# MULTICORE MYOPATHY

MUSCULAR DYSTROPHY CANADA
DYSTROPHIE MUSCULAIRE CANADA

## What is Multicore Myopathy?

Multicore Myopathy (MM) - also commonly known as Multi Minicore Myopathy - is a rare, inherited condition which causes degeneration of the muscle fibers. People with the disease experience generalized weakness along with muscle wasting. It affects both males and females. Close to half of Multicore Myopathy cases are caused by a defective gene in one of two genes: Selenoprotein N1 (SEPN1) and Ryanodine receptor 1 (RYR1). There are four types of Multicore Myopathy: the classic form; progressive form with hand involvement; antenatal form with arthrogryposis multiplex congenital; and the ophthalmoplegic form. Symptoms and progression differ with each type.

### What are Symptoms of Multicore Myopathy?

The main symptom of all Multicore Myopathy is muscle weakness and wasting. Other specific symptoms depend on the type of disorder. In some people, the disease may remain stable for a long period of time, while others may experience an increase in muscle weakness over time.

### What is the age of onset?

Multicore Myopathy is typically diagnosed at birth or within the first few months of life. On rare occasions, diagnosis is made in adulthood. One possibility for this late diagnosis could be a missed diagnosis in childhood (or misdiagnosis). Another possibility is that the condition moved from a mild, non-progressive phase to a progressive form where the symptoms became more evident.

### What causes Multicore Myopathy?

Most cases of Multicore Myopathy are inherited in an autosomal recessive pattern, meaning both parents must have a copy of the defective gene for offspring to develop symptoms. If only one parent has the defective gene, then their children are likely to become carriers and are at risk of passing the gene on to their children. Carriers generally do not have symptoms. Some cases of Multicore Myopathy are 'sporadic', meaning they occur with no known cause.

### What are the types of Multicore Myopathies?

*Classic Form.* Close to 75% of all Multicore Myopathy cases are the classic form. Symptoms typically present shortly after birth and include feeding problems, curvature of the spine (scoliosis), weak or floppy muscles (hypotonia), slow motor development, and

weak respiratory muscles causing breathing problems.

*Progressive form with hand involvement.* This form occurs in roughly 10% of all Multicore Myopathy cases and is less severe than the classic form. Scoliosis is less severe, and breathing problems are either minor or absent. Double-jointedness (hyperlaxity) is often seen in this form.

*Antenatal form with arthrogryposis multiplex congenita (AMC).* This occurs in fewer than 10% of Multicore Myopathy cases. The primary symptom at birth is contractures -- a chronic shortening of muscles or tendons around joints. People with the AMC version of the disease may have altered physical features such as an elongated head, low-set ears, and a short neck. Difficulties breathing range from moderate to severe.

*Ophthalmoplegic form*. This is the least common form of Multicore Myopathy. The main symptom is droopy eyelids (ptosis) due to weakness in the muscles around the eye. There is also weakness in the muscles that are closest to the trunk of the body.

## How is Multicore Myopathy Diagnosed?

Diagnosis of Multicore Myopathy is confirmed by use of a muscle biopsy. Here, a small piece of muscle is removed and studied under a microscope. Samples have a characteristic pattern that is not seen in healthy muscle tissue. Normally, muscle tissue is made-up of two different types of fiber: type 1 and type 2. However, in Multicore Myopathy the muscle consists of too many type 1 fibers and not enough type 2. Within the fibers, structures called 'cores' can be seen. The name of the disease refers to these cores.

## What treatments are available for Multicore Myopathy?

There is no cure for Multicore Myopathy. However, there are various treatments that help maintain general function and reduce pain. Key treatments include physiotherapy, exercise, ventilation, corrective surgery, and feeding tubes. The level of treatment needed depends on the severity of symptoms and the needs of an individual patient.

- **Physiotherapy**. Physiotherapists teach specific stretches or movements to help increase and maintain muscle function. This contributes to overall mobility.
- **Mild exercise** is believed to be helpful for some patients. However, some experts caution against strenuous or weight-bearing exercises, which may damage the muscles. Non-weight bearing exercise such as swimming, walking or using a stationary bicycle may be helpful because they provide the aerobic exercise needed for good heart health. Any exercise undertaken should be done under the supervision of a doctor.
- **Ventilation**. Commonly, people with Multicore Myopathy have breathing problems due to weak respiratory muscles. Respiratory function should be assessed frequently. For some people, use of a C-PAP (continuous positive air pressure) machine at night is an option. People using a C-PAP machine wear a small face mask while they sleep. The device which uses room air only, helps improve breathing, allowing for carbon dioxide to be more efficiently exhaled. Whether this is appropriate for a particular patient can be determined by a respirologist.
- **Surgery.** Curvature of the spine (scoliosis) is common among people with Multicore Myopathy. In severe cases, surgery to correct a curved spine may help improve quality of life. The surgery entails the insertion of rods, screws or wires to support the spine and related structures in the body.
- **Feeding Tube** (gastrosomy) or G-Tube: Some people with myopathies have difficulty swallowing. The placement of a feeding tube bypasses the need for swallowing and can ensure good nutrition is attained. Depending on the degree of swallowing difficulty, some people, when prescribed by a

swallowing specialist, are able to tolerate both a G-Tube and some swallowing by mouth.

## Special considerations

Caution should be taken in the use of anaesthetics and muscle relaxants. There is a relationship between Multicore Myopathy and malignant hyperthermia. Malignant hyperthermia is a potentially fatal condition. If surgery is being considered, it is important to inform doctors and anaethetists of this risk. A medical alert bracelet outlining the risk of adverse reactions to anesthesia is recommended.