



Congenital fibre type disproportion (CFTD)

Overview

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 who have 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, 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.

Symptoms

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 curvature 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.

Causes

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 would have inherited 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 has caused the condition. In such cases, all children of an affected parent have a 50 percent chance of inheriting 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. 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.

Diagnosis

Many of the features of Congenital fibre type disproportion (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 can be done of two ways: an open biopsy, where a small piece of muscle is taken under a general anaesthetic, or a needle biopsy, where a small muscle sample is removed through a small incision with a special needle, under local anaesthetic. The sample is then analysed under a microscope; muscle from people with CFTD has a characteristic pattern when viewed under a microscope.

Treatment

There is no cure for CFTD although much research is currently being conducted into the congenital myopathies. There is currently no treatment for CFTD, and no effective treatment to halt the progression, there are different ways to manage the symptoms of CFTD and maintain quality of life.

Physiotherapy. The primary aim of an individual with a muscle-wasting 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 muscle-wasting conditions.

Exercise. There is debate about whether people with muscle-wasting conditions should undertake strenuous physical exercise. Some say that putting additional strain on already weakened muscles will cause additional harm, while others believe 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 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 can be fitted during a short surgical procedure.

Corrective surgery. Scoliosis, or curvature of the spine, is common in CFTD. 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 from our Muscular Dystrophy UK 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 for children who do not walk, moulded seating may be used.

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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@muscular dystrophyuk.org.

Version: 03 / Date published: 1 March 2002 / Original author: Dr Ros Quinlivan, Consultant Paediatrician) Dr Heinz Jungbluth, Consultant Paediatric Neurologist / Updated: 1 October 2013 / Updated by: Dr Heinz Jungbluth, Consultant Paediatric Neurologist / Date of review: 1 October 2014