

Metabolic disorders that cause pain and/or weakness on exercise

Almost everybody experiences some muscle pain when they exercise more than usual or drive themselves to the limit in training, or suffer an actual 'sports injury'. However some people consistently develop muscle pain with 'ordinary' exercise and they may be found to have a 'metabolic disorder' of the muscles. If you have been found to have such a condition this information is designed to give you an understanding of it and why you experience pain on exercise. **However**, you must consult your doctor about your particular needs because the disorders described below vary considerably from person to person.

This Factsheet covers the following conditions:

- A** Disorders affecting the breakdown of glycogen or glucose into pyruvate, often termed **glycogen storage disorders**.
- B** Defects of fatty acid oxidation, some of which are known as **lipid storage myopathies**.
- C** Defects of the mitochondrial respiratory chain; these may be known as **mitochondrial myopathies**.

Why do I experience pain on exercise?

People who have a metabolic disorder which causes pain and or weakness when they exercise, have a fault somewhere in the mechanism which breaks up the 'fuels' in our food and converts them into energy. They experience pain when they exercise because the muscles are placing extra demands on the mechanism and, because there is a fault, it cannot cope; it breaks down.

What food do our muscles need to allow us to exercise?

In our diet we all consume carbohydrates (sugars and starches) and fat. Some of the carbohydrates and fat are used immediately for energy; most is stored for later use. Then, when we need energy, these stored 'fuels' are broken down in the body to provide it. This energy is used in all organs, for instance in the heart to allow the heart to pump blood, and in the brain to allow us to think and control our bodies. Muscles also use a great deal of this energy to allow us to exercise.

When we are sitting down, our energy use is quite low, but when we exercise the muscles demand a great quantity of energy very quickly. Therefore the carbohydrates and fat have to be broken down much more quickly. Which fuel is used, carbohydrate or fat is dependent upon the type and duration of the exercise undertaken. During short duration exercise of high intensity, for example sprinting, the main source of energy comes from carbohydrates. If exercise is more prolonged, for instance a three-mile jog or a full game of football, there is an initial use of carbohydrate with a gradual switch to fat as a major provider of energy. This happens because most of the carbohydrate reserves become depleted and the muscles have to start using stored fat to provide the energy.

An understanding of which fuel is required for different types of exercise is very important if we are to recognise the cause of the muscle pain or weakness on exercise, and for the treatment of metabolic disorders.

How are carbohydrates and fat converted into the energy we need to exercise?

Carbohydrates and fat are stored in different ways, and are broken down to provide energy by different processes.

Carbohydrates are broken down into glucose, which is stored, as a substance called 'glycogen'. Glycogen is stored in the muscles, liver, heart and brain. When we exercise, particularly when we exercise intensely in short bursts, the muscles call on the glycogen which is converted back into glucose and then into a substance called pyruvate. The pyruvate is then taken into 'mitochondria' (small particles in cells whose major function is to produce energy units known as 'ATP'). If the exercise is strenuous it can overload the system, which cannot function fast enough to convert all the pyruvate into energy via the mitochondria, and some of the pyruvate is turned into lactic acid instead.

Fat tends to be more of a storage fuel. It is stored in various places such as under the skin. When we use fat for energy, usually during prolonged exercise, the fat is brought out of the tissue where it is stored, goes into the blood and then into muscle, and finally continues into the 'mitochondria'. There the fats are broken down into carbon dioxide and water and energy (ATP), a process called 'fatty acid oxidation'.

A Glycogen storage disorders

People with glycogen storage disorders are unable to process carbohydrates into energy. They have a malfunction somewhere in the chain of chemical reactions by which glycogen should be broken down into pyruvate. This does not happen, and glycogen is therefore stored in excess quantities in muscle and other tissues. These 'unusable stores' may cause problems themselves, but in addition there is an acute lack of pyruvate when it is needed to provide energy. See diagram below.

What type of exercise will bring on symptoms?

Strenuous exercise such as sprinting or running for a bus tends to bring on symptoms. Swimming, for instance, may be painless if you are doing it gently, but painful if you are exerting yourself.

What are the symptoms and what should I do when I experience them?

Symptoms are mainly muscle pain and stiffness. They come on during exercise and stop you from continuing. The pain occurs in the working muscles, and therefore is often felt in the legs. Stop exercising when you feel much pain otherwise you will be damaging your muscles. You should consult your doctor about exercise in the future; some exercise is important to prevent your muscles becoming weak. Each patient is likely to have a type and variety of exercise that they can perform safely. Sometimes you may feel some pain during exercise, which then eases off; this is known as the 'second wind phenomenon'. It happens because you started off using

carbohydrates for energy and then switched to fat. You can safely carry on with your exercise in these circumstances.

If the pain is particularly bad, an energy crisis may occur. This leads to muscle break-down; a process called 'rhabdomyolysis'. It can cause the excretion of a muscle protein called myoglobin in the urine - 'myoglobinuria' - and may result in kidney failure. You will be warned of this if your urine is noticeably darker than usual (a brownish colour). If this is the case you should see your doctor **at once** because treatment may be required.

Most people are perfectly well between attacks but a few develop mild, permanent muscle weakness. This tends to be in the larger muscles; in the shoulder, hips, legs, and arms.

Does diet help?

There is now evidence that a drink of sucrose (75g) 30-40 minutes before exercise can improve exercise tolerance. There is also evidence that moderate aerobic exercise is an effective means of improving exercise capacity in patients with McArdle's disease by increasing circulatory delivery and mitochondrial metabolism of bloodborne fuels.

Two of the names of the glycogen storage disorders that cause pain on exercise are McArdle's disease and phosphofructokinase deficiency (PFK).

B Disorders of 'fatty acid oxidation' including lipid storage myopathies

People who have these disorders are unable to process fat into energy; the breakdown occurs at the point of 'fatty acid oxidation'. See diagram below.

What type of exercise brings on symptoms?

Generally, symptoms occur during prolonged exercise such as when you are playing a full game of football or going for a long run. The symptoms vary from person to person. Some people find they can sprint and weight-train with no ill effects. Others can play half a game of football, only developing symptoms in the second half. The important thing is to find your own limits and stay within them. Sometimes you will experience symptoms when you are feeling unwell or fasting. Fasting is another occasion when we use up the body's fat stores to provide energy.

What are the symptoms and what should I do if I experience them?

You may feel muscle pain, which can be an ache or very intense, both during and after exercise. (Sometimes the pain can come on a couple of hours after you stop exercising). When you feel pain you should stop exercising and should try to avoid continuing that form of exercise for such a long period in the future.

Diet is extremely important in alleviating the symptoms of these conditions. You should see a dietician and take expert advice.

If you are unwell, frequent glucose drinks should be taken to try and ensure that you never entirely rely on fat for energy.

The effects of some of these conditions on some people are potentially serious. They can lead to severe muscle weakness, which rapidly progresses to weakness of respiratory muscles, and can be life threatening. Diet can be an effective method of treatment and prevention, and it is important to remember this.

Are children who have these conditions affected in the same way?

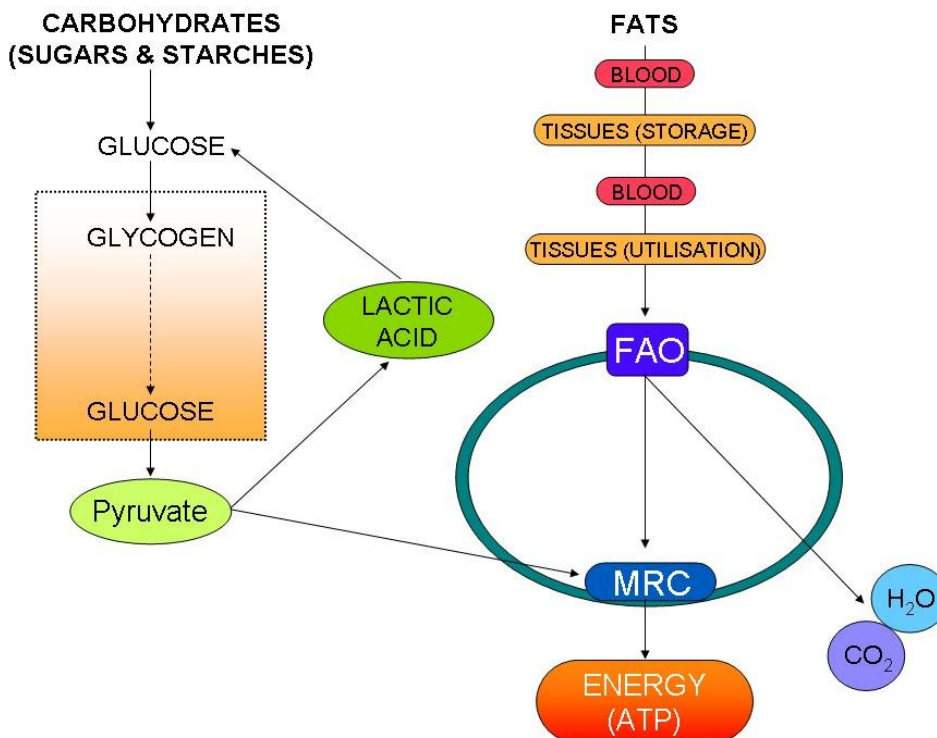
Youngsters rarely develop muscle pain on exercise but may develop severe muscle weakness at times of illness or stress when the child is not eating, especially after a cold or other viral infection. When this happens you should consult your doctor immediately.

The names of some of the disorders of fatty-acid oxidation are carnitine palmityl transferase (CPT) deficiency, acyl-CoA dehydrogenase deficiency and 3-hydroxy acyl-Co A dehydrogenase deficiency.

C Defects of the mitochondrial respiratory chain

If you have one of these disorders it means that there is a breakdown in the final stage of the chain which converts carbohydrates and fats into energy, at the point where pyruvate (from carbohydrates) or fatty acid (from fats) are converted into energy (ATP) in the mitochondria. See diagram below.

You may experience pain on exercise and if this happens, you should stop exercising and try and avoid such exercise in the future.



Other related factsheets by the Muscular Dystrophy Campaign:

- Mitochondrial Myopathies
- McArdle's disease

If you would like a copy please contact one of our Information Officers on **020 7803 4800** or freephone **0800 652 6352** or email: **info@muscular-dystrophy.org**

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Information point for mitochondrial disease:

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