teachers guides, classroom resources, videos, downloadable books, care guidelines and transition planning resources.

Muscular Dystrophy Association (US)

This website has excellent information about muscular dystrophy and neuromuscular conditions. You can find a full list of neuromuscular conditions, symptoms, causes and care options, as well as use the clinical trial finder tool.

Duchenne UK

This website has an extensive range of information for families living with DMD. You can download a DMD Family Folder, standards of care, emergency care advice and guides for employers, teachers and family/friends.

World Duchenne Organisation

World Duchenne Organisation is a worldwide organization dedicated to finding a cure and viable treatments for DMD, to promoting good standards of care, and to informing parents around the globe.

Support for DMD

Steroids and DMD

The following information is from the **Save our Sons Duchenne Foundation website**.

Steroids are proven to slow the progression of DMD, but they can cause a long list of worrying side effects, which makes the decision of 'if and when to start them' a tough one. It is important that you discuss any concerns you have with your doctor before making the decision to start steroid therapy.

Steroids have been prescribed in DMD for over 20 years and more than 90 percent of boys in Australia now take them. The types of steroids used are corticosteroids (also known as glucocorticoids). They are not the same as the anabolic steroids used by some body builders. It is thought that corticosteroids help in DMD due to their anti-inflammatory properties and by improving muscle regeneration.

Prednisolone and Deflazacort are the two types of corticosteroid prescribed for DMD. They each have pros and cons, which can be discussed with your doctor, but since prednisolone is much cheaper, this is usually tried first.

Steroids are usually started sometime between the ages of four and six, when motor skills have stopped improving, but have not yet started to decline. There is recent evidence that starting earlier is more effective and side effects are usually less problematic. Remember, 'one size does not fit all' and some children may not respond well to steroids and/or experience side effects which outweigh the benefits of the therapy.

Further information on Steroids and DMD

Steroids and Duchenne Muscular Dystrophy (DMD), Muscular Dystrophy UK

Care considerations for DMD

The Care Considerations for management of DMD provide detailed explanations about the optimal care that should be provided for a person living with DMD. These Care Considerations include:

- Steroid management-regimens, dosing, and side effects

- Endocrine management-growth, puberty and adrenal monitoring
- Bone health-monitoring and treatment of brittle bones
- Orthopedic management for monitoring for scoliosis and fractures
- **Physiotherapy** for maintaining physical strength and function, stretching programs and prevention of respiratory complications, such as the use of manual cough techniques when you get sick and cough-assist machines
- **Occupational therapy** to help with activities of everyday living and equipment and support for accessibility to home, school and the community
- Respiratory management for monitoring of breathing muscles including sleep studies and monitoring for need to use non-invasive ventilation
- Cardiac management for monitoring the heart including yearly check-ups and medication review
- Gastrointestinal management for seeing a **dietitian** for advice about nutrition, speech therapist for swallowing and other gastrointestinal issues
- **Psychology** and **counselling** to support mental health and adjusting to life with DMD
- Considerations for surgery
- Emergency care considerations and
- Transition of care through adulthood

Although this list might seem long and daunting, the Care Considerations are important to familiarise yourself with as a good starting point when talking to you or your child's medical team. Ask your child's doctor or local neuromuscular organisation about any of the information in the Care Considerations that you don't understand or need further explanation. **A Guide for Families-The Diagnosis and Management of Duchenne Muscular Dystrophy** is an easier-to-read summary of the Care Considerations.

Imperatives for Duchenne MD – A Guide for Providers is a single-page snapshot of the key components of comprehensive care for children with DMD described in the Care Considerations. This concise summary of care from diagnosis through early adolescence can be helpful for medical providers who may not be familiar with DMD.

The Adult Imperatives for Duchenne MD – A Guide for Providers is an excellent summary of the key components of care across late adolescence and adulthood which may be helpful for medical providers unfamiliar with caring for adult men living with Duchenne.

DMD Registry

The **Australian Neuromuscular Disease Registry (ANMDR)** aims to establish a database of all patients, children and adults in Australia diagnosed with neuromuscular disease. By creating this database, they hope to form a network of patients, clinicians, researchers, and industry to further research into neuromuscular diseases, ease the process in finding and selecting participants for clinical trials, and create an additional support for patients and their families. Joining the Registry is entirely voluntary and Australian families affected by DMD are encouraged to **register** or contact the Registry for more information via **anmdr@mcri.edu.au**.

Other helpful support services

The video is available at **https://youtu.be/BDaCNsd9R_s**.

- Your state or territory neuromuscular organisation can provide information and advice on what support they can offer such as local support groups, camps, programs, services in the local area, access to cough assist machines, advocacy or assistance in times of crisis. They can also provide an ear to listen if you need someone to talk to or guide you to get the assistance you are needing. Visit our page on **state or territory neuromuscular organisations** for more information.
- Seeing a psychologist, counsellor or social worker can be incredibly helpful if you, your child or other members of the family are having a tough time or struggling with negative thoughts and feelings. Learn more about what services are available and how to access them here. Visit our pages on **Counselling, Social Work** and **Wellbeing** for more information.

- Living with DMD may mean that you or your child may need some assistance for everyday activities. Find out more about how a disability support worker could help and how to engage this type of support. Visit our page on [**Disability Support Workers**](#) for more information.
- Other people and families living with DMD have also experienced the emotional rollercoaster that comes with a diagnosis of DMD and are able to understand exactly how you're feeling. You are not alone. Connect with other people and families living with DMD in The Loop Community on our [**Forum**](#).

Medical Alert Card

Muscular Dystrophy Queensland has developed a [**Medical Alert Card for DMD**](#). A Medical Alert Card can help you communicate your care needs in a medical emergency situation. If you would like a Medical Alert Card to keep in your wallet or purse, please call (07) 3243 9700 or complete the form on the Muscular Dystrophy Queensland [**website**](#).

How to have better conversations when communicating your needs

To learn how to have better conversations when communicating your needs, visit the following pages:

[**Employers**](#)

[**Educators**](#)

[**Living Life: Education**](#)

About DMD

Duchenne Muscular Dystrophy fact sheet

From Muscular Dystrophy New South Wales, 2017. This fact sheet includes considerations for future planning.

What is Duchenne Muscular Dystrophy? A Guide for Parents and Families

From Muscular Dystrophy Association, United States, February 2019. This guide includes what are the signs and symptoms of DMD, what should I know about DMD, how is DMD treated and a glossary of terms.

Save our Sons Duchenne Foundation

This organisation's website covers topics including genetics, diagnosis, living with the condition and steroids.

Resources for Families

A Guide for Families - The Diagnosis and Management of Duchenne Muscular Dystrophy.

This guide is based on the care considerations for DMD developed and endorsed by international experts. It summarises the medical care guidelines in plain language for people and families affected by DMD. The family guide is available in other languages through the [**TREAT-NMD Neuromuscular Network**](#).

Learning to Live with Neuromuscular Disease: A message for parents

The Duchess of Kent Children's Hospital at Sandy Bay. August 2017

Care Considerations

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Adult Imperatives for Duchenne MD – A Guide for Providers

This a summary of the key components of care across **late adolescence and adulthood** which may be helpful for medical providers unfamiliar with caring for adult men living with Duchenne.

Nutrition in DMD

The World Duchenne Organisation, together with experts on Duchenne and nutrition, has created two resources that contain information considering nutrition and supplements in DMD. On this website you will also find a series of videos of frequently asked questions concerning DMD and nutrition as well as a nutrition guide for adults with DMD, launched by DMD Pathfinders.

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