Scottish Muscle Network

Metabolic Myopathy patient Information Leaflet

You have been given this leaflet because your doctor thinks you have, or may have, a metabolic myopathy.

What is a metabolic myopathy?

A metabolic myopathy is a rare genetic condition. It affects the process by which muscles generate energy. There are many kinds of metabolic myopathy, with different genetic causes.

What are the symptoms of a metabolic myopathy?

These depend on the type of metabolic myopathy. Symptoms also vary from person to person, even with the same condition.

Most people will have pain, cramp, or fatigue**,** in their muscles, during or after exercise. This is described as exercise intolerance. Some people avoid exercise as a result.

Some people will have episodes of muscle weakness. Others will develop muscle weakness over time. However, many people will never have muscle weakness.

Occasionally, people develop a complication called rhabdomyolysis.

What is rhabdomyolysis?

This is acute muscle breakdown. When this happens, the person has very severe muscle pain. Some people describe this as like an intense “flu like” feeling in the muscles. It is very uncomfortable to move. Sometimes, the muscle is also weak. The urine may go dark black in colour or contain blood. Muscle proteins are released into the bloodstream from the damaged muscle. One of these, creatine kinase, will be very high when measured on a blood test. In severe cases, kidney function is affected. Rarely, this requires kidney dialysis.

What triggers rhabdomyolysis?

Triggers include very intense, prolonged, or unaccustomed exercise, fever, and other illnesses like viral infections. Some medicines can cause rhabdomyolysis including statins and herbal remedies. Alcohol in large amounts and recreational drugs like ecstasy can also trigger rhabdomyolysis.

What do I do if I get rhabdomyolysis?

You may have rhabdomyolysis if you experience very severe muscle pain and/or cramp and/or feel very unwell after exercise. If your urine is dark black in colour, or bloodstained, or if you are producing very small amounts of urine or no urine at all, go to a hospital accident and emergency department. Take this factsheet with you.

Is there a treatment for metabolic myopathy?

There is not usually a specific medicine to treat or cure metabolic myopathy. However, you can do a lot to manage your symptoms and prevent rhabdomyolysis from occurring or recurring. Your nutritional intake is crucial in providing you with the right amount of energy and will also help to manage your muscle symptoms. Sometimes referral to the nutrition team can be helpful to guide and advise you about the right proportion of carbohydrates and fats that you should be including in your diet.

How can I manage my symptoms?

Watch your diet. Eat regular meals. Eat a healthy, balanced diet. Avoid fasting. Drink plenty of water. Maintain a healthy weight. If you are unwell, for example with a fever, make sure you keep yourself well hydrated.

Plan your exercise. It is very important that you continue to exercise regularly if you can. Take a small carbohydrate snack half an hour before exercise. Avoid exercising when unwell, running a fever, fasting, or dehydrated. Avoid exercise if you have been drinking alcohol. Build up new exercise programmes gradually. Consider trying yoga, Tai Chi, pilates or similar.

Avoid*:* Overexertion, fasting, excessive or unaccustomed alcohol, recreational drug abuse, statin medicines. If you need medicine to lower your cholesterol, there are alternative preparations to statins that your doctor can prescribe for you.

What is the cause of metabolic myopathy?

There are several known causes of metabolic myopathy. Metabolic myopathy sometimes happens because of change(s) to the code of a gene that is involved in energy generation in the muscles.

How can I find out whether there is a genetic cause for my metabolic myopathy?

If your doctor thinks that your metabolic myopathy is due to a genetic problem, then it would be possible to do genetic testing on a blood test to look for genetic causes of metabolic myopathy. Other tests might also help, like muscle biopsy, muscle MRI and other blood tests. These tests often take a few years to complete.

Although finding the exact cause can take quite a lot of time, it can provide useful information both to you and your doctor.

Even after a lot of tests, it is only possible to find a genetic cause in around a quarter of people in whom a metabolic myopathy is strongly suspected. It is important to remember that the management of your muscle symptoms will not be significantly altered by knowing the precise genetic cause of your metabolic myopathy. Regular exercise and having a healthy diet and avoiding any triggers remain the mainstay of our advice to you.

Will other people in my family develop metabolic myopathy?

This will depend on the genetic cause of the condition. Your Neurologist can give you personalised information about this or refer you to the Clinical Genetics department.