Myotonic Muscular Dystrophy (DM)

Including 1 related type:

- Congenital Myotonic Dystrophy

Processing a new diagnosis

A diagnosis of DM can be a huge shock for yourself, parents, siblings, extended family members and friends. It is normal to feel an overwhelming mix of grief, confusion, anxiety, loneliness and helplessness as your life, and 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 your 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 number of 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 DM 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 DM. 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 DM have also experienced the emotional rollercoaster that comes with a diagnosis of DM and are able to understand exactly how you're feeling. You are not alone. Connect with other people and families living with DM in The Loop Community on our **Forum**.

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

Having a conversation with a loved one about a diagnosis of DM 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 many reasons why people may not want to have this conversation, mostly centred on wanting to protect their loved ones and themselves.

But avoiding the topic is not helpful. People may have noticed differences and changes that have occurred and/or heard the condition being discussed.

So it is crucial that you have a role in your loved one learning about their/your diagnosis. This will enable you to support them as they process the information and to be on hand to answer any questions they may have. If you are the person with DM it will provide you with support and someone to talk to. 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 they may have heard or read
- Show them that you have faith in their ability to handle difficult conversations.

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

Some important do's and don'ts:

- Do be positive but also realistic.
- Do reassure them that no one has done anything wrong and this is not their fault.
- Do tell them they/you will do many wonderful things in their/your life – they/you 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/they are having difficulty with your/their muscles?'

Remember, it's okay to be upset during the conversation but try to avoid breaking down as this will only cause greater distress. And if speaking with a child, remember children are resilient and they generally handle information of this nature far better than adults.

Understanding DM and how it's diagnosed

About DM

Myotonic Dystrophy (DM), sometimes called Steinert's Disease, is the most common form of adult muscular dystrophy (a group of diseases that cause your muscles to become progressively weaker). A key feature of DM is myotonia, difficulty relaxing a tightened muscle. DM also causes muscle weakness and a number of symptoms that affect other parts of the body.

The symptoms of DM may appear at any age, from birth to late adulthood and affects both males and females. There are 2 different types - DM1 or type 1 and the rarer DM2 or type 2. Both are genetic conditions but affect different genes.

Some babies are born with a severe form of the condition called congenital myotonic dystrophy.

What causes DM?

DM is caused by a specific change or fault (also called a mutation) in a gene (the parts of our cells that tell our cells what to do). The two forms of DM are caused by two different genes:

- DM1 (type1): a gene on chromosome 19 called DMPK
- DM2 (type2): a gene on chromosome 3 called ZNF9.

Although different genes are affected, both types of DM cause problems with messages being sent to the muscles, resulting in muscle weakness.

What are the types of DM?

DM1 is the most common form and occurs in about 1 in 8000 people worldwide. There are 3 categories of DM1:

- Congenital – a severe, early childhood form of DM1. Congenital means 'from birth' and symptoms are usually present at birth or shortly after. Most babies born with congenital DM will have a mother with DM type 1, although the mother's symptoms may be so mild she may not be aware of it.
- Childhood onset – typically presents before the age of 10. It is characterised by symptoms such as problems with learning, concentration and motivation, more than physical symptoms. Muscle weakness will usually develop to varying degrees as the child matures.
- Adult onset - the most common form of DM. Weakness begins in the muscles of the face, neck, fingers and ankles. The weakness is slowly progressive and will eventually include other muscles.

DM2 is generally a less common, milder form of the condition. Onset is usually between the ages of 30 to 60 years and it rarely begins in childhood. DM2 usually begins in the proximal muscles (those closest to the centre of the body) particularly around the hips. However, it often does include some finger weakness.

It is difficult to know exactly how many people are affected by DM2 as the symptoms are often very mild and many people do not seek treatment.

How is DM diagnosed?

Obtaining a diagnosis of DM can unfortunately take some time. Many doctors may be unfamiliar with the condition as it a rare disease and may focus on more common causes for your symptoms (eg. testing your eyes if you have problems with your vision).

The tests that your doctor may use to assist with the diagnosis of DM include:

- Physical examination – this may show a pattern of muscle wasting and weakness, as well as the presence of myotonia.
- Genetic blood test – this is the most precise test for dm. This type of blood test looks for the specific genetic changes that cause DM1 or DM2. Further information about genetic testing can be found on the **Healthdirect website**.
- Electromyogram (EMG) – this test involves inserting small needles into the muscles to measure their electrical activity. An EMG will detect the presence of myotonia in a high portion of people with DM.

People with DM1 can have quite a distinctive appearance due to weakness of their facial muscles and may be an early indicator that a person has DM. The facial muscle weakness can lead to people appearing to have a long and expressionless face.

The genetics of DM

DM is inherited in an autosomal dominant pattern. This means it takes only one DM gene inherited from either parent to cause the condition in their child. On average, half the children from a parent with the DM gene will have the condition.

There are very few cases of DM occurring spontaneously, meaning it is usually inherited from a parent with the condition.

Both males and females are equally likely to have the condition.

DM1 genetics

A molecule known as RNA (ribonucleic acid) is the key to causing DM1.

To understand DM1, we need to know how our cells work to make proteins (our body's building blocks):

1. In the core of the cell is the nucleus. The nucleus contains our genes, which are made up of lengths of DNA (deoxyribonucleic acid).
2. When a cell needs to make a certain protein, it 'switches on' the corresponding gene, or the portion of DNA that is responsible for that protein.
3. An exact copy of that portion of DNA is made, called RNA. RNA acts as a messenger, travelling out of the nucleus and instructing the rest of the cell to make the protein.

In DM1, the DMPK gene is abnormally long. This is caused by extra repeats of the DNA building blocks (cytosine, thymine and guanin, also abbreviated as CTG) in the DMPK gene. A normal DMPK gene has fewer than 35 repeats of CTG. In DM1, there can be hundreds or even thousands of CTG repeats in the DMPK gene. When this gene is 'switched on', the extra CTG repeats are also copied into RNA. The abnormally long RNA is now an unusual hairpin shape and get stuck in the nucleus. It then hooks onto certain proteins inside the nucleus, trapping them in clumps. The proteins held in these clumps are then unable to

perform their normal functions.

Generally, the more CTG repeats within the DMPK gene a person has, the younger they are when symptoms are noticed and the more severe the condition will be.

The number of CTG repeats in the faulty gene tend to increase in each generation of a family. This leads to increasing severity of the condition in each generation.

DM2 genetics

Type 2 is caused by extra pieces of repeated DNA in the ZNF9 gene (also known as the CNBP gene). A four letter DNA code (CCTG) is repeated many hundreds of times, rather than less than 30 times in healthy people. This mutation leads to faulty RNA being produced and getting stuck in the cell, meaning the cells do not function properly.

Unlike DM1 the repeats in the faulty gene do not increase with each generation of the condition.

Genetic testing

DM1 and DM2 are hereditary conditions that become increasingly severe in each generation. Therefore, relatives of people with DM can have genetic testing using a blood test to find out if they are at risk of developing the condition and/or passing it on to their children. This testing should be undertaken with **genetic counselling** to ensure a full understanding of a diagnosis. Genetic counselling is best commenced before testing to ensure people understand the impact of this decision, particularly if the test is positive. In some situations, such as obtaining travel and health insurance, you will be required to disclose this information.

Genetic testing of healthy young children is not recommended.

Long-term outlook

The course of DM varies widely between people who have the condition and even within families with the same condition. For the most part, muscles weakness and wasting progresses slowly. The majority of people with DM will maintain their ability to walk independently into late adulthood.

Generally, the older a person is when they first notice symptoms, the slower the condition progresses and usually causes less serious symptoms.

The condition tends to be more severe and have an earlier age of onset with each generation in a family. So, a grandparent might experience their first mild symptom at the age of 60, while their children may notice symptoms at 30 – 40 years of age, and grandchildren may be born with congenital DM.

Some babies born with congenital myotonic dystrophy may ‘outgrow’ some of the effects of the condition during childhood and their symptoms can improve. However all children with congenital DM will develop the adult form of DM when they reach adolescence or adulthood.

While no treatment exists that slows the progression of DM, management of its symptoms can greatly improve quality of life. See the Symptoms section below for more information.

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

- **Living Life**
- **Counselling**
- **Social Work**
- **Psychology**

Understanding and planning for changes

Learning about DM 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 DM 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 DM?

There are a wide range of symptoms, with varying severity, for people living with DM. These symptoms can first appear from birth to older age.

One of the first signs of DM is often myotonia or difficulty relaxing a muscle after use. You might notice muscle stiffness, trouble getting up and moving after sitting, or difficulty releasing your grip after holding an object or shaking hands. While this is common for people living with DM, it can be quite alarming when you first notice this symptom.

There are a range of other common early symptoms that can occur (see sections below). However, it is important to note this is not a checklist and not everyone will show all of these symptoms.

DM1

Early symptoms are weakness and wasting in the muscles of the face, neck, hands, forearms and feet. This weakness will cause difficulties when using the affected muscles such as when opening jars, writing and walking. Myotonia can also cause difficulty in relaxing these muscles. The large muscles of the legs are initially much less affected.

As the condition progresses symptoms can include:

- Mobility and balance issues, including falls
- Digestive problems leading to incontinence, constipation
- Issues with speech and swallowing, such as a weak voice, slurred speech or frequent choking on food or drinks
- Breathing difficulties
- Heart problems
- Cataracts
- Excessive daytime sleepiness
- Hormonal problems leading to reproductive difficulties, thyroid dysfunction and/or diabetes
- Difficulty blinking and shutting of the eyes, making sleep difficult and increasing the risk of eye injury.

Many people with DM1 will also have some form of learning difficulty, ranging from mild to severe. This includes problems with attention, memory and planning, lowered intellectual level, and personality changes. Learning and/or behavioural assessments might be needed to assist with identifying these symptoms.

DM2

DM2 is sometimes called PROMM (proximal myotonic myopathy) as the first muscles affected are the muscles close to the body, particularly the thigh muscles. This can cause difficulty when walking and when standing from a seated position.

People with DM2 may also experience:

- Cataracts
- Heart problems
- Diabetes
- Muscle pain.

Congenital DM

The symptoms of congenital DM are present at birth and include:

- Very weak muscles with low muscle tone (hypotonia). Babies with congenital dm often appear “floppy”.
- Difficulty breathing and babies may require breathing support, such as a ventilator
- Inability to suck or swallow properly, leading to difficulty feeding
- Facial muscle weakness, leading to lack of facial expression and an upper lip that comes to a point
- Turned in or club feet
- Delay in motor and intellectual milestones.

Progression and monitoring

The severity of DM differs vastly from person to person and within the same family. In general, symptoms progress slowly.

About half the people with DM will begin to show symptoms by 20 years of age. However, a significant number of people do not develop clear-cut DM symptoms until after the age of 50.

As the condition progresses it is recommended that people with DM are monitored for:

- Heart problems: annual heart checks are recommended and many people with DM will eventually require a pacemaker
- Breathing and lung problems: regular checks of your breathing and lung function are recommended as weak breathing muscles can lead to a lack of efficient sleep, increasing a person's tiredness and risk of developing pneumonia
- Mouth health and speech: as the condition progresses speech and mouth movements can become difficult, leading to a dry mouth, bad breath and difficulty with dental care. Seeing a **speech pathologist** and/or dentist can assist with these symptoms.

Congenital Myotonic Dystrophy

More babies with congenital DM are surviving the critical early infancy period as a result of advancements in treatment and management. A child with DM who makes it to their first birthday is likely to live to adulthood.

Generally, congenital DM improves through the early childhood years but can begin to deteriorate in late childhood and early adolescence, when the adult features of the condition begin to show.

Community Advice

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

The video is available at **https://youtu.be/I7labxjWv9w.**

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 DM

Myotonic Muscular Dystrophy fact sheet
From Muscular Dystrophy NSW, 2017. This fact sheet includes considerations for care and planning.

International organisations

Muscular Dystrophy Association
This US based website has excellent information regarding DM. You can find a full list of neuromuscular condition, symptoms, causes and care options, as well as use the clinical trial finder tool.

Myotonic Dystrophy Foundation
This US based organisation provides information about DM, toolkits and information about research, including:

- **Toolkit a Guide for Care**
- **Consensus-based Care recommendations for Adults with Myotonic Dystrophy Type 1**
- **Consensus-based Care recommendations for Adults with Myotonic Dystrophy Type 2**

Muscular Dystrophy UK
A UK based site that has useful information regarding DM.

Support for DM

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Treatment and management

At the moment there is no cure for DM but there is a lot you can do to improve your quality of life. Being a rare disease, it is often not well understood by the general medical community and you may find you need to work in partnership with, and help educate, your healthcare team.

Current treatment focuses on the management of symptoms and reducing the risk of medical complications. Treatment options include:

Medications

Medications can be used to:

- Control myotonia
- Manage muscle pain
- Alleviate daytime sleepiness
- Manage diabetes.

Therapy

A range of health professionals can help you manage your symptoms, including:

- **Physiotherapy** can treat muscle pain, weakness and stiffness.
- **Speech pathology** can help with swallowing and speech issues.
- **Psychology** and counselling can provide emotional support and assistance.
- Individualised support for learning difficulties.

Equipment and devices

There are many types of equipment and assistive devices that may be useful, for example:

- Arm and foot braces, walking sticks, walkers and electric scooters to assist with mobility
- Eye crutches for droopy eyes
- Cough assist machines to prevent respiratory issues
- Positive airway pressure machines to help with lung function.

Visit our page on **Equipment** for more information.

Dietary

There are ways to modify your diet to manage any problems with digestion, such as:

- Eating a high-fibre diet for constipation or diarrhea
- Nutritional supplements to manage weight gain or loss.

You can get advice about your diet and how to manage digestive issues from a **dietitian**.

Exercise

Movement and staying physically active is very important for people with DM. However, as many people with DM may have problems with their heart, seek advice from a health professional before starting. Exercise should focus on

- Strengthening exercises – to help maintain muscle strength
- Aerobic exercise – to help keep your heart and lungs heathy
- Range of motion and stretching exercises to help maintain mobility.

Surgery

Some people with DM may require surgery including:

- Orthopedic surgery, for bones, muscles and joints, to improve mobility and reduce contractures (shortening of muscles)
- Removal of cataracts to improve vision
- Eyelid surgery to fix droopy eyelids.

Care recommendations for DM

There are detailed care considerations for DM, outlining how to diagnose, treat and manage DM. These guidelines are evidence-based (based on best available current research) and have been developed and endorsed by international experts. Although these are written in medical terms and might seem daunting, they can be useful for discussions with your specialists, doctors, nurses and other health professionals:

- **Consensus-based Care Recommendations for Adults with Myotonic Dystrophy Type 1**
- **Consensus-based Care Recommendations for Adults with Myotonic Dystrophy Type 2**

Treatment and management of Congenital Myotonic Muscular Dystrophy

Children with congenital DM are usually managed by a multidisciplinary care team. The treatments and support required for a child with DM depends on their symptoms and may include:

- Respiratory support – children with breathing difficulties may require artificial ventilation, particularly at birth.
- Swallowing and feeding support – a child may require special feeding techniques including special bottles and food supplements. If necessary nasogastric feeding (feeding tube through the nose) or the use of a feeding tube directly into the stomach may be required to prevent choking and ensure adequate nutrition is achieved
- Club foot (turned in feet) – can be helped by physiotherapy but if severe may require surgery for a child to be able to walk.

If you need to have an anaesthetic

People with DM have a higher risk of an unwanted or potentially serious reaction to the medications used in anaesthetics. Potential problems, even in people with mild DM symptoms, include:

- Heart and lung complications
- Sensitivity to sedatives or painkillers
- Myotonia during surgery
- Abnormal reaction to muscle relaxing drugs.

Surgery can usually be undertaken safely with careful monitoring of the heart and lungs. If you require an anaesthetic, it is important to let the anaesthetist and your surgeon know about your DM diagnosis as early as possible.

You can use the following information when talking to your anaesthetist or doctor:

- **Myotonic Dystrophy Foundation – Practical Suggestions for the Anaesthetic Management of a Myotonic Dystrophy Patient**
- Medical Alert Card for DM - read more about this below.

Support with family planning

People with DM may have concerns about starting a family because of the risks of passing the condition to their children. Men and women with DM1 have an increased risk of having a child with congenital DM1. These issues should be discussed with a genetic counsellor as there are a number of options available. For more information, visit our page on **genetic counselling**.

DM 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 DM 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.
- 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 DM 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 DM have also experienced the emotional rollercoaster that comes with a diagnosis of DM and are able to understand exactly how you're feeling. You are not alone. Connect with other people and families living with DM in The Loop Community on our **Forum**.

Medical Alert Card

Muscular Dystrophy Queensland has developed a **Medical Alert Card for Myotonic MD Type 1 and Myotonic MD Type2** . 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 DM

Myotonic Muscular Dystrophy fact sheet
From Muscular Dystrophy NSW, 2017. This fact sheet includes considerations for future planning.

Care recommendations and toolkit

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- **Consensus-based Care recommendations for Adults with Myotonic Dystrophy Type 2**

Myotonic Dystrophy Foundation – Practical Suggestions for the Anaesthetic Management of a Myotonic Dystrophy Patient

Myotonic Dystrophy Foundation Toolkit

This comprehensive toolkit, from Myotonic Dystrophy Foundation, provides information for people with DM and families and medical professionals.

International Organisations

Myotonic Dystrophy Foundation
This US organisation provides information about DM, toolkits and information about research.

Muscular Dystrophy Association
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