What is congenital fibre type disproportion?

Congenital fibre type disproportion (CFTD) is so named because of the characteristic appearance of muscle fibres under a microscope. Muscle tissue has two different types of fibre: type 1 and type 2. In normal muscle, these are approximately the same size, and the proportion of them is even. In muscle of people affected by CFTD, type 1 fibres are visibly smaller than type 2 fibres, and there are often more type 1 fibres than type 2 fibres. In some people with CFTD, these changes remain the same throughout life; in others, additional features may arise that have been associated with other congenital myopathies such as nemaline myopathy (NM), multi-minicore disease (MmD) or centronuclear myopathy (CNM), indicating an overlap between CFTD and these conditions.

What are the causes of congenital fibre type disproportion?

CFTD is an inherited condition. In most instances, the myopathy is thought to be passed on in an autosomal recessive pattern, meaning that a child must inherit one copy of the mutation from each parent, although neither of them usually has any symptoms. There have, however, been cases reported of autosomal dominant inheritance, where only one copy of the mutation is needed to cause the condition. In such cases, all children of an affected parent have a 50 percent chance of also having the condition. For both patterns of inheritance, both males and females can be affected but there is also evidence that one form of CFTD may be more common in males than females and many sporadic cases have also been seen where there has been no previous family history.

CFTD has now been associated with mutations in several genes: the muscle α-actin (ACTA1) gene, the selenoprotein N (SEPN1) gene, the α-tropomyosin 3 (TPM3) gene, the skeletal muscle ryanodine receptor (RYR1) gene and the slow/β-cardiac myosin heavy chain (MYH7) gene. All of these genes have also been implicated in other congenital myopathies, again indicating a possible overlap between CFTD and these conditions.

How is congenital fibre type disproportion diagnosed?

Many of the features of CFTD are also associated with other conditions, in particular other congenital myopathies. For this reason, a diagnosis of CFTD must be confirmed through a muscle biopsy.

Muscle biopsy. This is done in one of two ways: either an open biopsy, where a small piece of muscle is taken under a general anaesthetic, or a needle biopsy, which is performed under local anaesthetic to remove a small muscle sample with a special needle through a small incision. The sample will be analysed under a microscope; muscle from people with CFTD has a characteristic pattern when viewed under a microscope.
Is there a treatment or cure?

There is currently no treatment for CFTD, but management of the condition is very important for maintaining quality of life. There is no cure for CFTD although much research is currently being conducted into the congenital myopathies. Although there is no effective treatment to halt the progression, there are different ways to manage the symptoms of CFTD and these are outlined below.

**Physiotherapy:** the primary aim of an individual with a neuromuscular condition is to increase or at least maintain muscle 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 (tightness of joints). It is important that the physiotherapist involved is familiar with the treatment of people with neuromuscular conditions.

**Exercise:** there is debate about 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 exercise may increase muscle strength. Insufficient evidence exists to support either, but it is believed that moderate non-weightbearing exercise such as swimming, walking or horse-riding 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.

**Night-time ventilation:** breathing problems are common in people with CFTD, and thus respiratory function should be regularly monitored. A decrease in oxygen intake can lead to headaches, breathlessness, poor appetite and disturbed sleep, among other things. Night-time ventilation involves the use of a face-mask attached to a small machine, which assists in breathing. This aids the muscles that control breathing, and allows a greater intake of oxygen. Night-time ventilation may be beneficial to people with CFTD, but this should be discussed fully with a consultant to determine whether it is appropriate. If there is a tendency for chest infections, it is worth considering Pneumovax and the flu vaccine.

**Feeding tube (or gastrostomy):** this is a tube that goes into the stomach through the stomach wall and enables a person to be given food and fluids by passing them directly into the stomach via the tube. People with a myopathy may have problems with swallowing, which can lead to choking and inhalation of food, and can result in chest infections. A feeding tube prevents this from happening. There are a number of different types of feeding tube which are available, and these are fitted by a short surgical procedure.

**Corrective surgery:** scoliosis, or curvature of the spine, is common in CFTD. 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 charity's Care 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?**

Progression of CFTD is highly variable, depending on the genetic background. Some people with CFTD show a progression, in others the condition remains static or may even show improvement over time.

CFTD becomes apparent at birth or in early childhood, and the initial signs are floppiness (hypotonia) and motor delay, such as delay in crawling and walking. External ophthalmoplegia (weakness of the muscles around the eye) may occur in some people and can lead to problems with eye movement and sometimes droopiness of the eyelids (ptosis). Generalised muscle weakness is seen and sometimes children are born with dislocated hip(s). Breathing problems can occur and these vary in severity, but can be life-threatening in some cases. Other characteristic features include curving of the spine (scoliosis), tightening of joints (contractures), deformities of the feet and a high arched palate. Problems with the heart are only rarely seen.

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Other related publications

This factsheet is to be used alongside the following publications:

- Myopathy
- Congenital myopathies
- Central core disease
- Minicore (multicore) myopathy
- Myotubular (centronuclear) myopathy
- Nemaline (rod) myopathies
- Mitochondrial myopathies
- Ocular myopathies
- Inheritance and the muscular dystrophies
- Muscle biopsies
- Surgical correction of spinal deformity in muscular dystrophy and other neuromuscular disorders
- Inheritance
- Adult Self-Management Pack
- An introductory guide for families with a child newly diagnosed with a neuromuscular condition
- Inclusive Education Guide
- Wheelchair Provision for Children and Adults with Muscular Dystrophy and other Neuromuscular conditions
- Muscle biopsies factsheet

The friendly staff in the care and support team at the Muscular Dystrophy Campaign’s London office are available 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 people who can help.

Contact our Freephone helpline on 0800 652 6352 or info@muscular-dystrophy.org

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