Myotonic Dystrophy FOUNDATION

TYPE 1

Myotonic Dystrophy Type 1

A Guide for Individuals and Families



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A GUIDE FOR INDIVIDUALS & FAMILIES

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Letter from the CEO

Dear Reader,

Welcome to the Myotonic Dystrophy Foundation (MDF) and to *Myotonic Dystrophy Type 1: A Guide for Individuals and Families*.

Whether you are newly learning about myotonic dystrophy (DM) or have been living with the condition for many years, we are honored to welcome you. Receiving a diagnosis, supporting a loved one, or navigating the ongoing challenges of DM can bring many questions. This guide was created to provide trusted information, practical resources, and a connection to a community dedicated to improving the lives of everyone affected by myotonic dystrophy.

Founded by individuals and families impacted by DM, MDF was built on the belief that no one should navigate this disease alone. That commitment continues to shape our mission and our work every day.

As the only global nonprofit organization dedicated exclusively to myotonic dystrophy Community, Care, and Cure, MDF brings together individuals and families, healthcare professionals, researchers, industry partners, and advocates to advance research, improve standards of care, expand education, and strengthen support for the DM community. Through collaboration, we are working to accelerate scientific progress, increase access to high-quality care, and improve quality of life for people living with DM.

This guide was developed in collaboration with leading clinicians, researchers, and members of the DM community living with or caring for someone with DM. It combines current scientific and clinical knowledge with the perspectives and experiences of individuals and families living with the condition. Myotonic dystrophy is widely recognized as one of the most variable diseases in medicine, with symptoms, age of onset, severity, and progression differing significantly from one person to another—even among members of the same family. Although each person's experience with DM is unique, we hope this resource provides clear, reliable information that helps you better understand the condition, make informed decisions, and identify resources that meet your needs.

When you connect with MDF, you become part of a global community committed to advancing knowledge, fostering connection, and supporting people affected by myotonic dystrophy. We are here to provide education, advocacy, community programs, and practical resources for individuals, families, caregivers, and healthcare professionals. We encourage you to explore our programs, connect with others, and reach out when you have questions or need support.

Thank you for allowing MDF to be part of your journey. It is our privilege to walk alongside you and your family today, as we work together to improve care, advance research, expand treatment options, and move closer to a future in which effective therapies—and ultimately a cure—are available for everyone living with myotonic dystrophy.

With warm regards,



Tanya Stevenson

Tanya Stevenson, EdD, MPH
Chief Executive Officer
Myotonic Dystrophy Foundation

1 Welcome & About

About the Myotonic Dystrophy Foundation

Welcome to *Myotonic Dystrophy Type 1: A Guide for Individuals and Families*, a resource developed by the Myotonic Dystrophy Foundation (MDF) for anyone whose life is touched by ***myotonic dystrophy (DM)***. Whether you are living with ***myotonic dystrophy type 1 (DM1)*** or are a caregiver or family member of someone living with DM1, this guide is meant for you. Remember, you are not alone – MDF is here to support you every step of the way!

Throughout this guide, you will notice terms that are ***bolded and italicized***. These terms are defined in the guide's glossary found on **page 97.**

Founded in 2007 by families affected by myotonic dystrophy, MDF's mission is to support and connect the myotonic dystrophy community, provide resources, advocate for care, and accelerate research toward treatments and a cure. Over the years, MDF has grown into the world's leading advocacy organization for the myotonic dystrophy community, serving tens of thousands of families in over 139 countries. We want to serve your family and be a supportive partner throughout your journey.

Because DM1 is a rare and complex disease, it is not widely known or understood by the medical community and even less so by the general public, including colleagues at work, peers at school, or even family members in your own home. You and your loved ones may find yourselves regularly introducing someone to DM for the first time, and, unfortunately, sometimes even educating your own medical and healthcare specialists! This guide is designed to help you become your own best advocate by helping you to recognize and ask for the proper care you deserve.

Who is This Guide For?

This guide is for you: a person living with DM1. Whether you've just received your diagnosis or have been managing life with DM1 for many years, this guide is intended to empower you with information, resources, and encouragement.

Although this guide has been designed with you in mind, it can also help your caregivers and your healthcare team understand more about living with and managing DM1. Ultimately, this guide exists to help you successfully navigate the challenges and obstacles that can be a part of life with DM1.

Throughout this guide, myotonic dystrophy is abbreviated as "***DM***", which stands for the universally used Latin name for the disease: ***dystrophia myotonica.***

Myotonic dystrophy type 1 is abbreviated as DM1, ***Juvenile-onset adult myotonic dystrophy*** as JOA, ***congenital myotonic dystrophy*** as CDM, and ***myotonic dystrophy type 2*** as DM2.

Designed for the DM1 Community

DM1 is a complex condition that can look radically different from person to person, even between family members. While this guide offers comprehensive and big-picture overviews on a wide range of DM1 related topics, you do not have to read this guide in a single sitting or cover to cover to find the answers you are looking for. Each section is clearly marked, so you may easily find the information that's most relevant to you at any given time.

Receiving a myotonic dystrophy diagnosis can be overwhelming. It's normal to experience a range of emotions, from confusion about your body to anxiety and depression about the future. By reading this guide, you are doing important work to learn more about DM1 and get the support you need. Here is a quick overview of what you'll find in this guide:

- **Understanding DM1:**
  Learn about the genetic mechanism, symptoms, progression, and management strategies.

- **Living with DM1:**
  Discover tips and strategies for daily life, including managing symptoms and maintaining wellness.

- **Sharing Your Experience:**
  Feel more confident having conversations with your healthcare providers about what's important to you in exploring and making healthcare decisions.

- **Support and Resources:**
  Find out how to connect with others, access support groups, and utilize MDF's myotonic dystrophy specific resources.

This guide is not a substitute for medical advice. You should always speak with your doctor before making any changes to your healthcare plan. Similarly, in some cases, this guide will provide information about medications or treatments that may help certain symptoms related to DM for some people. The information provided in this guide is not an endorsement of any specific medication or treatment.



2 What is Myotonic Dystrophy (DM)?

Overview

Myotonic dystrophy (DM) is a type of muscular dystrophy that can affect many parts of your body. The word "myotonic" comes from the word "***myotonia***," which means having trouble relaxing your muscles after they've tightened. "Dystrophy" is a condition where parts of the body, like a muscle or tissue, slowly become weaker over time. DM1 is a rare inherited ***genetic disorder***, and while there is currently no cure or specific treatment, there is a lot you and your healthcare team can do to improve your quality of life. Learning more about DM1 may help you to have better conversations with your doctors and allow those around you to better understand your experiences and ways to support you.

"Muscular dystrophy" describes a group of diseases that cause muscles to weaken and lose mass over time (Mayo Clinic, May 2025). ***Myotonic dystrophy (DM)*** is the most common form in adults, though it's also seen in children and young people.

When you talk about DM1, doctors who are unfamiliar with the condition may incorrectly assume you're referring to a different condition called ***diabetes mellitus type 1***, commonly known as ***type 1 diabetes***.

DM1 and type 1 *diabetes* are completely different conditions.

DM is a complex condition that can affect nearly all of your body systems. Its symptoms and disease progression can vary widely from person to person, even among members of the same family. For example, within a group of siblings with the same parents, one person might experience early-onset ***cataracts*** (Hamel, 2022) and mild muscle stiffness later in life, while a brother or sister may struggle with breathing difficulties (Hawkins et al., 2019; Henke et al., 2020) or develop heart rhythm irregularities (Freyermuth et al., 2016; Wahbi et al., 2016). This variability can make it challenging to predict just how DM1 will affect you and your loved ones.

Understanding that DM1 affects everyone differently can help you and your healthcare team focus on your unique experiences. By paying attention to your specific symptoms and how they change over time, you can work together to manage them effectively. To learn more about the various symptoms of DM, see **page 15** of this guide.

Types of Myotonic Dystrophy

There are two types of myotonic dystrophy: ***myotonic dystrophy type 1 (DM1)*** and ***myotonic dystrophy type 2 (DM2)***. DM1 is further categorized based on the age at which symptoms first appear.

Scientists and physicians often use Latin when naming different diseases or organisms. So, while the disease is called myotonic dystrophy – it is abbreviated "DM" after the Latin name, which is **dystrophia myotonica**. You might also hear other names used by doctors and researchers worldwide:

- ***DM:*** Myotonic Muscular Dystrophy (MMD)
- ***DM1:*** Steinert's Disease
- ***DM2:*** Proximal Myotonic Myopathy (PROMM)

Myotonic Dystrophy Type 1

The gene for DM1 was identified in 1992, with the first known recorded case traced back to 1909. DM1 is the most common form of myotonic dystrophy and may affect **as many as 1 in 2,100 people worldwide.** This means over 3 million individuals across the world may be affected or at risk of passing the disease on to their children (Johnson et al., 2021).

There are three categories of DM1. These are based on when symptoms of the disease first appear:

Congenital Myotonic Dystrophy (CDM)

"Congenital" means present at birth. In CDM, symptoms are noticeable from birth, such as difficulty breathing on their own or low muscle tone. Babies with CDM may face significant health challenges early on and often require specialized medical care right away. Delivery at a medical center with high-risk neonatal support may be recommended.

Juvenile-Onset Myotonic Dystrophy

For Juvenile-onset DM, symptoms usually appear during childhood or adolescence, before the age of 21. The first signs are often related to learning difficulties or challenges with intellectual development.

Adult-Onset Myotonic Dystrophy

Adult-Onset DM1 generally means symptoms did not appear in childhood or adolescence and the disease was diagnosed after the age of 21. Symptoms vary but commonly include muscle weakness, muscle wasting (loss of muscle mass), ***myotonia***, and stiffness. These symptoms can develop gradually.

Myotonic Dystrophy Type 2

DM2 was first identified in 1994, and its underlying CCTG repeat expansion was discovered in 2001. The exact number of people who may have this type of DM is currently unknown. In DM2, symptoms often show up in adulthood and can affect multiple body systems. The most common symptoms are muscle weakness, pain, and fatigue. MDF has a separate guide dedicated to those living with DM2 and their families, which can be found on ***https://myotonic.org***.

SCAN TO LEARN MORE about Myotonic Dystrophy Type 2

The Most Variable Disease in Medicine

Every individual's journey with DM is unique. The symptoms you experience, and their impact, are real. Throughout this guide, comparisons to potential differences in severity between types of DM are not meant to take away from the significance of your lived experience, but they may be useful when developing strategies to manage your symptoms. Additionally, learning about the most severe symptoms does not mean you will experience them. Instead, this information is intended to build your understanding and help prepare you for whatever direction your journey takes.

3 Where Does Myotonic Dystrophy Come From?

Scientific Definitions

DNA (deoxyriboneucleic acid)
The molecule that carries genetic information for the development and functioning of an organism.

RNA (ribonucleic acid)
A molecule that plays a crucial role in making ***proteins*** and regulating ***genes***.

Proteins
Large, complex molecules that play many important roles in the body. They are critical to most of the work done by cells and are required for the structure, function and regulation of the body's tissues and organs.

Gene
A segment of ***DNA*** that contains instructions for building a specific ***protein*** or ***RNA***.

Nucleotides
A basic building block of nucleic acids (***RNA*** and ***DNA***). They are represented by the letters C, A, T, and G in ***DNA*** and C, A, U, and G in ***RNA***.

RNA Splicing
A process in cells in which portions of a ***gene*** are joined in different combinations, leading to different copies of ***RNA*** that are used to produce different ***proteins***.

Mutation
A change in the ***DNA*** sequence of an organism.

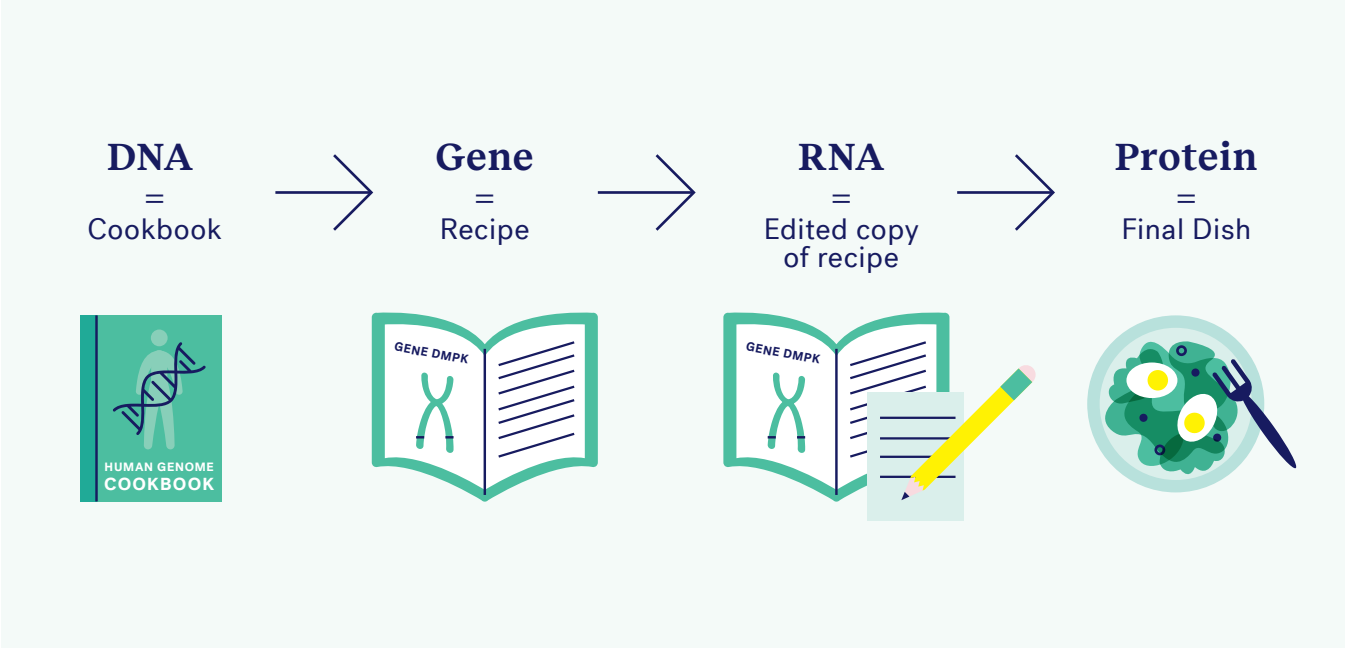
(National Human Genome Research Institute, 2025)

Myotonic dystrophy is an inherited disease that is passed from one generation to the next through a ***genetic mutation*** (a miscoded gene). It is not caused by an infectious agent such as a ***virus*** or ***bacteria***, or chemicals, biohazards, or similar exposures. This section will guide you through some basic principles of genetics, the genetics of DM1, and how DM1 is inherited.

The Basics of Genetics

To understand genetics, it may be helpful to think of a cookbook full of recipes. ***DNA* is like a master cookbook** that stores all the instructions our cells need. Each ***gene*** within the ***DNA*** is a recipe for making ***proteins***, which are essential for our bodies to function. When a cell needs a ***protein***, it makes a copy of the recipe called ***RNA***. Much like fine-tuning a recipe, ***RNA*** often needs to be edited after being copied during a process called ***RNA splicing***. To make sure ***RNA*** is edited correctly, the cell uses specialized ***proteins***. Once splicing is complete, the edited ***RNA*** is used to assemble ***proteins*** the body needs. **Think of *proteins* as the final dishes that keep our cells and bodies functioning by performing a variety of roles in the body.** Some ***proteins*** provide building blocks to help build strong muscles and bones. Other ***proteins*** make chemical reactions happen more easily, like helping our stomach break down food quickly. By understanding the flow from ***DNA*** to ***RNA*** to ***proteins***, we can appreciate how genetic instructions are used and executed.

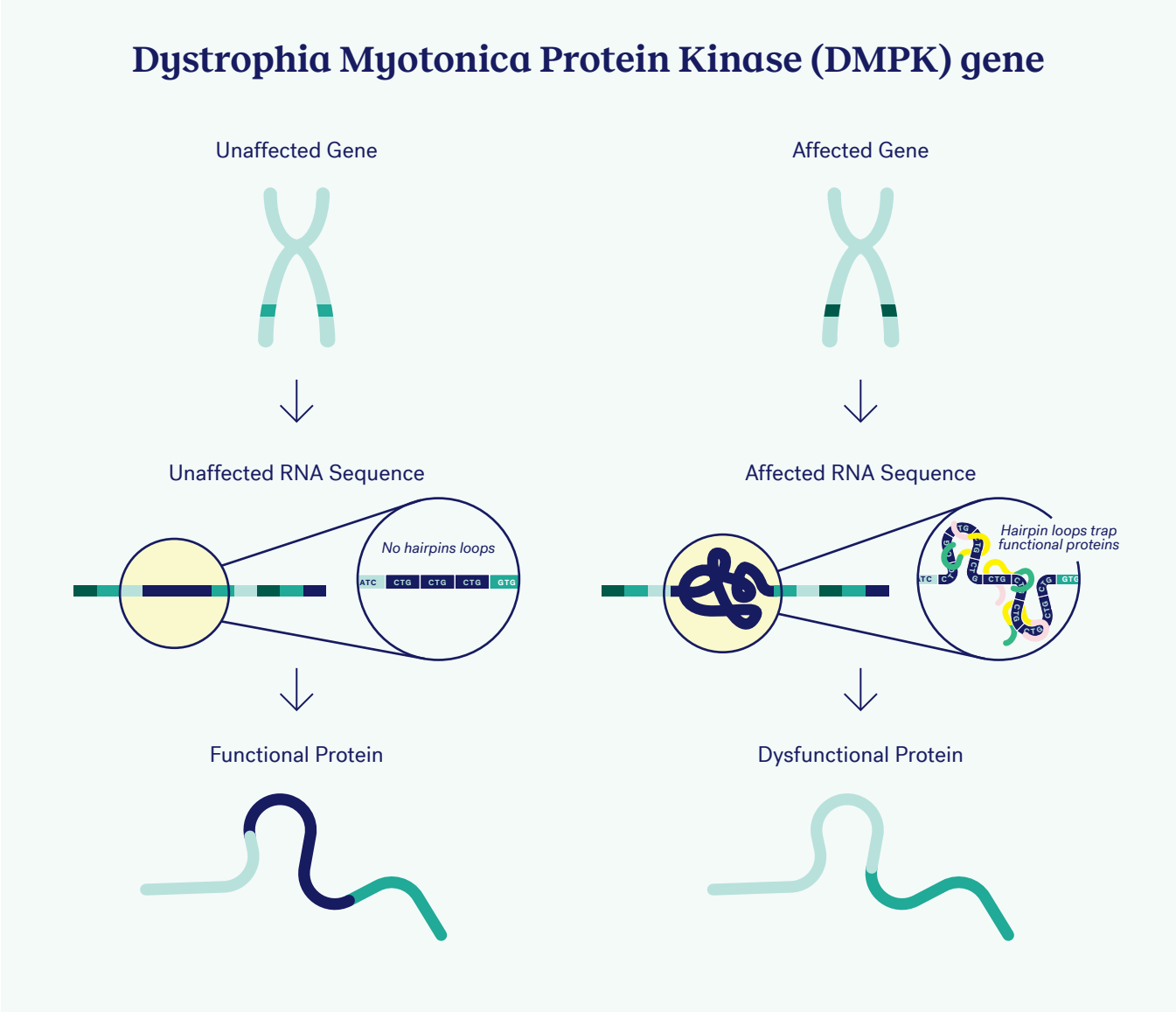
Sometimes, the ***DNA*** cookbook may contain typos or missing pages. These errors are called ***gene mutations***, and they can have various effects. *It is important to note that mutated genes don't always affect health.* For example, most mammals have hundreds of ***genes*** that code for ***olfactory receptors***, which are ***proteins*** that help us smell. Losing one of these ***genes*** likely won't impact the ability to smell (Genetics Science Learning Center, 2016). Some ***mutations*** can even help an organism survive and reproduce, and those ***mutations*** are passed on to the next generation. However, other ***mutations***, and the faulty ***proteins*** they produce, can cause significant changes that result in disease symptoms.



The Genetics of DM1

At the core of DM1 is a ***mutation*** in the ***Dystrophia Myotonica Protein Kinase (DMPK)*** gene. Normally, this gene has a small section where three ***DNA*** building blocks (***nucleotides***) – represented by the letters CTG – repeat a few times. However, in individuals affected by DM1, this CTG repeat is **abnormally expanded**, meaning it repeats many more times than it should. ***Genetic testing*** shows that people without DM1 usually have between 5-37 CTG repeats, while people with DM1 may have anywhere from 50 to more than 4,000 CTG repeats (Otero et al., 2021). When this expanded CTG sequence in the ***DMPK*** gene is copied, it creates abnormally long ***RNA***, which do not behave as they should.

Instead of properly making ***proteins***, these abnormally long ***RNA*** clump together inside of the cell nucleus and create something called ***hairpin loops***. This clumping causes ***RNA*** toxicity, meaning the ***RNA*** itself can harm the cell and its function. The clumps trap (or sequester) specialized ***proteins*** that are needed to process ***RNA***, and when they are trapped, they can't do their job. This disruption affects many processes within the cell and leads to the wide range of symptoms seen in DM1, such as muscle weakness, ***myotonia***, and other systemic issues.

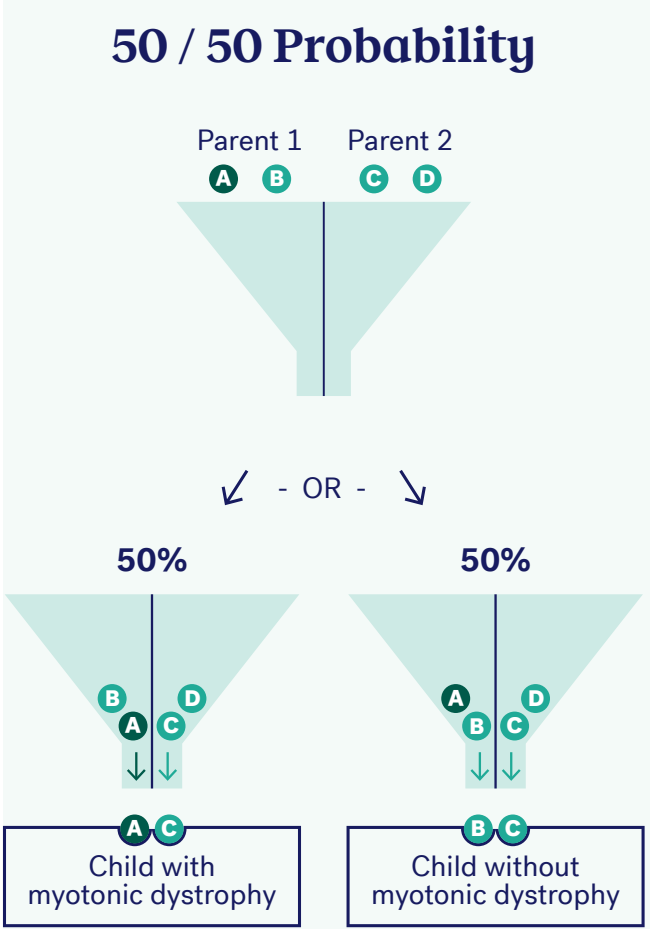


The genetic basis of DM2 is similar to DM1 in that they are both caused by expanded ***RNA*** repeats. However, in DM2 the expanded repeats occur in a completely different gene, called ***CNBP***, which carries the genetic information for cells to make a ***protein*** called CCHC-type zinc finger nucleic acid binding ***protein***. Alternative names for this gene include ZNF9 or ***CNBP1***. A person living with DM2 will have expanded CCTG repeats (instead of CTG repeats seen in DM1). MDF has a separate guide dedicated to those living with DM2, and their families, which can be found on ***https://myotonic.org***.

Throughout their lifetime, a person living with DM1 may have their CTG repeat number continue to expand. Because each cell carries a copy of every gene (its own "master cookbook"), the ***DNA*** repeats can expand differently in different cells. This process, called ***somatic mosaicism***, means the number of CTG repeats can vary between cells or tissues within the same person. For example, a blood cell may have a different number of ***DNA*** repeats than a leg muscle cell. This may help explain why DM symptoms differ in severity across different parts of their body. The exact repeat in each cell isn't needed for diagnosis or for most supportive treatments (see "How do I know if I have myotonic dystrophy?" section on **page 19** for more information about ***genetic testing***).

Inheritance of DM1

DM1 is passed from parent to child through an ***autosomal dominant mutation***. This means that if one parent has the condition, even if they don't show symptoms, each child has a 50% chance of inheriting the mutated gene and developing DM1 symptoms. *Autosomal* means that the gene responsible for DM1 is located on one of the 22 pairs of ***chromosomes*** that don't determine sex. *Dominant* means that only one copy of the mutated gene is needed for the symptoms of the disease to appear.

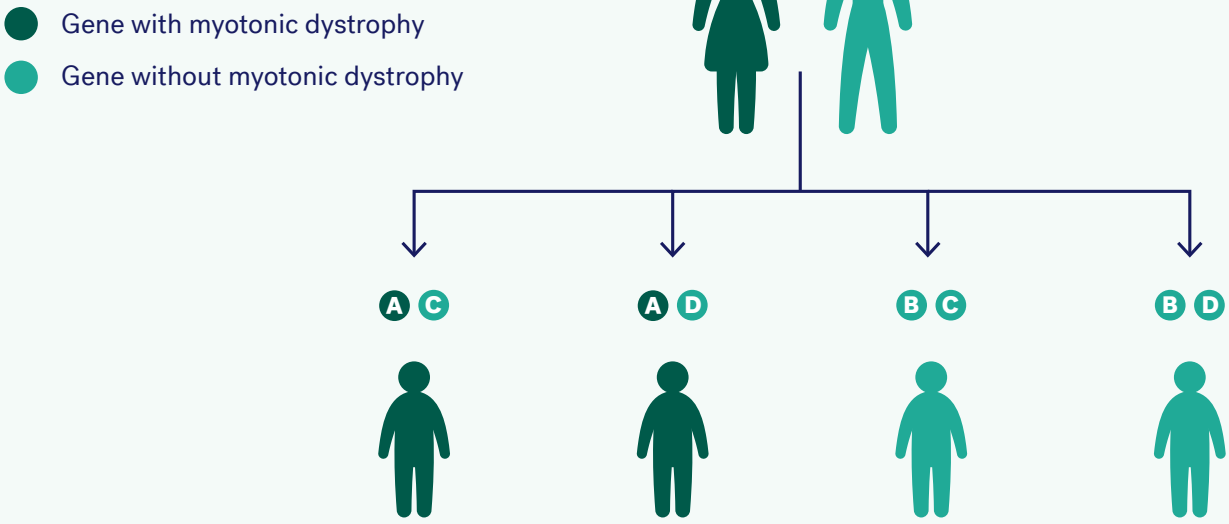


Gene with myotonic dystrophy

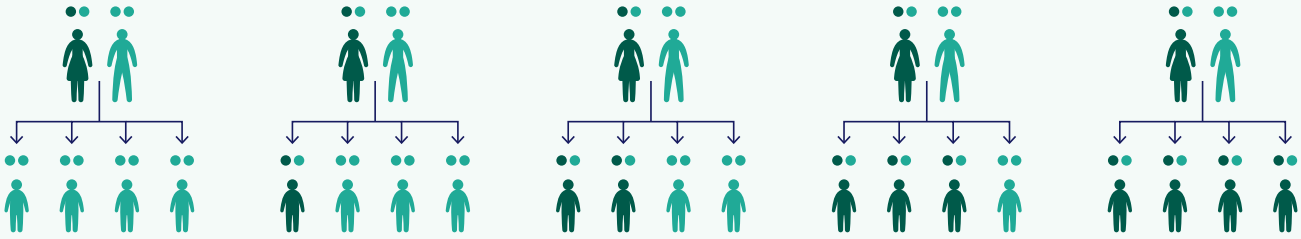
Gene without myotonic dystrophy

DM1 does not skip generations. If a child inherits one copy of the mutated gene, they will have DM1 and could pass it on to future generations. This is different from ***recessive*** genetic conditions, where a person would need to inherit two copies of the mutated gene (one from each parent) to show symptoms. DM1 can be passed to male and female children with equal likelihood, by either a male or female parent. Individuals who inherit the mutated gene will eventually show symptoms, although they may not appear for many years.

Autosomal Dominant Inheritance



Examples of Potential Inheritance Patterns



An important aspect of DM1 inheritance is a phenomenon known as genetic ***anticipation***. This means that with each successive generation within a family, DM symptoms may appear earlier and become more severe (Yum et al., 2017). ***Anticipation*** occurs because the number of CTG repeats in the ***DMPK*** gene is unstable and can expand with each generation. DM1 shows similar severity regardless of which parent it comes from. However, congenital myotonic dystrophy (CDM, the form of DM with the most severe symptoms defined on **page 9**) is most commonly inherited from the mother (Morales et al., 2015; Barbé et al., 2017; Chakraborty et al., 2016). When the mutated gene is passed on by a female, it tends to be more unstable and may expand further than when inherited from a male. Scientists think this may occur because CTG repeats significantly expand during the creation of ***egg cells*** but more research is needed (Dean et al., 2006).

4 How Do I Know If I Have Myotonic Dystrophy Type 1?

Myotonic dystrophy type 1 (***DM1***) is a complicated condition. This section will help you understand the signs and symptoms of DM1 as well as the process of ***genetic testing***.

Signs and Symptoms

The symptoms of DM1 and its progression can vary widely from person to person – even among members of the same family. This means it can be hard to predict how DM1 will affect you and your loved ones. For example, one person may have mild muscle stiffness or develop early ***cataracts***, while another person with the congenital form of DM1 may be born with serious breathing problems (Hawkins et al., 2019; Henke et al., 2020) or develop problems with the heart (Freyermuth et al., 2016; Wahbi et al., 2016). The most common effects of DM1 are muscle problems, including muscle weakness (***myopathy***), trouble relaxing a muscle (***myotonia***), and muscle wasting that gets worse over time (***atrophy***). Since muscles exist throughout almost every part of your body, DM1 can affect the heart, lungs, and the ***gastrointestinal (GI) system*** (including the mouth and throat). People living with DM1 can also experience non-muscle related complications with mental health, ***cognitive*** function, personality changes, and vision (Miller et al., 2021; Otero et al., 2021).

Not everyone with DM1 will have all or even most of the possible symptoms. For those with DM1 or a family history of the condition (who have not been tested), staying aware of and documenting signs and symptoms is essential to supporting informed care and proactive symptom management.

Common Symptoms of DM1

The three categories of DM1 (Congenital, Juvenile-Onset, and Adult-Onset) described on **page 9** of this guide show up in different ways but share many of the same symptoms. If you are experiencing the symptoms listed in the section below, you may consider following up with a doctor and/or seeking genetic counseling and testing for myotonic dystrophy. To learn more about symptoms, see Part 5 Body Systems and Effects on **page 22** of this guide. For more details on ***genetic testing***, see the Genetic Testing section on **page 19** of this guide.

Experiencing at least three of the following symptoms may suggest a DM1 diagnosis:

- Eyelid ***ptosis*** or drooping of the eyelid.
- Weakness of the fingers and wrists (not including stiffness).
- ***Myotonia*** or “stiffness” of muscles.
- Early onset ***cataracts*** (below 50 years of age).

Experiencing at least one of the symptoms above as well as the symptoms below OR having a family history of DM1 and experiencing the symptoms below may help point to a DM1 diagnosis.

- Signs that there may be issues with your heart, like ***heart palpitations***, chest pain, shortness of breath, difficulty breathing when lying flat, or lightheadedness and fainting.
- ***Irritable bowel syndrome*** (***IBS***).
- Elevation of any of the following liver ***enzymes*** (C. R. Heatwole et al., 2006; Perna et al., 2020; Peterson & Cooper, 2022).
  - → ***γ-glutamyltranspeptidase*** (***GGT***)
  - → ***Aspartate aminotransferase*** (***AST***)
  - → ***Alanine aminotransferase*** (***ALT*** or ***GPT***)
  - → ***Alkaline phosphatase*** (***ALP***)
  - → ***Lactate dehydrogenase*** (***LD***)



- ***Gallstones*** at a young age (below 30 years of age) (Cardani et al., 2008; Peterson & Cooper, 2022).
- Prolonged recovery from ***anesthesia***.
- ***Respiratory arrest*** following ***anesthesia*** administration (Bisinotto et al., 2010).
- ***Insulin resistance*** or ***diabetes***.
- ***Hypogonadotrophic hypogonadism***, when male ***testes*** or female ***ovaries*** produce minimal or no sex ***hormones*** due to problems with hormone regulation or glands.
- Decreased energy and/or ***excessive daytime sleepiness*** (***EDS***). This could look like falling asleep several times each day, even after a good night's sleep.
- Mild learning difficulty. For example, you may find it harder to plan, focus, or juggle multiple tasks at the same time.

Similarities and Differences Across DM1 Categories

Many symptoms of ***myotonic dystrophy type 1*** (***DM1***) are shared across all three categories: **Adult-Onset**, **Juvenile-Onset**, and **Congenital DM1** (**CDM**). The main differences are **when symptoms begin and how severely they show up**.

Muscle Weakness

Muscle weakness is a common symptom for people living with DM1, but it appears at different times and in different ways:

- **Adults**: Weakness often starts in the hands and feet and slowly gets worse. This might look like dropping objects, tripping, or needing handrails when climbing stairs (Bird, 1993).

- **Children and Teens**: Weakness may be milder at first but tends to increase over time.

- **Infants** (**CDM**): Muscle weakness or low muscle tone (sometimes called Floppy Baby Syndrome) is often clear from birth, leading to breathing and feeding challenges early in life. Because weakness is so strong in CDM, muscle stiffness (***myotonia***) can be less noticeable in infants, though it often becomes more noticeable in older children and adults.

Other Common Symptoms

Other symptoms show similar patterns across the DM1 categories:

- ***Cataracts***: Common in adults, but rare in children or babies because ***cataracts*** take time to form.

- **Heart Rhythm Problems** (***arrhythmias***): Can appear in any DM1 subtype but are more common as people get older.

Overall Pattern

DM1 symptoms are **variable**, meaning they can differ in both timing and severity from one person to another. While the same general symptoms appear across Adult Onset, ***Juvenile Onset***, and ***Congenital DM1***, the **speed** at which they develop and **how strongly** they affect someone can vary widely.

For more information on how each body system is affected and how symptoms progress over time in different subtypes, see **Part 5 of this guide** (on **page 22**).

Genetic Testing to Confirm a Diagnosis

Genetic testing plays an important role in confirming a myotonic dystrophy diagnosis. We understand that the path to diagnosis can feel overwhelming logistically, financially, and emotionally. If you are struggling to find answers, you are not alone. On average, it can take over six years to receive a diagnosis of DM1 (National Institute of Health, 2025), but for some people it can be over a decade. This is often due to the rarity of the condition and the overlap of DM symptoms with those of more common conditions. Testing may also be costly or hard to access depending on where you live.

Even with these challenges, obtaining a DM1 diagnosis can be a powerful step forward. For many, it opens doors to medical care and health insurance options, offers validation, guides future planning, and creates an opportunity to connect with a supportive community. MDF is here to help you along this journey. The resources below may offer guidance and support to make the process more manageable.

Who Might Get Tested and Why?

Usually, an individual seeking ***genetic testing*** is either someone with symptoms, someone with a clinical DM diagnosis (when your doctor believes your symptoms point to DM but this has not been verified through a genetic test), someone seeking a second opinion or confirmation, or someone without symptoms or a diagnosis but with a ***genetic predisposition*** (e.g., a history of DM in their family).

You may want to get tested for a variety of reasons. ***Genetic testing*** can be beneficial in the following situations:

- To eliminate the need for additional medical tests and reduce anxiety about the cause of symptoms.

To know if you should be concerned about the risks of ***anesthesia*** and alert your doctors should you ever need surgery (see **page 63** of this guide for more information about ***anesthesia*** and DM).

- To have a better understanding of yearly health monitoring such as visits with neurology and cardiology, even if you are not currently experiencing noticeable symptoms.

To make family planning decisions based on genetic risk (see "Family Planning" section on **page 80** of this guide for more information).

- To make decisions about ***prenatal testing*** (where the ***DNA*** of the fetus is checked for the presence of the myotonic dystrophy ***mutation***).
- To determine if a mother living with DM1 needs special monitoring during pregnancy and prepare for risks involved for a child born with congenital DM. Reference MDF's *Clinical Recommendations for People of Pregnancy Potential* with myotonic dystrophy for more information.

SCAN TO LEARN MORE about Clinical Recommendations for people of Pregnancy Potential

Genetic testing can not only diagnose DM1, but also tell you your repeat length (how many times the gene repeats in your ***DNA***). However, some genetic tests only look for the presence of a CTG repeat expansion, and do not measure or report the length of the expansion. Be sure to discuss with your ***genetic counselor*** and healthcare team to determine which test is best for you. Knowing your repeat length can be important for your cardiovascular care.

SCAN TO LEARN MORE about repeat length and cardiac risks

The Testing Process

Preliminary Investigation: Testing Before Genetic Confirmation

Getting a genetic test to confirm a diagnosis of DM1 isn't always the first step. Because ***myotonic dystrophy*** (***DM***) is so variable, many of its symptoms can also be caused by other, more common conditions. Doctors often begin by **ruling out those common causes first**.

Step 1: Review of Family and Medical History

Your doctor will usually start by:

- Reviewing your **personal and family health history**, including close and distant relatives.
- Asking about symptoms like muscle weakness, stiffness, or fatigue that may run in your family.

Step 2: Physical Examination

A physical exam helps the doctor look for signs that could suggest DM1, such as:

- Muscle weakness (***myopathy***)
- Muscle stiffness (***myotonia***)
- Drooping eyelids (***ptosis***)
- Early ***cataracts***

Step 3: Targeted Testing Based on Symptoms

The next tests depend on what symptoms you have:

- **If you have *cataracts***, your doctor may refer you to an ophthalmologist to see if they are the type often linked to DM1.
- **If you have high blood sugar or *diabetes***, your doctor may check whether the cause could be related to DM1 instead of another condition.

Step 4: Specialized Testing (If Needed)

If your results are still unclear, your doctor may suggest additional tests such as:

- ***Electromyography*** (***EMG***): This test measures the electrical activity in your muscles and nerves.
  → EMG can help detect ***myotonia*** (**muscle stiffness**), which is a key sign of DM1.

In short, before ordering a genetic test, doctors often:

1. Review your family and personal health history
2. Perform a physical exam
3. Order specific tests based on your symptoms
4. Use EMG or other specialized tools if more information is needed

These steps help rule out other conditions and decide whether ***genetic testing*** for DM1 is appropriate.

Genetic Testing

If you have a family history of ***myotonic dystrophy*** (***DM***), or if your symptoms include signs like early onset ***cataracts*** or difficulty relaxing your grip (***myotonia***), your doctor might suggest a genetic test, or you can ask for one. A genetic test uses a blood sample to check for the abnormal ***mutation*** that causes myotonic dystrophy.

Types of Tests

Genetic testing for DM uses standard ***DNA*** testing methods to check for the presence of the DM1 ***mutation***. The two types of ***genetic testing*** used to diagnose DM are called "***Southern blot***" and "***Polymerase Chain Reaction***" or "***PCR***" for short. These tests help doctors confirm whether or not someone has the DM1 ***mutation***. Generally, a ***Southern blot*** test is more expensive than a ***PCR*** test.

Genetic Counseling

Working with a ***genetic counselor*** can be helpful before, during, and/or after the testing process. ***Genetic counselors*** can assist with questions about insurance and potential financial implications, as well as explain the different types of tests, how they're done, how long results may take, and what the results mean. They can also help people make more informed decisions around family planning. To learn more about family planning while living with DM, see the "Family Planning" section on **page 80** of this guide and MDF's *Clinical Recommendations for People of Pregnancy Potential with Myotonic Dystrophy*.



International Resources for Genetic Counselors

Australia

Talk to your healthcare provider for national healthcare service referrals, or book private genetic counseling:

https://www.hgsa.org.au/Web/Web/HP-Resources/Clinical-genetics-services-by-state/National-Services.aspx

Canada

https://www.cagc-accg.ca/

Europe, Asia, and Middle East

Talk to your healthcare provider for national healthcare service referrals or visit *https://www.snpedia.com/index.php/Find_a_genetic_counselor*

Latin America

https://www.splagen.org/en/en/directory

United Kingdom

Talk to your GP for NHS Genetic Counseling services or book private genetic counseling: *https://geneticexperts.co.uk/*

To find a ***genetic counselor*** in the United States in your area search by zip code at *https://findageneticcounselor.nsgc.org/*. You can also ask your doctor for a referral to see a ***genetic counselor***.

SCAN TO FIND a Genetic Counselor

Where to Test and Costs

There are many laboratories that can provide a genetic test for DM, including those at university medical centers and private companies. The cost of testing can vary a lot, from a few hundred dollars to a few thousand, depending on the laboratory, insurance, and whether you're being tested for DM1 or DM2. Before getting tested, it's a good idea to do some research and talk with a ***genetic counselor***.

It might also help to call your insurance company to check if they will cover the cost of a lab "***Southern blot***" or "***polymerase chain reaction*** (***PCR***)" test for DM. Insurance companies might ask for details about your family medical history, symptoms, and past tests before they decide if they will cover ***genetic testing***.

If you're based in the United States, you can connect with the *Patient Advocate Foundation* for additional assistance navigating insurance coverage for ***genetic testing***.

SCAN TO LEARN MORE about the Patient Advocate Foundation

Other Considerations

If you're considering obtaining an official diagnosis through ***genetic testing***, you may want to consider a few things:

- In the United States, a diagnosis could make it difficult to get certain types of private insurance such as short- or long-term disability insurance (not disability provided through the US Government) and non-employer-based life insurance.
- You may also choose to avoid an official diagnosis due to the possibility of stigma or other kinds of reactions from friends, family, or others in the workplace or school environment.
- No less important is the mental health impact of being diagnosed with a disorder that does not currently have a cure or specific treatment.

Despite these potential barriers, there are laws to protect you. The Genetic Information Nondiscriminatory Act (GINA) is a federal law in the United States that prevents discrimination based on genetic information in employment and health insurance. The Americans with Disabilities Act (ADA) is an additional federal law in the United States that prohibits discrimination against individuals with disabilities.

Other countries and regions offer similar protections:

Australia	Disability Discrimination Act (1992)
Canada	Genetic Non-Discrimination Act (2017)
European Union	General Data Protection Regulation (GDPR)
United States	Genetic Information Nondiscrimination Act (GINA) (2008)
United Kingdom	Equality Act 2010; Genetic Insurance Moratorium

Other Routes

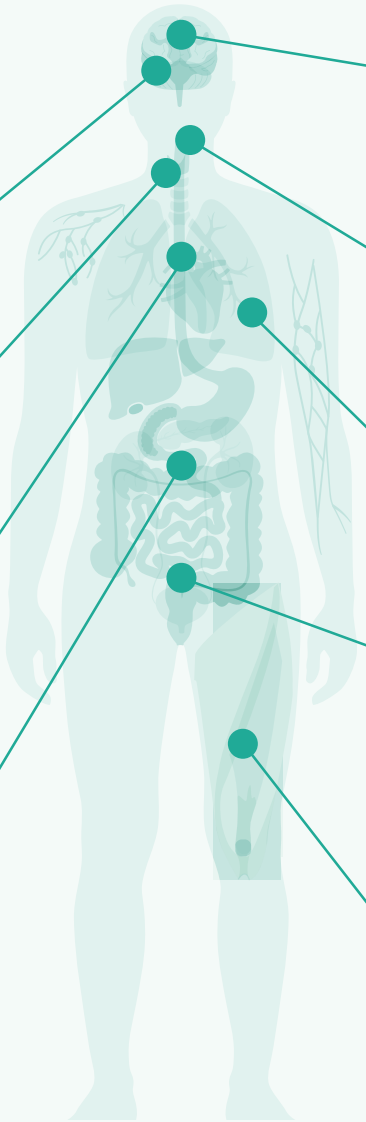
Genetic testing for DM is the only way to obtain an official confirmatory diagnosis. However, if testing is not possible because of cost or for some of the other reasons listed above, and you think you or someone in your family may have DM, you can still take steps to protect your health. For example, you can have yearly medical checkups recommended to those with DM, like heart health screening. You can also share MDF's ***Clinical Care Recommendations*** with your medical providers. Another option is to look into clinical trials that sometimes cover testing costs (see MDF Support section on **page 92** of this guide for more information).

SCAN TO READ MDF's Clinical Care Recommendations

5 How Does Myotonic Dystrophy Type 1 Affect People?

People living with myotonic dystrophy experience a wide range of symptoms that can affect many parts of the body. This section of the guide aims to provide information about the type of symptoms people living with myotonic dystrophy may experience, and how they relate to the different systems in your body. It also contains information which may help manage these symptoms and help those affected thrive in their daily activities. Below is a list of body systems, brief overviews of symptoms that someone living with DM1 may experience, and where you can find additional information about each body system in the guide.

How Myotonic Dystrophy Affects the Body



VISION *(page 59)*

- ***Cataracts***, blurred vision
- Damage to the ***retina***
- Drooping eyelids (***ptosis***)

ENDOCRINE *(page 51)*

- ***Diabetes***
- ***Insulin resistance***
- Low ***thyroid*** hormone levels
- Premature frontal balding in men

CARDIOVASCULAR *(page 28)*

- Heart rhythm problems (***arrhythmias***)
- Enlarged heart muscle (***cardiomyopathy***)
- Low blood pressure
- Sudden death

GASTROINTESTINAL *(page 37)*

- Difficulty swallowing
- Pain and bloating after meals
- Constipation, diarrhea, irritable bowel syndrome, reflux
- Poor nutrition and weight loss
- ***Gallstones***
- Enlarged colon
- Chronic infections

BRAIN *(page 42)*

- Difficulty with thinking and problem solving
- Emotional and behavior problems
- ***Excessive daytime sleepiness***
- Nerve damage in feet and hands

IMMUNE *(page 56)*

- Lower levels of ***antibodies*** in blood stream (***hypogammaglobulinemia***)
- Higher risk of benign skin tumor (***pilomatrixoma***)

RESPIRATORY *(page 33)*

- Breathing problems in newborns
- Frequent lung infections
- ***Aspiration*** of food or fluids into airways
- Inability to breathe in enough oxygen
- ***Sleep apnea***

REPRODUCTIVE *(page 47)*

- Males: low ***testosterone*** levels, low sperm count, small ***testes***, testicular failure, gonadal ***atrophy***
- Females: higher risk of ***miscarriage*** and ***stillbirth***, pregnancy and delivery complications, early ***menopause***
- Newborn complications

MUSCULAR *(page 23)*

- Muscle weakness (***myopathy***)
- Muscle pain, stiffness, trouble relaxing a muscle (***myotonia***)
- Muscle wasting that gets worse over time (***atrophy***)
- Severe muscle weakness, delayed development in newborns and infants

Understanding how myotonic dystrophy affects your body can help you manage symptoms and improve your quality of life. Because DM doesn't always look the same, the body systems it affects, how severe the symptoms are, and when they appear can vary a lot, even among members of the same family. That's why having support from your healthcare team, loved ones, and the DM community is so important as you navigate these challenges.

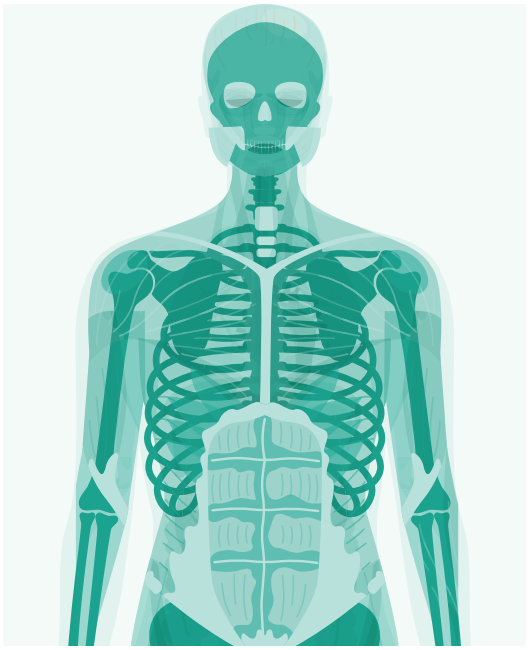
Skeletal Muscles



System Overview

The skeletal muscle system is responsible for voluntary movements, posture, and overall physical strength – helping you move, stand upright, and carry out everyday tasks. These are also called ***voluntary muscles*** and are under your conscious control, so you can move these muscles when you want. They are the muscles you use to make all the movements needed for physical activity. Muscles like your heart are called ***involuntary muscles*** and are not under your control, which means you can't make them contract when you want. Learn more about how DM can affect involuntary muscles in the Cardiovascular (heart) system section below **(page 28)**. In DM1, the skeletal muscle system is often one of the first systems affected. This section will discuss how DM1 affects the skeletal muscles, outline muscle-related symptoms in DM1, define the different muscle-related symptoms seen across types of DM1 (Congenital, Juvenile-Onset, Adult-Onset), and what may help manage these symptoms.

Common Symptoms: Skeletal Muscles



The most common skeletal muscle symptoms people living with DM1 experience are ***myotonia*** (a condition where muscles have difficulty relaxing after contracting) and muscle weakness and wasting, particularly in the face, neck, arms, legs, and especially hands (Hartman et al., 2024; Rimoldi et al., 2024).

Myotonia

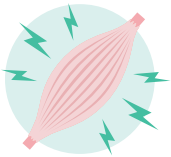
Myotonia means your muscles have a hard time relaxing after they contract (when a muscle squeezes or tightens). For instance, if you were to turn a doorknob or open a jar, you might notice that your fingers stay tense and don't immediately release. ***Myotonia*** sets myotonic dystrophy apart from other types of muscular dystrophies (Hamel, 2022).

Checking for Myotonia

Doctors will usually check for ***myotonia*** in one of two ways:

- ***Grip Myotonia:*** One common way to observe ***myotonia*** is by having an individual grip something tightly with their fingers, usually someone's hand, thumb, or a small device. If you are living with DM1, it may take noticeably longer – sometimes 20 seconds or more – to fully relax your hand after letting go (Chakraborty et al., 2016).
- ***Percussion Myotonia***: Another way to check for ***myotonia*** is by gently tapping a muscle with a reflex hammer to see if the muscle takes extra time to relax, signaling the presence of ***myotonia*** (Rimoldi et al., 2024).

How might myotonia affect me in everyday life?



Delayed muscle relaxation*:** After turning a key or gripping a coffee mug, your hand may stay partially clenched for a few seconds. This can make tasks like opening jars or turning keys a little tricky, but strategies like pausing for a moment to allow any ***myotonia to pass and your muscles to relax before using the same muscles again can help you adapt (Hamel, 2022). Alternatively, there are adaptive tools, like jar openers, that can be helpful to complete tasks.



Challenges with quick movements: Actions that require sudden bursts of movement, like hopping out of a car or getting up from a couch, can feel stiff at first. This stiffness usually eases after a few steps, but it might feel like your body takes a moment to "catch up" with your brain's commands. Planning for extra time can help make these transitions smoother.



Cold weather sensitivity: Cold weather can sometimes make ***myotonia*** worse (Hamel, 2022), causing your muscles to feel stiffer than usual. You might feel it when trying to button your coat or when your fingers don't want to cooperate with zippers or buckles. Dressing in layers that are easy to remove and keeping your hands warm can help minimize discomfort and make it easier to manage stiffness.



Exercise: Whether you're walking around the neighborhood or following a light exercise routine, you may notice that your muscles don't relax right away. Taking breaks and doing gentle, repetitive movements to 'warm up' can help your muscles settle down.



Face and jaw stiffness: *Myotonia* can sometimes affect facial muscles, making motions like smiling or activities like chewing a little slower or more difficult. This can make eating harder, especially if chewing requires a lot of effort. Choosing softer foods or smaller bites may help when your jaw feels tight. For additional strategies, see the Swallowing and Aspiration section of this guide on **page 70**.

Muscle Weakness and Atrophy

As we get older, our muscles naturally lose some strength. In DM1, this process can be easier to see, and you may notice muscle weakness in the neck, facial muscles, arms, legs, and especially hands (Hartman et al., 2024; Rimoldi et al., 2024). ***Atrophy*** is a word that clinicians and researchers use to describe when muscles shrink or waste away because they are not being used enough or, in this case, because it is caused by a disease like DM. This is different from simply feeling weak. Over time, the muscle itself becomes smaller and less able to do its job.

How Skeletal Muscle Symptoms May Change Over Time

If you are living with DM1, you might notice your muscles change slowly over time. These changes can affect how you stand and move, but they happen little by little and can often be managed with good care. One of the first signs to appear usually happens in the hands and fingers, making it harder to do small tasks like buttoning a shirt or using a fork. You may also notice weakness in your arms and shoulders, which can make carrying groceries or lifting objects overhead more difficult. Weakness in the legs and hips may cause tripping, difficulty climbing stairs, or more frequent falls. Later on, muscles in the face, neck, and back can also get weaker, which might cause slight changes in posture, like a curve in the spine, known as ***scoliosis*** (Schilling et al., 2013). However, everyone's experience with DM1 is different, and these changes don't happen all at once. Many people with DM1 stay active, continuing to do things they enjoy and that are meaningful to them.

Understanding the Different Forms of DM1 and How Muscles are Affected

Congenital DM1 (CDM)
People who are born with symptoms of DM may have weak muscles from the start, which can increase weakness in their mouth and breathing muscles as well as affect their bones as they grow. Babies and children living with CDM often look "floppy" (this is called ***hypotonia***) due to low muscle tone and may have tight joints that stay stuck in a bent position (this is called ***joint contracture***). As they get older, they might also develop a curved spine (***scoliosis***) or other body changes. Weak muscles can make it harder for them to reach milestones like rolling over, sitting up, or walking (Prasad et al., 2016; Fontinha et al., 2018).

Juvenile-Onset DM1
If you are living with Juvenile-Onset DM1, you may first notice weakness in your hands or feet, which can make everyday tasks feel harder. You might find yourself stumbling or catching your toe more often, struggling with writing, or having difficulty with other fine motor activities such as using scissors or doing up buttons. Over time, you may also feel these symptoms gradually spread to the muscles around your hips or shoulders (Canavese & Sussman, 2009). When that happens, you might have trouble lifting your arms, getting up from the floor, or climbing stairs.

Adult-Onset DM1
If you are living with Adult-Onset DM1, you may first notice weakness in your hands or feet, which can make everyday tasks feel harder. You might find yourself stumbling more often, struggling with writing, or having difficulty with other fine motor activities. Over time, you may also feel these symptoms gradually spread to the muscles around your hips or shoulders (Canavese & Sussman, 2009). When that happens, you might have trouble lifting your arms, getting up from the floor, or climbing stairs.

Although these symptoms can be challenging, various treatments and strategies outlined in the next section can help you manage them and maintain your health.

What Can I Do?

There are many options to help manage muscle problems in DM, and they often work best when used together. It's important to understand what these tools are and how you can use them on their own or combine them to help keep your muscles as healthy as possible. Remember, because DM1 can look very different from person to person, the management strategies below may need to be adapted to your specific needs. Getting help with physical therapy, adaptive devices, and proper medical management can significantly improve mobility and muscle function, helping you maintain an active life. **It is important to speak with your doctor before starting any new exercise or movement activities or routines.**

Exercise
Low- to moderate-intensity ***aerobic*** and ***resistance exercises*** are often recommended by healthcare providers to maintain muscle strength and function (Mackenzie et al., 2022) (see MDF's *Exercise Guide and Infographic* to learn more).

SCAN TO READ MDF's Exercise Guide and Infographic



Physical and Occupational Therapy

Regular therapy can help maintain mobility, dexterity, and daily functioning. Therapists can also recommend adaptive devices (such as braces, canes, and walkers to aid in mobility and prevent falls) and home modifications (Hamel, 2022) (see **page 65** of the guide for more information on adaptive devices and home modifications).



Speech and Swallowing Therapy

Over time, the muscles used for speech and facial expressions may weaken or become stiff, which can lead to difficulties with communication, chewing, and swallowing. Addressing muscle weakness early, through support like physical and speech therapy, can significantly help these symptoms before they worsen (see **page 70** of the guide for more information on speech and swallowing therapy).



Medications

For some people, a class of drugs called ***antiarrhythmics*** can sometimes be used to help manage ***myotonia*** (e.g., ***mexiletine***) (Rimoldi et al., 2024; Spillane et al., 2025). Speak with your primary physician and care team, as this medication should be used cautiously in individuals with cardiac (heart) involvement, which can also be affected by DM (see the cardiovascular system section on **page 28**).

Cold temperatures can cause or worsen skeletal muscle pain. To manage this pain, your doctor may prescribe pain medications (e.g., ***gabapentin***, ***NSAIDs***, etc.) (Rimoldi et al., 2024). If these medications do not help to control your pain, talk to your doctor about alternatives. There may be other options like cannabidiol (CBD), acupuncture, physical therapy, and ***cognitive behavioral therapy*** that can help with pain management (Solbakken et al., 2021).

Additionally, ***palliative care***, a type of medical care that focuses on ***holistic*** approaches to improving quality of life, can help to manage pain. It can be helpful to individuals of any age and at any stage living with DM (see **page 78** below for more information on ***palliative care***).

Regular Monitoring

Annual evaluations by a team of providers, including neurologists, cardiologists, and rehabilitation specialists, are important for comprehensive care. You can utilize MDF's *Find-A-Doctor Map* to search for community-recommended providers near you.

SCAN TO VISIT MDF's Find-A-Doctor Map.

How can I best communicate my needs with my doctor?



Prepare Ahead of Time: Keep a diary or journal to keep track of any symptoms, patterns, triggers, and effectiveness of treatments so that you can remember to discuss with your clinician. You can use MDF's *My Clinic Visit Planner* as a tool to track your symptoms.

My Clinic Visit Planner

SCAN TO VISIT MDF's My Clinic Visit Planner.

Reference the Latest Information: For example, with the right oversight exercise has been shown to be good for DM and for maintaining muscle health.



Request Referrals: Providers should be supporting your access to PT and OT, which may require them to battle with insurance companies (in the US) or specialty referrals (in other countries) to ensure you receive long-term PT care, not the usual 4-6-12 weeks of care only (Check out MDF's *Physical Therapy* and *Occupational Therapy* guides for more information).

Physical Therapy

Occupational Therapy

SCAN TO VISIT MDF's Physical Therapy or Occupational Therapy Guides.

Share Your Values: Sharing what is important to you with your provider can help them understand your quality of life aims and help you get there or adjust them to fit your ability You may share things like:

- I want to be able to do _____. I'm unable to do this now because _____. (especially share if this is due to pain or fatigue)
- Doing _____ is important to me; how can I make sure I am able to continue _____ as my muscles weaken?



Understand Potential Testing, Therapy, and Medication Options:

- Providers may use ***electromyography*** to look at muscle function and determine if the weakness is a result of myotonic dystrophy.
- Although not common anymore, doctors may order a muscle biopsy to confirm myotonic dystrophy and rule out other causes of muscle weakness.
- Ask for a referral to Physiotherapy/Physical Therapy. You can ask specifically for at-home physical therapy services or equipment.
- Possible Medications:
  → ***NSAIDs***: reduce inflammation and pain.
  → ***Antiarrhythmics***: reduce excessive electrical firing of nerves.
  → ***Anticonvulsants***: calm overactive electrical signals in nerves.
  → ***Tricyclic antidepressants***: increase neurotransmitters that dampen pain signals.

Cardiovascular (Heart) System



System Overview

The cardiovascular system includes your heart and blood vessels, which work together to circulate blood throughout your body. This system keeps you healthy and energized by delivering oxygen and nutrients to tissues and carrying away waste products like carbon dioxide. In DM1, the cardiovascular system can be affected or compromised, leading to issues such as ***arrhythmias*** (irregular heartbeats) and ***cardiomyopathy*** (enlarged heart muscle which leads to weakening of the heart muscle). This section will provide an overview of how DM1 affects the cardiovascular system, discuss common symptoms, define the different cardiovascular symptoms seen across types of DM1 (Congenital, Juvenile-Onset, Adult-Onset), and outline what you can do to help manage these symptoms and prevent serious complications.

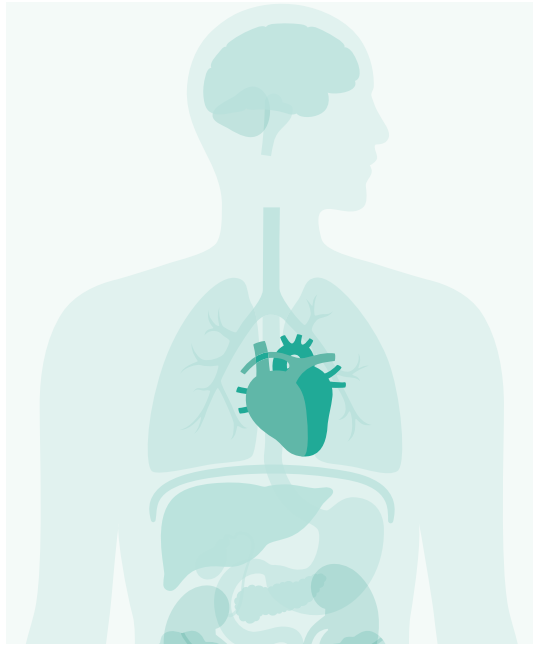
It's important to know that these symptoms are often silent in the early stages, and many people may not realize there's a problem until it becomes more serious. Therefore, this section places extra emphasis on making sure that you are regularly evaluated by a cardiologist because even if you feel fine, your heart health deserves ongoing attention.

For a more in-depth look at heart issues in DM1, refer to *Myotonic Dystrophy and the Heart: A Community Guide.*

SCAN TO READ Myotonic Dystrophy and the Heart: A Community Guide

Common Symptoms: Cardiovascular System

Heart problems are common for people living with DM1 and may show up in different ways for each person. Although these symptoms outlined below may sound alarming, regular monitoring (even if you are not experiencing symptoms now) and preventive care can greatly reduce the risk of serious complications and help you feel more confident in your heart health. Cardiovascular symptoms often include:



Arrhythmias

This is an irregular heartbeat that may include skipped beats, a racing heart, or longer pauses between heartbeats. You might notice your heart fluttering or experience dizziness or fainting. ***Arrhythmias*** are one of the earliest and most common symptoms in DM1 (Ashizawa et al., 2018; Itoh et al., 2024).



Conduction Defects

The heart's electrical signals can slow down or become blocked, causing a slow heart rate (***bradycardia***) or other more serious blockages. In severe cases, this can lead to the need for a ***pacemaker*** (Chakraborty et al., 2016; Wahbi et al., 2016).



Cardiomyopathy

This is when the heart muscle becomes enlarged and weaker, potentially leading to heart failure. You may feel unusually tired, short of breath, or notice swelling in your legs due to fluid buildup (Rimoldi et al., 2024). While ***cardiomyopathy*** can progress over time, early management can make a big difference in managing your heart health.



Pre-syncope or Syncope

Episodes of dizziness or fainting (***syncope***) can be caused by low blood pressure, or the heart's inability to efficiently pump enough blood to meet the body's needs. If you feel lightheaded or faint, it's important to seek medical attention. These episodes should be taken seriously, as they can be a sign that any conduction issues your heart may have may be worsening (Wahbi & Furling, 2020).



How Cardiovascular Symptoms May Change Over Time

Like many other aspects of DM1, heart problems can be different for each person. They might be more or less severe and may change over time at different rates. However, these factors are at least partly influenced by the CTG repeat length and age of onset of the individual (Itoh et al., 2024; Rimoldi et al., 2024; Yum et al., 2017). To learn more about the role that CTG repeat lengths play in DM, see the Genetics of DM section in Part 3 (**page 10**).

Paying close attention to your symptoms and how they show up is the first step to understanding what is normal for you. This helps doctors track any changes over time. If heart problems are not treated or if they get worse, they can lead to more serious health issues. So speak up, stay aware, and tell your heart doctor (cardiologist), neurologist, or primary doctor about any symptoms you notice.

In general, heart problems in DM1 usually get worse slowly and vary from person to person. Early on, you might feel mild symptoms like fatigue or occasional ***palpitations***. Over time, more serious issues such as heart failure or dangerous ***arrhythmias*** can appear. How quickly these problems increase depends on genetics and how early they're found and managed. It is essential that everyone living with DM1, whether they are experiencing symptoms or not, see a cardiologist on at least a yearly basis for consistent heart health monitoring (Bhakta et al., 2021).

Understanding the Different Forms of DM1 and How the Heart is Affected

Congenital DM1 (CDM)

For children born with CDM, heart problems are not usually seen in early childhood. However, as they grow older, it's important to keep an eye on heart health through regular checkups (Johnson et al., 2019). In adulthood, some people living with CDM may develop issues with how their hearts beat (***arrhythmias***) or problems with the heart's electrical signals (***conduction defects***), much like those seen in individuals living with Juvenile-Onset or Adult-Onset DM1.

Juvenile-Onset DM1

If you have Juvenile-Onset myotonic dystrophy, you may start to experience heart-related symptoms in early adulthood. These often involve irregular heartbeats (***arrhythmias***) or issues with your heart's electrical signals, called ***conduction defects***. As you get older, you may also develop ***cardiomyopathy***, a condition that affects your heart's ability to pump blood (Bassez et al., 2004; Stokes et al., 2019). Just like in congenital DM (CDM), regular heart checkups are important for you so that any new symptoms can be caught and treated early.

Adult-Onset DM1

Heart problems tend to be more common if you develop DM1 in adulthood. These can include irregular heart rhythms (***arrhythmias***) and issues with your heart's electrical system (***conduction defects***), which may become more serious over time. You may also develop ***cardiomyopathy***, a condition that affects how well your heart pumps blood. Because of these risks, regular checkups with a heart specialist are important for you to catch and manage any changes early (Itoh et al., 2024; Petri et al., 2012; Wahbi and Furling, 2020; Chakraborty et al., 2016; Wahbi et al., 2016).

Although these symptoms can be challenging, various treatments and strategies outlined in the next section can help you manage them and maintain your health.

What Can I Do?

Like with most symptoms in DM1, effectively managing your cardiovascular issues requires a multidisciplinary approach and regular monitoring. An overview of how you may improve your quality of life and reduce your risk of serious complications is outlined below. For more detailed information, see MDF's ***DM and the Heart: Community Guide.***

SCAN TO READ Myotonic Dystrophy and the Heart: A Community Guide.

Regular Cardiac Evaluations

It's important to begin with a comprehensive cardiac evaluation at the time of your diagnosis, which includes an ***electrocardiogram*** (***ECG***) to check your heart's electrical activity. After you've gotten your baseline heart activity around the time of your diagnosis, follow-up appointments with your cardiologist usually happen every 6-12 months. These appointments allow your doctor to watch for new or worsening symptoms (Wahbi et al., 2016). During these visits, keep your doctor informed about any changes, like feeling lightheaded or fainting, which may be signs that your heart's rhythm or function is changing.

It is recommended that you undergo a 24-hour Holter Monitor (a portable echocardiogram) every 3 to 5 years, or more frequently if an issue is identified (Rimoldi et al., 2024). This test allows your doctor to continuously monitor your heart over a 24-hour period heart abnormalities go unnoticed. It is a painless and non-invasive test where you wear the monitor at home and go about your daily activities for 24 hours.



Medications

There are medications that can help manage ***arrhythmias*** or heart failure symptoms, like the symptoms often seen in DM1. ***Antiarrhythmic medications***, including beta-blockers, work by blocking stress ***hormones*** that make the heart beat faster, thereby helping to maintain a more regular rhythm (Rimoldi et al., 2024). If you experience ***cardiomyopathy***, or heart failure, medications like ACE inhibitors or ***diuretics*** may be used to relax blood vessels and lower your heart's workload (Rimoldi et al., 2024).



Implantable Devices

In some cases, ***implantable devices*** may be necessary to protect your heart's function. ***Pacemakers*** are an available option that your cardiologist may consider if you're experiencing ***conduction defects*** or ***bradycardia*** that significantly affects your heart's rhythm. A ***pacemaker*** can help keep your heart steady. If your cardiologist determines that you're at risk for life-threatening ***arrhythmias***, then an ***Implantable Cardioverter Defibrillator (ICD)*** can help save your life by detecting and correcting dangerous rhythms before they cause sudden cardiac death (Itoh et al., 2024; Wahbi et al., 2016).



Lifestyle

In addition to medical interventions, there are lifestyle habits that can support your cardiovascular health. Low-intensity ***aerobic exercise***, done with your cardiologist's guidance, may help strengthen your heart without over stressing it. Likewise, tracking and reporting symptoms, like unusual fatigue, dizziness, ***syncope*** (fainting), or chest pain, can also be helpful in proactive management and help your healthcare team adjust your treatment as needed.



How can I best communicate my needs with my doctor?



Prepare Ahead of Time: Keep a heart health journal to share when you see your cardiologist. If you feel "funny" or experience symptoms like the ones above, note what you're doing when you feel them. This can help the doctor understand any additional influences such as worry/panic, stress, exercise, etc. which may be contributing to your heart health.



Share Your Values: Make sure your doctor understands what is important to you about your heart health. Share things such as fears you may have about your heart health and make sure to ask follow-up questions about their recommendations.

Request Referrals: Make sure your regular doctor understands the need for you to be referred to a cardiologist (heart doctor) and that they do so promptly, and advocate for any insurance issues related to this referral.

SCAN TO USE EDUCATIONAL MATERIALS

Share MDF's *Clinical Care Recommendations for Cardiologists*.

Clinical Care Recommendations for Cardiologists



Understand Potential Testing and Treatment Options:

- You should get an ***ECG*** every 6-24 (typically 12) months and cardiac imaging every 2-4 years (even without symptoms) (Bhakta et al., 2021).
- Possible Medications/Treatment:
  - → ***Antiarrhythmics:*** help the heart maintain a steady rhythm by controlling abnormal electrical signals that cause irregular beats
  - → ***Pacemakers:*** a device that regulates slow or irregular heart beats
  - → ***Implantable Cardioverter Defibrillator (ICDs):*** shock dangerous rhythms to prevent sudden cardiac arrest
  - → ***Beta blockers:*** slows heart rate, lower blood pressure, and reduces the heart's workload and stress response
  - → ***ACE Inhibitors (ACEI):*** Relaxes blood vessels, lowers blood pressure, and supports heart function, especially in heart failure

Respiratory System



System Overview

The ***respiratory system*** is responsible for bringing oxygen into your body and removing carbon dioxide. It includes your lungs, airways, and muscles that help you breathe – such as the ***diaphragm***. In DM1, these breathing muscles can weaken, which may lead to issues over time, such as shortness of breath, fatigue, or sleep-related breathing problems. Learning about the potential respiratory difficulty symptoms can help you work with your healthcare team to implement the best treatments and strategies for your ongoing respiratory health. This section will discuss how DM1 affects the ***respiratory system***, define the different symptoms seen across types of DM1 (Congenital, Juvenile-Onset, Adult-Onset), and introduce approaches to managing these respiratory challenges in DM1.

Common Symptoms: Respiratory System





Respiratory Muscle Weakness

Breathing requires both your ***diaphragm*** (for inhaling) and your abdominal muscles (for exhaling). In DM1, weakened breathing muscles make it harder for you to take full, deep breaths. Initially, this makes it hard to cough and clear mucus and can put you at risk of having longer and more severe respiratory illnesses like the ones described below that may require ongoing respiratory support (Hartog et al., 2021; Johnson et al., 2019).



Sleep-Related Breathing Disorders

As muscles become weaker, it can be harder to breathe in and out during sleep. Poor airflow during sleep can cause issues like ***sleep apnea***, where your breathing stops and starts repeatedly during the night. As a result, you may feel excessive sleepiness during the day or wake up with morning headaches (Cirignotta et al., 1987). As muscles become weaker shallow, slow breaths (***hypoventilation***) can also occur when you are awake.



Aspiration Risk

You may have difficulty swallowing, which can allow small amounts of food or liquids to enter your lungs instead of your stomach. This is called ***aspiration***, and it increases your risk of ***pneumonia*** (Alsaggaf et al., 2024).

How Respiratory Symptoms May Change Over Time

Respiratory system changes in DM1 often happen gradually. As your muscles weaken slowly, breathing can become more challenging over the years. However, by keeping track of your symptoms and monitoring them regularly, you can often stay ahead of problems and manage your respiratory health.

Understanding the Different Forms of DM1 and How Breathing is Affected

Congenital DM1 (CDM)
Babies born with CDM often struggle with breathing right from birth. They may require intensive care and specialized ventilatory support due to respiratory failure from weak lung muscles (Ho, 2015; Yum et al., 2017). Ongoing follow-up care is critical to ensure they get the support they need. Over time they may continue to need extra help with breathing day and night and a ***tracheostomy tube*** (a small tube in the windpipe) or nasal mask may be recommended (Johnson et al., 2019). In children living with CDM, respiratory infections can be life threatening (Johnson et al., 2019; Zapata-Aldana et al., 2018). Careful monitoring and support can help manage ***respiratory system*** issues.

Juvenile-Onset DM1
If you have Juvenile-Onset DM1, your muscle weakness can worsen as you continue to grow and develop, and this may lead to difficulty swallowing due to weakening throat muscles. This difficulty can cause foods or liquids to accidentally enter your airway, increasing your risk of ***pneumonia*** (Johnson et al., 2019). You may also experience breathing difficulties during sleep (***hypoventilation***), which can lead to snoring or morning headaches. If left untreated, this can cause a frequent or persistent cough. In addition, weakness in the muscles you use for coughing can make it hard to clear mucus from your lungs, which also increases your risk of ***pneumonia*** (Ashizawa et al., 2018; Johnson et al., 2019).

Adult-Onset DM1
If you have Adult-Onset DM, you may notice changes in your breathing, especially difficulty breathing at night (***hypoventilation***). These breathing issues can include loud snoring, abnormal breathing patterns, waking up with morning headaches, or feeling sleepy during the day (Ashizawa et al., 2018; Bianchi et al., 2014; Cirignotta et al., 1987). Over time, the muscles that help you breathe can become weaker, which may cause you to get out of breath more easily, feel tired much of the time, have difficulty coughing and clearing mucus, or catch lung infections more often (Alsaggaf et al., 2024; Hartog et al., 2021; Hamel, 2022).

Although these symptoms can be challenging, various treatments and strategies outlined in the next section can help you manage them and maintain your health.

What Can I Do?

Taking care of breathing problems with DM1 means acting early and working with doctors who specialize in managing breathing difficulties (like a pulmonologist, or a respiratory therapist) with neuromuscular diseases. Pay attention to any new symptoms and tell your healthcare team right away. With regular checkups, treatments, and planning, many breathing problems from DM1 can be managed well.



Sleep Studies
If you notice, told that you snore, are waking up tired, or experiencing morning headaches and decreased energy, talk to your doctor about an overnight sleep study (or ***polysomnography***), which can help to diagnose sleep disorders. During the sleep study doctors will record your breathing patterns, ***brain waves***, the oxygen levels in your blood, and your heart rate and breathing during sleep (Hamel, 2022).



Regular Monitoring and Evaluation

Pulmonary Function Tests: Scheduling lung function tests can help catch early signs of respiratory decline. Common assessments include the ***Forced Vital Capacity (FVC)*** and ***respiratory muscle strength tests*** (Ashizawa et al., 2018; Hamel, 2022; Henke et al., 2020). It is important for your doctor to measure your baseline respiratory function at the time of diagnosis and continue with semi-annual or annual examinations to monitor any progression of symptoms or potential complications.



Respiratory Support

Noninvasive Ventilation (***NIV***): If your lungs aren't able to move enough air in or out very well, your doctor may prescribe special masks that deliver pressurized air, or other devices like a continuous **positive airway pressure** (***CPAP***) or ***Bilevel Positive Airway Pressure*** (***BiPAP***) ***machine***, which help to support your breathing. This type of ventilation can support breathing during sleep and even during the day as symptoms progress (Bianchi et al., 2014; Spiesshoefer et al., 2019).

Cough Assist Device: This device is a machine that helps you cough when your muscles are too weak to do it on their own. It works by helping you breathe in deeply and then breathe out forcefully, like a strong, fake cough. This helps clear mucus from your lungs so you can breathe better and stay healthier.



Vaccinations and Preventative Care

Vaccinations: Because DM1 can put you at risk of more prolonged and severe infections, staying up to date on vaccines is a crucial step toward protecting your respiratory health. Annual flu and pneumococcal (***pneumonia***) vaccines are usually recommended (Rimoldi et al., 2024). In general, COVID-19 vaccines are also recommended if you have DM1. Always be sure to consult with your doctor to determine what is best for you.



Care Before, During, and After Surgery: ***Anesthesia*** and pain medications can make respiratory muscle weakness worse. Additionally, it is important to note that individuals affected by DM1 have increased sensitivity to ***anesthesia*** (see **page 63** of the guide for more information on ***anesthesia***). Always remember to remind your medical team of your DM1 diagnosis if ***anesthesia*** will be administered and provide them with the ***Anesthesia Consensus Based Care Recommendations*** for people with myotonic dystrophy (Mathieu et al., 2016).



SCAN TO VISIT MDF's Anesthesia Consensus Based Care Recommendations

Anesthesia Consensus Based Care Recommendations

Medications

Your doctor may prescribe medications to help manage respiratory issues. Some of the medications your doctor may prescribe include:

- ***Mucolytics:*** Break down thick mucus.
- ***Decongestants:*** Decrease swollen tissue.
- ***Antibiotics:*** Combat infections.
- ***Bronchodilators:*** Open the airways and may assist with airway clearance.



Lifestyle Modifications and Education:

It is critical to have an effective plan for airway clearance to use at the first sign of a respiratory illness. There are three ways of clearing the airways: cough, suction, or a ***cough assist device***. Learning clearance techniques to help you cough, like ***breath stacking*** or ***manual cough assist***, may help keep your lungs clear, maintain lung function and prevent infections. Talk with your doctor before using these techniques.

Breath Stacking: This technique helps expand the lungs and clear mucus by taking multiple small breaths in without exhaling. You can practice this technique by taking several small breaths in a row and then exhaling forcefully. You may notice that this helps you clear mucus more effectively.

Manual Cough Assist: This technique, also known as an "abdominal thrusts," allows someone to help provide gentle pressure on your upper abdomen as you cough to boost the strength of your cough and expel any buildup of mucus, making it easier to clear your airways. A respiratory therapist or other health care professional can show you how to do this safely and effectively.



How can I best communicate my needs with my doctor?



Mention Patterns or Changes:

- Talk to your doctors about snoring or breathing difficulty at night and whether you feel tired after sleeping through the night.
- Let your doctor know if it's hard to clear mucus or if your cough feels weak. This may be due to weakened breathing muscles and it is important for your doctor to know.

Explain the Impact on Daily Life: Whether it's walking, sleeping, or talking, your doctor needs to know how symptoms impact your routine.



Understand Potential Testing and Treatment Options:

- You can ask your doctor about ***Pulmonary Function Tests (PFT)*** to determine how well your lungs work.
- You can also ask for a ***Peak Cough Flow test*** which will assess cough strength, which can correlate with the body's ability to fight off future infection.
- You may benefit from a sleep study to evaluate for ***sleep apnea***. Ask for a ***BiPAP*** or ***CPAP machine*** to support your breathing if you do have ***sleep apnea***. *Note: If you have weak breathing muscles and you cannot fully exhale against CPAP pressure, you may require a BiPAP instead of a CPAP. Work with your doctor to determine the safest option for you.*
- If your doctor suspects you may have breathing weakness, they may order an at-home sleep test called ***nocturnal oximetry***. This test monitors your blood oxygen levels and heart rate through the night and can assess need for supplemental oxygen.
- **Possible Treatments: *Noninvasive Ventilation, mechanical cough assist device***, medication, supplemental oxygen

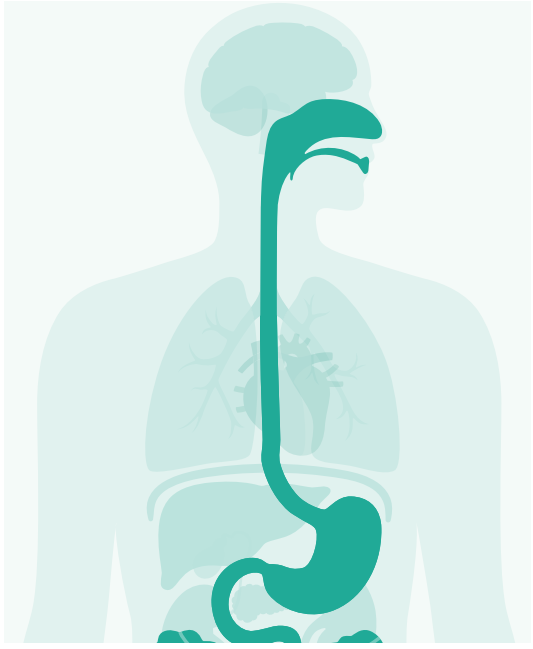
Gastrointestinal System



System Overview

The ***gastrointestinal (GI) system*** helps your body digest food, absorb nutrients, and eliminate waste. It includes organs such as the ***esophagus***, stomach, and ***intestines***. As food is digested, it is moved through your body by muscles that line your digestive organs. In DM1, the GI system can be significantly affected because when the muscles responsible for moving food through your body weaken, problems can arise (Peterson & Cooper, 2022). Depending on your age when you begin to notice symptoms, digestive issues may appear differently. In this section, we'll explore how DM1 impacts the GI system, define the different symptoms seen across types of DM1 (Congenital, Juvenile-Onset, Adult-Onset), and introduce strategies for managing these digestive issues.

Common Symptoms: Gastrointestinal System



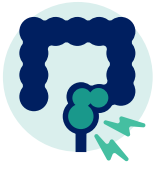


Dysphagia (Difficulty Swallowing)

Weak throat muscles can make it hard to move food or liquids down your throat. It might be difficult to transfer food from your mouth to your ***esophagus***, or it may feel like food is 'getting stuck' as you swallow (Hamel, 2022; Peterson & Cooper, 2022).

Gastroesophageal Reflux Disease *(GERD)*

Acid reflux and ***heartburn*** often result when the muscles that usually keep stomach acid in place weaken and don't close tightly, allowing stomach acid to move into your throat.



Constipation and Diarrhea

When the muscles in the ***intestines*** are weak or don't work well together, it can be harder for food and waste to move through the digestive system the way they should. This is known as ***dysmotility***, which means the muscles aren't coordinating properly. As a result, things might move too slowly or too quickly through the gut. This can lead to irregular bowel movements, like constipation and diarrhea, as well as stomach pain and bloating (Hilbert et al., 2017; Peterson & Cooper, 2022).



Gallbladder Dysfunction

This can occur when the muscles that help your ***gallbladder*** work become weak, which increases your risk of developing ***gallstones***, the hard deposits of bile that form in the ***gallbladder*** (Cardani et al., 2008). If this happens, you may experience symptoms such as pain in the upper right side of your abdomen, nausea, or discomfort after eating fatty foods.

How Gastrointestinal Symptoms May Change Over Time

Gastrointestinal symptoms in DM1 often start subtly and can worsen over time. At first, you might only notice a little bloating or mild swallowing difficulties. Over time, however, these symptoms may become more noticeable, leading to more significant challenges with digestion and swallowing. Some people may develop chronic constipation or severe ***acid reflux***/heart burn ***(GERD)*** that impacts their daily life. The severity of GI symptoms can vary widely between individuals and DM1 types, and not everyone will experience the same degree of symptoms. For instance, some people may only experience issues with swallowing and no issues with bloating or ***acid reflux***. While others may experience significant issues with constipation and little to no issues related to swallowing.

GI symptoms in DM1 are manageable. For some, GI symptoms may require lifestyle adjustments and medications. For others, more specialized treatments like ***feeding tubes*** may be required to ensure proper nutrition (Johnson et al., 2019; Peterson & Cooper, 2022).

Understanding the Different Forms of DM1 and How Digestion is Affected

Congenital DM1 (CDM)
Newborns with CDM often have relatively severe GI issues at birth, including difficulty swallowing, poor feeding, and ***aspiration*** (when food or liquids go into the lungs instead of the stomach) risks. These issues are primarily due to muscle weakness and may require treatments like temporary ***feeding tubes*** to ensure they get enough nutrients (Johnson et al., 2019). There is a treatment called ***dysphagia*** therapy that helps newborns get better at swallowing. Many babies with CDM who get this therapy early are able to start eating by mouth in their first year (Johnson et al., 2019).

Juvenile-Onset DM1
If you have Juvenile-Onset DM1, your GI issues may begin during childhood or adolescence, often as difficulty swallowing or problems like ***acid reflux*** and ***heartburn (GERD)***, or chronic constipation (Peterson and Cooper, 2022). Over time, you may also experience symptoms like bloating and other digestive discomforts.

Adult-Onset DM1
If you have Adult-Onset DM1, GI symptoms are fairly common. You may experience trouble swallowing (***dysphagia***), ***acid reflux (GERD)***, abdominal pain, bloating, or irregular bowel movements (Hamel, 2022). You may also have ***gallbladder*** issues, such as ***gallstones*** (Peterson and Cooper, 2022).

Although these symptoms can be challenging, various treatments and strategies outlined in the next section can help you manage them and maintain your health.

What Can I Do?

Managing gastrointestinal symptoms in DM1 can involve a combination of dietary adjustments, medications, and supportive therapies. Work with your healthcare providers to determine what symptom management strategies work best for you.

Dietary Adjustments

Below are some suggestions and guidelines for ways that you may make dietary adjustments to manage any symptoms you may experience. For more thorough information and guidance on dietary adjustments, please refer to MDF's ***Nutrition Guide for People Living with Myotonic Dystrophy.***

SCAN TO READ MDF's Nutrition Guide

Small, frequent meals: Eating smaller portions more often can help ease constipation, bloating, and help with difficulty swallowing.

Fiber-rich diet: Adding more fiber-rich foods into your diet – like the options listed below – may improve bowel regularity. Drinking enough water throughout the day is also crucial for softening stools. Some fiber-rich options include:

- **Whole Grains:** brown rice, quinoa, oats, whole wheat bread and whole grain pasta.
- **Fruits:** Apples (with the skin), pears, berries (like raspberries, blackberries, strawberries, and bananas).
- **Vegetables:** Broccoli, carrots, sweet potatoes (with the skin), and leafy greens like spinach and kale.
- **Legumes:** lentils, chickpeas, black beans, and kidney beans.
- **Nuts & Seeds:** Almonds, chia seeds, flaxseeds, sunflower seeds.

If you have trouble swallowing, smoothies are a great way to get *proteins*, fruits, vegetables, fiber, and dairy all at once. Sauces and gravies can help food not stick to your throat and pick up small food pieces like rice to reduce the risk of ***aspiration***.

Medications

Talk with your healthcare team about these medications and which option may be best for you.

Prokinetics: These medications can help boost GI function by strengthening contractions and helping muscles coordinate their movement, making it easier for food to move through your digestive system (Peterson & Cooper, 2022).

Antacids and Proton Pump Inhibitors (PPIs): These medications can help treat ***acid reflux*** and calm symptoms of ***GERD***.



Supportive Therapies

Talk with your healthcare team about these medications and which option may be best for you.

Speech and Swallowing Therapy: A speech-language pathologist can show you techniques that make swallowing safer, lowering your risk of ***aspiration*** (Hamel, 2022; Johnson et al., 2019; Zapata-Aldana et al., 2018). For more in-depth information, refer to the ***Dysphagia*** (**page 37**) and Speech Therapy (**page 72**) sections of the guide.



Physical Therapy: Strengthening your abdominal area and improving your overall mobility may help manage some of your GI symptoms (Hartman et al., 2024).

Nutritional Consultation: If you experience difficulty swallowing or experience weight-loss, consulting with a nutritionist or a nutrition specialist may help make sure your dietary needs are met.

Regular monitoring and Specialist Care



Routine GI Evaluations: Regular check-ups with a gastroenterologist are important to manage and prevent complications like ***bowel obstructions*** and severe constipation. Work with your doctor to track your symptoms and watch for complications (Hamel, 2022; Peterson & Cooper, 2022). It may be helpful to find a gastroenterologist who specializes in neuromuscular disorders. If you cannot find one in your area, look for someone who specializes in ***motility*** as they may have more resources and knowledge about GI issues related to DM.



Feeding Tubes*:** In some cases, ***feeding tubes may become necessary to ensure proper nutrition (Zapata-Aldana et al., 2018). There are several kinds of ***feeding tubes***, including ones that are easy to use and don't inhibit daily activities.

Always remember to talk to your anesthesiologist and your healthcare team about your DM diagnosis before any surgical procedure that requires ***anesthesia***. To learn more, refer to *MDF's DM Anesthesia Guide* and the ***anesthesia*** section on **page 63**.

SCAN TO READ DM Anesthesia Guide

How can I best communicate my needs with my doctor?



Request Referrals: Many regular doctors or even neurologists may not refer you to a GI specialist, believing not much can be done to help these issues. However, as just described above, there are options to support GI health. Additionally, if you are experiencing issues related to swallowing, you may request a referral to a speech therapist to improve swallowing.



Explain the Impact on Daily Life: It's important to stress to your doctor how and when these symptoms disrupt your daily life. Provide examples whenever possible and even consider keeping a journal to track symptoms with activities or be able to say for certain what was disrupted because of those issues.

Understand Potential Testing and Treatment Options:



- You can ask your doctor about a ***swallow study*** to assess whether you are at risk for ***aspiration*** or choking.
- Your doctor may suggest a **nutrition consult** to ensure proper dietary modifications and help you meet your nutritional goals.
- **Possible Medications:**
  - → ***Prokinetics:*** These medications can help boost GI function by strengthening contractions and helping muscles coordinate their movement, making it easier for food to move through your digestive system (Peterson & Cooper, 2022).
  - → ***Antacids and Proton Pump Inhibitors (PPIs):*** These medications can help treat ***acid reflux*** and calm symptoms of ***GERD***.

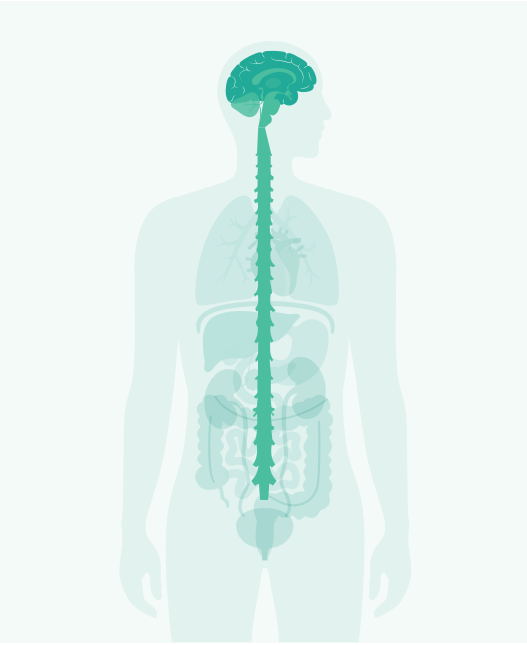
Central Nervous System



System Overview

The ***central nervous system*** (***CNS***) – made up of your brain and ***spinal cord*** – controls much of what you do, from moving and feeling to thinking and behaving. In DM1, the CNS is affected, leading to ***neurological*** and ***cognitive symptoms***. This section will explore the effects of DM1 on the CNS, outline some common symptoms, highlight the different symptoms seen across types of DM1 (Congenital, Juvenile-Onset, Adult-Onset), and discuss strategies to help manage these CNS-related challenges in DM1.

Common Symptoms: Central Nervous System



When the CNS is affected in DM1, you may notice changes in how you think, feel, or behave. ***Cognitive*** issues can include intellectual disabilities, speech and language delays, or ***visuospatial*** difficulties (trouble making sense of what you see around you). Behavioral changes can include ***apathy*** (not feeling motivated or interested, even in things that normally matter to you), depression, or social isolation and withdrawal. Because these changes can develop gradually, you may not always be aware of them, even though family members or close friends might notice them sooner. These symptoms can vary widely from person to person, but below are some of the more common ones you may experience:



Cognitive Impairment

You may find it harder to plan, focus, or juggle multiple tasks at the same time. You might notice more difficulty with organizing and planning daily activities or problem-solving. It may help to think about DM1's impact on your CNS like your brain's 'traffic lights' running a bit slower than they had in the past. For example, tasks like paying bills, keeping appointments, or managing a schedule may start to feel more demanding than before (Liu et al., 2021; Miller et al., 2021). It can be difficult to notice changes in symptoms in real time, because progression is gradual. However, being aware of your symptoms now can be helpful when you need a comparison in the future.



Mental Health Issues

People living with DM1 may experience depression or anxiety (Miller et al., 2021). Depression can be described as a persistent sadness and a lack of interest or pleasure in previously rewarding or enjoyable activities. Anxiety, on the other hand, is described as an excessive and persistent fear and worry about situations in everyday life. Depression and anxiety can sometimes lead to an increased risk in using drugs and/or alcohol in unhealthy ways. There are other mental health issues you may experience while living with DM1, like ***apathy***, difficulty concentrating, or ***executive function*** deficits (trouble

with planning, organizing, or staying focused on tasks). If you have any questions about how DM might affect mental health and what can be done to help manage these issues, refer to the ***Myotonic Dystrophy and Mental Health Handbook***.

SCAN TO READ Myotonic Dystrophy and Mental Health Handbook.



Behavioral and Personality Changes

You or a loved one may notice that you react to things differently over time, with changes in your behavior or personality that don't feel like the "old you". These symptoms and changes are related to how DM1 can affect the brain. These changes can make you feel that it's harder to feel motivated, start new tasks, or keep up with routines you used to enjoy (Meola et al, 2003; Gallais et al., 2015; Hamel, 2022; Miller et al., 2021; Thornton, 2014).



Excessive Daytime Sleepiness (EDS)

Because sleep may be disrupted in DM1, you might feel unusually tired during the day, potentially nodding off or falling asleep when you would normally be awake. This is referred to as ***excessive daytime sleepiness*** and can progress to a point where people living with DM1 have reported it affects daily activity and quality of life (Hamel, 2022; C. Heatwole et al., 2012; Miller et al., 2021), even in children living with CDM and childhood DM (Johnson et al., 2014). EDS can impact your work, responsibilities at home, social life, and can increase your risk of accidents (Laberge et al., 2013). A study that looked at many DM1 patients across many ages, showed that fatigue or tiredness can have an even greater impact on your quality of life as you age (Heatwole et al., 2012).



Anesthesia Sensitivity

Surgeries can be more complicated for people living with DM because they may be extra sensitive to ***anesthesia***, the medicine used to put you to sleep. This sensitivity means you might take longer to wake up, have trouble breathing, or react badly to the drugs (Ashizawa et al., 2018; Johnson et al., 2019). Because of these risks, it is very important to tell your surgical team that you have DM1 before any procedure. Even though researchers are still learning why this happens, changes in the brain and nervous system caused by DM1 likely play a role in this increased sensitivity. If you are preparing for a surgery or want to understand the causes of ***anesthesia*** sensitivity, how it affects someone living with DM, and what to share with your healthcare team, refer to the ***Anesthesia*** section in Part 6 of this guide, **page 63**.

How Central Nervous Symptoms May Change Over Time

DM1-related changes in your thinking and behavior usually happen slowly, although the pace can vary quite a bit from person to person, even within the same family. Generally, your ***cognitive*** and behavioral symptoms may worsen over time, especially without support such as ***cognitive behavioral therapy*** or structured routines like exercise and a regular sleep-wake schedule (Okkersen et al., 2018), which underscores the benefit of having a ***psychologist*** or ***psychiatrist*** as part of your care team. If you have CDM or Juvenile-Onset DM1, these ***cognitive*** and behavioral problems may appear earlier and progress more quickly. If you have Adult-Onset DM1, you might notice subtle shifts or changes in some symptoms, while noticing others haven't changed. For example, you may notice ***apathy***—your lack of interest—progressing or getting worse, while your feelings of tiredness or daytime sleepiness remain stable. On the other hand, you might meet someone else living with DM who has no ***apathy*** but has sleepiness that is getting progressively worse. Because the brain is so complex, scientists and clinicians continue to study exactly how DM1-related brain changes are connected to the overall progression of the disease (Nutter et al., 2023; Gliem et al., 2019).

Understanding the Different Forms of DM1 and How the Central Nervous System is Affected

Congenital DM1 (CDM)

- ***Cognitive Impairment:*** Babies and young children living with CDM often face significant learning and developmental hurdles, which can include learning difficulties, speech delays, and a higher likelihood of ***autism spectrum disorders*** (Johnson et al., 2019).
- **Behavioral Challenges :** If your child has CDM, you might notice they're more shy and withdrawn in social situations. You may even notice more aggressive behaviors in early childhood (Johnson et al., 2019). If your child is experiencing this, consider reaching out for extra help like behavioral therapy, social skills groups, or family counseling. Getting the help and support that you need can make a big difference (Johnson et al., 2019). To learn more about behavioral challenges and getting support, read *MDF's Mental Health Handbook.*

Juvenile-Onset DM1

- ***Cognitive* and Learning Challenges:** If you have Juvenile-Onset DM1, you may experience learning difficulties, such as struggling to concentrate, remember new information, or keep up with assignments. These symptoms often first appear during your school years (Hamel, 2022; Johnson et al., 2019). While these ***cognitive*** challenges can change over time, understanding them and seeking support can help improve your academic experiences and support meaningful social connections.
- **Behavioral Changes:** During adolescence (usually between 10-19 years old), you may also begin to notice symptoms such as anxiety, depression, or social withdrawal, or you may find that these symptoms become more noticeable over time. This is not uncommon in Juvenile-Onset DM1, but addressing these issues early can make a big difference later. To learn more about behavioral changes and getting support, read *MDF's Mental Health Handbook.*

Adult-Onset DM1

- **Subtle *Cognitive* Decline: *Cognitive symptoms*** in Adult-Onset DM1 may be mild for you at first but can gradually become more noticeable over time. These can include difficulties with memory, ***executive function*** (such as planning ahead or switching between tasks), and decision-making (Gallais et al., 2015; Hamel, 2022). These changes can, however, remain mild for many years.
- **Mental Health:** As physical symptoms become more obvious, you may experience feelings of anxiety, or feel depressed. This is not an uncommon experience – you are not alone. More information can be found in *MDF's Mental Health Handbook.*

Although these symptoms can be challenging, various treatments and strategies outlined in the next section can help you manage them and maintain your health.

SCAN TO READ SCAN TO READ Myotonic Dystrophy and Mental Health Handbook.

MDF's Mental Health Handbook

What Can I Do?

Good management of CNS symptoms involves a multidisciplinary approach with a variety of experts, such as neurologists, ***psychiatrists***, and therapists, who can coordinate to create the best plan for you (Hartman et al., 2024).



Neuropsychological Evaluations

Regular visits with a ***psychiatrist*** or neurologist can help catch early signs of symptoms you may experience, like ***apathy***, depression, or ***cognitive*** deficits, and help identify specific treatment strategies like ***cognitive behavioral therapy (CBT)*** (Ashizawa et al., 2018; Hamel, 2022; Liu et al., 2021; Miller et al., 2021).



Behavioral Interventions

Treatments like ***cognitive behavioral therapy (CBT)*** and other forms of counseling may help you manage anxiety, depression, and behavioral changes like ***apathy*** and decreases in ***executive function*** (the skills and processes that allow you to plan ahead or transition between tasks) (Ashizawa et al., 2018; Okkersen et al., 2018).



Medication

Talk with your healthcare team about medications that can help manage symptoms. Depending on your needs, physicians might suggest ***antidepressants***, anti-anxiety medications (***anxiolytics***), or ***stimulants*** for ***excessive daytime sleepiness*** (Annane et al., 2024; Hartman et al., 2024; Liu et al., 2021). Work closely with your healthcare team to find the right medication and treatment, as some medications may have side effects that need to be monitored to be used safely and effectively.



Supportive Strategies

Staying mentally active by engaging in social activities, maintaining structured routines (such as consistent exercise and a regular sleep-wake schedule), and doing mental exercises (such as relaxation techniques like ***autogenic training*** [quietly repeating calming phrases to yourself], breathing techniques, and meditation and mindfulness) may help maintain your ***cognitive*** function, manage mental health, and improve quality of life. For more information, see the *MDF Mental Health Handbook.*



Education and Support for Caregivers

Certain symptoms, like ***excessive daytime sleepiness*** or mood changes, can be so persistent and overwhelming that you may not fully realize their impact. In these cases, your family members or caregivers often can play a key role in identifying how these symptoms affect daily life and overall quality of living and can provide helpful feedback. Tools and training on how to cope with and support someone's mood changes, sleep issues, or anxiety can make a big difference for everyone involved. Your caregiver may want to consider joining one of MDF's *virtual caregiver support groups*, watching resources for caregivers on MDF's *Digital Academy*, or utilizing resources like MDA's *Guide for Caregivers*.



Preventive Care for Anesthesia Sensitivity

As an individual living with DM1, you can have an increased sensitivity to ***anesthesia***, which can lead to severe complications during surgical procedures (Ashizawa et al., 2018; Johnson et al., 2019). It is very important that you tell your healthcare providers, especially anesthesiologists and surgical care team, about your DM1 diagnosis. To learn more about what to share with your healthcare team, refer to the ***Anesthesia*** section on **page 63**.

How can I best communicate my needs with my doctor?



Prepare Ahead of Time: Make a list of mental health symptoms you've noticed (e.g., mood changes, anxiety, lack of motivation, memory issues) and how they affect your daily life.

Be Honest and Specific: Share how you're feeling emotionally and mentally, not just physically. Use clear examples, like trouble concentrating at work or withdrawing from social activities.



Mention Patterns and Changes: Let your provider know if symptoms have gotten worse, started recently, or changed over time.

Explain the Impact on Daily Life: Describe how your mental health symptoms are affecting your routines, relationships, sleep, or ability to manage your health.

Connect Mental and Physical Health: Mention if emotional symptoms seem to worsen with physical symptoms like fatigue or pain, or if they improve.



Use Educational Materials: Share written resources about DM and mental health (such as those from MDF) to help your provider better understand the condition.

Communicate Risks: Your doctor should be aware of the risks of ***anesthesia*** for people living with DM. To learn more about what to share with your healthcare team, refer to the ***Anesthesia*** section in Part 6 of this guide, **page 63**.

Follow Up: If your doctor doesn't address your concerns right away, bring it up again at a future visit or consider seeking a second opinion.



Bring a Support Person: If you feel nervous or unsure about how to talk about these topics, consider bringing someone you trust to help advocate for you.

Understand Potential Testing and Treatment Options:

- Your doctor should do routine neurology evaluations to get a baseline understanding of your health and determine if/how it's changing over time.
- If you're experiencing ***cognitive***, memory issues, or fatigue, ask your doctor for a ***neuropsychological evaluation***: a test that measures how a person's thinking, emotions, and behavior are working. The test starts with an interview, along with a review of medical history and background. You then take a series of standardized tests that look at how different areas of the brain function like attention, memory, language, problem solving, and personality or mood.
- Ask your doctor for a referral to outpatient therapy for mental health or behavioral problems.
- **Possible Medications/Treatments:**
  - → ***Antidepressants/anxiolytics:*** Balance brain chemicals to reduce anxiety or depression and calm overactive stress signals.
  - → **CNS stimulant:** increase wakefulness-related brain activity to reduce ***excessive daytime sleepiness***.
  - → **Nonpharmacologic *CBT*:** retrain brain-behavior patterns to improve fatigue, sleep, and coping without medications.



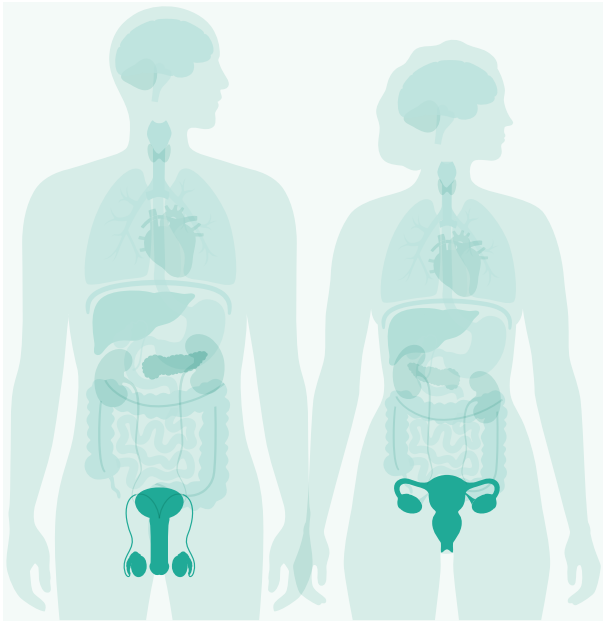
Reproductive System



System Overview

The ***reproductive system*** is made up of the organs, ***hormones***, and processes (such as producing mature reproductive cells like sperm or ***egg cells***) that allow people to have children. In men, these organs include the ***testes*** and ***prostate***; for women, they include the ***ovaries***, ***fallopian tubes***, ***uterus***, ***vagina***, ***clitoris***, and ***vulva***. In both men and women, ***hormones*** like ***testosterone*** and ***estrogen*** act like messengers, helping to regulate important functions like ***fertility***, sexual development, and pregnancy. In DM1, muscle weakness and hormone imbalances can affect how well this system works. Both men and women living with DM1 may experience challenges related to ***fertility***, sexual function, and hormone regulation. This section looks at how DM1 might affect men and women differently, how ***reproductive system*** is affected across types of DM1 (Congenital, Juvenile-Onset, Adult-Onset), and ways to manage these symptoms.

Common Symptoms: Reproductive System



DM1 can affect the ***reproductive system*** in distinct ways for men and women:

In Women

Reduced Ovarian Reserve **and Early** ***Menopause:*** If you are a woman living with DM1, you might have fewer eggs available than usual, and ***menopause*** (the stage when menstrual periods stop) may come earlier, which can shorten your ***fertility*** window (Srebnik et al., 2014). If you experience early ***menopause***, you may notice symptoms like hot flashes or mood changes at an age younger than you would expect.

Pregnancy Complications: DM1 can raise the risk of ***ectopic pregnancies*** (pregnancy that develops outside the ***uterus***), ***miscarriages***, ***preterm labor*** or ***stillbirths*** (Ashizawa et al., 2018; Ho, 2015). You may also be prone to producing too much ***amniotic fluid***, the fluid that cushions a fetus while it develops in the ***uterus*** (a condition known as ***polyhydramnios***), or heavier-than-normal bleeding after delivery (Yee et al., 2017).

Hormonal Imbalances: Your body may have a harder time regulating hormone levels, and you may experience ***irregular menstrual cycles***, changing energy levels, or mood swings (Hartman et al., 2024).

In Men

Lower ***Fertility*****:** Men living with DM1 may experience smaller ***testes*** or a decrease in their size over time, lower sperm counts, or lower ***testosterone***. These issues can make it more difficult for you to father a child naturally (Hamel, 2022).

Hormonal Imbalances: Your body may have a hard time producing certain ***hormones***, like ***testosterone*** (which helps maintain energy and sexual function), to the level they are needed. If you are experiencing lower level of ***testosterone***, you may notice unusual tiredness and a decreased sex drive (Hartman et al., 2024).

How Reproductive Symptoms May Change Over Time

Generally, your reproductive challenges become more noticeable as DM1 progresses. The severity and age of onset of your DM1 also play an important role: if you have a more severe or earlier-onset form of DM1, you may see these changes happen sooner and more dramatically. It's also important to note that pregnancy-related risks increase with the severity of DM1 symptoms in the mother, as well as if the fetus (developing infant) is affected (even if the woman is not affected).

Understanding the Different Forms of DM1 and How Fertility and Reproduction are Affected

Congenital DM1 (CDM)

Effect on Future Children: If a woman living with DM1 is pregnant with a child who has congenital DM (CDM), the pregnancy may come with added challenges. There is a much higher risk for extra ***amniotic fluid*** (***polyhydramnios***), ***stillbirth***, breech positioning (when a fetus is buttocks or feet first rather than head-down), and difficulty with fetal positioning during birth increasing the need for cesarean delivery (a surgical procedure used to deliver a baby through incisions in the mother's abdomen and ***uterus***). The baby may have trouble breathing or swallowing after birth, which means they might need extra medical support right away. Knowing about these risks ahead of time gives the healthcare team a chance to prepare and make sure the right care is in place as soon as the baby is born (Ashizawa et al., 2018; Brussé et al., 2020).

Juvenile-Onset DM1

Fertility*, Reproductive Challenges and Pregnancy Complications:** If you are living with DM1 (Juvenile & Adult-Onset), you may not have seen any signs or symptoms related to ***fertility during early childhood. However, you may notice symptoms related to reproductive health as you complete puberty and emerge from adolescence (usually between 10-19 years old) and enter adulthood (Ashizawa et al., 2018). Symptoms you might notice can include early signs of hormonal imbalances.

Adult-Onset DM1

- **In men,** you might notice smaller ***testes*** and your doctor may tell you that you have lower ***testosterone*** levels.
- **In women,** you may experience abnormal periods and you may have a higher chance for potential difficulties in future pregnancies, like shorter ***fertility*** window or pregnancy complications like preterm delivery or ***postpartum bleeding*** (Ashizawa et al., 2018; Brussé et al., 2020; Srebnik et al., 2014; Yee et al., 2017). With the right care and support, however, most people who want to become pregnant and parent a child can safely do so, although additional care, resources, and planning may be needed.

Although these symptoms can be challenging, various treatments and strategies outlined in the next section can help you manage them and maintain your health.

What Can I Do?

While DM1 can affect things like ***fertility***, pregnancy, and hormone levels, there are steps you can take to manage these challenges. Working closely with healthcare providers and making informed decisions can help support reproductive goals and improve health outcomes. The following section covers key topics such as preconception counseling, pregnancy management, and hormonal treatments to help you better understand your options and feel more confident in your care.



Fertility Planning & Assessment

You can talk to a doctor about tests to check egg and sperm health to help you understand your chances of pregnancy and explore family planning options. While it is not always possible to completely predict the total count of remaining, immature eggs (***oocytes***) in a woman's ***ovaries*** (***ovarian reserve***), there are ways to assess ***ovarian reserve***. There are also ways to assess sperm counts and motility. These can help provide an assessment of the probability of successful spontaneous conception (getting pregnant without help from a ***fertility specialist***) for most couples. Since DM1 is ***autosomal dominant*** and has a 50% chance of being passed on, (see Part 3 of this guide on **page 13** for more information), you may also consider working with a ***fertility specialist*** who can consult with you about options such as sperm banking or egg freezing. These conversations can help you better understand your options and make informed choices. For more details and guidance, see the Family Planning section on **page 80**.

It is also important to note that DM1 does not affect everyone equally, and there are people who will have no difficulty conceiving a child. Even with reduced sperm counts or ***ovarian reserve*** findings, the possibility of spontaneous pregnancy is typically not eliminated, only reduced. Therefore, for couples or people who do not want to have a child, DM1 alone should not be considered effective contraception. Couples that are not interested in having a child should use appropriate contraceptive methods regardless of ***fertility*** assessments.



Preconception Counseling

If you're thinking about starting a family, talking with a ***genetic counselor*** can be a helpful first step. ***Genetic counselors*** can explain the chances of passing on the DM1 gene and walk you through testing options that are available, such as ***preimplantation genetic diagnosis (PGD***) and ***prenatal testing*** (Ashizawa et al., 2018). For more information, see the Family Planning section on **page 80**.

Pregnancy Management

For people with DM1 who are pregnant, it's important to receive care from a ***high-risk obstetrician*** (also called a ***Maternal-Fetal Medicine doctor*** or ***Perinatologist***), someone who specializes in complex pregnancies and understands myotonic dystrophy. Extra monitoring during pregnancy may help catch and manage many potential issues like early labor or heavy bleeding after delivery (Brussé et al., 2020; Higgs et al., 2019). It is also important to note that there may be a risk of personal DM symptoms progressing during pregnancy, and preparing for delivery will need to include physical supports and planning (Johnson et al., 2015). For more detailed information and care recommendations, check out the *MDF Pregnancy Guide.*

SCAN TO READ MDF's Pregnancy Guide



Hormonal and Menstrual Care Treatments

Hormonal therapies may be used to manage symptoms like early ***menopause*** or low ***testosterone*** levels. They can also be used to improve menstrual symptoms and prevent pregnancy. For teens who are struggling with menstrual symptoms, such as ***anemia*** (not having enough healthy red blood cells) related to heavy periods or hygiene, many hormonal therapies may be used to improve symptoms. For older women who are not seeking to become pregnant, elimination of periods through both hormonal and surgical options may also be of benefit for preventing symptoms and improving quality of life. Talk with your care team to determine what treatments may be helpful for you.



How can I best communicate my needs with my doctor?



Bring up Symptoms: Issues like irregular or heavy periods, low ***libido***, or ***fertility*** concerns may be connected to DM and are important to mention.

Share Your Values: If you're thinking about having children now or in the future, sharing your goals with your doctor will help them offer the right support and resources.

Mention Patterns and Changes: Let your doctor know if you're noticing changes like fatigue, mood shifts, or other symptoms that may be tied to hormone levels.



Request Referrals: Ask for a referral to a ***reproductive endocrinologist***, a gynecologist who specializes in complex or high-risk care, ***fertility specialist***, or ***genetic counselor*** if you need more specialized care or advice.

Use Educational Materials: Bring educational resources (*like those from MDF*) to help explain how DM can affect reproductive health if your provider isn't familiar with the condition.



SCAN TO READ MDF's Educational Resources

Endocrine System



System Overview

The ***endocrine system*** is responsible for making and releasing ***hormones***, chemical messengers that help regulate many important functions in the body, like metabolism, growth, and reproduction. In people living with DM1, the ***endocrine system*** can be disrupted, leading to hormone imbalances that affect overall health. This section explains how DM1 impacts the ***endocrine system***, describes common symptoms, highlights how these symptoms can differ across types of DM1 (Congenital, Juvenile-Onset, Adult-Onset), and offers an overview of how these issues may progress and how they can be managed.

Common Symptoms: Endocrine System



In DM1, the ***endocrine system's*** involvement can lead to a variety of symptoms:



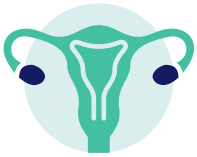
Insulin Resistance and *Diabetes*

If you're living with DM1, your body may become resistant to insulin, a hormone that helps keep your blood sugar in check (Hamel, 2022; Rimoldi et al., 2024). Without treatment, this ***insulin resistance*** can increase your chances of developing ***type 2 diabetes*** over time. Signs to watch for include feeling unusually tired after eating, craving sweets more often, or feeling unusually thirsty.



Thyroid Dysfunction

An underactive ***thyroid***, also called ***hypothyroidism***, is fairly common if you're living with DM1 (Rimoldi et al., 2024). It can cause symptoms like feeling very tired, gaining weight, or being more sensitive to cold. If you notice any of these changes, it's a good idea to talk to your primary care physician and/or endocrinologist.



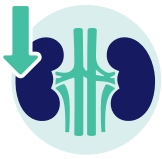
Reproductive Hormone Imbalances

DM1 can affect the ***reproductive system*** in both men and women. For men, it may lead to lower ***testosterone*** levels, which can cause symptoms like low energy, trouble with erections, or challenges with ***fertility*** (Hamel, 2022; Rimoldi et al., 2024). Women may experience irregular periods, early ***menopause***, or trouble getting pregnant. To learn more, refer to the ***Reproductive System*** section on **page 47**.



Adrenal and *Parathyroid* Gland Issues

In rare cases, DM1 can affect glands in the body that produce important ***hormones***, including the ***adrenal glands***, which help you manage stress, and the ***parathyroid glands***, which regulate your calcium levels (Winters, 2021). When these glands aren't working properly, you may experience symptoms like fatigue or problems with your bone health (Hlaing et al., 2019; Rimoldi et al., 2024).



Adrenal Insufficiency

Adrenal insufficiency can happen when your ***adrenal glands*** do not produce enough ***hormones***, such as cortisol, which helps your body respond to stress, illness, and injury. Normally, when your body has enough cortisol, your brain signals your ***adrenal glands*** to stop making it. However, long-term use of steroid medications can disrupt this balance, causing your brain to think you have enough cortisol when you don't. As a result, your ***adrenal glands*** may temporarily slow down or stop making cortisol, leading to adrenal insufficiency. This condition can cause fatigue, nausea, stomach pain, low blood sugar, and, in severe cases, a medical emergency called an adrenal crisis. To lower this risk, your steroid doses should be reduced gradually, your hormone levels should be monitored closely, and stress doses (emergency steroid doses) should be used when needed. This is especially important if you are living with myotonic dystrophy and take steroids long-term (F. Beuschlein et al., 2024).



Calcium Metabolism Issues

DM can affect how the body senses and manages calcium, a mineral that's important for strong bones, healthy muscles, and many other body functions. When your calcium levels aren't properly regulated, your body may mistakenly allow too much calcium to build up in the blood. This condition is known as ***hypercalcemia*** (Hlaing et al., 2019) and can result in increased thirst, frequent urination, digestive issues, bone pain and muscle weakness, and trouble focusing, confusion and fatigue (Mayo Clinic, 2024).



Osteoporosis and Fragility Fractures

You may have a greater risk of developing osteoporosis (a condition where your bones become thinner and weaker) and fragility fractures (bone breaks that happen from a minor fall or simple daily movement) if you are living with myotonic dystrophy. Many people with DM experience hormonal imbalances, such as low ***testosterone***, vitamin D deficiency, or problems with ***parathyroid*** hormone regulation; all of these can weaken your bone structure over time. In addition, your muscle weakness and loss of muscle mass can reduce your mobility, which decreases your bone strength and increases your risk of fragility fractures (F. Danckworth et al., 2014; SJ Winters et al., 2021).

In general, endocrine symptoms in DM1 can vary widely from person to person, but with proper care and attention, many of these endocrine issues can be managed effectively, with the help of your healthcare team.

How Endocrine Symptoms May Change Over Time

For many people with DM1, endocrine issues such as changes in hormone levels tend to develop gradually over time. You may experience ***insulin resistance***, which can become more noticeable as you age and may increase your risk of developing ***type 2 diabetes***. Hormone imbalances related to your ***reproductive system*** can also become more apparent in adulthood, sometimes showing up as irregular or missed periods, low energy, reduced sex drive, or difficulty getting pregnant. Changes in your ***thyroid*** and other hormone-producing glands can vary from person to person, which is why regular checkups and early treatment are important for you.

Understanding the Different Forms of DM1 and How the Endocrine System is Affected

Congenital DM1 (CDM)
Major hormone problems in very young children with DM1 are not common, but may emerge later in life, particularly in the transition through adolescence and into adulthood.

Juvenile-Onset DM1
For individuals with Juvenile-Onset DM1, signs of ***insulin resistance*** often begin to appear during adolescence. Around the same time, males may also show signs of reproductive hormone imbalance, which can affect puberty and ***fertility.*** If you have Juvenile-Onset DM1, signs of ***insulin resistance*** may begin to appear during adolescence (usually 10-19 years). Around the same time, if you are male, you may also show signs of reproductive hormone imbalance, which can affect puberty and ***fertility*** (Ashizawa et al., 2018; Winters, 2021).

Adult-Onset DM1
If you are living with DM1, it is common to experience endocrine symptoms, such as ***insulin resistance***, ***thyroid*** issues, and reproductive hormone imbalances (Hamel, 2022; Rimoldi et al., 2024; Winters, 2021). Because these changes can affect your overall health, regular monitoring and early intervention are important for managing your symptoms and preventing complications.

Although these symptoms can be challenging, various treatments and strategies outlined in the next section can help you manage them and maintain your health.

What Can I Do?

Taking care of your endocrine health while living with DM1 is an important part of managing the condition. Hormone changes can happen slowly and may be easy to miss, which is why regular checkups, healthy lifestyle habits, and the right medical care all play a role in staying well. This section will walk through steps you can take to stay on top of your hormone health, including routine evaluations, treatment options like medications or ***hormone therapy***, and when to see a specialist. A helpful first step, especially around the time of diagnosis, is to get a baseline for things like hormone levels, blood sugar control, and cholesterol. These numbers give your doctor a clearer picture of your health and help track any changes over time.



Regular Endocrine Evaluations
Screen for *Diabetes*: Getting yearly blood tests, like fasting blood sugar or ***HbA1c***, which shows your average blood sugar levels over the past few months, can help spot early signs of ***insulin resistance*** or ***diabetes*** (Ashizawa et al., 2018; Rimoldi et al., 2024).



Check *Thyroid* Function: Speak with your doctor about checking how well your body is producing ***thyroid hormones*** like ***thyroid-stimulating hormone (TSH)*** and ***free T4*** levels (Ashizawa et al., 2018; Hamel, 2022). Asking your doctor to check for these ***hormones*** at least every 3 years is important. Imbalances of these ***hormones*** can help in early detection and treatment of ***thyroid*** problems.



Monitor for Osteoporosis and Fragility Fracture Risk: It is important to assess fall risk, consider screening for hormonal deficiencies (***testosterone***, ***estrogen***, growth hormone (GH), Insulin-like growth factor 1 (IGF-1), ***parathyroid*** hormone, and insulin), measure vitamin D/calcium levels, and get bone density scans (***DEXA***) for prevention and treatment of osteoporosis and fractures.

Medical Management



Insulin and Oral Medication: If you are diagnosed with ***diabetes*** or ***insulin resistance***, your doctor may recommend insulin injections or medications to help maintain stable blood sugar levels (Rimoldi et al., 2024).



Hormone Replacement Therapy: ***Hypothyroidism*** can usually be managed with ***thyroid*** hormone replacement therapy, which gives your body the ***hormones*** it's not making on its own. This treatment can help improve energy levels, metabolism, and other functions that depend on healthy ***thyroid*** activity. For men with low ***testosterone***, ***testosterone*** therapy might also be helpful (Winters, 2021). These treatments may ease symptoms and help you feel more energetic.

Lifestyle Modifications

MDF's Exercise Guide



Diet and Exercise: A balanced diet and regular, low-impact physical activity like walking can help you manage ***insulin resistance*** and your overall metabolic health. For guidance on some low-impact physical activities and other exercises, take a look at *MDF's Exercise Guide for People Living with Myotonic Dystrophy.*

Weight Management: Managing a healthy weight can ease the stress on your ***endocrine system*** and reduce your risk of ***diabetes***.

Specialist Care



Endocrinologist: Working with an endocrinologist who is understanding, supportive, and knowledgeable about or is willing to learn about DM1 can ensure that all aspects of endocrine health are addressed, including more complex issues like adrenal or ***parathyroid*** gland dysfunction.

Fertility Specialist: If hormone imbalances or other issues are impacting your plans for a family, consultation with a ***fertility specialist*** can help you learn about available options.

Remember, you know your body best. *If something feels off – like sudden weight changes, extreme fatigue, or unexpected shifts in mood – reach out to your care team. Early detection and quick interventions can help you stay one step ahead.*

How can I best communicate my needs with my doctor?



Share Your Symptoms: Keep a list of symptoms, even if they seem unrelated. Endocrine issues can show up in subtle ways.

Share Your Family History: If others in your family have ***thyroid*** disease, ***diabetes***, or hormone imbalances, that information can guide your doctor's approach.

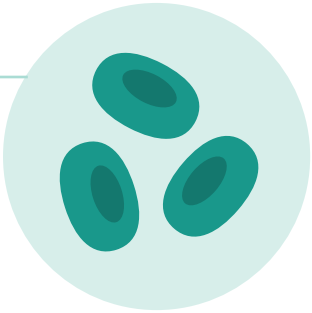
Understand Potential Testing and Treatment Options: If you have ***fertility***, ***libido***, or menstrual concerns, ask your doctor for testing to check your sex hormone levels (***testosterone, LH, FSH, estradiol***). Other testing you can discuss with your doctor includes:

- Checking Vitamin D levels and a bone density scan (***DEXA***) due to increased risk of osteoporosis.
- Testing fasting glucose and blood sugar (***HbA1C***) due to increased risk of ***insulin resistance***/***diabetes***.
- Checking ***TSH*** and ***free T4*** levels to check for ***hypothyroidism***.



- **Possible Medications:**
  - → ***Thyroid*** **Hormone Replacement:** restores metabolism and energy when the ***thyroid*** is underactive.
  - → **Oral *Diabetes* Medications:** lower blood sugar and improve body's insulin response to manage ***diabetes***.
  - → **Vitamin D:** strengthens bones and supports calcium balance when levels are low.

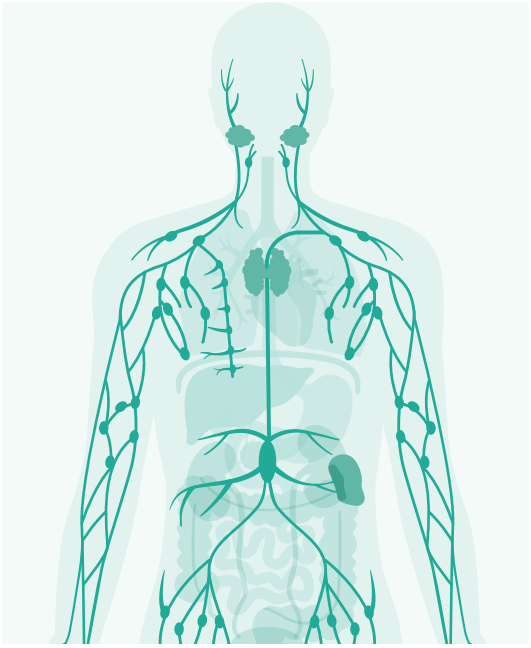
Immune System



System Overview

The ***immune system*** plays an important role in protecting your body from infections and other harmful invaders by producing important protective ***proteins*** called ***antibodies*** and other defense mechanisms. In DM1, the ***immune system*** can be subtly affected, usually through lower levels of the ***antibodies*** that circulate in the blood called ***immunoglobulins***. This section covers how DM1 might affect your ***immune system***, describes general symptoms related to the ***immune system***, discusses how these symptoms are seen across types of DM1 (Congenital, Juvenile-Onset, Adult-Onset), and outlines the progression and management strategies for your symptoms.

Common Symptoms: Immune System



In people with DM1, the main ***immune system*** issue is something called ***hypogammaglobulinemia***. This means there's a slightly lower amount of ***immunoglobulins***, or ***antibodies*** that help you fight off infections, in the blood. Although your body produces the normal number of ***antibodies***, they do not last as long in your blood, leading to lower overall levels. The reassuring part is that this drop doesn't lead most people with DM1 to get sick more often than is expected. Still, it's important to keep up with regular check-ups so any potential issues can be caught early.

How Immune Symptoms May Change Over Time

The way that DM1 affects the ***immune system*** is different from how it may affect other body systems. Any changes in immune function usually stay stable over time and don't tend to get worse as DM1 progresses. However, because DM1 can affect many body systems, the ***immune system*** can sometimes be indirectly impacted. For instance, if your lungs aren't working as well, you might be more likely to get respiratory infections. That's why it's important to focus on your overall health and stay on top of regular check-ups.

Understanding the Different Forms of DM1 and How the Immune System is Affected

Congenital DM1 (CDM)

Rare Immune System Involvement: Infants and young children living with CDM usually have little to no ***immune system*** problems, though it is still important to look out for any signs of infections or unusual symptoms. However, frequent respiratory infections may be a sign that the ***respiratory system*** may be affected by DM. To learn more about the ***respiratory system***, see **page 33**.

Juvenile-Onset DM1

Minimal Immune System Involvement: If you have Juvenile-Onset DM1, ***immune system*** problems are not common. Most children and adolescents with this form of DM1 do not experience major issues with immunity. However, it is still important for you and your caregivers to watch for signs of infections or any unusual symptoms, especially if they happen more often than expected. Frequent respiratory infections may be a sign that your ***respiratory system*** is affected by DM1. To learn more about breathing and lung health, see **page 33**.

Adult-Onset DM1

- **Decreased Blood Antibody Levels (*Hypogammaglobulinemia*) :** Most adults with DM1 do not experience more frequent infections (El-Wahsh et al., 2024). However, monitoring your blood for antibody levels can help your doctor know if you have an overall lower level of ***antibodies***. This is an area of ongoing research in the DM field.
- ***Pilomatrixoma*:** If you are living with DM1, you may be more likely to develop a benign (non-cancerous) skin tumor called a ***pilomatrixoma***. These tumors usually show up as a small, firm lump under the skin on the head or neck (Hamel, 2022; Thornton, 2014). While scary sounding, these benign tumors can be removed surgically. Always remember to talk to your anesthesiologist and your healthcare team about your DM diagnosis before any surgical procedure that requires ***anesthesia***. To learn more, refer to MDF's *DM Anesthesia Guide* and the ***anesthesia*** section on **page 63**.

SCAN TO READ DM Anesthesia Guide

Although these symptoms can be challenging, various treatments and strategies outlined in the next section can help you manage them and maintain your health.



What Can I Do?

Although the ***immune system*** is generally less affected in DM1 than other body systems may be, regular health monitoring and preventive care are key to maintaining overall well-being and addressing any issues that may arise. By being proactive, you can spot potential issues early to ensure you keep feeling your best.



Regular Monitoring

Check Antibody Levels: This can be done through simple blood tests to help you keep track of antibody levels, specifically: serum ***IgG, IgA, IgM, Gamma globulin, albumin,*** and ***lymphocytes*** (El-Wahsh et al., 2024).

Regular Skin Checks: Watch for unusual bumps or lumps and speak to a dermatologist or your primary care physician if you notice any.



Preventive Care

Stay up to date with Vaccines: Keeping current with shots like the flu vaccine is a great way to help protect yourself from preventable illnesses (Rimoldi et al., 2024). In general, COVID-19 vaccines are also recommended if you have DM1. However, a recent research study showed that after receiving the vaccine, people living with DM1 produced lower levels ***antibodies*** so always be sure to consult with your doctor to determine what is best for you (Saito et al., 2023).



Hygiene Habits: Maintaining good hygiene practices, like frequent handwashing and dental care, are essential to help prevent infections.

Healthy Lifestyle

Nutrition & Exercise: Balanced meals, regular gentle exercise, and adequate rest will help you support your overall immune function and help maintain your general health. For more information on how to do this effectively, check out the *MDF Cooks guide* for community recipes, and the *MDF exercise guide for people living with DM.*

MDF Cooks Guide

MDF Exercise Guide

Medical Management

Immunoglobulin Replacement: Although rare for DM1, immunoglobulin replacement therapy, which provides the body with ***antibodies*** it needs, may be considered if your antibody levels in your blood drop significantly (El-Wahsh et al., 2024). The effectiveness of ***antibody replacement therapy***, however, is still not entirely clear.

How can I best communicate my needs with my doctor?



Share Your Symptoms: Writing down when symptoms start, how long they last, and what makes them better or worse can help your doctor identify patterns. Mention any frequent or unusual infections. Let your doctor know if you're getting sick more often than usual.

Ask Questions:

- People with DM1 can have slightly lower antibody levels. Ask if you've ever been tested or if testing might be helpful.
- Ask if you're up to date on important vaccines (like flu, ***pneumonia***, or COVID-19), which can help prevent infections.
- Ask how to support your immune health. Inquire about steps you can take, like good nutrition, sleep, and exercise, that may help your ***immune system*** stay strong.

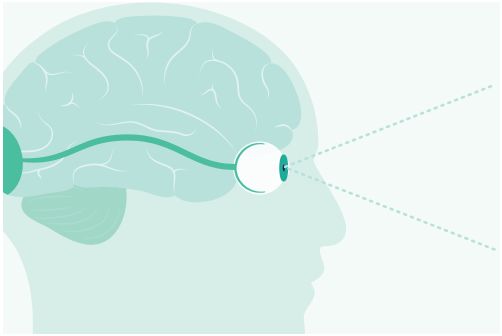
Visual System



System Overview

The visual system allows us to see and understand the world around us, by using the eyes, ***optic nerves***, and brain. In DM1, the visual system can be affected in several ways, potentially leading to a variety of eye-related issues. In this section, we'll explore how the visual system is influenced by DM1, discuss common symptoms and how they tend to develop, highlight what differences there may be in symptoms across types of DM1 (Congenital, Juvenile-Onset, Adult-Onset) and outline what you can do to manage these symptoms.

Common Symptoms: Visual System



If you are living with DM1, you may notice different kinds of vision changes, mostly due to changes in the lens of the eye and muscles that help our eyes move. While these symptoms may be challenging, they are manageable with the right care and intervention. The most common eye-related symptoms include:



Cataracts

Cataracts occur when the lens of your eye becomes cloudy, making it harder to see clearly, and is the most common eye-related complication in DM1. ***Cataracts*** in DM1 may develop earlier than usual, often before the age of 55 (Hamel, 2022). The different types of ***cataracts*** common in DM1 can occur together or independently, but both can be identified during an eye examination.

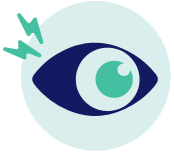
Stellate *Cataracts*: These ***cataracts*** appear as star-shaped patterns that develop in the lens of your eyes and can lead to glare or blurred vision (Moshirfar et al., 2022). They are one of the common forms of ***cataracts*** associated with DM1.

Christmas Tree *Cataracts*: Named after their appearance, these ***cataracts*** appear as multicolored, sparkling areas in the lens of your eye that can cause blurry vision (Pagoulatos et al., 2018). Like stellate ***cataracts***, they can contribute to visual issues.



Drooping Eyelids (*Ptosis*)

If the muscles in your eyelids weaken, they can droop or sag, sometimes blocking part of your vision (Hamel, 2022; Ho, 2015; Thornton, 2014). This may cause you to compensate by tilting your head to see better or using your forehead muscles to raise your eyelids.



Ocular Movement Abnormalities

Because DM1 can weaken certain muscles around the eyes, you may develop some difficulties with eye movements. Some people can develop a condition called ***strabismus*** where one eye is turned in a direction that's different from the other eye. Others may begin to have trouble focusing (Johnson et al., 2019; Zapata-Aldana et al., 2018).



Dry Eyes

Reduced blinking and incomplete eyelid closure can make your eyes feel dry and irritated. This increased dryness can make you more sensitive to light and more prone to infections.

How Vision Symptoms May Change Over Time

In DM1, vision problems getting worse over time is usually influenced by how severe the overall condition is and which type of DM1 a person has. You may not notice any changes to your vision for many years. However, over time, ***cataracts*** may worsen, leading to more significant vision impairment. Droopy eyelids and eye movement issues may also progress, which can lead to challenges with daily activities. Regular monitoring and early interventions can help manage symptoms effectively and help with potentially slowing the progression of visual issues.

Understanding the Different Forms of DM1 and How Vision is Affected

Congenital DM1 (CDM)

Eye Movement Concerns: Babies with myotonic dystrophy may show signs of ***strabismus***, where the eyes turn inward, outward, or don't move together. This misalignment can affect how a child sees, but early treatment can prevent long-term vision problems (Ho, 2015; Zapata-Aldana et al., 2018).

Juvenile-Onset DM1

Developing *Cataracts:* If you have Juvenile-Onset DM, you may develop ***cataracts*** earlier than what is typical for age-related ***cataracts***. These ***cataracts*** can continue to progress over time and may affect your vision if they are not treated (Hamel, 2022; Moshirfar et al., 2022; Pagoulatos et al., 2018).

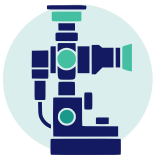
Adult-Onset DM1

Prominent *Cataracts*: *Cataracts* are very common if you have Adult-Onset DM1 and may develop in your 30s, 40s, or 50s, which is much sooner than typical age-related ***cataracts***. Your ***cataracts*** may have a unique appearance, such as stellate (star-shaped) or Christmas tree-like patterns, and they can affect your vision enough to require surgery if they are not treated (Moshirfar et al., 2022; Hamel, 2022; Pagoulatos et al., 2018; Ashizawa et al., 2018).

Although these symptoms can be challenging, various treatments and strategies outlined in the next section can help you manage them and maintain your health.

What Can I Do?

Effective management of visual symptoms in DM1 involves regular monitoring and appropriate care. By staying proactive with regular eye care and addressing visual symptoms as they arise, you can effectively protect your eye health throughout your life.



Regular Eye Exams

Annual eye exams at your ophthalmologist are important and should include a ***slit lamp examination*** (an exam using a specialized magnifying microscope). They allow your doctor to catch ***cataracts***, eyelid droop, alignment issues, and other eye-related issues early. Make sure to ask your doctor for an Optical Coherence Tomography (OCT) test, which is an eye exam that will help avoid misdiagnosis of other treatable eye conditions (Rimoldi et al., 2024).



Cataract Surgery

If ***cataracts*** interfere significantly with your vision during everyday tasks, like reading labels or driving, surgery to remove the opaque lens and replace it with a new lens can restore your visual clarity (Moshirfar et al., 2022). Before getting a procedure, it may be helpful to talk with your provider about posterior capsule opacification (PCO), the clouding of the thin membrane (posterior capsule) that holds the artificial lens in place after cataract surgery, to understand potential risks (Garrott et al., 2004). Local ***anesthesia*** is often preferred due to the potential risks associated with general ***anesthesia*** in DM1. For more information on DM and ***Anesthesia***, see **page 63** of this guide.



Management

If droopy eyelids (***ptosis***) start to block your vision, there are helpful tools that might make things easier. For example, eyelid "crutches" attach to your glasses and gently hold your eyelids up so you can see better (Burnstine et al., 2020). Oxymetazoline eye drops may also be helpful in lifting lids. Doctors usually try simple, non-surgical options like these first to avoid any unnecessary risks. Surgery to manage ***ptosis*** may be useful, but if you also have weakness when trying to close your eyelid, be sure your doctor understands that, so lid closure is not negatively affected by lid-lifting surgery. It is important to note that lifting eyelids worsens dry eyes, especially when you experience weakness when trying to close your eyelid, or you have dry eyes in general. Surgical options may be considered if the droopy eyelids block your vision to maintain visual function and activities of daily life but not for cosmetic reasons given the risk of worsening your dry eyes. Before any surgery, be sure to refer to information on DM and ***Anesthesia***, found on **page 63** of this guide.



Treatment for Dry Eyes

Think of your eyes as your car's windshield and your eyelids as your car's windshield wipers. When the wipers are turned on and there's no rain on the windshield, the wipers can skip and scratch the glass. Likewise, if your eyes are dry and your eyelids open and close, they may rub against your eye and cause injury. Eye drops, or ophthalmic lubricants, can help reduce dry eye symptoms and protect the cornea (a transparent part of the eye that covers your iris and pupil) from damage or complications associated with dry eyes (Hamel, 2022).

How can I best communicate my needs with my doctor?



Prepare Ahead of Time: Keeping a short log of when vision changes happen can help your doctor better understand what is going on.

Share Your Symptoms: Mention if your eyelids feel heavy or droopy, or if you have eye movement issues. Let them know if you've been diagnosed with ***cataracts*** and if they seem to be worsening.



Ask About Aids: Inquire about non-surgical options like eyelid crutches or specialized glasses that might help you see better.

Explain the Impact on Daily Life: Whether it's reading, driving, working, or watching TV, let your doctor know what's becoming harder to do.



Understand Potential Testing and Treatment Options:

- Obtain a regular ophthalmology check-up (every 1-2 years).
- If you are experiencing vision/eye problems, ask your doctor about ***slit lamp evaluation***, a test used to look for ***cataracts***.
- If you have ***cataracts***, your doctor should evaluate to see if you may be a good candidate for surgery.
- You can ask your doctor about eye drops for dry eyes.
- If you have droopy eyelids, your doctor may suggest management strategies like eyelid "crutches."

6 Anesthesia Risk

Individuals with myotonic dystrophy may need surgery or other medical procedures that use ***anesthesia***, which is a medicine that reduces pain and anxiety during surgery. However, ***anesthesia*** can bring special risks for people living with myotonic dystrophy because they are more sensitive to ***sedatives*** (medications that make you feel relaxed) and ***analgesics*** (painkillers).

General ***anesthesia***, which makes a person unconscious during a procedure or surgery, can be especially dangerous for people with DM because it can make muscle weakness worse and can affect a wide range of body systems.

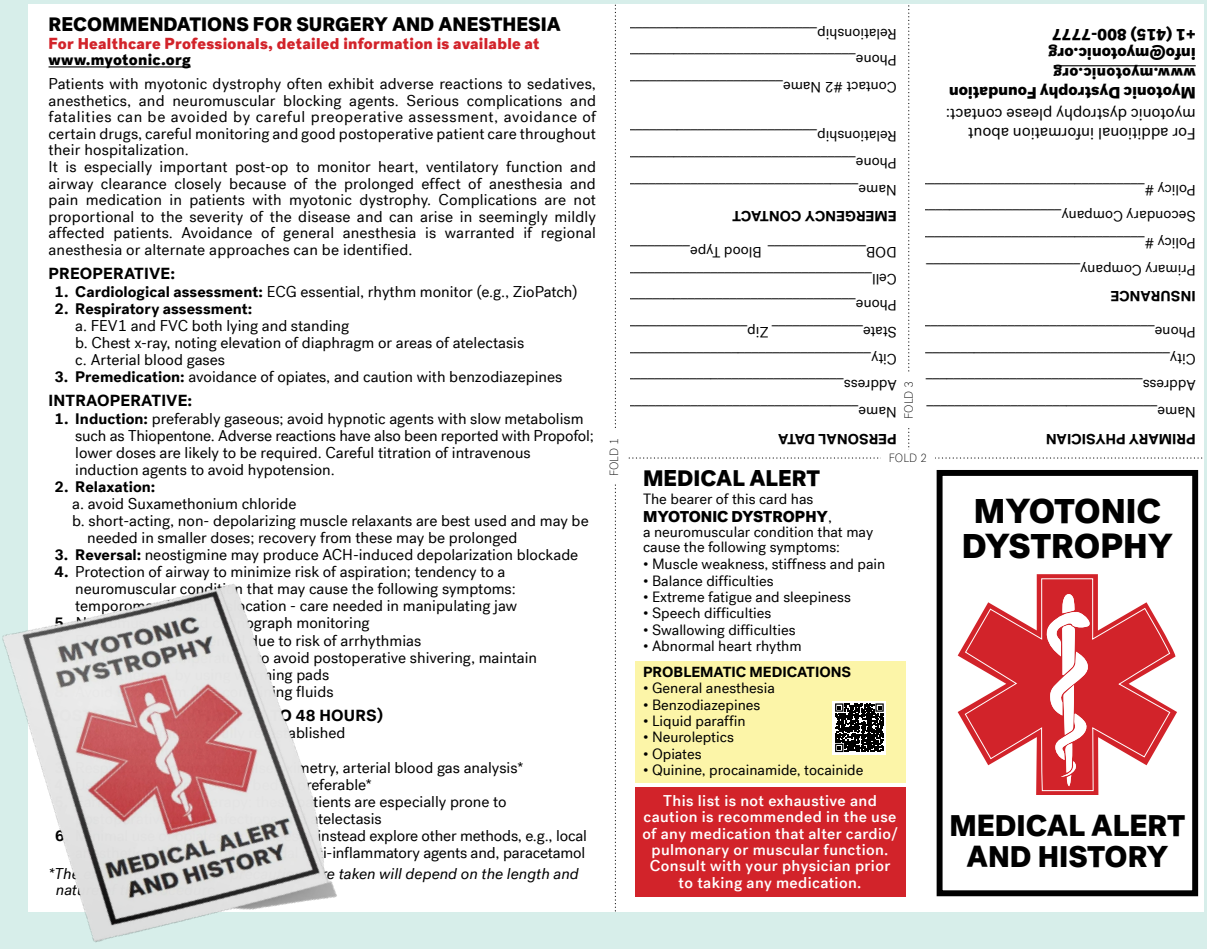
BODY SYSTEM	POTENTIAL RISK WITH ANESTHESIA
Heart	• Rhythm (heartbeat) or electrical problems called ***arrhythmias***. • For more information on DM and the heart, refer to MDF's *Myotonic Dystrophy and the Heart: A Community Guide* found here:
Lungs	• Weak respiratory muscles can lead to weak cough or breathing, lung collapse (***atelectasis***), low volume of air inhaled (***poor inspiratory capacity***), infrequent breathing (***alveolar hypoventilation***), and ***pneumonias*** (lung infections). All can cause low oxygen (***hypoxemia***) and high carbon dioxide (CO2) (***hypercapnia***) in the blood, which is very dangerous. • Respiratory risks are higher during surgeries around the upper abdomen, chest, spine, and for older individuals.
Muscles	• Inability to relax muscles (***myotonia***) triggered by cold, low potassium, cautery (tool to stop bleeding), and electrical stimulation. • Increased sensitivity to muscle relaxant medications used during ***anesthesia***. • ***Succinylcholine*** (a commonly used depolarizing muscle relaxant) should be avoided in patients with DM. • ***Non-depolarizing muscle relaxants*** (medicines that help relax muscles during surgery without causing big muscle twitches) may be necessary to allow the surgeon to have adequate conditions for your type of surgery. • The degree of muscle relaxation is constantly monitored during surgery by the anesthesiologist. They will also administer a medication to reverse the effects of the muscle relaxant at the conclusion of the operation.
Nervous System	• People living with DM are more sensitive to sedative medications, and opiates (a type of painkiller) can cause a variety of problems throughout the body.
Airway	• ***Aspiration pneumonia*** caused by weak airway muscles and an incompletely empty stomach, difficulty swallowing (***dysphagia***), and ***sleep apnea*** (breathing stops and starts repeatedly during sleep).
Digestive	• Issues with GI muscles can cause reduced contractions called gastrointestinal ***dysmotility*** that can result in total shutdown called ***ileus***. This can lead to nausea, vomiting, constipation, reaching fullness easily, and a distended stomach.
Brain	• Confusion from brain dysfunction called ***encephalopathy*** and difficulty waking up.
Endocrine	• Problems with glucose and insulin metabolism that can lead to abnormal blood sugar.

Despite these concerns, with proper care and communication, individuals with DM can undergo surgery with limited risk. Give your anesthesiologist and medical team MDF's *Anesthesia Guidelines* and/or *Anesthesia Quick Reference Guide*.

SCAN TO READ about Anesthesia

Reminder: Carry a *Myotonic Dystrophy Medical Alert Card* that indicates you have DM in case of emergency surgery.

SCAN TO ACCESS MDF's Medical Alert Card



RECOMMENDATIONS FOR SURGERY AND ANESTHESIA

For Healthcare Professionals, detailed information is available at www.myotonic.org

Patients with myotonic dystrophy often exhibit adverse reactions to sedatives, anesthetics, and neuromuscular blocking agents. Serious complications and fatalities can be avoided by careful preoperative assessment, avoidance of certain drugs, careful monitoring and good postoperative patient care throughout their hospitalization.

It is especially important post-op to monitor heart, ventilatory function and airway clearance closely because of the prolonged effect of anesthesia and pain medication in patients with myotonic dystrophy. Complications are not proportional to the severity of the disease and can arise in seemingly mildly affected patients. Avoidance of general anesthesia is warranted if regional anesthesia or alternate approaches can be identified.

PREOPERATIVE:

1. **Cardiological assessment:** ECG essential, rhythm monitor (e.g., ZioPatch)
2. **Respiratory assessment:**
   a. FEV1 and FVC both lying and standing
   b. Chest x-ray, noting elevation of diaphragm or areas of atelectasis
   c. Arterial blood gases
3. **Premedication:** avoidance of opiates, and caution with benzodiazepines

INTRAOPERATIVE:

1. **Induction:** preferably gaseous; avoid hypnotic agents with slow metabolism such as Thiopentone. Adverse reactions have also been reported with Propofol; lower doses are likely to be required. Careful titration of intravenous induction agents to avoid hypotension.
2. **Relaxation:**
   a. avoid Suxamethonium chloride
   b. short-acting, non- depolarizing muscle relaxants are best used and may be needed in smaller doses; recovery from these may be prolonged
3. **Reversal:** neostigmine may produce ACH-induced depolarization blockade
4. Protection of airway to minimize risk of aspiration; tendency to a neuromuscular condi[illegible]n that may cause the following symptoms: temporom[illegible]cation - care needed in manipulating jaw
5. N[illegible]ograph monitoring
   [illegible]due to risk of arrhythmias
   [illegible]o avoid postoperative shivering, maintain
   [illegible]ing pads
   [illegible]ing fluids

[illegible]O 48 HOURS)

[illegible]ablished

[illegible]metry, arterial blood gas analysis*
[illegible]referable*
[illegible]tients are especially prone to
[illegible]telectasis
6 [illegible]instead explore other methods, e.g., local
[illegible]i-inflammatory agents and, paracetamol

*Th[illegible] nat[illegible] *[illegible]e taken will depend on the length and*

MYOTONIC DYSTROPHY
MEDICAL ALERT AND HISTORY

FOLD 1

PERSONAL DATA
Name
Address
City
State Zip
Phone
Cell
DOB Blood Type

EMERGENCY CONTACT
Name
Phone
Relationship
Phone
Contact #2 Name
Phone
Relationship

FOLD 3

PRIMARY PHYSICIAN
Name
Address
City
Phone

INSURANCE
Primary Company
Policy #
Secondary Company
Policy #

For additional information about myotonic dystrophy please contact:
Myotonic Dystrophy Foundation
www.myotonic.org
info@myotonic.org
+1 (415) 800-7777

FOLD 2

MEDICAL ALERT
The bearer of this card has **MYOTONIC DYSTROPHY**, a neuromuscular condition that may cause the following symptoms:
- Muscle weakness, stiffness and pain
- Balance difficulties
- Extreme fatigue and sleepiness
- Speech difficulties
- Swallowing difficulties
- Abnormal heart rhythm

PROBLEMATIC MEDICATIONS
- General anesthesia
- Benzodiazepines
- Liquid paraffin
- Neuroleptics
- Opiates
- Quinine, procainamide, tocainide

This list is not exhaustive and caution is recommended in the use of any medication that alter cardio/ pulmonary or muscular function. Consult with your physician prior to taking any medication.

MYOTONIC DYSTROPHY
MEDICAL ALERT AND HISTORY

To learn more about the potential risks of ***anesthesia***, how to support safe ***anesthesia*** before, during, and after your procedure, as well as what to expect during care and how to communicate with your medical team, reference MDF's *Practical Advice for Anesthesia for Individuals with Myotonic Dystrophy and Their Families* resource.

SCAN TO READ MDF's *Practical Advice for Anesthesia*

7 Daily Life and Modifications

Living with DM1 can be challenging, but there are many ways you can improve your comfort, safety, and independence. This section gives you practical tools to help you manage daily activities at home, at work, and in your community. You will learn about improving independence, accessing ***occupational therapy***, physical therapy and exercise, nutrition and swallowing, speech, sleeping difficulties, and work considerations.

Improving Independence

Living with DM1 may make day-to-day activities more difficult, such as preparing and eating meals, personal care and hygiene, performing household tasks, and mobility. For example, individuals living with DM1 that experience weak hand and/or feet muscles may struggle with tasks involving grip or be impacted by ***foot drop***.

Occupational therapy, sometimes called ***ergotherapy***, is a health profession that promotes health and well-being in everyday life (CAOT Position Statement on Everyday Occupations and Health, 2003). ***Occupational therapy*** can help you participate in activities and everyday tasks that give meaning and purpose to your life and discover ways to live as independently as possible.

When working together, occupational therapists will typically explore three options with you:
1. Changing the task
2. Using assistive devices
3. Adapting your environment

For example, if preparing meals is difficult due to fatigue or hand ***myotonia***, an occupational therapist may suggest using pre-cut vegetables (altering the task), adaptive utensils (assistive devices), or sitting while preparing ingredients (environmental modification). You can also use this same problem-solving method on your own to address personal challenges you experience at home or in the community.



1. Changing the task



2. Using assistive devices



3. Adapting your environment

The following sections provide examples of ***occupational therapy*** suggestions for eating, kitchen activities, dressing and grooming, bathing and toileting, and assistive devices. For more information on ***occupational therapy***, you can view this session from ***MDF's 2025 Conference on Occupational Therapy and Home Health***.

SCAN TO WATCH MDF's 2025 Conference session on Occupational Therapy and Home Health

You can also view ***MDF's Ask the Expert Life Hacks Community Panel*** session to hear from community members about different tools that have helped them.

SCAN TO WATCH MDF's Ask the Expert Life Hacks Community Panel



Eating

Myotonia in your hands can make holding utensils more difficult. Increasing the size of utensil handles allows you to use larger muscle groups instead of relying on small hand muscles, which can make eating easier. One simple option is adding soft foam handles to your existing utensils. Other helpful tools may include positioning aids, splints, or universal cuffs, which help secure utensils to your hand and reduce strain.



Dressing and Grooming

Similar to eating utensils, the handles of grooming tools may be adapted so they are easier for you to hold and use. For example, using an electric toothbrush with a bigger handle may be easier for you than a traditional toothbrush. Extended handles or silicone universal cuffs on brushes, combs, or other grooming tools may also help make this easier for you.



Dressing may also become challenging for you, especially tasks requiring fine motor coordination such as zippers and buttons. Making small adaptations may help:

- Attaching a key ring to your zipper reduces the need to pinch small objects.
- Using a button hook with a large handle can make fastening your buttons easier.
- Wearing clothing with Velcro or magnetic closures can be alternatives to traditional buttons.
- Using step-in shoes or shoes with elastic laces to help with taking shoes on and off.
- Exploring strategies like avoiding tight clothing, adjusting your body position while dressing, or using adaptive equipment such as reachers or dressing sticks.



Kitchen Activities

You may find tasks like opening jars or chopping ingredients can be hard due to grip weakness and hand ***myotonia***. Tools that allow you to use your whole arm instead of just your hand may be especially helpful. Examples include:



- Jar openers that stabilize the lid and let you use arm strength rather than hand strength.
- Non-slip material placed under the jar to stabilize it or on the lid to help with grip.
- Electric can openers, which reduce the effort needed to open canned food.
- Kitchen scissors, including spring-loaded versions that automatically reopen after each cut, can be safer and easier to use than knives.
- Some examples of tools that can help you prepare meals with less fatigue and lower risk of injury include: a one-handed cutting board (a cutting board with a suction/non-slip base, food-holding prongs, and/or corner guard).



Bathing and Toileting

Making small adjustments in the bathroom may significantly improve your safety and comfort. Examples of supports that may reduce fall risk, improve independence, and make daily self-care more manageable include:



For bathing, helpful tools include:

- A shower seat or tub transfer bench
- Grab bars
- Long-handled sponges
- Wash mitts



For toileting, helpful tools include:

- An elevated toilet seat or toilet stool
- Grab bars for support
- An attachable bidet
- Hygiene assist tools (e.g., wiping aids)

These supports may reduce fall risk, improve independence, and make daily self-care more manageable.



Assistive Mobility Devices

Assistive mobility devices may help support your independence, safety, and energy reserves. They may also help prevent falls or injuries. Examples include:



- Braces
- Canes
- Walkers
- Crutches
- Manual or power wheelchairs
- Golf carts and other power-driven mobility devices

Using mobility devices is a proactive way to keep yourself moving safely and confidently.



Home Modifications

Home modifications may reduce strain and prevent injuries during your daily activities. For example:



- Non-slip materials like Dycem may make gripping doorknobs or turning light switches easier.
- Key turners may make locking and unlocking your doors less strenuous.
- Stair safety improvements such as removing loose runners, adding non-slip strips, improving lighting, using contrasting stair colors, installing a stair lift, or adding additional rails may help with navigating stairs.
- Replacing round doorknobs with lever handles may make doors easier to open.
- Rubber threshold ramps or other aids may help with smooth movement throughout your home.
- Voice activated assistance, smart home devices, and app-based environment controls can help with managing tasks around your home.



These modifications may help support your independence and reduce your risk without requiring major home renovations.



Conserving Energy

Fatigue is a common part of living with myotonic dystrophy, and learning how to pace yourself can make daily activities more manageable. Occupational therapists can help you use energy conservation strategies, such as:

- Breaking tasks into smaller steps
- Taking planned rest breaks
- Alternating harder tasks with easier ones
- Prioritizing what needs to be done each day

Simple changes can help you use your energy more efficiently and reduce exhaustion, like:

- Sitting instead of standing for tasks
- Gathering supplies before starting
- Spreading activities throughout the day

Over time, pacing yourself can help you do more of what matters to you without feeling worn out.

Accessing Occupational Therapy

Occupational therapy may be a helpful service for managing DM. An occupational therapist can help you identify your goals, values, challenges, and priorities; teach adaptive strategies; recommend interventions and provide training and support to help you live as independently and safely as possible. You can find an occupational therapist through your usual medical providers or at a private clinic or through MDF's ***Find-A-Doctor Map***. Occupational therapists do not have to be experts in myotonic dystrophy; however, they should be familiar with DM to provide you with the highest quality care.

Find-A-Doctor Map

Consensus-based Care Recommendations

During your first meeting with your occupational therapist, you can give them the ***Consensus-based Care Recommendations for Adults with Myotonic Dystrophy Type 1*** and MDF's ***Occupational Therapy Suggestions for the Management of a Myotonic Dystrophy Patient***. Additionally, it may be helpful to write down the ways DM impacts your daily living before meeting with a therapist and bring that list with you to your first meeting.

Occupational Therapy Suggestions

Physical Therapy and Exercise

Physical therapy can play an important role in helping you maintain mobility, strength, and comfort while living with myotonic dystrophy. ***Physical therapists (PTs)***, also called ***Physiotherapists*** in Canada, the UK, Australia, New Zealand, India, and other countries around the world, specialize in assessing movement and function and can design individualized exercise programs tailored to your needs. They may also recommend orthotics (such as braces or splints) or adaptive equipment to support your balance, safety, and independence. In addition, PTs often recommend assistive mobility devices to help support your independence, safety, and energy reserves and to reduce the risk of falls and injuries. These devices include canes, walkers, crutches, manual or power wheelchairs, golf carts, and other power-driven mobility devices. Using these type of mobility devices is a proactive way to keep yourself moving safely and confidently.

Role of Physical Therapy

Because symptoms vary widely among people with DM, your physical therapy plan may look different from someone else's. PTs often work closely with other members of your medical team to coordinate care. If you're looking for a PT familiar with neuromuscular conditions, you can search MDF's ***Find-A-Doctor Map***. During your first visit, it may be helpful to share MDF's ***Role of Physical Therapy in the Assessment and Management of Individuals with Myotonic Dystrophy*** article. It may also be helpful to have a caregiver attend appointments with you to learn the recommended exercises with you.

Research shows that moderate exercise can be safe in people with DM and support overall health without worsening disease progression (Voet NB, et al., 2019; Cup EH, et al., 2007; Orngreen MC, et al., 2005; Roussel MP, et al., 2019). However, it is important not to overexert yourself. Before starting any new exercise program, individuals with DM should consult with a neurologist, physical medicine and rehab physician, and/or cardiologist who has knowledge about DM or neuromuscular diseases for any recommended screening or testing. Having an evaluation by a PT who is familiar with DM or other neuromuscular conditions can be a great next step for initiating an exercise program that is tailored for you as an individual. For many people with DM, exercise can help maintain joint health, support heart and lung function, improve balance, and even reduce certain types of pain you may experience related to muscle imbalance or tightness. Together with your PT, you will develop individualized goals that match your symptoms, lifestyle, and priorities.

MDF Exercise Resources

For more information about the benefits of exercise, types of exercise, how to get started, monitoring, motivation, and technology and exercise aids check MDF's ***Exercise Guide*** written specifically for individuals and families with DM and reference MDF's ***Exercising with Myotonic Dystrophy*** infographic.



You can also join MDF's ***Myotonic Dystrophy in Motion*** movement and celebrate each July during Myotonic Dystrophy in Motion Awareness month by participating in movement activities, educational webinars, and more.

Myotoic Dystrophy in Motion

Nutrition

Nutrients provide the fuel your body needs to function and stay healthy. While research has not identified a specific diet that reduces DM symptoms, general healthy eating guidelines apply to most people living with myotonic dystrophy. Along with guidance from your clinical care team, the following are some general nutrition guidelines that may be helpful.

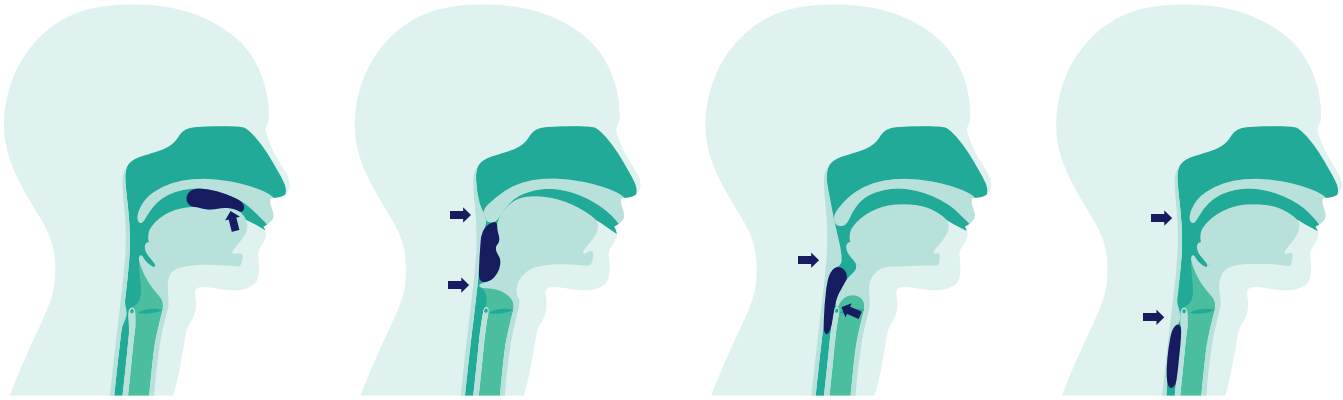
- Eat a well-balanced diet with ***protein***, whole grains (like oatmeal or whole grain bread), unsaturated fats (like avocado, salmon, nuts, or olive oil), and water. Avoid sugary drinks, too much caffeine, and saturated fats (such as fried foods, processed foods, deli meats).
- Add fruits and vegetables where you can.
- Do not skip meals.
- Take a daily multivitamin if recommended, especially if you take medications that may cause nutrient loss (for example, steroids may lower calcium and vitamin D levels). Always consult your doctor before starting new supplements.
- Spread out meals/snacks throughout the day.
- Drink at least 6-8 glasses of water a day.

MDF Nutrition Resources

For more information on nutrition and DM including examples of mini-meals and snack lists, managing constipation, being over or underweight, managing swallowing problems, modified foods, ***feeding tubes***, supplements, and where to get answers see MDF's ***Nutrition Guide for People Living with Myotonic Dystrophy***. For recipe ideas check out ***MDF Cooks***, a recipe book put together by the MDF community tailored towards people with swallowing difficulties.

Swallowing Issues

Myotonic dystrophy can result in weakness and/or ***myotonia*** in the mouth, tongue, and throat, making it harder to bite, chew, and swallow. Swallowing has four major phases:



1.
Oral preparatory phase: chewing food and gathering it, or liquid, together in your mouth to swallow.

2.
Oral phase: Starting to move food or liquid from the mouth to the throat.

3.
Pharyngeal phase: moving food or liquid through the throat (neck area) towards the ***esophagus***.

4.
Esophageal phase: moving food or liquid from throat to stomach through the ***esophagus***. Issues with this phase can sometimes cause chest pain due to ***acid reflux*** from the stomach.

Aspiration Risk

If any of the muscles used in swallowing are weak (e.g., tongue and throat muscles), you may misdirect food/liquid into your windpipe. This is called ***aspiration***. The food/liquid that is aspirated could reach your lungs. If this happens regularly or in a large quantities, this can increase your risk for ***aspiration pneumonia***. This can be a serious medical concern for those living with myotonic dystrophy.

Difficulty Swallowing (Dysphagia)

Difficulty swallowing is known as ***dysphagia***. People with DM may have difficulties in any swallowing phase. If you experience ***dysphagia***, you may notice:

- Difficulty chewing
- Trouble moving food/liquid in your mouth
- Food or liquid leaking from your nose or mouth
- Taking multiple swallows for food or liquid, or feeling there is leftover food/liquid in your mouth or throat
- Food or liquid “sticking” in mid chest area
- Pain when swallowing
- Coughing during or after meals
- Changes in breathing while eating
- Avoiding particular foods, especially if they are ones you used to be able to eat
- Unintended weight loss or dehydration
- Difficulty with cold foods or drinks
- Fatigue during meals
- Meals taking a long time to eat



Evaluating Dysphagia

If you are experiencing swallowing challenges, your provider may recommend a clinical swallowing evaluation. In most cases, this is a referral to a speech-language pathologist (SLP). SLPs assess and treat swallowing and communication disorders. If you are referred to an SLP they may:

- Ask about your symptoms.
- Perform a cranial nerve exam looking at movement and sensation of muscles of face, lips, and tongue.
- Observe you swallowing different textures.
- Recommend further tests to identify the cause of your swallowing issues and what can be done to help ease your symptoms. For instance the SLP may recommend a ***Modified Barium Swallow Study*** or ***Fiberoptic Endoscopic Evaluation of Swallowing*** to visualize the swallow action.

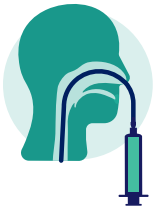


Treatments and Interventions

The types of treatments and interventions for ***dysphagia*** vary. Work with your health care provider to determine the best options for you, your specific symptoms, and the food you like to eat. Treatments might include the following:









- Diet modification (soft, moist solids; liquids, avoiding cold items, liquid supplements).
- Aid of equipment or utensils (e.g., cups or straws that can slow the flow or amount of liquid or help with good head position).
- Training on maneuvers (e.g., effortful swallow maneuver where you push your tongue against the roof of your mouth as you swallow hard) and postural/position techniques (e.g., chin tuck where you tilt your chin down while you swallow, eat/drink in an upright position). The SLP will teach you the maneuvers and postures that were helpful during the testing.
- Pacing and eating strategies, such as small bites and sips; alternating foods and liquids; smaller, more frequent meals; eating harder to eat foods when least fatigued, etc.
- Limiting foods that are difficult to swallow such as popcorn, nuts, or other hard/crunchy foods.
- Education/counseling on ***aspiration*** precautions and methods to reduce the risk of ***aspiration pneumonia*** (e.g., maintain good oral hygiene, maintain mobility).
- Counseling on alternative means of nutrition such as a ***feeding tube***, a tube that provides nutrition to the digestive system. ***Feeding tubes*** can be temporary, or long-term:
  - → Nasogastric tube: A temporary, soft, flexible tube inserted through the nose, down the ***esophagus***, and into the stomach to provide nutrition, administer medication, or drain stomach contents.
  - → Gastrostomy tube: A long-term ***feeding tube*** inserted through a surgical opening in the abdomen directly into the stomach to deliver nutrition, fluids, and medication.

While ***dysphagia*** and ***aspiration*** can add challenges to daily life, there are many proven approaches to help keep you safe and supported. By learning what works best for you and staying connected with your medical team, you may continue to enjoy food and maintain your well-being.

Speech Therapy



As someone living with DM1, you may experience ***dysarthria***, a group of speech disorders characterized by abnormalities in the strength, speed, range, steadiness, tone, or accuracy of movements required for various aspects of speech production. You may notice ***dysarthria*** symptoms such as a quieter voice, limited range of pitch, slurred or unclear speech sounds, nasally sounding speech, and speaking slower. You may also notice tongue and jaw ***myotonia*** and slower syllable repetition.

If you think you might be experiencing a speech disorder and/or facing challenges with verbal communication, you may benefit from a speech evaluation and longer-term speech therapy management. A speech evaluation is often recommended by a neurologist or primary care physician. This type of evaluation may include any or all of the following:

- An interview (questions about how you or your caregiver/loved ones feel about your speech).
- A ***cognitive*** assessment assessing attention, memory, problem solving, planning, sequencing, and other ***cognitive*** functions.
- A cranial nerve exam looking at movement and sensation of muscles of face, lips and tongue.
- Reviewing recommendations and creating goals.
- Providing educational materials.

Depending on the outcome of this evaluation, speech therapy may be recommended for you. Speech therapy has unique benefits for each person and can even help improve communication between you and your caregivers or loved ones. Speech therapy may include use of speech strategies (such as repeating a message/word/phrase, over-articulating, increasing loudness, changing rate of speech, rephrasing, or spelling key words), education and counseling for you and your caregivers/loved ones, and learning alternative forms of communication as a backup. Speech therapy may also provide support to help you and your loved ones improve the overall communication exchange.

Be sure to give your neurologist, ***primary care provider***, and/or speech therapist MDF's *Care Guidelines for Speech and Language Pathologists Treating Adults and Children with Myotonic Dystrophy*.

Sleeping Difficulties



Overview

Difficulties related to sleeping are a common issue experienced by individuals with DM. As of December 2025, 83% of individuals in MDF's Myotonic Dystrophy Family Registry living with DM1 indicate that they experience daytime sleepiness due to DM. In fact, in a study where patients and their caregivers were asked to determine the symptoms that most impacted their quality of life, nearly 92% of patients reported decreased energy and ***excessive daytime sleepiness*** (C. Heatwole et al., 2012). You may experience a variety of symptoms that indicate sleep issues related to DM, including:

- Greater than normal need for sleep because of poor quality sleep.
- Inability to wake up quickly.
- Snoring, dry eyes, dry mouth during sleep.
- Fragmented sleep patterns/disrupted sleep.
- Excess or abnormal sleep movements.
- Sleep paralysis, sleep-related hallucinations, or excessively vivid dreams.

If you are struggling with symptoms related to sleep, talk to your care team. Your provider will give recommendations on potential medications or devices. They can also help you decide if it may be beneficial to see a sleep medicine doctor who may request a polysomnogram/sleep study (see below). Your healthcare provider may use screening measures like the ones below to decide if you should get more help or be referred to a sleep lab:



- **Sleep diaries**: Recording your sleep habits to help you and your physicians get a better understanding of your sleep patterns.

- **Actigraphy**: A device that records your movement to measure your sleep habits and shows how many hours you are inactive or asleep over several days.

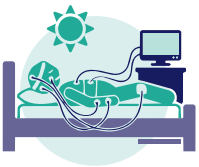


- ***Nocturnal oximetry*:** Performed at home to measure how well you breathe during sleep. It helps determine if shallow breathing, ***sleep apnea***, or other nighttime breathing problems are contributing to poor sleep and daytime sleepiness.

If you do enroll in a sleep study, a variety of tools may be used to help evaluate your sleep and your overall symptoms:



- ***Polysomnography***: A one night test that measures your breathing, oxygen levels, heart rate and movements while you sleep to determine whether you have a sleep disorder that could lead to unrefreshing sleep such as ***sleep apnea***.



- **Multiple Sleep Latency Test (MSLT)**: A test that measures the time it takes to repeatedly fall asleep over the course of 5 naps during one day. This test is best performed after a standard night's sleep monitored with a ***polysomnography***. MSLT is often essential for the diagnosis of hypersomnia/***excessive daytime sleepiness*** in myotonic dystrophy.



- **Structural assessments using *magnetic resonance imaging* (*MRI*)**: Brain scans using ***magnetic resonance imaging (MRI)*** can show changes in the brain. Another type of scan, called a ***positron emission tomography (PET)*** scan, can show decreased blood flow in certain parts of the brain.



- **Neuropsychological assessment:** An assessment typically done as a separate evaluation after a sleep study to determine a person's ***cognitive*** strengths and weaknesses and how sleep quality may be impacting them. For children with DM1, these tests should be done regularly if there are early signs of learning or developmental challenges. The tests should be adjusted to account for any physical challenges, like hearing loss or speech difficulties, and should help tell the difference between physical issues and actual ***cognitive*** challenges.

  Tests include both ***cognitive*** skills test (how well you think, remember, and solve problems) and tests for other neuropsychological functions (how well your brain handles things like memory, attention, and problem solving):

  Cognitive **skills tests:**
  - Age-appropriate Intelligence Quotient (IQ) (e.g., The Wechsler Preschool and Primary Scale of Intelligence [WPPSI] or the Wechsler Intelligence Scale for Children [WISC])
  - ***Executive function*** and higher cognition skills
  - Visual-spatial ordering skills
  - Visual perception/construction/memory skills
  - Attention skills
  - Verbal abstract reasoning skills
  - Temporal-sequential ordering skills

  Tests for other neuropsychological functions:
  - Attention-deficit/hyperactivity disorder (ADHD)
  - Energy levels
  - Social skills and general behavior
  - Emotional faculty (such as evaluation of anxiety, withdrawal, depression, conduct disorders)

Common Sleep Issues

Excessive Daytime Sleepiness (EDS) is common in people with myotonic dystrophy and can happen at any age. EDS is different from regular fatigue because it can make you fall asleep often, and sometimes without warning, during the day even if you sleep a normal amount or more at night. It is important to recognize EDS and understand what is causing it. You may want to talk with your provider about being evaluated using a simple questionnaire, such as the Stanford Sleepiness Scale, which is commonly used to assess daytime sleepiness in people living with DM1. You may also ask your provider about medicines called wakefulness promoting agents, such as Armodafinil/Nuvigil or Modafinil/Provigil, which may help treat ***excessive daytime sleepiness*** for some people.



Sleep Apnea: You may also have trouble sleeping because of ***sleep apnea***, which means you stop breathing for short periods while you sleep. When you do not get enough airflow, your oxygen levels can drop to dangerous levels and carbon dioxide can build up in your blood. Mild ***sleep apnea*** can cause disrupted sleep, excessive fatigue, and morning headaches. Severe ***sleep apnea*** can lead to high blood pressure, heart rhythm problems, and even heart attack. If you think you have ***sleep apnea***, working with a pulmonary specialist can help. They can evaluate your symptoms and recommend ways to improve your sleep and breathing. This may include using machines that help you breathe, such as a ***continuous positive airway pressure*** (***CPAP***) device or a ***bilevel positive airway pressure*** (***BiPAP***) device.

Employment with DM

Finding and maintaining a job can be difficult when living with myotonic dystrophy; however, many in the DM community successfully hold jobs they excel at and love. For a detailed walkthrough of the employment process for individuals living with myotonic dystrophy in the United States, reference MDF's ***Employment Access Toolkit***. This section contains an overview of the Employment Access Toolkit's key points:

- Assess yourself and your ability to work - consider the physical and mental challenges different jobs might bring and ask yourself if you feel prepared.
  → E.g., Poor vision might affect your ability to look at a computer screen or write in a small font, heart problems might cause you to take frequent breaks, or muscular issues might make it difficult to sit or stand for long periods of time.

- Think about the type of job you might excel at. Note your strengths and what you bring to the workforce.

- Think about what type of job you want. Consider how many hours you'd like to work, consult MDF's ***Employment Access Toolkit*** for more detail and for categorized list of jobs you might consider (pg. 9-12).

- Write a resume and cover letter and prepare for an interview. MDF's Employment Toolkit walks you through how to write a resume and cover letter and what to expect before, during, and after an interview in Parts 3 and 4.

- Consider what types of health and other insurance that potential employers offer, as well as transportation to and from your work.

- Consider work accommodation. You can ask for reasonable accommodations at any time during the application process or your period of employment. Asking for reasonable accommodations is recommended if your DM symptoms are significantly interfering with your work.
  → Your reasonable accommodation request and process must be kept confidential by your employer according to disability nondiscrimination laws.
  → MDF's Employment Guide includes a sample accommodation request letter on **page 48**.



Because myotonic dystrophy can cause ***cognitive*** symptoms like ***apathy*** and daytime sleepiness, you may find it hard to stay motivated to find or keep a job. **It is critically important to remember that these symptoms are part of the disease and are not a sign of personal or moral laziness.** To learn more about how myotonic dystrophy affects mental health, reference MDF's ***Myotonic Dystrophy and Mental Health Handbook***.

8 Building Your Care Team

You deserve the best care possible as you navigate your DM journey. Because myotonic dystrophy affects many parts of the body, it can be helpful to have a team of different specialists who work together to support your care. Building a care team allows you to play an active role in managing your health and can help prevent potential complications. Below is a list of specialists and health professionals you may want to include on your care team and the symptoms they can help you manage.

MEDICAL SPECIALIST	DM MEDICAL ISSUES	RECOMMENDED VISIT FREQUENCY
Neurologist	Supports you with muscle weakness, stiffness, chronic pain, and changes in thinking or concentration. They help track how DM affects your muscles and nervous system over time.	Every 6 months
Cardiologist	Helps monitor and treat heart rhythm changes, heart weakness, and symptoms like fainting. Routine heart checkups can help detect changes early and support ongoing heart health.	Every 6 months to annually
Primary Care Physician or General Practitioner (GP)	Helps manage your overall health, including concerns like low energy, trouble sleeping, ***excessive daytime sleepiness***, dizziness, or general wellness. Often serves as your main coordinator of care.	Every 6 months
Pulmonary Specialist/ Pulmonologist	Helps you with breathing concerns, such as ***sleep apnea***, frequent colds that don't go away, shortness of breath, or other issues related to nighttime breathing problems. They help protect your lung health and sleep quality.	Every 6 months to annually
Ophthalmologist/ Neuro-Ophthalmologist	Monitors your vision and eye health, including early signs of ***cataracts***, eyelid drooping, or eye muscle weakness. Regular check-ins help protect and preserve your vision.	Annually
Anesthesiologist	Plays an important role if you ever need surgery or a procedure that requires ***anesthesia***. They help keep you safe before, during, and after the procedure by using DM-informed ***anesthesia*** practices.	As needed
Audiologist	Evaluates and treats hearing changes. They can help if you notice trouble hearing conversations or need hearing support.	As needed
Dermatologist	Examines and treats skin changes, including noncancerous (benign) tumors of hair follicles called ***pilomatrixomas***, which are common for those living with DM.	As needed
Endocrinologist	Helps with hormone-related concerns, such as ***insulin resistance*** or ***thyroid*** changes. They support healthy metabolism and energy levels.	As needed
Gastroenterologist	Helps manage digestive symptoms like diarrhea, constipation, stomach pain, swallowing difficulties, or ***gallstones***.	As needed

MEDICAL SPECIALIST	DM MEDICAL ISSUES	RECOMMENDED VISIT FREQUENCY
Genetic Counselor	Supports family planning, pregnancy, and questions about passing DM to children. They help you understand options such as ***prenatal testing*** or in-vitro fertilization.	As needed
High-Risk Obstetrician	Specializes in pregnancies that are higher risk.	As needed
Occupational Therapist	Promotes health and well-being in everyday life through helping you participate in activities and complete everyday tasks.	As needed
Orthopedic Surgeon	Helps with bone or joint concerns such as foot deformities or curvature of the spine. They assist with mobility and comfort.	As needed
Palliative Care Team	A form of specialized medical care for people living with serious illness with the aim of providing relief from symptoms and stress of the illness.	As needed
Pediatrician	Provides general and DM-informed care for children. They help monitor symptoms such as low muscle tone, learning differences, or behavior changes.	As needed
Physical Therapist	Helps you manage movement, balance, mobility, and muscle strength. They can design exercises that support your independence and safety.	As needed
Plastic Surgeon & Oral Surgeon	Helps with facial, jaw, or mouth structure concerns that affect chewing or speech.	As needed
Podiatrist	Helps with foot health, walking difficulties, and muscle changes that affect how you move.	As needed
Psychiatrist/ Psychologist or another Mental Health Provider	Supports emotional and mental well-being, including symptoms like depression, anxiety, ***apathy***, or attention difficulties.	As needed
Rheumatologist	Helps with issues affecting joints, muscles, bones, and the ***immune system*** (e.g., arthritis, ***autoimmune diseases***).	As needed
Sleep Medicine Specialist	Specialists that diagnose and treat sleep disorders (such as snoring, insomnia, restless leg syndrome, narcolepsy, etc.).	As needed
Speech Language Pathologist	Helps with swallowing challenges and communication changes. They can teach strategies that make eating and speaking easier and safer.	As needed
Urologist and Reproductive Endocrinologist	Supports reproductive and urinary health, including ***fertility*** concerns, low ***testosterone***, pregnancy challenges, and ***miscarriage*** risk.	As needed

Find Doctors and Specialists

Finding medical professionals and specialists who understand and have experience with myotonic dystrophy is one of the most important things you can do to help manage the unique symptoms of DM. While MDF does not endorse any specific healthcare provider, we have developed a Find-A-Doctor Map, an interactive tool with community recommended specialists (recommended by people living with DM or their caregivers). To find a doctor near you, visit our ***Find-A-Doctor Map***. This interactive tool allows you to search for community recommended healthcare providers nearest you, and filter for specialties or community recommendations for subtypes of DM such as ***congenital*** or ***juvenile onset***. The map also features badges to help identify healthcare providers who are part of the Myotonic Dystrophy Clinical Research Network (DMCRN) and those who have previously been speakers at an MDF event, such as a conference or webinar.

Find-A-Doctor Map

Palliative Care



Palliative care is a form of specialized medical care for people living with serious or chronic illness. Its aim is to provide relief from symptoms and stress of the illness. A ***palliative care*** team consists of physicians, nurse practitioners, nurses, social workers and, sometimes, other specialists, like chaplains and music therapists. Doctors and nurses in this field have extra training and expertise and focus on individuals and their goals.

In the United States, ***palliative care*** is not the same as hospice care. ***Palliative care*** is available at any age and at any stage of illness. It is guided by your needs, not by your age, diagnosis, or how advanced your condition is. ***Palliative care*** does not replace any of your other treatments but instead involves working hand in hand with your existing care team. ***Palliative care*** can take place in your home, in a clinic, in a nursing facility, or in a hospital. Early incorporation of palliative medicine has been shown to improve quality of life and symptom burden (Haroen et al., 2025; Oliver, 2025).

Note: In many countries, including the UK, both hospice and end-of-life services are often included under the broader term ***palliative care***.

You may find adding ***palliative care*** to your care plan useful, especially if you are experiencing:

- Difficult symptoms (e.g., shortness of breath/needing help breathing, trouble swallowing, frequent hospitalizations, falls, etc.).
- Emotional distress (e.g., significant anxiety, depression, or feelings of being overwhelmed).
- Decline in function (e.g., trouble with routine daily tasks or activities of daily living).
- Need for planning (e.g., needing help with care coordination, understanding treatment options, making medical decisions, or advance care planning).

At a ***palliative care*** visit, your ***palliative care*** provider will take time to assess your symptoms and pain levels. They will also discuss your goals and values to ensure treatment choices are aligned with what is most important to you.

Palliative care specialists can also help you with advance care planning and communication. They seek to understand what matters most to you and help to ensure that your values and preferences are clearly expressed to ensure the care you receive aligns closely with your beliefs and wishes. For more information about planning for the future, see **page 84** of this guide.

United States Palliative Care

If you would like to begin working with a ***palliative care*** team, talk to your general practitioner or neurologist as many institutions have ***palliative care*** experts as part of their overall services. You can also visit ***getpalliativecare.org*** (United States only) or the ***International Association for Hospice & Palliative Care***, type in your location, and see ***palliative care*** programs in your area.

International Palliative Care

Mental Health Support



Living with myotonic dystrophy can affect more than just your physical health; DM can also influence how you think, feel, and interact with others. While research in this area is still ongoing, mental and emotional changes are a real part of living with DM. You may notice shifts in your mood, motivation, energy, or social comfort, and these experiences are valid and common in the DM community.

There are many ways to support your emotional well-being. Taking an active role in your mental health can make a meaningful difference in your day-to-day life, no matter the severity of your symptoms. MDF's ***Myotonic Dystrophy and Mental Health Handbook*** provides a detailed, community-focused overview of common mental health symptoms and treatment options, along with recommendations from mental health and neuropsychology experts.

Mental Health Handbook

Talking with a mental health professional can help you understand your emotions, improve coping skills, and strengthen relationships. Different types of therapy can help you in different ways, such as managing stress, understanding your thoughts and feelings, changing unhelpful thinking patterns, or handling emotions better. The purpose of therapy is to help you feel better and support you in your daily life. Therapists may include ***psychologists, psychiatrists***, licensed counselors, marriage and family therapists, clinical social workers, psychiatric nurses, or other licensed mental health providers. Before starting therapy, it can be helpful to ask about a provider's training, experience, and areas of focus. Most importantly, you should feel comfortable, respected, and supported by the therapist you choose.

Note: Caregivers can take on multiple roles (appointment schedulers, drivers, home health aides, etc.), leading to high stress levels due to the significant responsibility of maintaining their loved one's health and wellbeing. As a result, caregivers may neglect their own health and therefore need support in place to help manage stress and build resilience. Support groups, mindfulness and meditation, as well as respite care are all effective options for caregivers to receive support.

Mental health is an important part of living well with myotonic dystrophy, and you do not have to navigate it alone. With the right support, many people find renewed confidence, better coping strategies, and a greater sense of connection in their daily lives. Reaching out for help is a sign of strength, and exploring the tools available to you can open the door to a more balanced, fulfilling future. One place you can find support is through MDF support groups. Learn more on ***MDF's website***.

MDF Support Groups

9 Family Planning

Living with myotonic dystrophy can shape how you think about having children and planning for the future. This section is designed to support you as you consider family planning decisions, including genetic considerations, timing, and personal goals. It offers information and reassurance to help you make informed choices that reflect your values, priorities, and individual circumstances.

Family Planning

If you are living with myotonic dystrophy, you may have questions or concerns about starting a family. Learning about your options and talking with a ***genetic counselor*** can help you make informed decisions that feel right for you. A ***genetic counselor*** can explain how myotonic dystrophy is inherited, and review testing and family planning options in a clear and supportive way. For more information about genetic counseling, see Part 4 of this guide on **page 15**.

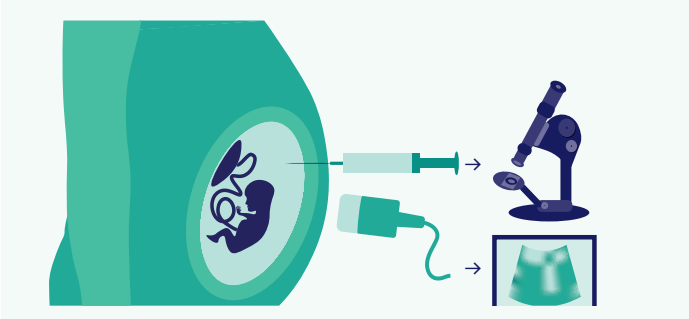
If you are considering having children, a ***genetic counselor*** may discuss different testing options with you:

- ***Preimplantation genetic diagnosis (PGD): Preimplantation genetic diagnosis*** is a way to test for myotonic dystrophy before pregnancy. This testing is done using ***in vitro fertilization*** where a doctor collects eggs from a woman, which are then fertilized with sperm from a man to create embryos in a laboratory. The embryos are tested for myotonic dystrophy at an early stage. Only embryos without myotonic dystrophy are implanted into the ***uterus***.



- **Prenatal diagnosis**: If you want to know during pregnancy whether a fetus has inherited the myotonic dystrophy ***gene***, ***prenatal testing*** may be an option. Two types of prenatal tests are commonly used.

  → ***Amniocentesis*** involves taking a small sample of fluid from the ***uterus*** that contains cells from the fetus. These cells are tested for myotonic dystrophy. This test is usually done around 15 weeks of pregnancy, and results are often available within two to three weeks.



- → ***Chorionic Villus Sampling (CVS)*** involves removing a small sample of tissue from the ***placenta***, an organ in the ***uterus*** of a pregnant woman that helps nourish the fetus throughout pregnancy. The tissue sample contains the same genetic information as the fetus and can be tested for myotonic dystrophy. CVS is usually done earlier in pregnancy, around 10 weeks, and results are often available within one to two weeks.



See Part 5 of this guide for additional information on fertility on **page 22**.

Some people choose ***genetic testing*** to learn whether they are at risk of passing myotonic dystrophy on to their children. Others may choose adoption, egg or sperm donation, or decide to have children without ***genetic testing***. There is no single right choice. What matters most is choosing the option that aligns with your values, goals, and circumstances.

If you have a family history of myotonic dystrophy or symptoms of the condition, it is important to tell your obstetrician. This allows your care team to prepare for any possible complications, including those related to congenital myotonic dystrophy. If you are pregnant or considering becoming pregnant and living with DM1, be sure to share MDF's ***Pregnancy Guide*** with your health care providers.

SCAN TO READ MDF's Pregnancy Guide

Talking with a ***genetic counselor*** can help you understand your options and feel more confident as you make family planning decisions that are right for you and your family.

10 Intimacy and Sexuality

For many people, intimacy and sexuality are an important part of life. Living with myotonic dystrophy may bring changes to how you experience intimacy, but it does not mean intimacy and sexual connection are no longer possible. This section offers an overview of factors that can influence sexuality, how DM may affect sexual health, and strategies that may help improve comfort, confidence, and enjoyment.

Sexuality is shaped by many factors, including physical health, emotional well-being, relationships, and social expectations. Myotonic dystrophy can affect some of these areas. For example, you may notice changes related to movement, strength, or energy that make activities like undressing or positioning more challenging. In addition, DM1 can directly affect sexual function; you may experience concerns such as erectile dysfunction, pelvic floor muscle weakness, lower sexual desire (***libido***), or pain during sexual activity. Other symptoms that can impact sexuality include muscle weakness, fatigue, changes in mobility, bladder or bowel issues, and heart or breathing challenges.

There are many strategies you can try to support intimacy and sexual well-being. What works best will depend on your body, your symptoms, and your comfort level:



- Staying physically active, within your abilities, can support both physical and emotional health. You can also try stretching before sexual activities to help avoid injury.
- Finding ways to work around muscle weakness, such as modifying sexual positions or practicing a position ahead of time may also help.
- Determine when your energy level is highest and plan sexual activity around it. You can also plan for a rest period before and after sexual activities.
- If you experience erectile difficulties, your healthcare provider may recommend options to help, such as adjusting your diet (Bauer et al., 2020), reducing alcohol or tobacco use, and increasing physical activity. Working with a ***physical therapist*** or ***occupational therapist*** to strengthen your pelvic floor or communicating with your partner about what feels supportive may also help.
- If you experience vaginal dryness, trying different lubricants may improve comfort. Be sure to consult a healthcare provider if you have persistent or painful dryness.
- If pain is a concern, you may find it helpful to plan intimacy when pain is better controlled or when medications are most effective. Relaxing before and after sex, changing positions, and working with a physical or occupational therapist may also reduce discomfort.

- If bladder or bowel concerns cause anxiety, emptying your bladder (or ostomy bag, if applicable) before sexual activity and using protective bedding may help you feel more at ease.

- If breathing feels difficult during intimacy, avoiding sexual activity soon after eating, limiting alcohol beforehand, choosing a comfortable environment, and using breathing techniques like slow, deep breathing may be helpful.

- Using sex aids such as a wedged pillow, penis sleeve, slings, straps, or other body assists may be useful. You can find more information at ***The Loop: Your Neuromuscular Resource Hub***.



The Loop

- Communicate your needs or desires with your partner and healthcare provider in a way that advocates for you and your sexual health. Communication is an important component of sexual activity and intimacy.

- If you have questions or concerns related to your symptoms or sexual health, talk with your doctor or another trusted member of your care team. They can help you explore options and find solutions that support your comfort, confidence, and well-being.

11 Planning for the Future: Legal Decision Making

Note: The legal information in this section is specific to the United States and is intended to provide a general overview of common legal planning options. Laws, terminology, and available supports vary by country. If you live outside the United States, similar options may exist, but they may have different names or legal requirements. For information specific to where you live, contact your local government agency responsible for ***guardianship*** or public trustees, or a disability advocacy organization. Many countries also have public guardians, public trustees, or offices of the public advocate that provide free information about legal decision-making options.

Planning for the future can feel overwhelming, but taking steps early can help you feel more prepared and supported. Thinking ahead about legal decisions can give you greater peace of mind and help protect your independence, health, and well-being over time. If you are a parent of a child living with ***Congenital Myotonic Dystrophy*** (***CDM***) or Juvenile-Onset myotonic dystrophy, you can find more information about planning for your child's future in MDF's guide *Planning for Adulthood: A Guide for Families and Caretakers of Children with CDM.*

As an adult, you have the right to make your own medical, financial, and personal decisions. However, some people with myotonic dystrophy find it helpful to plan for times when symptoms may make decision-making more difficult. Planning ahead allows you to choose who can support you and how decisions should be made if you ever need help.

In the US, there are several legal options that may help, depending on your needs and preferences. Talking with an attorney who specializes in estate planning or disability law can help you understand what is right for you.

International Resources for Legal Decision Making

Canada

https://aidecanada.ca/resources/learn/asd-id-core-knowledge/aide-canada-consent-and-capacity-toolkit

UK

https://www.gov.uk/make-decisions-for-someone/making-decisions

Australia

https://www.disabilitygateway.gov.au/sdmhub



Guardianship or ***conservatorship*** may be considered if you are unable to safely make certain decisions on your own. They are legal arrangements that allow a court to give someone the authority to make decisions for another person who cannot safely make certain decisions on their own. This may include decisions about healthcare, living arrangements, or money. A guardian is usually responsible for personal and health-related decisions, such as where someone lives or the medical care they receive. A conservator is generally responsible for financial matters, such as managing money or property. In some cases, one person may serve as both guardian and conservator. Each US state uses these terms a little differently. Some states use only the term ***guardianship*** for both roles, while others use separate terms for personal and financial decisions.

Power of attorney is another option. This is a legal document that lets you choose someone you trust to help with medical decisions, financial matters, or both if you ask for help or become incapacitated (physically or mentally unable to manage affairs). ***Power of attorney*** must be set up before you are incapacitated and allows you to keep your rights unless you give consent for someone to help or you become incapacitated and unable to make decisions on your own.

Supported decision-making is a more flexible option for people who want help thinking through choices but do not want someone else to make decisions for them. You choose trusted supporters, such as family members or friends, who help you understand options and make decisions. ***Supported decision-making*** does not require court approval, and someone can have supported decision making in place and later add ***guardianship*** or ***power of attorney*** if additional support becomes necessary.

Planning ahead gives you a voice in how decisions are made and helps ensure your wishes are respected. By planning for the future and putting supports in place, you give yourself more control, flexibility, and security. With the right information, professional guidance, and community support, you can move forward with confidence and create a future that reflects your values, goals, and needs.

12 Financial Resources & Insurance Information

If you are living with DM in a country without a national healthcare system, like the United States, you may run into problems getting insurance to cover the care and support you need. This can include ***genetic testing***, procedures, medicines, durable medical equipment (such as wheelchairs, walkers, etc.), and more. It can also be hard and frustrating to figure out what your insurance plan covers and what it does not. In some cases, you may need to get approval before you can receive certain services or treatments.

This section of the guide explains disability programs, financial support, and health insurance in the United States. It also covers different types of insurance and common insurance problems that people and families affected by DM may face, and what you can do about them.

Disability Programs in the United States

If you have DM and are unable to work due to a DM-related disability and/or other conditions, you may be entitled to Social Security Disability Insurance (SSDI) benefits or Supplemental Security Income (SSI) benefits available through the US Social Security Administration (SSA). This section provides a brief overview of the differences between SSDI and SSI.

For a deeper dive on applying for social security disability benefits, view MDF's Applying for Social Security Disability Benefits resource.

SSI vs SSDI

The Social Security Administration (SSA) oversees the largest of several federal programs that provide assistance to people with disabilities, including SSDI and SSI.

Social Security Disability Insurance (SSDI) provides cash benefits to replace some of the income you can no longer earn due to disability. You must have sufficient work history (or, for minors, have a parent with sufficient work history) and meet disability criteria to be entitled to these benefits.

Supplemental Security Income (SSI) provides cash benefits to assist people who are elderly, blind, and/or disabled who have limited means. You do not need to have a work history to qualify, but you must have little or no income and resources. For children under the age of 18, benefits are based on their parent(s') income and resources.

While SSI and SSDI provide different benefits, the SSA uses the same disability determination process for both. You can even qualify for both at the same time. SSA field staff use a publication called the Blue Book to help make decisions about disability claims. The Blue Book includes a list of medical impairments with detailed requirements for when SSA staff should judge a medical condition to be disabling. The official name of the Blue Book is ***Disability Evaluation Under Social Security***.

SCAN TO READ Disability Evaluation Under Social Security

SSA doesn't include adult-onset myotonic dystrophy on the list of chronic illnesses or impairments provided in its Blue Book. Adult-onset DM is not included in the Blue Book in part because the disease is highly variable, and not all people living with DM are unable to work, making a broad determination impossible, and partially because DM is such a difficult disease to understand. However, if any of the following, or other common DM symptoms, prevent you from working, you might qualify for Social Security disability benefits:

- Depression
- Difficulty with walking and other motor skills
- Difficulty seeing
- Difficulty concentrating or completing simple tasks
- Inflammatory bowel disease, including digestive disorders/weight loss and pseudo-obstructions
- Difficulty remembering
- Extreme fatigue, regardless of sleep
- Speech impairment
- Trouble breathing or lung infections
- Persistent or recurrent heart problems, including ***arrhythmias***
- Side effects of medication(s)

Financial Security

Planning for long-term financial security can help you feel more confident about the future. Consider meeting with a financial planner, benefits counselor, or other professional who has experience working with people with disabilities or chronic health conditions. They can help you understand available benefits, create a long-term financial plan, and make informed decisions about topics such as employment, insurance, savings, and estate planning. You may also find helpful tools and information through the ***National Disability Institute's Financial Resilience Center***, which offers resources on financial wellness, public benefits, taxes, employment, and money management for people with disabilities, or the ***Patient Advocate Foundation***, which provides case management, financial aid resources, and assistance navigating health insurance and medical debt.

Financial Resilience Center

Patient Advocate Foundation

Note, as of 2017, CDM is on the Social Security Administration (SSA)'s Compassionate Allowance List (CAL). The Compassionate Allowances initiative helps the SSA quickly identify conditions that always meet Social Security disability standards. This is not a separate program from SSDI or SSI but instead moves applicants with a condition on the Compassionate Allowance List to the front of the line for a disability decision. When you apply for benefits for your child, be sure to include the name "congenital myotonic dystrophy" where it asks for your CAL allegation/condition so your application can be fast tracked.

Common financial planning tools in the United States include ***wills***, ***trusts***, and savings accounts designed for people with disabilities.

A ***will*** lets you decide what happens to your money and belongings after you pass away and who will carry out your wishes.

A ***trust*** is a legal arrangement where a person or financial institution (called the "trustee") manages money or property for someone else (the "beneficiary"). A ***Special Needs Trust (SNT)*** is a type of trust that helps support a person with disabilities without putting their government benefits at risk. With a SNT, benefits like SSI or Medicaid are not affected because the beneficiary does not directly own the money or property in the SNT. The trustee of a SNT can use the trust to pay for extra needs of the beneficiary that benefits don't cover like respite care, transportation, home care, or dental and medical costs not paid for by Medicaid or Medicare.

An ***Achieving a Better Life Experience (ABLE) Account*** is another option for saving money for disability-related expenses without affecting certain benefits. ABLE accounts have contribution limits and specific rules, so it is important to understand how they work before opening one.

Working with an attorney or financial planner who understands disability planning can help you choose the tools that best fit your situation.

Financial Resources

Other Financial Resources

When a family or individual is faced with a progressive condition such as DM, they are often presented with additional financial expenses as well as loss of a steady income. Check out MDF's webpage on ***Myotonic Dystrophy Financial Resources*** for more support and information.



Types of Insurance in the United States

The two main categories of health insurance are private and public insurance.

Private Health Insurance

There are various types of private health insurance. Private plans are often sponsored by an employer or may be purchased directly from an insurance company or from insurance exchanges in participating states. The two main types of managed care plans within private health insurance are **HMO (Health Maintenance Organization) and PPO (Preferred Provider Organization)**.

HMO plans offer healthcare services through a network of healthcare providers. A "provider" may mean anything from doctor to ***physical therapists*** to hospitals to pharmacies, among other deliveries of healthcare services. Under an HMO plan, you must choose a ***primary care provider*** **(PCP)** within your network. The PCP is responsible for referring you to other specialists within your HMO network when needed. HMO plans only cover healthcare costs that are within the network. Since DM specialists are rare, it is unlikely that a person living with DM will have a specialist within their HMO network. Therefore, if you want to visit a DM specialist who is outside of your HMO network, you will need to get a referral to be seen out of network or pay for this visit out-of-pocket.

A PPO plan offers a more extensive network of healthcare providers to choose from. PPO plans generally offer more flexibility for seeing out-of-network specialists than HMO plans, but premiums tend to be higher. PPO plans typically require higher monthly payments in exchange for increased flexibility. In addition, with a PPO, you don't need to have a PCP. PPO plans often provide more freedom to visit DM specialists with a lower likelihood of having to pay out-of-pocket.

Additional health insurance plans include an EPO (Exclusive Provider Organization), POS (Point of Service), and HDHP (High-Deductible Health Plans), which are often used in conjunction with Health Savings Accounts (HSA), and Catastrophic Health Plans. For more information about how these plans work, visit ***HealthCare.gov***.

HealthCare.gov

Public Health Insurance

Federal and State programs that offer insurance, most commonly **Medicare** and **Medicaid**, are forms of public health insurance. These programs are for people who qualify based on their age, income, employment status, and/or whether or not they live with a disability.

Note: Some people may be eligible for both Medicare and Medicaid. This is known as "Dual Eligibility." Medicare pays covered medical services first for dual eligible beneficiaries because Medicaid is generally the payer of last resort. To learn more about the difference between Medicare and Medicaid, visit the ***Medicare Learning Network Factsheet***.

Medicare is a US federal health insurance program for people aged 65+ and those with certain disabilities. Someone living with DM doesn't automatically qualify for Medicare. They must receive Social Security Disability Insurance (SSDI) benefits for 24 months to be eligible and then, generally, they will be automatically enrolled.

For more information about Medicare and how to enroll, visit the official ***Medicare.gov*** website. You may also reach out to the **State Health Insurance Assistance Program (SHIP)** in your state.

Note: Unlike Medicaid, Medicare does not pay for long-term care.

Medicare.gov

Medicaid is a joint US federal and state program that helps cover medical costs for people with limited income and resources. In many states, children who get SSI qualify automatically for Medicaid. Medicaid waiver programs help people who need a higher level of support receive home and community-based services (HCBS), like healthcare and daily living assistance. In some states, waivers allow kids to receive Medicaid regardless of parents' income. Each state has its own rules about who qualifies. You can contact your state's Medicaid office to find out if you qualify.

Spousal Impoverishment Protections

For married couples, the cost of long-term care and support services can create significant financial concerns, especially when one spouse needs Medicaid to help pay for services. Medicaid has special rules, known as spousal impoverishment protections, that are designed to help protect the income and resources of the spouse who does not need Medicaid. These protections can help prevent the necessity to spend the couple's savings or income for the applicant's spouse to qualify for Medicaid-funded long-term services and supports.

It is important to speak with a Medicaid benefits specialist, benefits counselor, or attorney who understands Medicaid and disability planning before making major financial decisions. Do not assume that having too much income or savings automatically means a married person cannot qualify for Medicaid long-term services and support. Ask specifically whether spousal impoverishment protections apply to the situation and what resources may be protected.

If your income is too high for Medicaid, your child may still qualify for the Children's Health Insurance Program (CHIP). This program covers medical and dental care for uninsured children and teens up to age 19. CHIP qualifications are different in every state. In most cases, they depend on income. You can apply for CHIP through your state.

Other health insurance options include coverage under the Veterans Administration (VA) or TRICARE and Indian Health Services (IHS), if eligible.

For more information about insurance, visit ***MDF's website***.

Insurance Considerations

Common Insurance Issues for DM Families

If you are living with DM in the United States, you may be denied coverage for medications and services that are not listed in your health plan's ***insurance formulary*** (a list of generic and brand-name prescription medications covered by a health plan). Because DM is a rare disease, insurance companies are often not aware of the symptom management strategies that are medically necessary for people living with it. Health insurers often deny claims because they think that certain services are not medically necessary, or because they view them as experimental or investigational for people with DM.
Below is a list of medical services that are commonly denied for DM care:



Medications

- Modafinil (Provigil) and Armodafinil (Nuvigil): Though not approved specifically for DM, these medications may sometimes be prescribed to treat ***excessive daytime sleepiness*** and ***sleep apnea***.



Durable Medical Equipment

- ***CPAP/BiPAP***: helps with breathing and treating ***sleep apnea***.
- Electric lifts (toilet seat lift, lift chair, lift for bathtub, etc.): Helps manage ***myotonia***, ***atrophy***, and/or ***myopathy***.
- Braces (leg, knee, foot, etc.): Helps manage ***myotonia***, ***atrophy***, and/or ***myopathy***.



Genetic Testing

- People showing DM symptoms often have difficulty getting coverage for ***genetic testing*** to confirm diagnosis.
- Family members of people diagnosed with DM often struggle with getting coverage for ***genetic testing*** to determine whether they have DM as well. This can occur because their symptoms may not be as prominent as their family member who was diagnosed.



Specialists

- Depending on the type of insurance an individual has, it can be difficult to get coverage for specialists, especially if that specialist is outside of your insurance network (out-of-network).



In Vitro Fertilization

- Many people living with DM experience ***fertility*** difficulties but may have difficulty getting ***in vitro fertilization*** covered by their insurance company.

What You Can Do

To get necessary services covered, you can initiate a claims process to appeal the decision. For more information about this process see MDF's resource ***Health Insurance Considerations for People Living with Myotonic Dystrophy in the United States***. The ***Patient Advocate Foundation*** also has resources to help with navigating insurance issues, including case managers to help with the appeals process in the United States.

Patient Advocate Foundation

Health Insurance Considerations

13 MDF Support

Support systems are essential for navigating the changes and new developments that accompany living with myotonic dystrophy. A support system can consist of family, friends, religious and community groups, psychotherapists, and healthcare professionals, as well as other members of the DM community who live with the disease and have life experiences similar to your own. This section of the guide describes the support you can find through the Myotonic Dystrophy Foundation.

MDF is the world's largest DM patient advocacy organization, connecting people living with DM in more than 139 countries around the world. If you're living with DM and/or caring for someone who is, you may sometimes feel overwhelmed, isolated, and in need of support. MDF aims to create safe spaces to network, learn, and share through a variety of support systems.

Connect with the DM Community

Support Groups

MDF has over 25 in-person and virtual support groups around the globe with participants from over 10 countries. MDF support groups create safe spaces to network, learn, and share. You can find support groups based on your region (e.g., Mountain West Regional Support Group, California Regional Support Group), language (English, German, Spanish, or French), or on specific topics (e.g., DM2, Caregiver Support Group).

Support Groups

Support Group Facilitators

Visit ***MDF's website*** to find the best support group(s) for you.

Support groups are led by more than 45 trained volunteer community members. Volunteers respect the privacy and confidentiality of anything shared in these groups and can refer you to MDF's trusted credible information and resources.

You can learn more about MDF's Support Group Facilitators (SGFs) on ***MDF's website***.

Warmline

MDF offers professionally staffed support for individuals requesting resources, support, and referrals. Feel free to call MDF at +1 415-800-7777 anytime Monday through Friday 9am to 5pm PST or leave a message after hours. You can also email ***info@myotonic.org*** and an MDF staff member will get back to you within 1-2 business days.



MDF Conferences

MDF hosts conferences every year for people living with DM, family members, caregivers, clinicians, researchers, industry partners, and advocates. Our conference program includes two types of events that take place in alternating years:

- **MDF International Conference:** Held every other year, this multi-day event brings the whole DM community together in one place. It includes sessions for people and families living with DM, along with a professional track for clinicians and researchers. The event is designed to create opportunities for learning, connection, and collaboration across the full community.

- **MDF Regional Conferences**: In the alternating years, MDF hosts several one-day regional conferences in different locations. These events bring education, resources, and community connection closer to home for people and families affected by DM.

Both conference formats offer updates on research and care, practical information for daily life with DM, and opportunities to connect with others who understand the DM journey. Through presentations, panel discussions, and community conversations, MDF conferences help strengthen support, share knowledge, and move progress forward for everyone impacted by DM. You can learn more about upcoming conferences on ***our website***.

Upcoming MDF Conferences

Continue Learning and Stay Informed

Other MDF Publications

As you've seen throughout this guide, MDF has a wide range of other toolkits, handbooks, and publications written for individuals affected by DM (including information on DM and the heart, ***anesthesia***, mental health, exercise, and more) as well as health care providers (including various clinical care guidelines). You can access them all for free on ***MDF's website*** or email ***info@myotonic.org*** or call the Warmline at +1 415-800-7777, to get a hard copy mailed to you.

MDF Resources

Ask-the-DM-Expert

Webinars

Ask-the-DM-Expert

MDF's Ask-the-DM-Expert webinar series is a virtual, interactive program that connects individuals and families affected by DM with experienced DM clinicians, therapists and experts across a range of specialties. This series gives the community a direct opportunity to ask questions, gain trusted insights, and receive practical guidance from experts who understand the complexities of DM. Visit ***MDF's website*** to learn more and register for an upcoming webinar.

Meet-the-DM-Drug-Developers

MDF's Meet-the-DM-Drug-Developers webinar series connects the myotonic dystrophy community with leading biotechnology and pharmaceutical partners developing treatments and potential cures for DM. These live sessions offer participants the chance to hear updates directly from drug developers and ask questions in real time. Registration is required to join and submit questions in advance. Visit ***MDF's website*** to learn more and register for an upcoming webinar.

Meet-the-DM-Drug-Developers

You can access past recorded webinars and conference sessions on ***MDF's Digital Academy***.

E-Newsletter "My Dispatch"

The MDF monthly e-newsletter, or My Dispatch, provides you with up-to-date information on research activities, DM daily living strategies, upcoming events, stories from MDF community members, and much more. It is the best way to stay current on news in the DM community. ***Sign up for the monthly newsletter!***

Sign up for MDF's newsletter

Social Media

Follow MDF on social media to receive information about MDF and DM as well connect with other DM community members for support and friendship as you navigate DM.

- Instagram: ***@myotonicstrong***
- X: ***@myotonicstrong***
- Facebook: ***Myotonic Dystrophy Foundation***
- LinkedIn: ***Myotonic Dystrophy Foundation***



Find-A-Doctor Map

Find Care and Resources

Find-A-Doctor Map

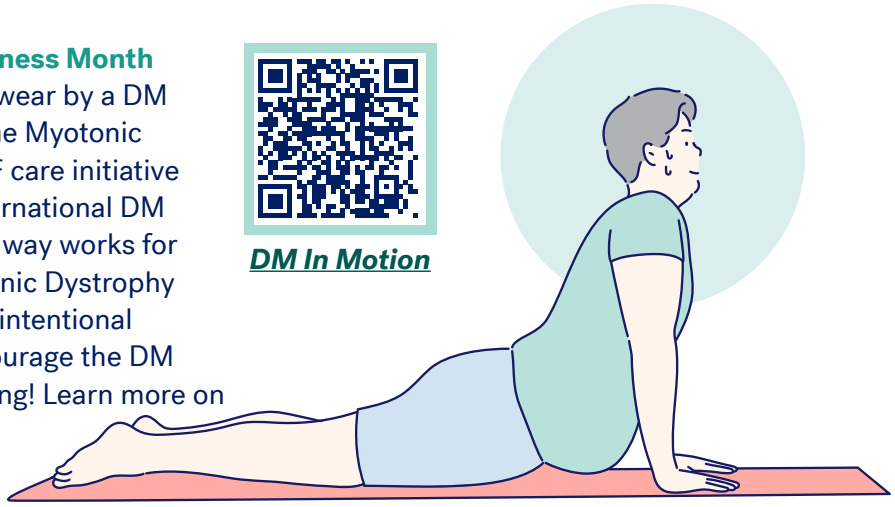
While MDF does not endorse any specific healthcare provider, we have developed a ***Find-A-Doctor Map***, an interactive tool with community recommended (recommended by people living with DM or their caregivers) specialists. To find a doctor near you visit our Find-A-Doctor Map. This interactive tool allows you to search for community-recommended healthcare providers nearest you, and filter for specialties or community recommendations for subtypes of DM such as congenital or juvenile onset. The map also features badges to help identify healthcare providers who are part of the Myotonic Dystrophy Clinical Research Network (DMCRN) and those who have previously been speakers at an MDF event.

Get Involved

Myotonic Dystrophy in Motion Awareness Month

What started as an idea for MDF activewear by a DM community member has evolved into the Myotonic Dystrophy In Motion Movement: A MDF care initiative to unify, inspire, and encourage the international DM community to start moving in whatever way works for them. Each July, MDF celebrates Myotonic Dystrophy in Motion awareness month by hosting intentional movement-based programming to encourage the DM community to get active and keep moving! Learn more on ***MDF's website***.

DM In Motion



MDF Family Registry

The MDF Family Registry is a patient self-reported online database gathering information from individuals affected by myotonic dystrophy type 1 (DM1; including CDM, JOA, Childhood, and Adult onset) and type 2 (DM2). The Myotonic Dystrophy Family Registry is unique in that it not only helps researchers find new, effective treatments and identify possible participants for upcoming clinical trials and research studies, but also allows anyone who is registered to have access to the anonymous data, including individuals and families living with DM. The Registry gives all DM stakeholders, from researchers and pharmaceutical partners to family members, a better understanding of the disease, the DM community, and current research and advocacy efforts. In 2026, MDF published an ***academic paper on results from the DM Family Registry***.

Family Registry Academic Paper

Community Connections

Many people living with DM experience social isolation and find it hard to stay connected. Through Community Connections, you can brighten someone's day by sending a heartfelt letter, drawing, or story—and remind them that they are not alone. You can also nominate a community member to receive a special MDF Care Package filled with kind messages from across the DM community. Learn more about Community Connections on ***MDF's website***.

Community Connections

Fundraise

Fundraise with MDF

MDF is the leading global advocate for those impacted by DM. With your help, we can fuel our mission of Community, Care, and a Cure. You can start your own fundraiser to empower our work to support the DM community, provide essential resources, and accelerate research toward treatments and a cure. Learn more about how you can fundraise on ***MDF's website***.

Become an Advocacy Champion

Nobody knows this disease better than those living with it. As someone living with DM or a caregiver, you have unique insights and experiences that are invaluable. With the right help and resources, you can become an effective champion and advocate for yourself and the greater DM community. Advocacy can take many forms, from sharing your story with others, to participating in research, to influencing policy throughout the country.

Advocacy webpage

Contact your representative

MDF has many ways for the DM community to become advocates. You can start by exploring our ***advocacy webpage*** which provides an overview of MDF's advocacy efforts and how you can get involved such as ***contacting your representatives*** through MDF's Voter Voice campaign or using our ***toolkit to request a proclamation*** in the US or Canada for International Myotonic Dystrophy Awareness Day.

Proclamation Request Toolkit

Awareness Day

Celebrate International Myotonic Dystrophy Awareness Day

September 15th is International Myotonic Dystrophy Awareness Day. Established in 2021, Awareness Day is an amazing opportunity to garner the attention of the wider general public, policy makers, regulators, biopharmaceutical representatives, researchers, health care professionals, and anyone with an interest in changing the future of myotonic dystrophy. Learn more about how to participate in Awareness Day on ***MDF's website***.

Looking to the Future

Dozens of DM researchers, scientists, rare disease advocates, MDF staff, and, most importantly, DM community members are working tirelessly towards a better future for myotonic dystrophy.

DM Drug Development

MDF partners with a variety of biopharmaceutical companies to keep the DM community up to date on new developments and to foster relationships between drug developers and real people living with myotonic dystrophy.

Check out ***MDF's Drug Development Pipeline*** for information on current and ongoing DM programs, including the company developing the therapy, the drug name and approach (e.g., gene therapy, small molecule), the development phase, and the target disease subtype.

Drug Development Pipeline

Clinical Trials

There are many opportunities to get involved in ongoing clinical trials. MDF has a list of current studies and trials that individuals with DM can participate in, as well as find answers to some frequently asked questions, and a training video with more information on clinical trials on ***our website***.

Clinical Trials

DM Research

MDF is committed to helping advance DM research to improve quality of life and advance treatments for myotonic dystrophy. We offer multiple funding opportunities and resources to strengthen scientific progress in the field. This includes funding research fellows, early career scholars, pilot grants, and other funding mechanisms. You can learn more about MDF grants and funding opportunities via ***our website***.

The future of myotonic dystrophy is being shaped by a global community of researchers, clinicians, advocates, industry partners, and people living with DM who are working together toward better treatments and, ultimately, a cure. While progress takes time, advances in research, growing awareness, and increasing opportunities to participate in studies and advocacy offer real reasons for hope. At the time of this guide's publication (2026), we expect drug approvals and better diagnostics in the near future. Every person who shares their experience, participates in research, joins a clinical trial, or raises awareness helps move the field forward and brings us one step closer to a brighter future for everyone affected by DM.

MDF Grants and Research Funding Opportunities

Living with myotonic dystrophy comes with real challenges, but it also comes with options, support, and a strong community. You do not have to navigate this journey alone. The Myotonic Dystrophy Foundation is here to support you with trusted information, programs, and connections to others who understand your experience. With the right tools, thoughtful planning, and ongoing support, you can move forward with confidence, make informed decisions, and continue building a life that reflects your values, goals, and hopes for the future.



Glossary

ACE inhibitors (ACEI): Short for angiotensin-converting enzyme inhibitors; medications that relax blood vessels and lower blood pressure by blocking the formation of a hormone that narrows them.

Achieving a Better Life Experience (ABLE) Account: A savings and investment account for disability-related expenses that allows people with disabilities to save money without losing eligibility to most federally funded need-based benefits.

Acid reflux: The backward flow of stomach acid into the esophagus, which can cause a burning sensation.

Adrenal glands: Small glands located above the kidneys that produce hormones involved in stress response, metabolism, and other functions.

Aerobic exercises: Physical activities, such as walking or swimming, that increase heart rate and breathing over a sustained period to improve cardiovascular fitness.

Alanine aminotransferase (ALT or GPT): An enzyme concentrated primarily in the liver and commonly used as a marker of liver injury.

Albumin: A protein made by the liver that helps maintain fluid balance in the blood.

Alkaline phosphatase (ALP): An enzyme originating from the liver, bones, and other tissues, with abnormal levels pointing to possible liver or bone disorders.

Amniocentesis: A prenatal medical procedure where a small sample of fluid is withdrawn from the amniotic sac to test for genetic disorders, chromosomal abnormalities, and birth defects.

Amniotic fluid: The fluid that surrounds and protects a fetus in the uterus.

Anemia: A condition in which the blood has a lower-than-normal number of healthy red blood cells, leading to fatigue and weakness.

Anesthesia: The use of medication to block pain or induce loss of sensation, awareness, or consciousness during medical or surgical procedures.

Antacids : Medications that neutralize stomach acid to relieve heartburn or indigestion.

Antiarrhythmic medications : Drugs used to prevent or treat irregular heart rhythms.

Antibiotics: Medications used to treat infections caused by bacteria.

Antibodies : Proteins produced by the immune system to identify and neutralize harmful substances such as bacteria and viruses.

Antibody replacement therapy: A treatment that provides antibodies to individuals whose immune systems do not produce enough on their own.

Anticipation: A phenomenon where the signs and symptoms of a genetic disorder become more severe and appear at an earlier age in each successive generation.

Anticonvulsants: A class of medication used to calming overactive electrical signals in the nerves.

Antidepressants: Medications used to treat depression and certain other mental health conditions.

Anxiolytics: Medications used to reduce symptoms of anxiety.

Apathy: A lack of interest, motivation, or emotional response.

Arrhythmias: Irregular heartbeats caused by disruptions in the heart's normal electrical signaling.

Aspartate aminotransferase: An enzyme present in the liver and muscle tissue whose blood levels rise when either organ is damaged.

Aspiration: The accidental inhalation of food, liquid, or saliva into the lungs instead of the stomach.

Aspiration pneumonia: A lung infection caused by inhaling something other than air into the lungs.

Atrophy: The wasting away or shrinking of muscle tissue, often resulting in decreased muscle size and strength.

Autism spectrum disorders: A group of developmental conditions characterized by differences in communication, social interaction, and behavior.

Autogenic training: A relaxation technique that uses self-guided statements to reduce stress and promote physical relaxation.

Autoimmune diseases: Conditions in which the immune system mistakenly attacks the body's own healthy tissue.

Autosomal dominant: A genetic inheritance pattern where a single copy of an altered gene from one parent causes a trait or disorder.

Bacteria: Microscopic living organism that has only one cell.

Beta blockers: Medications that slow the heart rate and reduce blood pressure by blocking the effects of certain stress hormones on the heart.

Bilevel positive airway pressure (BiPAP) machine: A non-invasive breathing device that uses two air pressure levels to help you get enough oxygen and clear carbon dioxide.

Bowel Obstruction: A blockage that prevents food or waste from passing normally through the intestines.

Bradycardia: An abnormally slow heart rate.

Brain waves: Patterns of electrical activity in the brain that can be measured to assess brain function, including during sleep.

Breath stacking: A technique in which a person takes multiple breaths without fully exhaling in order to increase lung volume before coughing.

Bronchodilators: Medications that relax and widen the airways to make breathing easier.

Cardiomyopathy: A disease of the heart muscle that affects its ability to pump blood effectively.

Cataracts: A clouding of the lens inside the eye that makes vision blurry, hazy, or dim. It can develop gradually and, if it interferes with daily activities, is often treatable with surgery.

Central nervous system: The brain and spinal cord, which control most bodily functions and process information from the rest of the body.

Chorionic Villus Sampling (CVS): A prenatal medical procedure where a small sample of tissue from the placenta is removed to test for genetic disorders and chromosome abnormalities.

Chromosomes: Long strands of DNA wrapped around proteins; a way for cells to organize and store DNA.

Clitoris: A small, highly sensitive erectile organ in female anatomy whose primary function is providing sexual pleasure.

CNBP: A gene that has expanded repeats in individuals with DM2. Also called ZNF9 or CNBP1.

Cognitive: Relating to the mental processes of thinking, learning, remembering, and understanding.

Cognitive Behavioral Therapy (CBT): A type of talk therapy that helps individuals identify and change unhelpful thought patterns and behaviors.

Cognitive impairment: A decline in mental functions such as memory, attention, or reasoning.

Conduction defects: Disruptions in the heart's electrical signaling pathway that can cause irregular or abnormal heart rhythms.

Congenital myotonic dystrophy (CDM): A sub-type of DM1 where symptoms are noticeable from birth.

Conservatorship: A legal arrangement that allow a court to give someone authority to make decisions for another person who cannot safely make certain decisions on their own.

Cough assist device: A machine that helps clear mucus from the lungs by simulating a natural cough using changes in air pressure.

CPAP machine: A device that delivers a continuous stream of air pressure through a mask to keep the airway open during sleep.

Decongestants: Medications that reduce swelling in nasal passages to relieve congestion.

Delayed muscle relaxation: A slower-than-normal return of a muscle to its resting state after it has been tightened or contracted.

DEXA scan: An imaging test that measures bone density to assess bone strength and risk of fracture.

Diabetes: A chronic disease marked by high blood sugar levels, stemming from the body's inability to produce or effectively use insulin.

Diaphragm: A dome-shaped muscle located below the lungs that contracts and relaxes to enable breathing.

Diuretics: Medications that help the body eliminate excess fluid and salt through increased urine production, often used to reduce blood pressure or swelling.

DNA: Short for deoxyribonucleic acid; the genetic material present in nearly all cells that contains the instructions for how the body grows, develops, and functions.

Dysarthria: A group of speech disorders characterized by abnormalities in the strength, speed, range, steadiness, tone, or accuracy of movements required for various aspects of speech production.

Dysmotility: Abnormal movement or contraction of muscles within the digestive tract, affecting the passage of food.

Dysphagia: Difficulty swallowing.

Dystrophia myotonica: The universally used Latin name for myotonic dystrophy.

Dystrophia myotonica protein kinase (DMPK): A gene that has expanded repeats in individuals with DM1.

Ectopic pregnancy: A pregnancy that occurs outside the uterus, most often in a fallopian tube, which can be life-threatening.

Egg cell: The female reproductive cell, also called ovum.

Electrocardiogram (ECG): A test that records the electrical activity of the heart to help detect abnormal rhythms or damage.

Electromyography (EMG): A diagnostic procedure that records the electrical activity of muscles to assess nerve and muscle function.

Endocrine system: The network of glands that produce and release hormones to regulate body functions.

Enzymes: Proteins that speed up chemical reactions in the body, such as those involved in digestion and metabolism.

Ergotherapy: A health profession that promotes health and well-being in everyday life.

Esophagus: The muscular tube that carries food from the throat to the stomach.

Estradiol: A form of estrogen that plays a key role in female reproductive health.

Estrogen: A hormone primarily produced in the ovaries that plays a key role in female reproductive development and function.

Excessive daytime sleepiness (EDS): A persistent feeling of tiredness or an increased need for sleep during the day, despite adequate nighttime rest.

Executive function: A set of mental skills that includes planning, organizing, and managing tasks.

Fallopian tubes: Thin, muscular channels in the female reproductive system that connect the ovaries to the uterus.

Feeding tube: A medical device used to deliver nutrition directly into the stomach or intestines for individuals who cannot eat safely by mouth.

Fertility: A person's ability to conceive a child.

Fertility specialist: A healthcare provider who focuses on diagnosing and treating difficulties with conceiving a child.

Foot drop: Difficulty lifting the front part of the foot.

Forced vital capacity (FVC): A measurement of the total amount of air a person can forcefully exhale after taking the deepest breath possible.

Free T4: A blood test that measures the level of active thyroid hormone circulating in the body.

FSH: Short for follicle-stimulating hormone; a hormone that plays a key role in regulating reproductive function, including egg and sperm production.

Gabapentin: A medication commonly used to treat nerve-related pain and certain seizure disorders.

Gallbladder: A small organ beneath the liver that stores and releases bile to aid in digestion.

Gallstones: Hardened deposits that form in the gallbladder, which can cause pain or blockage if they obstruct the flow of bile.

Gamma globulin: A group of proteins in the blood that includes antibodies, important for immune function.

Gastrointestinal (GI) system: The group of organs, including the esophagus, stomach, and intestines, responsible for digesting food and absorbing nutrients.

Gene: A segment of DNA that contains instructions for building a specific protein or RNA.

Genetic counselor: A healthcare professional who helps individuals understand their risk of inherited conditions and the implications of genetic testing.

Genetic mutation: A miscoded segment of DNA.

Genetic predisposition: An increased likelihood of developing a particular condition due to specific inherited genetic factors, though not a guarantee the condition will occur.

Genetic testing: A laboratory analysis of DNA used to identify changes or mutations linked to inherited conditions.

GERD: Short for gastroesophageal reflux disease; a chronic condition in which stomach acid frequently flows back into the esophagus, causing irritation.

Grip myotonia: Difficulty releasing one's grip quickly after tightly grasping an object, due to delayed muscle relaxation.

Guardianship: A legal arrangement that allow a court to give someone authority to make decisions for another person who cannot safely make certain decisions on their own.

Hairpin loops: Abnormally long portions of RNA clumped together inside of the cell nucleus.

HbA1c: A blood test that measures average blood sugar levels over the past two to three months.

Heart palpitations: A sensation of a rapid, fluttering, or pounding heartbeat.

Heartburn: A burning sensation in the chest caused by stomach acid backing up into the esophagus.

High risk neonatal: Refers to a newborn baby who needs extra medical monitoring or specialized care due to health complications present at birth, such as breathing difficulties, low birth weight, or feeding problems.

High risk obstetrician: A doctor who specializes in managing pregnancies with an increased risk of complications.

Holistic: An approach to care that considers the whole person, including physical, emotional, and social well-being, rather than focusing on a single symptom or condition.

Hormone therapy: Treatment that involves supplementing or adjusting hormone levels to manage a medical condition.

Hormones: Chemical messengers produced by glands that regulate various body functions.

Hypercalcemia: A condition involving abnormally high levels of calcium in the blood.

Hypogammaglobulinemia: A condition in which the body has lower-than-normal levels of antibodies, increasing the risk of infection.

Hypogonadotrophic hypogonadism: A condition in which the reproductive organs produce insufficient sex hormones because of reduced hormonal signaling from the brain.

Hypothyroidism: A condition in which the thyroid gland does not produce enough hormone, leading to symptoms such as fatigue and weight gain.

Hypotonia: Reduced muscle tone, resulting in decreased resistance to movement and a floppy or limp appearance of the limbs.

Hypoventilation: A condition in which breathing is too shallow or slow to adequately exchange oxygen and carbon dioxide.

IgA: A type of antibody found in mucous membranes, such as those in the gut and respiratory tract, that helps protect against infection.

IgG: A type of antibody that provides long-term immune protection and is the most common antibody in the blood.

IgM: A type of antibody that is the first produced by the immune system in response to a new infection.

Immune system: The body's network of cells and organs that work together to defend against infection and disease.

Immunoglobulins: Another term for antibodies, proteins produced by the immune system to fight infection.

Implantable cardioverter defibrillator (ICD): A device placed inside the body that detects dangerous heart rhythms and delivers an electrical shock to restore a normal heartbeat.

Implantable devices: Medical devices placed inside the body, such as pacemakers or defibrillators, used to monitor or regulate bodily functions like heart rhythm.

In vitro fertilization: A fertility treatment in which an egg is fertilized by sperm outside the body before being implanted into the uterus.

Insulin resistance: A condition in which the body's cells fail to respond normally to insulin, causing blood sugar levels to rise.

Insurance formulary: A list of generic and brand-name prescription medications covered by a health plan.

Intestines: The long tube-like organs in the digestive system where nutrients are absorbed and waste is processed.

Involuntary muscles: Muscles that function automatically without conscious control, such as those in the heart and digestive system.

Irregular menstrual cycles: Menstrual periods that occur at unpredictable intervals or vary significantly in length.

Irritable bowel syndrome (IBS): A chronic disorder affecting the large intestine that causes symptoms such as abdominal pain, bloating, and changes in bowel habits.

Joint contracture: A permanent tightening or shortening of muscles, tendons, or other tissue around a joint, leading to reduced flexibility and limited range of motion.

Juvenile-onset adult myotonic dystrophy (JOA): A sub-type of DM1 where symptoms appear during childhood or adolescence, before the age of 21.

Lactate dehydrogenase (LD): An enzyme distributed widely throughout body tissues, released into the blood when cells are injured or destroyed.

LH: Short for luteinizing hormone; a hormone that plays a key role in regulating reproductive function.

Libido: A person's overall sexual drive or desire.

Lymphocytes: A type of white blood cell that plays a key role in the immune system's response to infection.

Magnetic resonance imaging (MRI): A scan that creates pictures of the inside of your body.
Manual cough assist: A technique in which another person applies pressure to the chest or abdomen to help clear mucus from the lungs.

Maternal-Fetal Medicine doctor: A physician who specializes in caring for pregnant women with high-risk conditions and their fetuses.

Menopause: The natural end of a woman's menstrual cycles, typically occurring around midlife.

Mexiletine: A medication used to reduce muscle stiffness by affecting electrical signaling in muscle cells.

Miscarriage: The loss of a pregnancy before the fetus can survive outside the womb.

Motility: The movement of food and waste through the digestive tract via muscle contractions.

Mucolytics: Medications that thin and loosen mucus, making it easier to cough up.

Mutation: A change in the DNA sequence of an organism.

Myopathy: A disease of the muscle tissue that causes weakness, but is not caused by a problem with the nerves that control the muscles.

Myotonia: A condition characterized by delayed relaxation of muscles after they contract, resulting in prolonged stiffness or tightness.

Myotonic dystrophy (DM): An inherited neuromuscular disorder that affects many parts of the body.

Myotonic dystrophy type 1 (DM1): An inherited neuromuscular disorder that primarily shows up in adulthood and can affect multiple body systems. The most common symptoms are muscle weakness, pain, and fatigue.

Myotonic dystrophy type 2 (DM2): An inherited neuromuscular disorder that primarily affects muscle strength, function, and control, but can affect multiple systems of the body. It is caused by a genetic mutation that leads to progressive muscle weakness and myotonia.

Neurological: Relating to, affecting, or involving the nervous system.

Neuropsychological evaluation: A test that measures how a person's thinking, emotions, and behavior are working.

Nocturnal oximetry: A test that measures blood oxygen levels during sleep.

Noninvasive ventilation (NIV): A method of providing breathing support using a mask rather than a tube inserted into the airway.

NSAIDs: Short for nonsteroidal anti-inflammatory drugs; a class of medications used to reduce pain, inflammation, and fever.

Nucleotides: A basic building block of nucleic acids (RNA and DNA). They are represented by the letters C, A, T, and G in DNA and C, A, U, and G, in RNA.

Occupational therapy: A health profession that promotes health and well-being in everyday life.

Olfactory receptors: Proteins that help a person smell.

Oocytes: Immature egg cells found in the ovaries.

Optic nerves: The nerves that transmit visual information from the eyes to the brain.

Ovarian reserve: The quantity and quality of a woman's remaining eggs.

Ovaries: A pair of female reproductive glands that produce eggs and sex hormones.

Pacemaker: A small implanted device that sends electrical signals to help regulate an abnormal heart rhythm.

Palliative care: Specialized medical care focused on providing relief from symptoms and improving quality of life for individuals with serious illness.

Parathyroid glands: Small glands near the thyroid that regulate calcium levels in the body.

Peak cough flow test: A test that measures the speed of air expelled during a cough to assess the effectiveness of clearing the airway.

Percussion myotonia: Prolonged muscle contraction triggered by a brief tap or strike to the muscle, often tested during a physical exam.

Perinatologist: Another term for a Maternal-Fetal Medicine doctor, specializing in high-risk pregnancy care.

Physical therapist (PT): A healthcare professional who evaluates and treats patients to improve movement, manage pain, and recover from injuries or surgeries.

Physiotherapist: A healthcare professional who evaluates and treats patients to improve movement, manage pain, and recover from injuries or surgeries.

Pilomatrixoma: A noncancerous skin tumor that develops from hair follicle cells.

Placenta: An organ that develops in the uterus during pregnancy to provide oxygen and nutrients to the fetus.

Pneumonia: Inflammation and fluid in the lungs caused by infection.

Polyhydramnios: A condition involving an excessive amount of amniotic fluid during pregnancy.

Polymerase chain reaction (PCR): A laboratory technique that generates multiple copies of a targeted DNA segment for further analysis, frequently used in genetic testing.

Polysomnography: An overnight test that records brain waves, breathing, heart rate, and other body functions to diagnose sleep disorders.

Positron emission tomography (PET): A test using a safe, injectible radioactive chemical tracer that helps detect diseased cells.

Postpartum bleeding: Heavy bleeding that occurs after childbirth.

Power of attorney: A legal document that allows someone else to act on a person's behalf.

Preimplantation genetic diagnosis (PGD): A procedure used during in vitro fertilization to test embryos for genetic conditions before they are implanted in the uterus.

Prenatal testing: Medical testing conducted during pregnancy to evaluate the health and development of the fetus, including screening for genetic conditions.

Preterm labor: Labor that begins before 37 weeks of pregnancy. Babies born too early may have health problems because they have not fully developed.

Primary Care Provider: A general health professional who serves as the main ongoing point of contact for routine medical care, illness treatment, and disease prevention.

Prokinetics : Medications that help speed up movement through the digestive tract.

Prostate: A small gland in the male reproductive system that adds fluids to the semen and helps semen exit your body.

Protein: A molecule made by the body that performs essential functions, including building and repairing tissue, enabling chemical reactions, and supporting the structure and function of muscles and organs.

Proton pump inhibitors (PPIs): Medications that reduce the amount of acid produced by the stomach.

Psychiatrist: A medical doctor who specializes in diagnosing and treating mental health conditions, including through medication.

Psychologist: A mental health professional trained to assess and treat emotional, behavioral, and cognitive issues through therapy, but who typically cannot prescribe medication.

Ptosis: The drooping of the upper eyelid, which can partially or fully cover the eye.

Pulmonary function tests (PFT): A group of tests that measure how well the lungs take in and release air.

Ranolazine: A medication primarily used to treat chest pain related to heart conditions, and sometimes studied for its effect on muscle stiffness.

Recessive: A genetic inheritance pattern where a trait or condition is only expressed when an individual inherits two identical copies of that gene - one from each parent.

Reduced ovarian reserve: A decrease in the number or quality of a woman's remaining eggs.

Reproductive endocrinologist: A doctor who specializes in hormone-related fertility issues.

Reproductive system: The organs involved in producing offspring, including those responsible for hormone production and pregnancy.

Resistance exercises: Physical activities that involve working muscles against an opposing force, such as weights or resistance bands, to build strength.

Respiratory arrest: The complete stopping of breathing.

Respiratory muscle strength tests: Tests that measure the strength of the muscles used for breathing, including the diaphragm.

Respiratory system: The group of organs, including the lungs, airways, and diaphragm, that work together to bring oxygen into the body and remove carbon dioxide.

RNA: Short for ribonucleic acid; a molecule that copies genetic instructions from DNA and uses them to direct the production of proteins in the cell.

RNA splicing: A process in cells in which portions of a gene are joined in different combinations, leading to different copies of RNA that are used to produce different proteins.

Scoliosis: An abnormal sideways curvature of the spine.

Sleep apnea: A disorder in which breathing repeatedly stops and starts during sleep.

Slit lamp examination: An eye exam that uses a specialized microscope with a bright light to closely examine the structures of the eye.

Somatic mosaicism: A condition where some cells in the body carry new, unique changes to genetic codes.

Southern blot: A laboratory method used to identify specific DNA sequences within a sample, often applied to detect genetic mutations.

Special Needs Trust: A type of trust that helps support a person with disabilities without putting their government benefits at risk.

Spinal cord: A bundle of nerves that runs through the spine, carrying signals between the brain and the rest of the body.

Stillbirth: The loss of a baby after 20 weeks of pregnancy before or during birth. Causes can vary and are sometimes unknown.

Stimulants: Medications that increase alertness and activity in the brain, often used to treat attention-related disorders.

Strabismus: A condition in which the eyes are not properly aligned and point in different directions.

Supported decision-making: A process that allows people with disabilities to make choices about their own lives with support from a team of people they choose.

Swallow study: A diagnostic test that evaluates how safely and effectively a person swallows food and liquid.

Syncope: A temporary loss of consciousness caused by a sudden drop in blood flow to the brain, commonly known as fainting.

Testes: The pair of male reproductive glands responsible for producing sperm and hormones.

Testosterone: A hormone primarily produced in the testes that plays a key role in male reproductive development and function.

Thyroid: A butterfly-shaped gland in the neck that produces hormones controlling metabolism, energy, heart rate, and body temperature.

Thyroid-stimulating hormone (TSH): A hormone that regulates thyroid gland activity, measured to assess thyroid function.

Tracheostomy tube: A tube inserted through an opening in the neck directly into the windpipe to help a person breathe.

Tricyclic antidepressants : A class of medications originally developed to treat depression that are also used to manage certain types of pain.

Trust: A legal arrangement in which the owner of property gives it to another to manage and use for the benefit of a designated person.

Type 2 diabetes: A chronic condition in which the body becomes resistant to insulin or does not produce enough of it, leading to high blood sugar levels.

Uterus: The hollow, muscular female reproductive organ where a fertilized egg implants and grows into a fetus.

Vagina: The muscular, stretchy canal inside the female body that connects the outer genitals to the uterus.

Virus: Microscopic germ that infects a host (like a human, animal, or plant).

Visuospatial: Relating to the ability to perceive and understand the visual and spatial relationships between objects.

Voluntary muscles: Muscles that a person can control consciously, such as those used for walking or gripping an object.

Vulva: The collective name for all the external, visible parts of the female genital organs.

Will: A legal document that states a person's wishes for distributing their property and managing their estate after death.

γ-***glutamyltranspeptidase (GGT):*** Short for gamma-glutamyl transferase; an enzyme produced mainly in the liver, elevated levels of which can signal liver disease or bile duct problems.

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We provide resources and advocate for care.
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