



## Central core disease

Central core disease falls under the umbrella of congenital myopathies which are characterised by muscle weakness and wasting. It is a rare condition, and symptoms are usually present at birth or during early infancy. These include hypotonia (floppiness), delayed motor developments, and muscle weakness and cramps. It is generally thought to be non- or slowly progressive, and most children affected are eventually able to walk. Some children show improvement over time, and increased muscle tone.

Central core disease is closely associated with malignant hyperthermia, a serious acute reaction to general anaesthetics which is potentially fatal. It is triggered by muscle relaxants and inhaled anaesthetics.

### 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 foetus. The condition is generally non- or slowly progressive and people affected usually have a normal life span.

### What causes it?

In some families, central core disease is caused by an error in the ryanodine receptor (RYR1) gene, located on chromosome 19q13. This gene produces a protein which is involved in calcium release in muscle. It is not known exactly how errors 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 recessive inheritance with no previous family history. Autosomal dominant inheritance means if a parent has the condition, there is a 50% chance that each child will have the condition also. Either parent can pass on the error, and both male and female children can be affected.

More information on genetic inheritance is available in our factsheet 'Inheritance and the Muscular Dystrophies'.

### What are the common features?

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 round 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.

### **How is it 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 precipitate MH) 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/or molecular tests, in order for a diagnosis of central core disease to be made. A fact sheet on *Muscle biopsies* is available from the Information and Support Line.

**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.

### **What other tests are available?**

**Prenatal diagnosis** is available for families where the mutation has been identified as being in the RYR1 gene. The technique is described in the section Molecular testing, but there are two ways to obtain samples for testing:

- Amniocentesis is traditionally performed at 15 to 17 weeks into the pregnancy. Using ultrasound to visualise, a needle is inserted through the abdominal wall, and a sample of the fluid surrounding the baby (amniotic fluid) is taken.
- Chorionic villus sampling (CVS) is carried out at 10 to 11 weeks. This involves taking a sample of tissue from the placenta. Results are available earlier using this technique than amniocentesis, but the rate of spontaneous abortion is slightly higher.

### **How will it progress?**

Central core disease is generally thought to be non- or very slowly progressive. Sometimes progression is seen in adulthood, but some people actually show an improvement over time, with reduced weakness and increased mobility.

### **Is there a treatment?**

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

**Physiotherapy.** The primary aim of an individual with a neuromuscular disorder 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 disorders.

**Exercise.** There is debate over whether people with neuromuscular disorders should undertake strenuous physical exercise. Some say that putting additional strain on already weakened muscles will cause additional harm, whilst 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 peddling 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 realigning 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 from the Information and Support Line. 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.

### **Is there a cure?**

Currently there is no cure for central core disease although much research is being conducted into all of the neuromuscular disorders. Although there is no effective treatment, there are a number of different ways in which to manage the symptoms of central core disease and these are outlined above.

### **What research is currently being done?**

Researchers world-wide are exploring many avenues in an attempt to develop more effective treatments and hopefully a cure. The research department at the Muscular Dystrophy Campaign regularly monitors research advances in congenital myopathies, and produces releases, which are sent to members when significant scientific advances occur.

The research department can be contacted by email or by telephone.

Email: [research@muscular-dystrophy.org](mailto:research@muscular-dystrophy.org)

Tel: **020 7803 4800**  
Freephone: **0800 652 6352**

### **Planning for the future?**

Since central core disease is generally non- or slowly progressive, the needs of a person affected will not vary greatly over time. Depending on the severity of the condition there are things which may have to be considered, such as:

- Education
- Adaptations
- Holidays

Further information on these subjects can be obtained from the Information and Support Line.

### **Other things to consider**

**Anaesthetics and muscle relaxants.** As mentioned, there is an association between central core disease and a condition called malignant hyperthermia, which is triggered by the administration of certain general anaesthetics and muscle relaxants. It is important that this is brought to the attention of the consultant and the anaesthetist if surgery is being considered.

**Medical alert card.** It is very important that health professionals are aware of your condition should you require treatment. There are often issues they will have to consider. Many companies are able to provide a Medic Alert Card, which can be carried to advise of any medical condition. These come in the form of bracelets, pendants etc and carry essential information. In the case of central core disease, the risk of malignant hyperthermia should be clearly displayed.

### **Where can I get help?**

#### **Muscular Dystrophy Campaign**

61 Southwark Street

London SE1 0HL

Tel: **020 7803 4800** (all departments)

Free phone: **0800 652 6352**

Email addresses:

Information and Support Line: [info@muscular-dystrophy.org](mailto:info@muscular-dystrophy.org)

Research: [research@muscular-dystrophy.org](mailto:research@muscular-dystrophy.org)

#### **Contact a Family**

209-211 City Road,

London EC1V 1JN

Tel: **020 7608 8700**

Helpline: **0808 808 3555** or Textphone: **0808 808 3556**

Email: [info@cafamilly.org.uk](mailto:info@cafamilly.org.uk)

Web: [www.cafamily.org.uk](http://www.cafamily.org.uk)

### Other MDC factsheets that may be useful

- Myopathy
- Congenital myopathies
  - Congenital fibre type disproportion myopathy
  - Minicore (multicore) myopathy
  - Myotubular (centronuclear) myopathy
  - Nemaline (rod) myopathies
- Mitochondrial myopathies
- Ocular myopathies
- Anaesthetics
- Carrier detection tests and prenatal diagnosis of inherited neuromuscular conditions
- Inheritance and the muscular dystrophies
- Muscle biopsies
- Surgical correction of spinal deformity in muscular dystrophy and other neuromuscular disorders

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