Hamilton Health Sciences
PATIENT EDUCATION

# Myotonic Dystrophy Type 1

## What is myotonic dystrophy type 1?

Myotonic dystrophy is a medical term for a condition that affects muscle function. The term myotonic comes from several words in Greek:

- 'myo' means muscles (muscles are affected)
- 'tonia' meaning tension (the tension of the muscle is affected)

Myotonic dystrophy type 1 (DM1) is one of several types of muscular dystrophy.

## What are the signs and symptoms of DM1?

### Muscle weakness

- DM1 commonly affects the muscles of the lower legs, hands, neck and face.
- People with DM1 often have prolonged myotonia (sustained muscle contractions), and are not able to relax certain muscles after use. For example, an individual may have difficulty releasing their grip on a door knob, or temporary locking of their jaw with biting or chewing.
- The muscles may become smaller (muscle atrophy) over time.

**Other common signs or symptoms include:**

- unable to completely close eyes during sleep
- droopiness of the eyelids (ptosis)
- male pattern baldness
- feet slapping/heavy footsteps due to weakness (foot drop)
- gastrointestinal problems (constipation and/or diarrhoea)
- trouble swallowing/choking
- early-onset cataracts (before 55 years of age)
- diabetes
- excessive daytime sleepiness
- heart irregularities (known as “heart block”)

DM1 is usually a chronic or life-long disease that affects people in different ways. The spectrum of people with DM1 range from mild, to classic, to onset at birth (congenital onset).

## Types of DM1

1. **Mild** – early onset cataracts, increased risk of heart block, may not experience muscle weakness, some may have grip myotonia.

2. **Classic** – childhood or adolescent onset muscle weakness, grip myotonia, early onset cataracts, male-pattern balding.

3. **Congenital** – floppiness/severe generalized weakness at birth, difficulty breathing at birth, intellectual disability is common.

## What causes DM1?

DM1 is a genetic condition which is typically inherited from one parent. The abnormal gene can be inherited from a mother or a father. Having the abnormal gene means that you have the condition. This is called “autosomal dominant inheritance”. This condition can change and become more severe as it is passed down from generation to generation. This is known as “anticipation”. Sometimes a parent’s condition is so mild that they do not realize they have this genetic condition until it is diagnosed in another family member.

## How do you know I have DM1?

Your doctor or specialist can tell if you have DM1 from:

- talking with you about your symptoms and family history
- checking the strength of your muscles
- reviewing the results of a genetic test for DM1

### Genetic tests

- All individuals with DM1 have an expansion (extra information) of genetic material near a gene known as *DMPK*. The extra material consists of a single genetic “word” of three letters, CTG, which is repeated over and over. Having more than 50 CTG repeats fits with a diagnosis of DM1. An individual’s CTG repeat number can go up to over 1000 repeats. This can be measured using a special genetic test in your blood.

### Electromyography (EMG)

- This test measures the electrical activity in your muscles and nerves. An acupuncture needle is inserted into a muscle and listens for a distinct sound known as “myotonic discharges” which occur in the muscles of an individual with myotonic dystrophy.

## How is DM1 treated?

DM1 is usually managed with a combination of methods.
Your treatment/management plan will be designed to meet your needs.

**The goal of treatment is to prevent or lessen your symptoms and help you retain the strength in your muscles.**

### Heart

- Monitoring your heart on a yearly or bi-yearly basis is important to check for a heart irregularity known as “heart block”.
- An ECG is done to check for changes in the rhythm of the heart and other potential cardiac symptoms.

### Mobility

- It is important to have neurological and strength assessments regularly to monitor the progression of your DM1.
- Assistive devices may be considered, depending on your abilities. This can include ankle-foot orthoses, Rollator walker, wheelchairs, or other assistive devices.

### Exercise

- Exercise is always helpful to help build and maintain your muscle strength! We can provide strength monitoring and exercise advice with a DVD and exercise hand-outs.
- We recommend slowly increasing exercise tolerance starting off slowly and gradually to eventually attain 3 to 4 sessions each week of mixed endurance (such as biking, rowing, walk, jog, elliptical) and strength (such as weights, elastic bands, pilates) exercise. However, the most important thing is to do a type of exercise you enjoy.

### Eyes

- Checking eyes for cataracts on a yearly or bi-yearly basis.
- Removal of cataracts if vision is impaired.

### Nutrition

- Your diet is also important to your treatment. Individuals with DM1 are at an increased risk to develop type 2 diabetes.
- Refer to **Eating Well with Canada's Food Guide** to make sure you have enough protein and calories in your diet. Ask your family doctor for a copy or visit Health Canada's website.
- We recommend that you take a multivitamin and Vitamin D (2000 IU a day for adults and 400 IU a day for children).
- A supplement of Creatine Monohydrate can also help improve strength in conjunction with exercise. The amount you should take is based on your weight. You can add it to a glass of juice or sprinkle it on cereal or yogurt.

### Gastrointestinal

- Some people my experience digestive issues, such as diarrhea or constipation. People experiencing these symptoms can undergo a test known as a "Breath Hydrogen Test" to see if these symptoms are caused by bacterial overgrowth in the intestines. This can be treated with antibiotics.

### Hormones

- It is common for males with DM1 to have low testosterone. Your testosterone levels may be checked and treatment recommended.

## How will having DM1 affect my life?

Each person responds differently to DM1. Outcome depends on the severity of your condition.

You can continue to have follow-up appointments at the Neuromuscular and Neurometabolic Clinic where the health care team will plan your treatment/management and provide your care.

Living with a chronic condition can be challenging and emotional. Here are some ideas for living well with a chronic disease.

### Educate yourself

- Learn as much as you can about the disease and how to manage symptoms. If you have questions, ask your health care providers. This will help you make informed decisions about your care.

### Take part in your care

- Work closely with your health care providers to follow your management plan. Keep track of your symptoms and your response to treatment/management. Tell your health care providers how you are doing.

### Take care of yourself

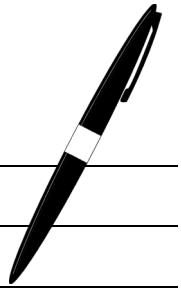
- Learn to listen to your body. Rest and conserve your energy when you feel tired. When you are active, go at your own pace.

### Talk about your feelings

- It is normal to have mixed emotions when you are living with a chronic illness. At times, you may feel sad, angry or helpless.
- It can help to talk about your feelings with your partner, someone close to you or another person who has faced a similar situation. Remember that your health care providers can also help.

**If you have any questions about DM1, please speak with your doctor, genetic counsellor, or nurse at the Neuromuscular and Neurometabolic Centre.**

## Notes and questions



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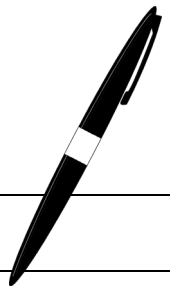

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