



Central core disease

What is central core disease?

Central core disease falls under the category of congenital myopathies, which are a group of conditions characterised by muscle weakness and wasting. Central core disease is a rare condition and symptoms usually become apparent at birth or early infancy, although cases have been reported where symptoms are present in the developing foetus. The condition is generally non-progressive or slowly-progressive and people affected usually have a normal life span.

What causes central core disease?

In some families, central core disease is caused by a mutation in the ryanodine receptor (RYR1) gene, located on chromosome 19. This gene carries the instructions for a protein which is involved in calcium release in muscle. It is not known exactly how mutations in this gene cause the condition. In many other families, the genetic cause has not been determined.

Central core disease is inherited in an autosomal dominant pattern, although many cases occur sporadically, sometimes caused by autosomal recessive inheritance with no previous family history. Autosomal dominant inheritance means if a parent has the condition, there is a 50 percent chance that each child will also have the condition. Either of the parents can pass on the error, and both male and female children can be affected.

How is central core disease diagnosed?

Muscle biopsy. Generally, diagnosis is made through a muscle biopsy. A sample of muscle is taken and examined under a microscope. This is done in one of two ways: either a small piece of muscle is taken under general anaesthetic (avoiding the drugs which may cause malignant hyperthermia – see below) or a needle biopsy is performed to remove a small sample. Muscle from people affected by central core disease has a distinctive pattern with core structures centrally located within the muscle cells. It is important to note that these structures are also seen in other unrelated conditions. For this reason, the muscle sample must be considered along with the physical signs and symptoms and/or molecular tests, for a diagnosis of central core disease to be made.

Molecular testing. In families where the mutation is known to occur in the RYR1 gene, molecular testing is available. This involves taking a blood sample and analysing the DNA for the presence of a mutation. This process can take up to several months to complete.

Is there a treatment or cure?

Currently there is no treatment or cure for central core disease, but management of the condition is very important.

Physiotherapy. The primary aim of managing a neuromuscular condition is to increase or at least maintain function and mobility. Physiotherapy can assist in doing this, and it can also maintain breathing capacity, delay the onset of curvature of the spine (scoliosis) and



help prevent the development of contractures. It is important that the physiotherapist involved is familiar with the treatment of people with neuromuscular conditions.

Exercise. There is some debate over whether people with neuromuscular conditions should undertake strenuous physical exercise. Some say that putting additional strain on already weakened muscles will cause additional harm, while others believe that the exercise may increase muscle strength. Insufficient evidence exists to support either, but it is believed that moderate non-weight-bearing exercise such as swimming, walking or pedalling may be the best solution. This sort of aerobic exercise helps to maintain a healthy cardiovascular system and a steady weight. It is however important that this is discussed fully with a clinician.

Corrective surgery. Scoliosis, or curvature of the spine, is common with central core disease. Spinal surgery aims to correct the posture by re-aligning the spinal column, and involves the insertion of rods, screws or wires. There are benefits and risks associated with this surgery, and more information is available if you call the Information and Support helpline. As with other treatments, it is very important that the options are discussed fully with a consultant or specialist, before a decision is made. In young children, a spinal brace may be used and in children who do not walk, moulded seating is used.

What is the prognosis?

In most cases symptoms become apparent at birth or shortly after, and include hypotonia (floppiness) and weakness of the muscles closest to the trunk of the body. There is often a delay in achieving motor milestones, but the majority of people affected should eventually be able to walk. Muscle cramps are common and mild facial weakness has been seen in some cases, specifically involving the eyes. Weakness around the hips can lead to hip dislocations or tightening of the joints (contractures), particularly the knees and hips. Curvature of the spine (scoliosis) may also occur. Generally the heart and respiratory function are not affected.

Malignant hyperthermia (MH) is an acute reaction triggered by certain general anaesthetics or muscle relaxants (which are used for general anaesthesia). Symptoms of MH include high fever, muscle rigidity, dark brown colouration of urine and acute kidney failure. MH is potentially fatal if not treated immediately with a drug called dantrolene. MH can be prevented by avoiding the triggering anaesthetic agents with alternative drugs. Local anaesthetics are quite safe. Both MH and central core disease are associated with abnormalities in the RYR1 gene thus it is important to inform the consultant surgeon or anaesthetist if surgery is being considered.

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We're here for you at the point of diagnosis and at every stage thereafter, and can:

- ▶ give you accurate and up-to-date information about your or your child's muscle-wasting condition, and let you know of progress in research
- ▶ give you tips and advice about day-to-day life, written by people who know exactly what it's like to live with a muscle-wasting condition
- ▶ put you in touch with other families living with the same muscle-wasting condition, who can tell you about their experiences
- ▶ tell you about – and help you get – the services, equipment and support you're entitled to.

If you would like your GP or other health professional to have more information about Central core disease, we have some relevant materials. We've developed an online training module for GPs, as well as one for physiotherapists working with adults with muscle-wasting conditions. Contact our helpline or email us to find out more.

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Here for you

The friendly staff in the care and support team at the Muscular Dystrophy UK's London office are available on **0800 652 6352** or **info@musculardystrophyuk.org** from 8.30am to 6pm Monday to Friday to offer free information and emotional support.

If they can't help you, they are more than happy to signpost you to specialist services close to you, or to other people who can help.

www.musculardystrophyuk.org